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Parental and carer responses to Angelman syndrome and Prader-Willi syndrome

Allyson K. Thomson
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**Parental and carer responses to Angelman syndrome and
Prader-Willi syndrome**

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Submitted: 30-06-2010
Re-submitted: 25-10-2011

USE OF THESIS

The Use of Thesis statement is not included in this version of the thesis.

Abstract

The research project undertaken as part of this thesis was designed to assess family stress levels and the use of coping strategies among the carers of people with an intellectual disability caused by Angelman syndrome (AS) or Prader-Willi syndrome (PWS). Both syndromes are genomic imprinting disorders that arise from disruptions in genes located within human chromosome 15q11-q13. Although the disease phenotypes are quite distinct, the genetic mechanisms involved are common to both syndromes but involve paternally-derived mutations in PWS as opposed to maternal mutations in AS.

Previous investigations in Western Australia (WA) indicated that people with AS and PWS experience substantial ill health over the life course, and require more frequent hospital-based care than their typically-developing peers (Thomson, 2005; Thomson, *et al.*, 2006a; b; Thomson, *et al.*, 2007). A high proportion of the people identified in the earlier study were resident in the family home and many relied on family carers for assistance with activities of daily living. Studies of the families of people with intellectual and developmental disabilities (IDD) from other causes have shown that family carers often experience considerable stress through the caring role, especially as their offspring reach adulthood and beyond (e.g., Baxter, *et al.*, 2000; Benson & Karlof, 2009; Glidden & Natcher, 2009). Information regarding the family carers of people with AS and PWS is limited and therefore this study examines the family aspects of caring for individuals with these disorders.

Participants were recruited by means of invitation letters sent by staff of Disability Services WA and Genetic Services WA, and by presentations by the candidate to the Western Australian branches of the Angelman Syndrome Association and the Prader-Willi Association. Data were collected using postal questionnaires and by face-to-face interview with family carers. The detailed information collected at individual level on people with AS (n = 11) or PWS (n = 5) included demographic data, the nature and extent of their care needs, and their clinical and behavioural profiles. Family carers (n = 21) also provided personal demographic information, rated their own life satisfaction and health levels, and participated in the Family Stress and Coping Interview (FSCI).

There was widespread variation in the responses provided by family carers with regard to their levels of stress and/or satisfaction, degrees of ‘normality’, and coping strategies. Their relatively high scores on the FSCI Scale (mean = 32.7 of a possible 96) indicated that many carers had experienced significant stress over extended time-periods. A substantial level of anger and frustration was expressed and many carers felt that the care demands they faced were almost ceaseless, leaving insufficient time to meet their own needs and those of other family members. Additional concerns included managing the financial wellbeing and appropriate accommodation for their offspring in future years.

Participants demonstrated a diverse range of coping mechanisms. However, the limited number of participants meant it was not possible to test for correlations between type of coping strategy and the level of reported stress. There were examples of carers utilising both ‘resources’ and ‘perceptions’ coping methods to deal with the same problem, indicative of the complexity of the relationships between these different types of coping strategy. Support group membership was viewed as beneficial by many respondents, resulting in both practical and emotional support from their peers and an increased sense of belonging. In particular, younger carers valued the interaction with other people facing similar issues to them, and they felt encouraged by the support provided by other family carers.

Family, friends, and dedicated support groups are the predominant current sources of practical and emotional support for carers. However, given the increasing life expectancy of people with AS/PWS, there will be continued reliance on medical professionals, service providers, and Disability Services WA to assist this under-recognized and often under-served section of society.

Declaration

I certify that this thesis does not, to the best of my knowledge and belief:

- (i) *Incorporate without acknowledgment any material previously submitted for a degree or diploma in any institution of higher degree or diploma in any institution of higher education;*
- (ii) *Contain any material previously published or written by another person except where due reference is made in the text of this thesis; or*
- (iii) *Contain any defamatory material.*
- (iv) *Contain any data that has not been collected in a manner consistent with ethics approval.*

The Ethics Committee may refer any incidents involving requests for ethics approval after data collection to the relevant Faculty for action.

Signed:

Date: 20th Feb 2012

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Abbreviations

ADHD	Attention Deficit Hyperactivity Disorder
AIHW	Australian Institute of Health & Welfare
AS	Angelman Syndrome
ASD	Autism Spectrum Disorder
CES-D	Center for Epidemiological Studies Depression Scale
CHIC	Confidentiality in Health Information Committee
COPE	Coping Orientation to Problems Experienced
DLU	Data Linkage Unit
DNA	Deoxyribonucleic acid
DSC	Disability Services Commission
ECU	Edith Cowan University
EEG	Electroencephalogram
EU	European Union
FRPQ	Food-Related Problems Questionnaire
FSCI	Family Stress and Coping Interview
GAP	Group Action Planning
GSWA	Genetic Services WA
HREC	Human Research Ethics Committee
IDD	Intellectual and Developmental Disability
IQ	Intelligence Quotient
KEMH	King Edward Memorial Hospital for Women
LAC	Local Area Co-ordinator
PIN	Planned Individual Network
PLAN	Planned Lifetime Advocacy Network
PMH	Princess Margaret Hospital for Children
PSI	Parenting Stress Index
PWS	Prader-Willi Syndrome
QRS-SF	Questionnaire on Resources and Stress-Short Form
UK	United Kingdom
USA	United States of America
WA	Western Australia
WCQ	Ways of Coping Questionnaire

Chapter 1 Introduction

1.1 Significance

Over the course of the last twenty years, there has been considerable research into the long term effects on family members of the role of caring for an individual with intellectual and developmental disability (IDD) (e.g., Crnic, *et al.*, 1983; Greenberg, *et al.*, 1993; Dyson, 1997; Heller, *et al.*, 1997; Woodford, 1998; Pruchno & Patrick, 1999; Baxter, *et al.*, 2000; Hastings, *et al.*, 2002; Baronet, 2003; Minnes & Woodford, 2004; Cantwell-Bartl, 2006; Kandel & Merrick, 2007; Kenny & McGilloway, 2007; Lopes, *et al.*, 2008; McConkey, *et al.*, 2008). As defined by the American Association on Intellectual and Developmental Disabilities (2011) (formerly the American Association on Mental Retardation):

Intellectual disability is a disability characterized by significant limitations both in intellectual functioning and in adaptive behavior, which covers many everyday social and practical skills. This disability originates before the age of 18.

People with IDD have been estimated to constitute 1-3% of the general population in developed countries and up to 8% in underdeveloped countries (Roeleveld & Zielhuis, 1997) and they are living significantly longer than in the past (Janicki, *et al.*, 1999; Hogg, *et al.*, 2001; Bittles, *et al.*, 2002; Gustavson, *et al.*, 2005). In developed countries, individuals diagnosed with IDD but with higher levels of intellectual function now have a median life expectancy of 74 years (Bittles, *et al.*, 2002), and those with severe IDD around 60 years (Patja, *et al.*, 2000; Patja, *et al.*, 2001b; Sutherland, *et al.*, 2002). People with IDD are also prone to premature ageing, thereby experiencing the effects of senescence much earlier than the general population and individuals may live for many years in this biological phase (Cooper, 1998; Fisher & Kettl, 2005; van Schroyenstein Lantman-de Valk, 2005; Torr & Davis, 2007). As increased life expectancy is associated with a greater probability of developing disease and disability, for many individuals with IDD and high care needs this equates to needing specialist care for a considerable proportion of their life-span.

The family unit is recognised as a vital component in the life of individuals with IDD, offering support and constancy to persons who usually require assistance in all aspects of their daily life. However, raising a child and then adult with IDD can impose

a number of limitations on the lifestyle of other family members, and often these limitations are demanding and life-long (Swenson, 2005). Elevated stress levels, low self-esteem, and social difficulties have all been shown to be significant consequences of the carer role (Dyson, 1997; Ben-Zur, *et al.*, 2005; Bauer, *et al.*, 2009). Long-term carers are assumed to be particularly affected as age-related health issues of the individual with IDD, and of the carers themselves, limit their ability to provide appropriate assistance.

There are many factors which affect the degree of satisfaction felt by a person caring for a family member with IDD, and these include personal feelings of empowerment, having access to a variety of supports, using effective coping styles, and feeling emotionally connected to their relative (Seltzer, *et al.*, 1995; Knox, *et al.*, 2000; Hastings, *et al.*, 2002). A knowledge and understanding of the specific aetiology of the IDD has been reported to impact positively on the psychological well-being of family members (Lenhard, *et al.*, 2005). Additionally, it has been noted that stress factors in families with a person with IDD may be partly related to the specific cause of the disability, most notably in conditions which are characterised by behavioural disorders, such as Prader-Willi syndrome (PWS) or autism spectrum disorders (Hodapp, *et al.*, 1997; Eisenhower, *et al.*, 2005).

In common with members of the general population, the health care needs and phenotypic profiles of people with IDD vary across the lifespan. These needs have significant implications with regard to their support requirements and the resultant economic costs (Edgerton, *et al.*, 1994; Janicki & Dalton, 1995; Haveman, *et al.*, 1997; Cooper, 1998; Thompson & Reid, 2002). In many instances, however, family carers of children and adults with IDD have inadequate resources to provide for dependent family members at home as they age (Fujiura, 1998; Parish, *et al.*, 2004; Ben-Zur, *et al.*, 2005; McConkey, 2005). Parental carers often harbour feelings of loss, trauma and stress over many years (Hodapp, *et al.*, 1997; Hogg, *et al.*, 2001; Cantwell-Bartl, 2006), and the extra burden associated with ageing family members with IDD necessitates increasing levels of personal support, including respite services, to assist parents in fulfilling their caring role (Hoare, *et al.*, 1998; Freedman, *et al.*, 1999; McConkey, 2005). In many cases parents become physically incapable of providing for an adult son or daughter with IDD at home, in which case a sibling may assume the responsibility (Orsmond & Seltzer, 2000; Dew, *et al.*, 2004). Alternatively, assistance with the transition towards

out-of-home living for the individual may be provided, so that from the perspective of both the person with IDD and their parents, the move may be looked upon as a sign of natural progression in development and increasing independence (Bigby, *et al.*, 2002; Llewellyn, *et al.*, 2004; Todd & Jones, 2005; Heller & Caldwell, 2006).

Much of the research dedicated to addressing the particular needs of carers of people with IDD has been devoted to Down syndrome and, more recently, Autism Spectrum Disorder, (e.g., Crombie & Gunn, 1998; Bailey, *et al.*, 2003; Carr, 2005; Heiman & Berger, 2008; Van Der Veeck, *et al.*, 2009; Kayfitz, *et al.*, 2010; Keenan, *et al.*, 2010). Two syndromes that have been under researched in this area are Angelman syndrome (AS) and Prader-Willi syndrome (PWS). At the time of the commencement of the current study, there were few published reports referring to the needs of the carers of people with these disorders (Hodapp, *et al.*, 1997; van den Borne, *et al.*, 1999), however several related publications have since been released (Wulffaert, *et al.*, 2010; Griffith, *et al.*, 2011). These reports and their findings will be discussed within the framework of the stress and coping model presented in Section 3.2. Although PWS and AS have genetic similarities, they are distinct in their physical presentations and behavioural profiles. However people with both disorders experience significant age-related disability and require ongoing care throughout their lifespan which may, in turn, account for the high stress levels reported by their family carers (van den Borne, *et al.*, 1999; Thomson, 2005; Thomson, *et al.*, 2006a; b; Thomson, *et al.*, 2007).

1.2 Genomic imprinting in Angelman syndrome and Prader-Willi syndrome

Angelman syndrome and Prader-Willi syndrome are examples of genomic imprinting disorders. Both syndromes result from failure of imprinting or one of several gene mutations within the same region of chromosome 15q11-q13. The normal expression of imprinted genes is dependent on the parent of origin: some are solely expressed from the paternally inherited allele, whereas others only express when maternally inherited. If the affected segment of chromosome 15q11-q13 is maternally derived then AS occurs, whereas PWS results if the paternal chromosome is altered. Usually there is no obvious gender bias in either syndrome, although more boys than girls are identified with PWS before adolescence, possibly due to the more subtle phenotypic changes in prepubescent girls (Smith, *et al.*, 2003a). Duplications within the same area of chr15q11-q13 have been associated with some autism spectrum disorders,

and these conditions share a number of the phenotypic characters of AS and PWS (Dykens, *et al.*, 2004).

1.2.1 Angelman syndrome

Angelman syndrome was first described in 1965, with the phenotype characterised as a combination of severe intellectual disability, seizures with a specific electroencephalogram (EEG) pattern, absent speech, jerky ataxic movements, and a generally happy sociable disposition (Angelman, 1965). Consensus diagnostic criteria were subsequently published in 1995 and revised in 2006 (Williams, *et al.*, 1995; Williams, *et al.*, 2006), and the first laboratory diagnostic tests for AS were introduced in 1987 (Clayton-Smith & Laan, 2003).

The major clinical and developmental signs of AS are listed in Table 1.1. As shown, there is extensive variation in the reported prevalence of many of these features. In part, this can be attributed to inconsistent data collection practices or to a lack of certainty on the part of respondents. The phenotypic profiles reported within AS also appear to be dependent on the specific genetic mechanism involved (Dan, 2009), and may change with increasing age (Didden, *et al.*, 2009). One constant is the absence of typical speech development by a person with AS. This may, however, be offset by use of augmentative and alternative communication (AAC) systems (Calculator & Black, 2010). Family members feel that use of these techniques and devices allows their relative to develop a voice of their own.

Formerly, the diagnosis of AS was generally based on clinical findings, but more recently cytogenetic or DNA testing has been used to confirm 80-85% of cases (Laan, *et al.*, 1999; Williams, *et al.*, 2010). Most cases of AS are caused by either a large-scale deletion or a mutation of the *UBE3A* gene on the maternal chr15q11-q13. The availability of detailed clinical criteria and genetic diagnostic tests has led both to confirmation of diagnoses and a reduction in the average age at diagnosis, with many affected individuals in developed countries now identified before the age of one year (Minassian, *et al.*, 1998; Ruggieri & McShane, 1998; Valente, *et al.*, 2003).

Table 1.1 Clinical signs in individuals with Angelman syndrome

Characteristic	Occurrence (%)	References
Developmental delay (mean age)	Sit = 12-20 m Walk = 33-72 m	Laan <i>et al.</i> (1999); Moncla <i>et al.</i> (1999a; 1999b); Thomson <i>et al.</i> (2006a)
Absent/minimal speech	82-100	Moncla <i>et al.</i> (1999b); Thomson <i>et al.</i> (2006a)
Ataxia	79-96	Laan <i>et al.</i> (1999); Beckung <i>et al.</i> (2004); Thomson <i>et al.</i> (2006a)
Happy disposition/ inappropriate laughter	76-100	Laan <i>et al.</i> (1999); Moncla <i>et al.</i> (1999b); Thomson <i>et al.</i> (2006a)
Microcephaly	44-93	Moncla <i>et al.</i> (1999a; 1999b); Thomson <i>et al.</i> (2006a)
Epilepsy	79-88	Minassian <i>et al.</i> (1998) Moncla <i>et al.</i> (1999b) Thomson <i>et al.</i> (2006a)
Abnormal EEG	82-100	Moncla <i>et al.</i> (1999b); Thomson <i>et al.</i> (2006a)
Feeding difficulties in infancy	21-77	Smith <i>et al.</i> (1996); Moncla <i>et al.</i> (1999b); Thomson <i>et al.</i> (2006a)
Strabismus	21-57	Laan <i>et al.</i> (1996); Moncla <i>et al.</i> (1999b); Thomson <i>et al.</i> (2006a)
Hypopigmentation	0-64	Moncla <i>et al.</i> (1999a; 1999b); Thomson <i>et al.</i> (2006a)
Hypotonia	48-79	Smith <i>et al.</i> (1996); Thomson <i>et al.</i> (2006a)
Scoliosis	25-70	Moncla <i>et al.</i> (1999b); Clayton-Smith (2001); Thomson <i>et al.</i> (2006a)
Obesity	15-50	Moncla <i>et al.</i> (1999a; 1999b); Clayton-Smith (2001); Thomson <i>et al.</i> (2006a)

Adapted and updated from Thomson *et al.* (2006a)

1.2.2 Prader-Willi syndrome

This condition was first described by Prader, Labhart and Willi in 1956. Symptoms common to all patients are intellectual disability and behaviour problems, hyperphagia and obesity, and delayed sexual development (Prader, *et al.*, 1956; Holm, *et al.*, 1993; State & Dykens, 2000; Young, *et al.*, 2006). The genetic basis of PWS was clarified in the late 1980s, with consensus diagnostic criteria published by Holm and others in 1993. A comprehensive picture of the phenotypic profile of PWS and current management practices is provided by Butler, Lee & Whitman (2006b).

There are generally considered to be age-dependent phenotypic profiles exhibited by individuals with PWS (Butler, *et al.*, 2010). The first, from birth to 2 years old, is characterised by hypotonia and failure to thrive. This is followed by a period when the child starts to gain weight, often from 1-3 years old, and culminates in the mature untreated phenotype of hyperphagia and short stature (Crinò, *et al.*, 2009; Butler, *et al.*, 2010).

Table 1.2 Clinical signs in individuals with Prader-Willi syndrome

Characteristic	Occurrence (%)	References
Hypotonia	83-100	Whittington <i>et al.</i> (2002); Thomson <i>et al.</i> (2006b); Lin <i>et al.</i> (2007a)
Feeding difficulties in infancy	81-100	Webb <i>et al.</i> (2002); Thomson <i>et al.</i> (2006b); Lin <i>et al.</i> (2007a)
Weight gain, obesity	73-93	Whittington <i>et al.</i> (2002); Thomson <i>et al.</i> (2006b); Lin <i>et al.</i> (2007b)
Hypogonadism	Male = 91-100 Female = 58-100	Webb <i>et al.</i> (2002); Whittington <i>et al.</i> (2002); Thomson <i>et al.</i> (2006b)
Developmental delay (mean age)	Sit = 12 m Walk = 24-31 m	Webb <i>et al.</i> (2002); Whittington <i>et al.</i> (2002); Thomson <i>et al.</i> (2006b)
Hyperphagia	66-90	Whittington <i>et al.</i> (2002); Thomson <i>et al.</i> (2006b)
Short stature	48-83	Whittington <i>et al.</i> (2002); Thomson <i>et al.</i> (2006b)
Hypopigmentation	32-81	Whittington <i>et al.</i> (2002); Thomson <i>et al.</i> (2006b); Lin <i>et al.</i> (2007b)
Epilepsy	14-46	Butler <i>et al.</i> (2002); Thomson <i>et al.</i> (2006b)

Adapted and updated from Thomson *et al.* (2006b)

People with PWS usually show mild or moderate levels of intellectual disability (Lindmark, *et al.*, 2010), but the characteristic food obsession and attendant weight gain pose substantial problems for family carers. In addition, other physical conditions such as a lack of gonadal development and poor muscle tone are prevalent in this group of people (Table 1.2).

It is considered that a deletion or mutations in one or more imprinted genes within the PWS region of chr15q11-q13 disrupts the function of the hypothalamus

and/or pituitary gland, which could account for the cardinal features of PWS (Holland, *et al.*, 2003; Miller, *et al.*, 2008). In fact, a number of genes affecting the development of the paraventricular nucleus in the hypothalamus have been identified (Buiting, *et al.*, 2007; Miller, *et al.*, 2009b), but to date there has been no evidence to connect any specific gene or genes and the development of the PWS phenotype.

1.3 *Purpose of the present study*

Earlier research (Thomson, 2005; Thomson, *et al.*, 2006a; b; United Nations, 2006; Thomson, *et al.*, 2007) identified broad health issues faced by individuals in Western Australia with AS or PWS and their family carers. It appeared that specific aspects of each condition resulted in a reduced quality of life for people with the disorders. It was inferred from these findings that a reduced quality of life for their family-carers would also occur, especially when the period of care was long-term and parents had their own health issues.

People with PWS and AS and their families need access to evidence-based information to assist them in decision-making and appropriate care management as they all grow older. Knowledge of what to expect in terms of the timing of health support needs, the acquisition of adequate resources, and understanding the expectations of the carer role are all important tools to assist in coping with the day-to-day management of a family member with AS or PWS. An understanding of the needs of carers and how best these requirements might be met by service providers and support funders is also necessary. The project thus serves to provide a basis against which the needs of people with other genetically-determined disorders associated with IDD and their family-carers can be assessed.

1.4 *Overview of the thesis structure*

Following this introductory chapter will be two chapters summarising the literature reviewed in the preparation of the document. The first of these will consider some aspects of life with an intellectual disability, with special reference to individuals with AS or PWS. The second will concentrate on the effect on family members of the caring role, and include a discussion of stress and coping theory and its relevance to the caring role. The research questions will be derived from the material covered in these two chapters and presented at the end of Chapter 3.

Chapter 4 will contain an overview of the research design and procedures, some ethical issues, participant recruitment, the survey instruments, and a presentation of the focus groups that were conducted prior to the main study.

The results of the study will be presented in two separate sections: Chapter 5 will contain the detailed methods and the results of the survey section of the study, while Chapter 6 will be devoted to the methods and results of the interview component.

The results will be drawn together and their significance discussed in Chapter 7. The discussion will be followed by a Bibliography and by Appendices containing material unsuited for inclusion in the body of the thesis.

1.5 Chapter summary

The average life expectancy of people with IDD is increasing, with a consequent extension of their requirement for care from parents and other family members. Over this extended life course, many stressful situations arise for family carers and they will engage various coping strategies to manage that stress.

Issues that will be investigated within this study include: the degree of satisfaction engendered by the caring role; any perceived stress associated with the caring role; the use of supports and resources to assist with the caring role; and the coping methods used most often and/or effectively by family carers. Any specific features of the AS and PWS that appear to affect the caring role may also form part of this investigation.

By recruiting people with Angelman or Prader-Willi syndrome and their family carers as models for other genetic disorders associated with intellectual and developmental disability, this project seeks to identify the specific coping mechanisms used by carers, and the efficacy of these strategies in terms of the reduction of perceived stress.

Chapter 2 An overview of intellectual and developmental disability

2.1 *Literature sources*

Material on which both the literature review and research project are based was collected over a number of years. The primary sources of published papers were database searches and automatic content alerts. The original searches used a metasearch facility, available through the University library, which simultaneously explores multiple databases such as Academic OneFile, Health & Medical Complete, and PubMed. Generalised search terms used included Angelman, Prader-Willi, intellectual disability/mental retardation, stress and coping, caregiver/carer. These were refined with queries such as transition, health, residence, ABCX. Additional papers and books were identified from the reference lists of papers of interest. Content alerts were received from a number of relevant journals (see Appendix I), and from article lists using the keywords Angelman and Prader-Willi generated by the National Center for Biotechnology Information (NCBI).

The catalogue of the Edith Cowan University library was searched using similar keywords to source books of interest, and again using keywords related to methodology and data analysis e.g., mixed methods, qualitative research, research methods. All references were managed using the Endnote software program.

The review is arranged in two main sections. Chapter 2 begins with a brief introduction to intellectual and developmental disability followed by presentation of aspects of IDD, with reference to AS and PWS, that may be regarded as having an impact on the lives of not only affected people but also their family carers. The following Chapter discusses stress and coping theory, with especial attention to family carers, and the stresses and satisfactions attached to that role.

2.2 *Definitions of intellectual and developmental disability*

Historically, there has been variability in the terminology applied to the classification of IDD used by investigators and service providers from different parts of the world (Haveman, 1996), with no common, agreed standard. However, from 1959 onwards IDD has been defined as concurrent deficiencies in both intelligence and adaptive behaviour skills, with onset before the age of 18 years (Heber, 1959; Grossman, 1983; American Association on Mental Retardation, 2002). The medical

model of disability, with its focus on aetiological diagnosis and organic impairment or deficiency, has been largely superseded by a multidimensional social representation which examines the correspondence between a person's capacity and their functional environment (Gross & Hahn, 2004; Wehmeyer, *et al.*, 2008; Anastasiou & Kauffman, 2011). The increased emphasis on functional limitations, as opposed to IQ scores, in more recent definitions can be attributed to the social interpretation of IDD. As explained by Schalock *et al.* (2007), substitution of the term 'intellectual and developmental disability' for 'mental retardation' does not affect the overall construct, i.e., the abstract idea of what IDD actually represents, and as a descriptor the former is preferable to the latter. Learning disability is an expression that has been used to encompass people with IDD, especially in the UK. However, the phrase can also apply to individuals with Attention Deficit Hyperactivity Disorder (ADHD), dyslexia, and other conditions which affect learning abilities, and therefore may not offer the same categorical precision as the term intellectual and developmental disability.

Both the Diagnostic & Statistical Manual of Mental Disorders (DSM-IV) (American Psychiatric Association, 2000) and the International Classification of Diseases (ICD 10) (World Health Organization, 2007) use ranges of IQ values to rate individuals with IDD from mild disability to severe/profound disability (ICD: F70-F79; DSM-IV: Axis II). However, there are indications that the revised DSM-5, due to be released in 2013, will include a slightly different definition of intellectual disability, possibly more reflective of the interaction between the IQ level and the adaptive profile of an individual with IDD advocated by the social model of intellectual disability (American Psychiatric Association, 2011).

The current (2011) definition of IDD (formerly mental retardation in the USA) formulated by the American Association on Intellectual & Developmental Disabilities also discards the use of IQ ranges to identify the different levels of handicap and instead requires the assessment of adaptive functioning across three skill types:

- Conceptual skills—language and literacy; money, time, and number concepts; and self-direction.
- Social skills—interpersonal skills, social responsibility, self-esteem, gullibility, naïveté (i.e., wariness), social problem solving, and the ability to follow rules/obey laws and to avoid being victimized.
- Practical skills—activities of daily living (personal care), occupational skills, healthcare, travel/transportation, schedules/routines, safety, use of money, use of the telephone.

(American Association on Intellectual and Developmental Disabilities, 2011).

However, below-average intellectual function, i.e., a measured IQ of 75 or less, is still a necessary component of the definition (Luckasson & Reeve, 2001; American Association on Intellectual and Developmental Disabilities, 2011).

2.3 *International estimates of prevalence*

Worldwide there is a broad range of reported prevalence figures for IDD (Table 2.1), with 4.3-35.6 people/1000 diagnosed with IDD in different countries (Christianson, *et al.*, 2002; Leonard, *et al.*, 2003; Australian Institute of Health and Welfare, 2004; van Schrojenstein Lantman-de Valk, *et al.*, 2006; Wullink, *et al.*, 2007). In China alone, it was estimated in 1987 that there were 10.2 million people with intellectual impairment (Pierini, *et al.*, 2001). In part, these diverse figures may be due to inconsistent methods of ascertainment, variation between birth prevalence and population prevalence, and changes in the definition of IDD over time and locality (see 2.2). Regardless of which estimate is accepted as most appropriate, the number of individuals with IDD across the globe is considerable. A majority of research studies relating to IDD have been conducted in developed countries, although progress is being made with regard to the developing nations of the world.

Table 2.1 Reported prevalence of IDD from various studies

Location (Year)	Prevalence per 1000 persons	Reference	Sources of case ascertainment
Western Australia (1983-92)	14.3	Leonard <i>et al.</i> (2003)	Educational and administrative sources
Rural South Africa (1993-6)	35.6	Christianson <i>et al.</i> (2002)	Comprehensive screening and assessment
Western Australia (1996)	8.3	Bower <i>et al.</i> (2000)	Registered for services: born between 1980-89
Australia (1998)	27.0	Australian Institute of Health and Welfare (2004)	Included dementias and other learning difficulties
Finland (2000)	7.0	Westerinen <i>et al.</i> (2007)	Identified from health and social benefit registers
People's Republic of China (2001)	9.3	Xie <i>et al.</i> (2008)	Screening and assessment
The Netherlands (2001)	7.0	Wullink <i>et al.</i> (2007)	Identified from GP dataset, service registries
Norway (2008)	4.4	Søndenaa, <i>et al.</i> (2010)	Administrative sources

2.4 *Australian policies on IDD support services*

As a signatory to the Convention on the Rights of Persons with Disabilities (United Nations, 2006), Australia has adopted the general principles laid out in that document:

(a) Respect for inherent dignity, individual autonomy including the freedom to make one's own choices, and independence of persons;

(b) Non-discrimination;

(c) Full and effective participation and inclusion in society;

(d) Respect for difference and acceptance of persons with disabilities as part of human diversity and humanity;

(e) Equality of opportunity;

(f) Accessibility;

(g) Equality between men and women;

(h) Respect for the evolving capacities of children with disabilities and respect for the right of children with disabilities to preserve their identities.

Responsibility for the provision of residential accommodation and support services for people with IDD in Australia rests with the State and Territory governments under the National Disability Agreement (Australian Institute of Health and Welfare, 2005; Disability ACT, 2009). The desired outcomes included in the current Agreement, which derive from the Convention on the Rights of Persons with Disabilities, state that:

- a) people with disability achieve economic participation and social inclusion
- b) people with disability enjoy choice, wellbeing and the opportunity to live as independently as possible
- c) families and carers are well supported (Disability ACT, 2009)

Accordingly, the trend over time in Australia has been for fewer people with IDD to live in institutions, with corresponding increases in the numbers living in group homes, receiving support to live in the community, or residing with their family. By 2002, of the 36% of people with IDD who were accessing accommodation support services, approximately 51% lived in a group home, almost 20% in their own home, and the remainder were dispersed between nursing homes, hostels or institutions, and the

formal residential services system (Australian Institute of Health Welfare, 2003; Australian Institute of Health and Welfare, 2005). More than three years later, 56% of adults with IDD in a regional area of Australia lived in the family home, many cared for by an ageing mother (Eley, *et al.*, 2009a; Eley, *et al.*, 2009b); this may be indicative of insufficient accommodation support outside the major urban centres in Australia. It has also been noted by Bigby & Fyffe (2009) that people with more severe functional limitations have been generally placed in more restrictive accommodation than their peers with higher functional abilities, and this situation seems to be slow to change. An appraisal of six case study accommodation options in various parts of Australia reported that it was possible to achieve suitable 24-hour support housing in the community for people with disabilities (Fisher, *et al.*, 2009). However, no detail was supplied of the type and/or severity of the functional limitations of the residents.

Over the last 30 years there have been considerable changes in the delivery of services to people with IDD and their family carers in Western Australia (WA). A form of individualised funding in which the Local Area Coordinator is responsible for funds management was established in the 1980s (Bartnik & Chalmers, 2007). More recently, this has been superseded in many cases by direct funding, where the funds are managed by the individual or their family carer, allowing greater flexibility and personal control of the services and supports accessed (Laragy & Ottmann, 2011). The system of direct care payments is reported to be greatly valued by people with IDD and their families, and is considered more responsive to their requirements. However, there remain some issues concerning the availability of service options and about the systems involved in the administration of funding (Fisher, *et al.*, 2010; Laragy & Ottmann, 2011). Among the projects in WA that utilise the direct funding approach is the Community Living Initiative in which a facilitator adopts a person-centred focus to assist an individual with IDD and their family to achieve a 'flexible, individualised accommodation option based on personal choice' (Fisher, *et al.*, 2010). These developments in the delivery of services and funds for people with IDD are working towards the provisions of the UN Convention on the Rights of Persons with Intellectual Disabilities (United Nations, 2006), as they encourage independence and autonomy while maintaining respect and reduce discrimination.

Indigenous Australians are almost twice as likely as the rest of the population to be registered with an intellectual and developmental disability (Glasson, *et al.*, 2005;

Australian Institute of Health and Welfare, 2007). Low socio-economic status and poor overall health status are associated with high rates of intellectual dysfunction in other countries (Durkin, *et al.*, 2000; Australian Institute of Health and Welfare, 2007; Emerson & Hatton, 2007a; 2008) and these factors are likely to be associated with higher rates of intellectual disability among Indigenous Australians compared to the non-indigenous population.

2.5 *Living with intellectual and developmental disability*

2.5.1 *Growing up: transitions and the future*

2.5.1.1 *Introduction*

In developed countries there is increased emphasis in attitudes to people with IDD on the principles of normalisation (Wolfensberger, 1972) or social role valorisation (SRV) (Wolfensberger, 2000):

‘The key premise of SRV is that people’s welfare depends extensively on the social roles they occupy: People who fill roles that are positively valued by others will generally be afforded by the latter the good things of life, but people who fill roles that are devalued by others will typically get badly treated by them’ (Wolfensberger, 2000).

Inclusion or encouraging participation, by children with IDD in mainstream schooling, and by adults with IDD in the wider community through opportunities in employment, leisure activities and residential options, is one of the primary effects of social role valorisation (Stella, 1996; Young, *et al.*, 1998; Aspray, *et al.*, 1999; Department of Health, 2001; Cinamon & Gifsh, 2004; Duvdevany & Arar, 2004; Yazbeck, *et al.*, 2004; Fillary & Pernice, 2005). The expectation is that there will be accommodation, services and employment available for older people with IDD to allow individuals to be included into the wider community. However, it has been reported that support staff who state a firm belief in SRV as a philosophical approach may not necessarily accept it’s practicality for people with more severe limitations (Bigby, *et al.*, 2009). Into such a group would fall most people with AS and even some individuals with PWS.

2.5.1.2 *Transitions*

As children with IDD grow into adolescence, more detailed consideration may be given to their future. The processes of transition, such as from school to work, or

from the family home to a community-based residence, are perceived as less challenging if the family feel connected to other parents in similar situations, receive ample pertinent information, and engage in appropriate forward-planning (Llewellyn, *et al.*, 2004; Heslop & Abbott, 2007). The shift from childhood social roles to those of adulthood can be adversely affected by severe physical or intellectual impairment and by deficiencies in the specific types of skills that are important to employers (Goupil, *et al.*, 2002; Van Naarden Braun, *et al.*, 2006b).

Appropriate planning and co-ordination between schools, families, support services, and potential employers can greatly facilitate the process of transition to post-school options (Goupil, *et al.*, 2002; Docherty & Reid, 2009). Generally, parents consider themselves to be vital components of transition planning for their offspring who is moving to adult services; although they also often feel they are required to fight for their adolescent's future (Smart, 2004; Hudson, 2006; Docherty & Reid, 2009). In some instances, there is considerable discrepancy between the opinions of parents and of service personnel regarding the important factors to be considered during transition planning procedures. Parents believe that there should be more emphasis on opportunities for social and leisure activities, and alternative residential arrangements, while professionals tend to promote options for further education, and skills for independent living (Ward, *et al.*, 2003; Clegg, *et al.*, 2008). To achieve the optimum outcome and work-life balance for all concerned, it is important that both perspectives are considered.

Among older individuals with IDD there may be the need for a further transition: from group home or independent living to an aged care facility (Bigby, *et al.*, 2010), as well as 'retirement' from employment or day activities (Ashman, *et al.*, 1995; Bigby, *et al.*, 2004). As people with IDD are now achieving greater life spans, these issues are likely to increase in importance over the next one or two decades. It would be necessary to conduct specific research to determine if there are any particular risk factors for retirement issues among people with AS and PWS.

2.5.1.3 *Residence*

A substantial proportion of individuals with IDD, e.g., in the Republic of Ireland and Northern Ireland (McConkey, *et al.*, 2006; MacDonald, *et al.*, 2007), Western Australia (Thomson, *et al.*, 2006a; b), and the USA (Murphy, *et al.*, 2007) live at home

with their families for most or all of their lives. Studies indicate that there are advantages for both the person with IDD and their family in residential placement, especially if the new home is readily accessible to the family home, and if family members regard the change as a natural 'launching' process rather than a 'leaving' (Essex, *et al.*, 1997; Blacher, *et al.*, 1999; Baker & Blacher, 2002). At present, however, few jurisdictions can supply sufficient supported accommodation opportunities along with the requisite funding to enable individuals with IDD to live away from the family home (Essex, *et al.*, 1997; Department of Health, 2001; Walsh, 2002; Lakin, *et al.*, 2003; Young & Ashman, 2004; Stancliffe, *et al.*, 2005; Heller & Caldwell, 2006; Mansell, *et al.*, 2006; MacDonald, *et al.*, 2007; Robertson, *et al.*, 2007b).

Mutual support may become more frequent as parent-caregivers age, so that in time people with IDD undertake some care duties on behalf of their parents (Rimmerman & Muraver, 2001; Williams & Robinson, 2001). Often a sibling of an adult with IDD will take over the care-giving responsibility from their ageing or deceased parents, a move which is facilitated by prior planning and effective communication between all family members (Griffiths & Unger, 1994; Davys & Haigh, 2008).

Out-of-home placement for people with IDD comes in various forms, e.g., group home, larger-scale residential setting, and independent living with minimal support. The cost of residential accommodation differs according to the type of facility, and to the intellectual and behavioural characteristics of the individual (Knobbe, *et al.*, 1995; Stancliffe & Keane, 2000; Hallam, *et al.*, 2002; Knapp, *et al.*, 2005; Chou, *et al.*, 2008). People with high levels of challenging behaviour are difficult for the family to cope with at home, and also harder to place in supported accommodation due to their increased support needs (Janssen, *et al.*, 2002; Plant & Sanders, 2007). However, the benefits of residential placement, in the form of better community access and improved quality of life, can be considerable (Knobbe, *et al.*, 1995).

There are a number of programmes aimed at developing and sustaining a network of family, friends and others, such that all members of the network are familiar with the needs, desires and service history of the individual with IDD. Each programme differs slightly, but a universal aspect is the acknowledgement that a person with IDD has the right to be part of a family, a neighbourhood, and a community (Turnbull, *et al.*,

1996). The Planned Lifetime Advocacy Network (PLAN) is based in Canada (www.plan.ca), while Kansas hosts a Group Action Planning (GAP) network (www.beachcenter.org), and Planned Individual Networks (PIN), which is fashioned on the PLAN model, has been established in Western Australia (www.pin.org.au). Carers Australia has been active in recommending that Australian Federal, state and territory governments all adopt a regional respite and succession support service to assist with future planning for people with IDD and their ageing parents (Carers Australia, 2005).

2.5.1.4 *Social interactions*

Highly variable levels of participation in social and leisure activities are reported among people with IDD (Floyd & Gallagher, 1997; Felce, *et al.*, 1998; Nøttestad & Linaker, 1999; Bigby, *et al.*, 2004). The generalised adoption of the principles of normalisation and social role valorisation has changed the way that people of all ages with IDD are regarded and treated, although there continue to be areas that can be and need to be improved in this regard.

For example, considerable numbers of people with IDD continue to have limited involvement in community activities, regardless of their living arrangements, with few social contacts outside their family or circle of similarly affected persons (Modell, *et al.*, 1997; Baker, 2007; Beadle-Brown, *et al.*, 2007; Kampert & Goreczny, 2007). The degree of social inclusion experienced by each individual depends largely on their level of functional competence, age, type of education, and place of residence (Modell, *et al.*, 1997; Felce & Emerson, 2001; McConkey, 2006; Van Naarden Braun, *et al.*, 2006a). Inadequate social skills and/or high levels of challenging behaviour can restrict employment opportunities, reduce quality of life, limit community inclusion, and decrease feelings of satisfaction for people with IDD (Beadle-Brown, *et al.*, 2005; Abbott & McConkey, 2006; Nota, *et al.*, 2007).

A substantial number of individuals with IDD in a US study expressed a desire for increased opportunities for community participation, such as going to church or going to a restaurant (Kampert & Goreczny, 2007). In Northern Ireland, adults with IDD identified three main aspects of their residential circumstances which improved their sense of satisfaction: social networks, feeling included, and reciprocal relationships (Barr, *et al.*, 2003). Individuals who are supported to live in a home of their own are more likely to participate in community-based activities than their peers residing in

group homes (Emerson, *et al.*, 2001; Chou, *et al.*, 2008), although this may be thought to reflect their relatively higher functional level. More recently, the system of direct payments in the UK, under which either people with IDD or their caregivers are given funding to arrange appropriate services, generally leads to greater flexibility for the recipients, and many young people with disabilities use the funds to promote greater involvement in non-specialized community activities (Blyth & Gardner, 2007). Conversely, 'out-of-area' residential placement reportedly has an adverse effect on feelings of community inclusion (Mansell, *et al.*, 2006), as do negative staff attitudes and transport concerns (Verdonschot, *et al.*, 2009).

Children with IDD who attend 'special' school or day centres may have fewer opportunities for inclusive leisure activities than their peers attending mainstream classes (Modell, *et al.*, 1997). Although some parents of children with IDD who are included in mainstream education have concerns about the capability of general teachers to successfully handle a child with special needs (Leyser & Kirk, 2004), a majority of parents support classroom inclusion, and both the children with IDD and typically developing children can benefit from this type of placement (Myklebust, 2006). Typically developing students in one New Zealand school exhibited more positive attitudes to disability after participating in combined sporting events with children with IDD (Townsend & Hassall, 2007). However, a report from Hong Kong concluded that the highly competitive nature of the educational culture in that country was not supportive of the introduction of children with IDD into classrooms (Wong, 2008).

The degree of personal choice available to people with IDD regarding socialisation has been improved by the movement away from large institutions, with the highest levels of freedom available to those competent to live in dispersed community residences with minimal support (Mansell, 2006; Young, 2006; Nota, *et al.*, 2007; Chou, *et al.*, 2008). Specific aspects of life may involve varying degrees of choice: for example, the selection of personal items and indoor activities often entails free choice, but decisions regarding where and with whom to live, and staff selection are generally more restricted (Robertson, *et al.*, 2001; Chou, *et al.*, 2008).

2.5.1.5 *Employment and retirement*

Post-school life for many individuals with IDD includes choices for vocational activity, e.g., in supported work, or a sheltered workshop or day centre, if these services

are available (Ford, *et al.*, 1995; Kraemer & Blacher, 2001; Pierini, *et al.*, 2001; Braddock, *et al.*, 2004; Metzel, *et al.*, 2005; Heslop & Abbott, 2007; Robertson, *et al.*, 2007a). Integrated employment is still comparatively rare, with employment in the open market for people with IDD often confined to manual and/or unskilled work, and generally restricted to those with mild IDD (Kraemer & Blacher, 2001; Moore, *et al.*, 2004a; Hensel, *et al.*, 2007; Hogg, *et al.*, 2007).

People with mild IDD in the Netherlands who are engaged in supported employment, place great emphasis on feeling useful, included and valued in their workplace (Cramm, *et al.*, 2009). Contrary to the findings of Rimmerman (1998) a decade before, many Dutch participants did not want special allowance made for their limitations, although it was appreciated when required (Cramm, *et al.*, 2009). A report from Taiwan found an improvement in the rates of supported employment (Wilson, 2003), but people with IDD were generally offered fewer hours of work and received lower wages than members of the general population, and these factors may contribute to their insecure financial status (Su, *et al.*, 2008). In the USA the fear of losing access to Medicaid funding and insurance can be a major contributory factor to the low rates of employment and/or fewer hours of employment common among people with disabilities of all kinds (Braddock, *et al.*, 2004; Rehm & Bradley, 2005; Taanila, *et al.*, 2005).

It is still rare for people with IDD who are members of the paid workforce to be offered assistance with the move to retirement (Su, *et al.*, 2008). As more individuals with IDD reach official retirement ages in countries where these exist, there is an increased necessity for the development and dissemination of specific programs to facilitate this major life change, along similar lines to the school-work transition planning. Within Australia there are a number of people intensively studying this particular issue and bringing it to the attention of policy makers (Bigby & Ozanne, 2004; Cartwright & Parker, 2004; Bigby, 2008b; Bigby, *et al.*, 2010).

2.5.1.6 *Summary*

It is widely accepted in the developed world that the principles of normalisation and social role valorisation represent the ideal for people with IDD. However, it is apparent that there are often insufficient services and supports in place for this ideal to be realised, particularly for the transition periods associated with adulthood. Individuals

with severe functional limitations or behavioural problems are less likely to be included in mainstream society than persons with mild or moderate impairments.

2.5.2 Access to health services

2.5.2.1 Introduction

In many countries, the general health system only provides care for those who request it, which can marginalise people with IDD (Walsh & Kastner, 1999; Leeder & Dominello, 2005; Melville, 2005; Melville, *et al.*, 2005; Krahn, *et al.*, 2006), as they and their carers may not recognise symptoms that would cause unease in others, or they may be unable to adequately articulate their needs (Evenhuis, 1997; Janicki, *et al.*, 1999; Hogg, 2001; Lunskey, *et al.*, 2002; Ruddick, 2005). Another source of concern is diagnostic overshadowing (Reiss, *et al.*, 1982; Reiss & Szyszko, 1983): where aspects of the disability ‘overshadow’ associated behavioural, physical or mental health issues and affects the conduct of diagnostic or investigative testing (Jopp & Keys, 2001; Rush, *et al.*, 2004; Centre for Developmental Disability Studies, 2006; Wood & Tracey, 2009). In addition, General Practitioners and hospital staff may be poorly educated in the recognition, care & management of many of the rarer causes of IDD, both in Australia and overseas (Minnes & Steiner, 2009).

2.5.2.2 Social and physical obstacles

Suitable access to health care for people with IDD may have been hindered by the general move from institutional living to community-based residence during the last few decades (Fisher, *et al.*, 2005). Although health care in large institutions was reported to be sub-optimal in the past (Chaney & Eyman, 2000), physicians who were usually experienced in the care of people with IDD were directly responsible for the health of residents and examinations were regularly conducted. Currently, people with IDD who live in the community may see a General Practitioner untrained in the specific problems associated with IDD, and who may experience difficulty communicating with an affected individual (Lennox, *et al.*, 2000; Iacono & Johnson, 2004; Lennox & Eastgate, 2004). Over a decade ago, an Australian study found that more than half of the health problems of individuals with IDD were not adequately managed, and a large percentage of these problems had not been diagnosed prior to the study (Beange, *et al.*, 1995). Corroboration of these results has since been published in studies conducted in the UK and USA (Timehin & Timehin, 2004; Fisher, *et al.*, 2005; Hayden, *et al.*, 2005).

It should be noted that people with mild or moderate levels of IDD are capable of making independent healthcare decisions (Allan, 1997; Wong, *et al.*, 2000; Parish, *et al.*, 2008), and may have definite ideas of what is, and is not, acceptable service from medical staff (Fender, *et al.*, 2007). Accessible and appropriately presented written information, including information about adverse reactions to medication, details of treatment plans, or other aspects of the care programme, should be made available to the individual and/or their carer to take home for reference (Unwin & Deb, 2007). The services of a health advocate, whether a parent, staff member or trained advocate, will often assist in expediting appropriate medical service delivery (Iacono, *et al.*, 2003; Lennox, *et al.*, 2004; Fisher, *et al.*, 2005; Ouellette-Kuntz, *et al.*, 2005), and many health care providers suggest the maintenance of a detailed health journal by carers to assist in health management (Hogg, 2001; Ziviani, *et al.*, 2004). Health advocates also may help to address the poor compliance often encountered among people with IDD (Edgerton, *et al.*, 1994; Iacono, *et al.*, 2003; Lennox, *et al.*, 2004).

2.5.2.3 *Health screening*

Health screening among people with IDD has been found to be deficient in many instances. Screening tests for cancers of many kinds are reported to be much rarer in people with IDD (Pearson, *et al.*, 1998; Hall & Ward, 1999; Hanna, *et al.*, 2011). However, some studies have reported that tests such as cholesterol levels and urine analysis are performed more frequently for people with IDD than in the general population (Beange, *et al.*, 1995; Martin, *et al.*, 1997; Roy, *et al.*, 1997; Wells, *et al.*, 1997; Durvasula & Beange, 2001; Janicki, *et al.*, 2002; Fisher, 2004; Iacono & Sutherland, 2006; Parish, *et al.*, 2008), suggesting that in some instances careful and continuous monitoring of patients is possible.

It is recognised internationally that the health of individuals with IDD is likely to be improved by receiving the same diagnostic procedures and the same treatment as the general population (Prasher & Janicki, 2002). Although the specific needs of people with IDD may be different to the wider population, it is mainly a question of the degree rather than type of physical health problem requiring identification and therapy.

A number of countries have either tested or introduced an annual health check for individuals with IDD (Traci, *et al.*, 2002; Cooper, *et al.*, 2006; Felce, *et al.*, 2008a; Felce, *et al.*, 2008b). In Australia, a randomised control trial of the Comprehensive

Health Assessment Program found it improved the rate of identification of health issues in people with IDD (Lennox, *et al.*, 2007). The introduction in July 2007 of an annual health assessment item specifically for use with people with IDD into the Australian Medicare Schedule aims to provide sufficient incentive for General Practitioners to perform a range of screening tests, as recommended in the guidelines produced by the Centre for Developmental Disability Studies (2006).

The cost-effectiveness of a similar screening intervention in Glasgow, Scotland has been examined by Romeo *et al.* (2009), and the health benefits to the participants in the screening investigated by Cooper *et al.* (2006), with encouraging results. In Wales, Felce and colleagues (2008a; 2008b) uncovered similar numbers of newly-identified morbidity problems at repeat health checks to the figures found at the original examinations, indicating a continued decline in the overall health of the participants with IDD. They therefore recommended that regular health checks continue to be conducted for this population.

Studies from Australia, the USA, Taiwan, and Canada have reported higher usage rates of both emergency and inpatient hospital services by children and adults with IDD when compared with the general population (Walsh, *et al.*, 1997; Balogh, *et al.*, 2005; Williams, *et al.*, 2005; Thomson, *et al.*, 2006a; b; Loh, *et al.*, 2007). Utilisation of hospital services by people with IDD is influenced by economic factors as well as by the health status of the individual (Hayden, *et al.*, 2005), as families with low incomes or limited access to health funds tend to use emergency departments more often than those who can attend general practice clinics (Iacono & Davis, 2003; Lin, *et al.*, 2004; Lin, *et al.*, 2006).

2.5.3 Health issues

2.5.3.1 Introduction

The development of chronic disease states in the general population, especially in older persons, can be predicted by many factors, e.g., low educational attainment, female gender, low income, reduced access to health services, paucity of close personal contacts, and restricted levels of physical activity (Strawbridge, *et al.*, 1996; Leveille, *et al.*, 1999; Walsh, 2002; Sudore, *et al.*, 2006). People with IDD usually fall into at least two of these risk categories, and so these predictors are often relevant to individuals with limited intellectual capacity (Holland, 2000; Temple & Walkley, 2003; Ouellette-

Kuntz, *et al.*, 2005; Stanish, *et al.*, 2006). An estimated 24-31% of the increased risk of poor health experienced by young people with IDD in the UK has been attributed to socio-economic disadvantage (Emerson & Hatton, 2007a; b), as signified by a range of indicators of socio-economic position and social capital. These findings suggest that the lives of children and adults with IDD need to be improved in the socio-economic domain in order to produce long-term benefits to health on an individual and community level. Supports that allow improved opportunities for family members to engage in paid employment appear to promise the most beneficial results (Emerson, *et al.*, 2010).

The following health-related topics are all of significance to people with AS and PWS, and also tend to present in other people with IDD. Several of the sections are related to recent health objectives of the Australian government: cardiovascular disease and diabetes (related to obesity), and musculoskeletal conditions (National Health and Medical Research Council, 2007b); mental health, and ageing and health (National Health and Medical Research Council, 2010). Information on people with a variety of disorders causing IDD has been included, where available, as a form of comparison.

2.5.3.2 *The Effects of Obesity*

A substantial percentage of individuals with PWS or AS are reported to be either overweight or obese, with excessive weight gain in persons with PWS generally obvious by the age of 4 years (Brambilla, *et al.*, 1997; Thomson, 2005; Dykens, *et al.*, 2007; Thomson, *et al.*, 2007; Miller, *et al.*, 2008). Indeed, the phenotypic profile of PWS includes hyperphagia and obesity, with only individuals who have received growth hormone treatment being likely to retain a healthy weight (Dudley, *et al.*, 2008; Cassidy & Driscoll, 2009). Among a small sample of Australian adults with IDD (aged 19-63 years), 78% of females and 62% of males were reported to exceed the normal weight for height range (Moore, *et al.*, 2004b), and a number of international studies, although often only involving only small groups of people or individuals from limited backgrounds, report that up to 50% of adults with IDD are obese (Marshall, *et al.*, 2003; Emerson, 2005; Rimmer & Yamaki, 2006; McGuire, *et al.*, 2007; Melville, *et al.*, 2007). Even three-year-old children with developmental delays are significantly more likely to be obese than their peers who exhibit normal development, and this trend tends to intensify as children grow older (Stewart, *et al.*, 2009; Emerson & Robertson, 2010; Rimmer, *et al.*, 2010). However, in one study from the UK, a higher proportion of

adults with IDD were underweight compared with population norms (Emerson, 2005) and more than 16% of individuals with IDD surveyed in Northern Ireland were underweight (Marshall, *et al.*, 2003).

Obesity leads, in turn, to numerous secondary morbidities, such as reduced mobility, increased cardiovascular disease, leg ulcers, and diabetes mellitus. Obesity-related health problems, including hypertension, osteoporosis, and scoliosis are common among individuals with PWS and as a result they often require hospital-based care (Butler, *et al.*, 2002; Butler, *et al.*, 2006a; Thomson, *et al.*, 2006b; Thomson, *et al.*, 2007; de Lind van Wijngaarden, *et al.*, 2008; Shim, *et al.*, 2010). High proportions of individuals with PWS have as the reported cause of death some complication of obesity, such as cardiovascular disease and respiratory disease (Smith, *et al.*, 2003b; Schrandt-Stumpel, *et al.*, 2004; Vogels, *et al.*, 2004b; Grugni, *et al.*, 2008).

Generally, people with IDD undertake insufficient exercise to regulate their weight and maintain bodily health, possibly due to insufficient support and supervision from family or paid carers, difficulties with transport, or financial limitations (Draheim, *et al.*, 2002; Emerson, 2005; Butler, *et al.*, 2007; McGuire, *et al.*, 2007; Temple, 2007; Temple & Walkley, 2007; Peterson, *et al.*, 2008; Finlayson, *et al.*, 2009). Additionally, they receive less community education advice on avoiding obesity and are seldom targets of 'healthy living' campaigns. The poor muscle tone commonly associated with PWS results in reduced energy expenditure compared with other obese persons (Butler, *et al.*, 2007) but overall physical function in these individuals can be improved by supervised daily exercise (Vismara, *et al.*, 2010). Epilepsy, which is common in people with AS, has been identified as a predictor of reduced physical activity levels (Finlayson, *et al.*, 2009), and the ataxia exhibited by people with AS also affects their ability to take part in sustained activity.

2.5.3.3 *Sleep disorders*

Sleep disorders are experienced by as many as one-third of all people with IDD, although some carers may be unaware that aberrant sleep behaviours constitute a health problem (Hoare, *et al.*, 1998; Gunning & Espie, 2003; Robinson & Richdale, 2004; Cotton & Richdale, 2006). Among the types of disturbances reported are: abnormal sleep/wake cycles, daytime sleepiness, refusal to settle at night, night-time waking and sleep apnoea (Sarimski, 1996; Montgomery, *et al.*, 2004; Doran, *et al.*, 2006; Gombos,

et al., 2011). Daytime sleepiness, sleep maintenance issues, and/or obstructive sleep apnoea are commonly reported in people with PWS, while settling difficulties and/or sleep maintenance problems are more often reported in people with autism and Smith-Magenis syndrome (Didden, *et al.*, 2004; Camfferman, *et al.*, 2006; Carpizo, *et al.*, 2006; Cotton & Richdale, 2006; Festen, *et al.*, 2006; Maas, *et al.*, 2009; Maas, *et al.*, 2010). Individuals with AS, especially those with poorly-controlled epilepsy, have high rates of settling difficulties and often get insufficient sleep (Pelc, *et al.*, 2008b; Conant, *et al.*, 2009). Both the physical and mental health of mothers of babies with sleep problems have been shown to be affected, and this is also true for the parents of individuals of all ages with IDD and associated sleep problems (Robinson & Richdale, 2004; Bayer, *et al.*, 2007; Foster, *et al.*, 2010).

Treatment with melatonin has been reported by two groups to be effective in improving sleep parameters in individuals with AS (Zhdanova, *et al.*, 1999; Braam, *et al.*, 2008), although the samples in both studies were small. Further trials on greater numbers of individuals with AS, and with due attention to their epilepsy type, are warranted.

The sleep disorders of children with PWS were reported by Haqq *et al.* (2003) to benefit from growth hormone therapy, although the results did not reach significance, while the infants studied by Miller and co-workers (2009a) presented mixed results. The infants who were later diagnosed with upper respiratory infections or gastroesophageal reflux tended to experience an increase in obstructive hypopnoea during GH treatment compared with those without upper respiratory problems whose incidence of hypopnoea decreased during treatment. However, other studies have reported no overall effect of GH treatment on apnoea/hypopnoea in children with PWS (Festen, *et al.*, 2006; Salvatoni, *et al.*, 2009).

2.5.3.4 *Mental health issues*

Dual diagnoses, the co-occurrence of IDD and mental health problems, are reported to be relatively common in people with reduced intellectual function (Cooper, 1999; Cooper, *et al.*, 2003; Cowley, *et al.*, 2005; Edwards, *et al.*, 2007; Myrbakk & von Tetzchner, 2008), although this co-occurrence is generally less prevalent in people with milder forms of IDD (Whitaker & Read, 2006). Psychiatric inpatient or outpatient services are accessed by individuals with IDD for a number of conditions, including

psychosis, affective disorder, depressive disorder, and dementia (Cowley, *et al.*, 2005; Thomson, *et al.*, 2006a; b; Cooper, *et al.*, 2007). The most commonly used manuals for the diagnosis of mental disorders in people with IDD are the British 'Diagnostic Criteria for psychiatric disorders for use with adults with Learning Disabilities/mental retardation' (DC-LD) and the American 'Diagnostic Manual -- Intellectual Disability' (DM-ID) (Fletcher, *et al.*, 2007): both guides should be used in conjunction with the ICD-10 (World Health Organization, 2007) and DSM-IV (American Psychiatric Association, 2000) manuals.

Problem behaviours, which have been linked to the subsequent development of psychiatric disturbance, are reported at higher rates in children with IDD than those without IDD (de Ruiter, *et al.*, 2007) and are common in individuals with PWS (Clarke & Boer, 1998; Maas, *et al.*, 2010). A range of behavioural problems, including perseveration, repetitive behaviour, and temper outbursts (Greaves, *et al.*, 2005; Woodcock, *et al.*, 2009; Ho & Dimitropoulos, 2010), are often found in people with PWS and they are considered to be at an increased risk of developing mental health problems including psychosis and bipolar disorder (Beardsmore, *et al.*, 1998; Verhoeven, *et al.*, 2003a; b; Vogels, *et al.*, 2004a). Obsessive-compulsive tendencies, including hoarding food, skin-picking, and resistance to change, are also common (Moss & Howlin, 2009; Moss, *et al.*, 2009; Morgan, *et al.*, 2010). These mental and behavioural issues in individuals with PWS can adversely affect the well-being of other family members.

A substantial number of people with AS have been reported to present with a range of autistic traits such as insistence on sameness, and ritualised and repetitive behaviours, albeit at lower rates than are reported in a number of other syndromes (Oliver, *et al.*, 2010). Compulsive behaviours and a dislike of changes are more commonly found in individuals with PWS, as these particular characteristics are central to the PWS phenotype involving food and compulsive eating (Moss & Howlin, 2009; Moss, *et al.*, 2009).

Holden and Gitlesen (2009) have highlighted the difficulty untrained people may have in differentiating between challenging behaviour and psychiatric symptomology, a problem that may have a significant effect on the reported rates of mental health issues in people with IDD. An expert assessment based on the criteria of

DSM-IV (American Psychiatric Association, 2000) is the most effective method for distinguishing between behavioural problems and mental health issues.

Treatment with appropriate antipsychotic medication appears to be more effective in reducing challenging behaviour than the administration of antidepressant drugs, although there have been relatively few randomised controlled trials conducted to determine the most beneficial medications for behavioural problems in people with IDD (Deb, *et al.*, 2007; Sohanpal, *et al.*, 2007; Unwin & Deb, 2007). A number of studies have concluded that psychotropic medications overall may be over-prescribed for people with IDD, especially by General Practitioners, and that monitoring of treatment efficacy is generally inadequate (Holden & Gitlesen, 2004; Parish, *et al.*, 2008).

2.5.3.5 *The effect of secondary conditions*

A range of other secondary health problems are found at high frequencies in people with IDD at all ages. These include respiratory disorders, gastrointestinal problems, thyroid dysfunction, and epilepsy (Evenhuis, 1997; van Schrojenstein Lantman-de Valk, *et al.*, 1997; Beange & Lennox, 1998; Kapell, *et al.*, 1998; Patja, *et al.*, 2001a; Janicki, *et al.*, 2002; Morgan, *et al.*, 2003; Hogg, *et al.*, 2007). People with AS are often subject to intractable epileptic episodes (Valente, 2006; Davies & Mackie Ogilvie, 2007; Pelc, *et al.*, 2008a), and people with PWS experience high rates of both respiratory disorders and gastrointestinal problems (Butler, *et al.*, 2002; Wilson, *et al.*, 2006; Tauber, *et al.*, 2008).

Other health issues, such as visual impairment and dental disorders are also widely reported in people with PWS and AS (Butler, *et al.*, 2002; Scardina, *et al.*, 2007). Indeed, vision disorders form part of the clinical criteria for both AS and PWS (Holm, *et al.*, 1993; Williams, *et al.*, 2006). Oral health has also been reported to be compromised in a significant proportion of the IDD population (Ouellette-Kuntz, *et al.*, 2005; Owens, *et al.*, 2006; Nachshen, *et al.*, 2009). This is may be due to a combination of factors including poor oral hygiene, low socio-economic status, and reduced compliance with treatment regimes (Conyers, *et al.*, 2004; Miyawaki, *et al.*, 2004). A recent Western Australian study reported limited use of dental services by young children with IDD (Wilkins, *et al.*, 2010).

There is some evidence that people with PWS and, to a lesser degree, AS are prone to scoliosis and other musculoskeletal malformations (Clayton-Smith, 1993;

Guerrini, *et al.*, 2003; Nagai, *et al.*, 2006; de Lind van Wijngaarden, *et al.*, 2008; Shim, *et al.*, 2010). Physical limitations such as scoliosis, osteoporosis, and balance difficulties can all lead to greater dependence on mobility assistance and an increased risk of falls in people with AS and PWS (Capodaglio, *et al.*, 2010), all of which may also adversely impact the health and well-being of the family carer. Surgical treatment of scoliosis in PWS has been reported to result in complications at an unacceptably high rate when compared to non-surgical intervention (Weiss & Goodall, 2009), so that conservative treatment is indicated unless the degree of curvature is more than 70°.

Ageing people with IDD generally have similar patterns of ill-health to the wider population, but occurring at a younger chronological age (World Health Organization, 2001; Burt, *et al.*, 2005). Furthermore, because individuals with more severe IDD usually die earlier than persons with higher functional abilities, the profile of surviving people with IDD tends to converge with that of the general population (Patja, *et al.*, 2001b; Hogg, *et al.*, 2007). Secondary conditions, i.e., disorders that are not attributable to the primary cause of disability, which are common in older members of the wider community, such as cardiovascular disease and cancer, therefore become more frequent among similarly aged people with IDD (Turner & Moss, 1996; Butler, *et al.*, 2002).

2.5.3.6 Hospitalisation in AS and PWS

Previous Western Australian research investigating the physical ill-health associated with both AS and PWS identified unique patterns for both syndromes in terms of hospitalisation patterns and specific health issues (Thomson, 2005; Thomson, *et al.*, 2006a; b; Thomson, *et al.*, 2007). This research included a total of 80 people with either AS (n = 34) or PWS (n = 46) previously or currently resident in Western Australia (Thomson, 2005; Thomson, *et al.*, 2006a; b; Thomson, *et al.*, 2007). As shown in Figure 2.1, infants and adolescents with AS in Western Australia were noticeably more likely to require hospital-based care than their peers in the general population (Thomson, *et al.*, 2007). During infancy this was mainly due to a failure to thrive and feeding difficulties, while the increase in epileptic episodes in adolescence also required more hospital-based care (Thomson, *et al.*, 2007).

In the first four years of life, individuals with PWS were admitted to hospital much more frequently than members of the general population or people with AS. A

number of serious health issues, such as failure to thrive, feeding difficulties and undescended testes, are common in infants with PWS. Admission frequency also increased for people with PWS during adolescence primarily due to hyperphagia-induced obesity that affects many aspects of their overall health (Figure 2.1).

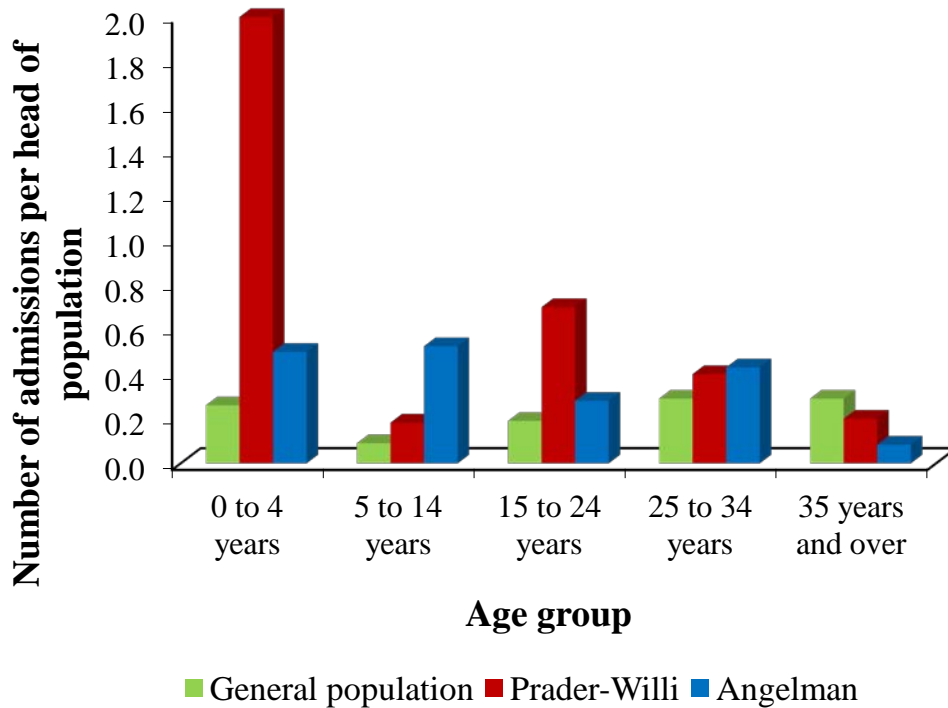


Figure 2.1 Comparison of hospital admissions per head of population between people with Angelman syndrome, Prader-Willi syndrome, and the general population in Western Australia (1998-2001).*

*Adapted from Thomson *et al.* (2007)

Similarly, an earlier Western Australian study found that, up to the age of five years, children with IDD experienced approximately quadruple the number of hospital admissions compared to children without IDD (Williams, *et al.*, 2005). A substantial proportion of these admissions involved the central nervous system, mental issues or congenital abnormalities.

The highest proportion of admissions among people with AS across the lifespan was for the treatment of epilepsy, followed by gastrointestinal disorders. People with PWS were more likely to be admitted for respiratory and breathing disorders, and undescended testes in young males (Thomson, *et al.*, 2007). Members of both groups required frequent hospitalisation for dental treatment, as dentists commonly experience

difficulty in carrying out scaling or filling procedures in people with IDD without the use of a general anaesthetic (Miyawaki, *et al.*, 2004; Faulks, *et al.*, 2007; Thomson, *et al.*, 2007).

2.5.3.7 *Summary*

People with AS or PWS, in common with most people with IDD, tend to experience more health problems than their typically-developing peers throughout the life span. Some illnesses are associated with the specific disorders; others reflect the health problems of the IDD population and may appear with an earlier onset of symptoms. Most individuals with AS and PWS therefore will require significant levels of health service provision throughout their lifetime.

2.6 *Chapter summary*

Many people with AS and PWS require some degree of assistance with daily activities throughout their lifespan. The amount and type of care needed varies between individuals and across the different life stages. People with IDD are generally less healthy, require more medical assistance, and have reduced financial security when compared to members of the general population, and individuals with AS and PWS are no exception. Although improvements have been made and continue to be introduced, measures of inequality still exist in the areas of social and employment inclusion, and in terms of leisure participation for people with AS and PWS, and for other people with IDD in Australia. Determinations of the care needs, service usage, and living situations of people with AS and PWS in WA are among the poorly researched issues relating to this particular population.

However, it is not only persons with AS and PWS themselves who are deprived of opportunity: their parents and siblings are also disadvantaged in terms of reduced free time of their own, external social pressure, and increased stress. Lack of speech and epilepsy in people with AS, and obesity and food obsession in individuals with PWS may restrict work opportunities and increase feelings of stress, among other potentially-harmful effects of the caring role on the family carers of people with these disorders. These issues will be examined in Chapter 3.

Chapter 3 Family care-giving; stresses and rewards

3.1 Introduction

The role of family carer is regarded by many parents as both long-term and open-ended, however, it can also offer substantial satisfaction and rewards (Hastings & Taunt, 2002). There is also recognition that the role is not, and should not be, exclusive to mothers or other female family members. Regardless of gender, it is rare that family carers regard the structure and function of their family as 'normal' (Nachshen, 1999; Rehm & Bradley, 2005; Kenny & McGiloway, 2007; McConkey, *et al.*, 2008), with most acknowledging that there exist considerable differences between their own families and those without a member with IDD. Nevertheless, Todd and Jones (2005) reported some mothers of people with IDD in Wales preferring to think of themselves as 'ordinary mothers'.

There tends to be a degree of stressful feelings associated with the loss of normality commonly experienced in the role of carer. Lazarus and Folkman (1984) describe stress as being experienced when the perceived burden of an event exceeds the resources available to ensure successful management of that event. According to Folkman, *et al.* (1986), the two central processes involved are:

- a) Primary appraisal; determination of a situation or event as posing a threat or being a potential stressor
- b) Secondary appraisal; assessment of the coping reserves at hand and their sufficiency to manage the threat or stressful situation

Within this model of cognitive-behavioural theory, coping is described as the cognitive and behavioural processes utilised by carers who are attempting to deal with a stressful situation (Taanila, *et al.*, 2002b). Coping appears to act as a mediating influence and can modify the emotional effect of a potentially stressful event. Given the diversity of coping responses to different stressors, the range of possible outcomes is also highly varied (Lazarus, 1999). Research involving the parents of typically-developing children and of children with disabilities indicates that some coping strategies do indeed moderate stress (Seltzer, *et al.*, 2004; Lopes, *et al.*, 2008), and that the act of coping in itself may lead to a more positive appraisal of the event and its consequences (Epel, *et al.*, 2009).

3.2 *Stress and coping models*

The ABCX (Hill, 1958) and the Double ABCX (McCubbin & Patterson, 1983) models of stress and coping are widely used in studies of families who care for a person with IDD (Nachshen, *et al.*, 2003; Jones & Passey, 2004; Minnes, *et al.*, 2007; Devonport, *et al.*, 2008). These models have been informed by transactional theory which views the interactions between events, resources, appraisals, and coping as affecting the well-being of individuals (Folkman, *et al.*, 1986; Folkman & Lazarus, 1988; Lazarus, 2000).

Within the ABCX model 'A' represents stressor events which are subject to 'primary appraisal' (Folkman, *et al.*, 1986; Folkman & Lazarus, 1988), with the perception (C) of the stressor as more or less of a problem. An event perceived as stressful may then be subjected to secondary appraisal; with consideration of the resources and supports available (B) to reduce the effects of the stressor. Coping responses are engaged in an attempt to manage the stressful situation, followed by appraisal of the outcome in terms of (X), which in the context of the current study represents carer strain or carer adjustment.

Each of these steps is interrelated, and there may be multiple cycles over time as the effectiveness of coping strategies and the availability of resources change (Brannan & Heflinger, 2001). These reiterative processes lead to the Double ABCX model (Figure 3.1) as described by Nachshen and Minnes (2005). Within this model there is a build-up of stressors, either over time or from different sources (aA); perception of the stressor(s) as a problem and appraisal of previous coping efforts (cC); use of personal and family resources and supports (bB); and positive or negative outcomes (xX). This model has been used extensively in research into the stresses associated with caring for a family member with IDD (Saloviita, *et al.*, 2003; Jones & Passey, 2004; Nachshen & Minnes, 2005; Minnes, *et al.*, 2007).

The current project was based on the Double ABCX model of stress and coping (Figure 3.1) and assessed the stressors acting on carers, their personal and family resources, the comparative use of formal and informal supports, and the reported outcomes.

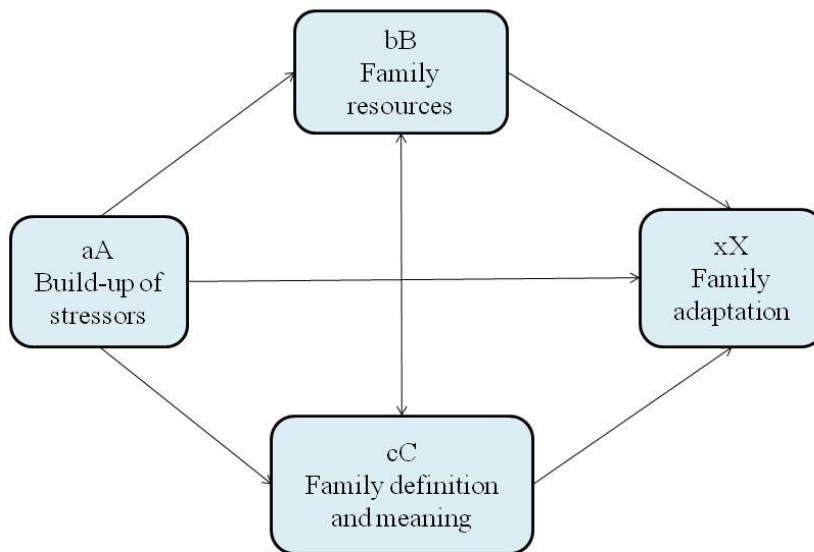


Figure 3.1 The Double ABCX model of stress and coping*

*Adapted from Nachshen & Minnes (2005)

3.2.1 Components of the stress and coping model

The presence in the family of a child or adult with IDD is often attended by considerable amounts of stress and worry. Although there have been few studies examining the stress felt by the family carers of people with AS and PWS, all report stress levels higher than the general population and, frequently, higher than similar parents with offspring with IDD from other causes (Hodapp, *et al.*, 1997; van den Borne, *et al.*, 1999; Wulffaert, *et al.*, 2010; Griffith, *et al.*, 2011).

Within the framework of the stress and coping literature the specific components of the model presented in Figure 3.1 can be identified. These components share many features with those experienced by members of the wider population; however there are some aspects that are specifically relevant to family carers of individuals with IDD. Various coping mechanisms appear to be part of either the resources, or the perceptions components of the model (Hastings, *et al.*, 2002; Hassall, *et al.*, 2005; Gold, *et al.*, 2008), and for this reason an examination of the coping literature is also included in this section.

3.2.1.1 *Stressor events: child characteristics (a)*

The parents of a child with IDD often experience considerable emotional trauma during the diagnostic period (Hogg, *et al.*, 2001; Cantwell-Bartl, 2006), and may undergo even greater distress if no definite diagnosis is provided (Graungaard & Skov, 2007). The stress is initially related to the actual process of obtaining a specific diagnosis, next to the realisation that the child is unlikely to have a 'normal' life, and finally leads to feelings of guilt and grief (McConkey, *et al.*, 2008). Once given a clear causality for their child's condition, most parents report a reduction in stress levels (Lenhard, *et al.*, 2005; Skotko, 2005; Graungaard & Skov, 2007). Clear and accurate information regarding their child's diagnosis and prognosis is considered to be a vital component of the adjustment process for the parents of a child recently diagnosed with IDD (Skotko, 2005; Graungaard & Skov, 2007; Kenny & McGilloway, 2007). Information of this nature may be provided by professionals, or by other parents who have experienced similar circumstances, for instance, the members of a relevant support group.

The personal characteristics and limitations of the child can greatly influence the stress felt within a family (Khamis, 2007). High levels of stress tend to correlate with the presence and severity of behavioural problems, which are common among people with IDD, and with communication limitations and a high level of daily care needed by the child (Chen, *et al.*, 2001; Raina, *et al.*, 2004; McConnell & Llewellyn, 2006; Neece & Baker, 2008; Norizan & Shamsuddin, 2010). For example, people with AS tend to exhibit high levels of aggressive behaviours and of overactivity and impulsivity (Oliver, *et al.*, 2010; Arron, *et al.*, 2011), and rarely attain more than a few words of vocabulary (Thomson, *et al.*, 2006a; Williams, *et al.*, 2006). Self-injury and underactivity are common features of PWS, and impulsivity and self-injury of Cri du Chat (Oliver, *et al.*, 2010; Arron, *et al.*, 2011). Overall, individuals with PWS and Williams syndrome are reported to present with greater behavioural disturbance than people with Down syndrome or Fragile X (Einfeld, *et al.*, 1999; Einfeld, 2005). In many cases, challenging behaviour seems to be triggered by external factors, such as changes of routine or refusal of food (Woodcock, *et al.*, 2011).

Periods of transition, such as from home to school, or from school to post-school options, are regarded by many parents as very stressful. There is often a perceived gap in services as their offspring move from one milieu to another (Hayden & Heller, 1997;

Maes, *et al.*, 2003), and families may experience all the trauma of re-organising routines and processes, not only for their child with IDD but for the entire family. Older parents often express feelings of concern about the future, especially the possible consequences of their own disability or death (Kenny & McGilloway, 2007; Mirza, *et al.*, 2009), as this will mean another potentially traumatic transition for their offspring.

3.2.1.2 *Stressor events: other sources (A)*

The quantity and quality of community assistance for people with disabilities is suboptimal in many countries, especially in terms of recreational opportunities and residential options (Essex, 2002; Minnes & Woodford, 2004; Davys & Haigh, 2008). As noted previously, there is a general lack of out-of-home residential places for children and adults with IDD, and access to accommodation may be preceded by lengthy delays, or be dependent on carer/s reaching a crisis point (Wodehouse & McGill, 2009). Other specific problems relate to staff co-ordination difficulties, the inconsistency of service delivery, and the limited effectiveness of some recommended interventions (McGill, *et al.*, 2006b; Wodehouse & McGill, 2009). Information regarding the available services and supports may not be accessible for all families (Redmond & Richardson, 2003; Graungaard & Skov, 2007).

Parents sometimes report they have little faith in the reliability and availability of services for their son or daughter with IDD, and feel that their knowledge of the needs and wishes of their offspring is often disregarded (Lefley, 1997; Redmond & Richardson, 2003; McGill, *et al.*, 2006b; McConkey, *et al.*, 2008). Education and support systems for family carers are also reported to be limited (Essex, 2002). In developing countries some of these problems are magnified, due to a paucity of medical and intervention services, limited family support, social stigma, and lack of community education (Goldbart & Mukherjee, 2001; Mirza, *et al.*, 2009).

Many families feel socially isolated and fail to function well (Rehm & Bradley, 2005), and some suffer considerable financial strain, partly due to the associated reduced employment capacities of family carers (Olsson & Hwang, 2006). There may also be feelings of guilt at being unable to provide the social and recreational opportunities for the person with IDD that carers feel are necessary (Sanders & Morgan, 1997). In addition, family carers, particularly those in the older age groups, may have

physical and/or emotional difficulties that affect their ability to provide the care they feel is required for their offspring.

3.2.1.3 *Resources: personal and family (b)*

Various characteristics of personal and family resources can reduce psychological stress and improve family adjustment. The use of specific coping mechanisms such as acceptance, and problem-solving (Grant & Whittell, 2000; Taanila, *et al.*, 2002b; Norizan & Shamsuddin, 2010), and feelings of self-mastery or efficacy (Jones & Passey, 2004; Paczkowski & Baker, 2007) have been associated with stress reduction among the carers of individuals with IDD. For example, the acceptance of negative aspects of the caring role generally reduces maternal feelings of stress, depression and anxiety (Lloyd & Hastings, 2008). In essence these coping mechanisms and stress moderators can all be described as forces for resilience, a poorly-defined construct which has been discussed at length by Rutter (2006) and Grant *et al.* (2007).

Parent/family support groups, whether specific to particular disorders such as Down syndrome or encompassing all conditions, can be a source of considerable advantage for the participants (Solomon, *et al.*, 2001). Members often develop a sense of self-efficacy, expand their knowledge of available resources, and gather relevant information about both their child's condition and the processes involved in accessing services (Chadwick, *et al.*, 2001). They also welcome being part of a like-minded group and experiencing the feelings of inclusiveness and belonging (Solomon, *et al.*, 2001; Hale, *et al.*, 2005).

Participation in the paid workforce is reported to have a positive effect on the parents of individuals with IDD, and may be perceived either as a break from the day-to-day requirements of their caring role, or as confirmation of their value as contributors to society (Olsson & Hwang, 2006; 2008). In reality, it is unusual for either parent to be employed full-time, and the mothers of children with IDD generally have low rates of employment, both in terms of the likelihood of having a job and the number of hours worked (Seltzer, *et al.*, 2001; Parish, *et al.*, 2004; Olsson & Hwang, 2006). Indeed, an Australian study reported slightly higher rates of part-time work in mothers of children with a disability when compared to mothers of typically-developing children, as well as lower rates of full-time work (Gordon, *et al.*, 2007). While managing family equilibrium is easier when there are sufficient available financial resources (Khamis,

2007), this situation may conflict with the amount of time parents believe they need to devote to their offspring with IDD and therefore family carer feelings about their own employment may be ambivalent.

3.2.1.4 *Supports: formal and informal (B)*

Aspects of the social environment, such as emotional support from family members and the broader community (Greenberg, *et al.*, 1997; Hong, *et al.*, 2001; Ben-Zur, *et al.*, 2005; Guralnick, *et al.*, 2008), tend to increase both maternal well-being and a sense of satisfaction among carers (Heller, *et al.*, 1997; Olsson & Hwang, 2003; Gordon, *et al.*, 2007; Plant & Sanders, 2007). Some studies, however, have found no significant mediating effect of support on the perceived stress of carers (Seltzer & Krauss, 1989; Minnes, *et al.*, 2007), and this aspect of support needs to be researched further across different carer groups.

Families who are no longer able to successfully support their relative with IDD in the family home require access to suitable facilities and assistance with their progeny's transition to alternative accommodation (Hayden & Heller, 1997; Heller, *et al.*, 1999; McConkey, 2005). Once the person with IDD has moved to supported accommodation the family may feel considerable relief, although this is often intertwined with feelings of guilt and loss (Blacher & Baker, 1994; Ward, *et al.*, 2003; Carers Australia, 2005). Service providers in developed countries increasingly recognise that transition planning and support are highly effective ways of assisting families in dealing with these changes (Bigby, *et al.*, 2002).

The use of respite care services has been reported to enhance the well-being of family carers (Freedman, *et al.*, 1999; Caldwell & Heller, 2003; Burt, *et al.*, 2005; Caldwell, 2007; MacDonald, *et al.*, 2007), although some parents feel that the process of obtaining respite is unnecessarily difficult and overly time-consuming (Doig, *et al.*, 2009). Consumer-directed support programmes in which families exercise greater control over the provision of respite and other assistance for their family member with IDD have been associated with higher levels of satisfaction with services, improved opportunities for the parents to participate in paid employment, and greater involvement in community-based activities for the individual with IDD (Caldwell & Heller, 2003).

3.2.1.5 *Perceptions and attitudes (cC)*

During the last few decades there has been increased study of the positive aspects and attitudes related to caring for a family member with IDD. These attitudes can assist in moderating the effects of stressful events, as they can alter the emotional response to these situations (Rimmerman & Muraver, 2001; Hastings, *et al.*, 2002; Hassall, *et al.*, 2005; Greer, *et al.*, 2006; Rapanaro, *et al.*, 2008).

The perceptions and beliefs of carers can act as mediators of the effects of any challenging behaviour exhibited by their child (Judge, 1998; Kim, *et al.*, 2003; Murphy, *et al.*, 2007; Plant & Sanders, 2007; Lopes, *et al.*, 2008). For instance, it has been reported that positive attitudes held by carers regarding the normality of transition processes, such as their adult offspring with IDD leaving home, can have a significant benefit on the carer's level of adaptation (Blacher & Baker, 1994; Baker & Blacher, 2002; Bigby, *et al.*, 2002; Goupil, *et al.*, 2002; Knox & Bigby, 2007; Docherty & Reid, 2009).

3.2.1.6 *Outcomes: distress or positive affect (xX)*

As noted previously, family carers of people with IDD generally experience higher levels of stress than the parents of typically-developing children, and the parents of children with AS and PWS are no exception. Van den Borne *et al.* (1999) reported that the parents of children with AS and PWS tended to feel they had less control over their lives, and that the mothers of a child with AS were more likely to show signs of depression than the fathers of either group. Parental self-esteem levels were not significantly affected by having a child with either disorder. One clear message from these parents was a need for more and better information on the disorders and the future prospects for their child (van den Borne, *et al.*, 1999). Another recent publication compared stress levels in family carers of children with AS and PWS in the Netherlands (Wulffaert, *et al.*, 2010), and examined factors that influenced the stress levels. Mothers of children with AS (58%) and PWS (26%) were much more likely to report high or very high stress levels when compared to the population norms (15%). Similarly, Griffith *et al.* (2011) reported significantly higher stress in the mothers of children with AS than in those with a child with Cornelia de Lange syndrome or Cri du Chat syndrome. However, the affected children in that study all exhibited substantial levels of challenging behaviour and therefore the findings may not extend to families where

the child does not have the same behavioural issues. Challenging behaviour also affected the stress levels of parents of children with PWS, and these parents overall reported more stress than the parents of children with mixed aetiology IDD (Hodapp, *et al.*, 1997).

The extent of maladaptation or bonadaptation (positive adaptation) resulting from both individual and family responses to stressor events have been measured by means of a large variety of instruments, as have the coping methods employed by family carers. Some of the common measures for both these concepts that have been adopted by the research community are listed in Table 3.1a & 3.1b.

Many of these instruments are commonly administered in conjunction with other measures, for example, the Questionnaire on Resources and Stress–Short Form (QRS-SF: Friedrich, *et al.*, 1983) and the Ways of Coping Questionnaire (WCQ: Folkman, *et al.*, 1986), both formed part of the data collection schedule for a study comparing cross-cultural variation/similarity in the stress felt by mothers bringing up a child with IDD (McConkey, *et al.*, 2008). Two subscales, pessimism and parent/family problems connected to the child are delimited by the QRS-SF, however there was a high correlation between the subscales and the sum across all items was used in the analysis by McConkey and co-workers (2008). Factor analysis of the QRS-SF by Scott (1989) extracted four domains of the scale: family and parent problems arising from the individual with IDD; pessimism or parent affect; child characteristics; and child incapacity, albeit with ambiguous placements for a number of items. A different version of the QRS, used by Dąbrowska and Pisula (2010), examined three domains by eliminating the pessimism/parent affect subscale. These studies taken together may indicate that the QRS in some of its short forms does not necessarily represent fixed domains (Table 3.1a). Within the ABCX model, the QRS would appear to measure stressors (A) and an attitude (C).

The Ways of Coping Questionnaire has been used for more than two decades to measure coping strategies (Dunkel-Schetter, *et al.*, 1987; Billings, *et al.*, 2000; McConkey, *et al.*, 2008). The major domains have variously been identified as: active coping, behavioural avoidance, cognitive avoidance, and social coping (Billings, *et al.*, 2000); problem-focused and emotion-focused coping (Dunkel-Schetter, *et al.*, 1987; McConkey, *et al.*, 2008); cognitive reframing, emotional respite, and direct assistance (Wineman, *et al.*, 1994); and eight individual subscales (Glidden & Natcher, 2009).

Table 3.1a Overview of some stress and coping studies

Paper (Year)	Scale	Participants	Description of scale used	Comments
Scott (1989)	Questionnaire on resources & stress – short form (QRS-SF)	66 married families including a child with DD (33), all typically developing children (33)	54 items; 4 Factors	Factor analysis of the Friedrich Short Form
McConkey <i>et al.</i> (2008)	QRSF Ways of coping (WCQ)	Mothers of children with IDD: Taiwan (98), Northern Ireland (62), Jordan (49)	Two subscales, 20 & 11 items respectively; Yes or No Two sub-scales, each 21 items: Likert 0-3	Compared scores between locations, and correlations of support, stress and coping
Dąbrowska & Pisula (2010)	QRS-SF (Polish translation)	Parents of children with autism (51), Down syndrome (54), typically developing (57)	11 sub-scales, 3 domains: True or False	Kuder-Richardson values ranged from 0.53 to 0.80: measure of stress level
Folkman <i>et al.</i> (1986)	WCQ	Married couples, woman 35-45y, with a child at home	66 items: eight sub-scales	α -values ranged from 0.61 to 0.76
Dunkel-Schetter <i>et al.</i> (1987)	WCQ (situational)	75 married couples, woman aged 25-35	67 items: Likert 0-3	Eight factors: α -values ranged from 0.61 to 0.79
Wineman <i>et al.</i> (1994)	WCQ	Adults with multiple sclerosis or spinal cord injury (655)	Eight sub-scales	Derived three factors
Billings <i>et al.</i> (2000)	WCQ	HIV+ carer (86); HIV- carer (167)	Seven sub-scales: Likert 4-point scale	Reduced to four scales by combination and splitting: α -values ranged from 0.78 to 0.86

Table 3.1b Overview of some stress and coping studies (cont'd)

Paper (Year)	Scale	Participants	Variation of scale	Comments
Glidden & Natcher (2009)	WCQ	Married couples; 35 birth parents of child with DD, 33 adoptive parents of child with DD	66 items: eight sub-scales: 4-point scale	α -values ranged from 0.60 to 0.82: quite small sample size
Carver <i>et al.</i> (1989)	Coping orientations to problems experienced (COPE)	College students, various numbers	13 sub-scales: Likert 1-4	Dispositional and situational uses
Miquelon (2006), Miquelon & Vallerand (2008)	COPE (short version, situational)	Undergraduate students (240)	'Vigilant'; 4 items: 'Avoidant'; 8 items: Likert 0-6	Vigilant α =0.70: Avoidant α =0.74
Sheppard & Crocker (2008)	COPE (situational)	102 mothers of families in need and not in receipt of support	Six of possible 13 sub-scales	Relationship with Locus of Control
Smith <i>et al.</i> (2008)	COPE (dispositional)	151 mothers of toddlers and 201 mothers of adolescents with ASD	Six of possible 13 sub-scales: Likert 1-4	α values ranged from 0.60 to 0.86
Nachshen <i>et al.</i> (2003)	Family stress & coping interview (FSCI)	Parents (106) of people with IDD: wide range of ages for both groups	22 items: Likert scale 0-4	α =0.89; r =0.80; face and discriminant validity good
Minnes <i>et al.</i> (2007)	FSCI	Parents (80) aged >50yr, caring for an adult with IDD at home	24 items: Likert scale 0-4	α =0.90: quantitative and qualitative data
Lopes <i>et al.</i> (2008)	FSCI WCQ (short form)	Parents (29) of children with DD; parents (17) of children without DD	22 of 24 items 31 items: Likert 0-3	Stress level Coping methods

Such variation in domains and the number of sub-scales employed for various studies makes comparisons between studies difficult (Table 3.1a).

The Coping Orientations to Problems Experienced Scale (COPEs) has been tested for reliability, internal consistency, and both discriminant and convergent validity over the course of the development of the instrument (Carver, *et al.*, 1989). It has been used in a number of versions in various studies. Miquelon and Vallerand employed an academic version of the scale to assess coping in undergraduate students (2006; 2008); Sheppard and Crocker explored the influence of locus of control on the use of various coping mechanism identified by the COPEs (2008); and six subscales were selected in a comparison of mothers of toddlers and adolescents with autism (Smith, *et al.*, 2008). The reported α -values indicated good internal consistency (Table 3.1b).

One of the perceived advantages of the Family Stress and Coping Interview (FSCI) (Nachshen, *et al.*, 2003) is the inclusion within a single instrument of both a measure of perceived stress and a qualitative assessment of the coping strategies used by the participants (Minnes & Nachshen, 1997; Minnes & Woodford, 2004; Minnes, *et al.*, 2007). However, some studies using the FSCI have utilised the quantitative measurement without drawing on the qualitative potential of this instrument (Lopes, *et al.*, 2008; Tehee, *et al.*, 2009). The FSCI and its predecessors were specifically designed for use with the families of people with IDD and the reported psychometric properties are also good (Table 3.1b).

Although most carers express negative emotions and describe their lives as stressful, a majority also report that caring for their offspring with IDD has some positive effects on their own physical and emotional health, and on family function. Hayden and Heller (1997) found that older parents of adults with IDD used their social networks to better manage their carer role, and that they felt less burdened than parents under the age of 55 years. The parents of individuals with IDD generally feel considerable positive attachment to their offspring (Hong, *et al.*, 2001).

Indicators of the intensity of psychological stress or distress, and the ensuing biological burden faced by the long-term carers of people with IDD have been reported by numerous authors (Mak & Ho, 2007; Murphy, *et al.*, 2007; Trute, *et al.*, 2007; Rapanaro, *et al.*, 2008; Bauer, *et al.*, 2009). In a study of mothers caring for a chronically-ill child, physiological changes at the cellular level mediated via reduced

telomerase activity and truncated telomeres, i.e., shortened ends of chromosomes, were associated with increased psychological stress (Epel, *et al.*, 2004). Both of these changes are considered to be signs of biological ageing, and may be precursors of cardiovascular disease (Epel, *et al.*, 2006; Martin, 2007).

Systemic reactions to chronic stress of this nature could be considered analogous to the reduction of adaptability in response to environmental challenges that appears to occur during normal ageing (Bauer, *et al.*, 2009). There seems little doubt that similar physical effects would be found in the carers of individuals with IDD, which raises important questions as to how best carers can be assisted to continue to function effectively in their role, especially if their own health is impaired (Epel, *et al.*, 2004; Epel, *et al.*, 2006). In some countries where private health insurance is common, potential difficulties may arise regarding the provision of appropriate life and health insurance cover for carers of people with IDD, with higher premiums imposed in recognition of the stressful home circumstances (Flood, 2005; Benson, *et al.*, 2007).

3.3 *Coping strategies*

Two categories of coping strategies are predominantly recognised throughout the literature, and both may include strategies that involve either behavioural, physical actions, or cognitive, mental reactions (Folkman, 1984). The first, problem-focus, is aimed at changing the situations arising from the problems or behaviour of the individual with IDD. The other, emotion-focus, concentrates on the reduction or management of the subjective assessment by the carer of the emotional effects of stress (Jones & Passey, 2004; Garland, 2007; Kenny & McGilloway, 2007; Van Der Veek, *et al.*, 2009).

3.3.1 *Problem-focused coping*

The adoption of problem-focused strategies is reported to have a positive impact on maternal well-being (Smith, *et al.*, 2008), and many of the predominant coping strategies tend to belong to this category (Minnes, *et al.*, 2007; Lopes, *et al.*, 2008). Specific strategies generally attributed to this category are presented in Table 3.2. As can be seen from the descriptions, there is a certain amount of overlap between these categories. This will be discussed further in 3.3.3.

Table 3.2 Concepts and definitions from the problem-focused coping category

Coping concept	Operational definition
Active	Taking direct action to remove the stressor or reduce its effect, increasing one's efforts
Confrontive	Strenuous efforts to deal with the situation
Planning	Making a plan of action, thinking about what steps to take
Suppression	Putting off other activities to concentrate on the task at hand, trying to avoid being distracted from the task at hand
Accommodative	Adjustment of goals
Restraint	Waiting for the appropriate time to act, not acting prematurely
Positive re-interpretation and growth	Looking for the good, accentuating the positives, creating positive meaning
Seeking support (instrumental), seeking social support	Seeking help, advice, or knowledge from professionals, family and peers, mobilizing agency or professional assistance
Mobilizing professional help	Getting assistance from professional services and agencies
Problem-solving	Having a regular structure or routine, talking things over with others, setting priorities, thinking about the problem and devising strategies

Sources: Carver, *et al.* (1989), Hayden & Heller (1997), Woodford (1998), Saloviita, *et al.* (2003), Hastings, *et al.* (2005b), Lloyd & Hastings (2008), Glidden & Natcher (2009)

3.3.2 Emotion-focused coping

Emotion-focused coping is reportedly both rarer and less effective in reducing stress among parent carers (Smith, *et al.*, 2008), although the fathers of children with IDD in one study gained more benefit from emotion-focused strategies and used them more often than the mothers in the same study (MacDonald, *et al.*, 2007). Other studies have concluded that active emotion-focused strategies, e.g., creating positive meaning, have adaptive effects whereas avoidance emotion-focused strategies, e.g., acting as if the stressor is not real, do not (Schnider, *et al.*, 2007; Glass, *et al.*, 2009). A range of the concepts and definitions often associated with emotion-focused coping are presented in Table 3.3. There are some categories where the definitions used by different authors are substantially different from each other. The implications of this are discussed in 3.3.3.

Table 3.3 Concepts and definitions from the emotion-focused coping category

Coping concept	Definition
Reframing, positive appraisal	Redefining events to make them more manageable, creating positive meaning
Positive coping	Using humour, looking for the positive, seeking emotional comfort
Seeking social/emotional support	Seeking understanding, sympathy, moral support, seeking support from family and friends
Acceptance, passive appraisal	Accepting the reality of the situation, acknowledging that it may not be amenable to change, avoiding a situation
Positive re-interpretation and growth	Looking for the good, accentuating the positives, creating positive meaning
Turning to religion	Using religion as emotional support, to assist with positive re-interpretation, seeking spiritual support
Self-control	Attempting to regulate feelings
Self-blame	Taking responsibility, acknowledging role in problem
Wishful thinking	Hoping for change, behavioural efforts to avoid or escape the problem
Mental disengagement, distraction	Attempting to mentally detach oneself, reduce the significance of a situation, distraction (day-dreaming, TV/movies, drugs), saving time for oneself
Behavioural disengagement	Giving up attempts to manage the situation, concentrate on the stressor situation
Denial	Refusing to believe the situation is occurring, acting as if the stressor is not real
Focus, venting	Concentrating on the source of the stress, expressing emotions
Avoidance	A combination of venting, behavioural and mental disengagement
Managing meaning	Looking for the positive, self-belief, not blaming the person, seeing the funny side of a situation, accepting the situation, doing what has to be done

Sources: Carver, *et al.* (1989), Kramer (1993), Hayden & Heller (1997), Grant & Whittell (2000), Saloviita, *et al.* (2003), Hastings, *et al.* (2005b), Lloyd & Hastings (2008)

3.3.3 Variations on a theme

Definitions of specific coping strategies can be widely variable when compared from author-to-author: indeed the same coping mechanism can function in either a problem- or an emotion-focused way, or even both simultaneously (Carver, *et al.*, 1989). This can make comparisons of the effectiveness of various approaches highly

problematic. For example, within the problem-focused category (Table 3.2) confrontive coping as used by Glidden & Natcher (2009) contains elements of both the active and suppression coping strategies of Seltzer *et al.*, (1995), and problem-solving can be interpreted to incorporate both making a plan and taking direct action (van den Borne, *et al.*, 1999; Glidden & Natcher, 2009). Some authors divide seeking support into instrumental, i.e., obtaining practical assistance, especially from professionals and service providers, and social, i.e., getting help from family, friends, and people in a similar situation (Hayden & Heller, 1997; Woodford, 1998). By comparison, Carver *et al.* (1989) separated social support into instrumental, i.e., getting practical advice and assistance, and emotional, i.e., expressing feelings or getting sympathy.

Similar contentions occur within the emotion-focused category. Venting, self-blame and behavioural disengagement are all included in the category of active avoidance by Hastings *et al.* (2005b) and Lloyd & Hastings (2008). Mental disengagement comprises some aspects included in active avoidance (Hastings, *et al.*, 2005b; Lloyd & Hastings, 2008), but also in managing meanings (Grant & Whittell, 2000). Positive coping (Hastings, *et al.*, 2005b) would seem to encompass parts of reframing, seeking emotional support, and mental disengagement (the use of humour to distract the mind) using this model.

Glass *et al.* (2009) followed Schnider *et al.* (2007) in placing the active emotion-focused subscales of the Brief COPE, namely venting, positive reframing, humour, acceptance and emotional support, within their version of the problem-focused coping category. This was done with the purpose of combining the adaptive subscales together, leaving the less adaptive subscales to form the emotion-focused coping category (Schnider, *et al.*, 2007; Glass, *et al.*, 2009). However, this strategy may have been productive of further complication in the coping literature.

A number of studies have identified three coping categories derived from the WCQ (Folkman & Lazarus, 1988): problem-oriented, perception-oriented, and emotion-oriented (Daniels, 1999; Lopes, *et al.*, 2008). Within this scheme, problem-oriented involves active coping and direct assistance; perception-oriented coping includes changing the appraisal of the problem by asking advice or gaining knowledge and cognitive reframing; and emotion-oriented entails emotional respite by use of distraction or denial tactics (Wineman, *et al.*, 1994; McColl & Skinner, 1995). Perception-oriented coping in this schema thus includes some aspects that are generally

included in problem-focused coping and others normally related to emotion-focused coping.

Hastings *et al.* (2005b) derived four coping categories from the Brief COPE scale (Carver, *et al.*, 1989) by using factor analysis. These are active avoidance coping, problem-focused coping, positive coping, and religious/denial coping. However, these factors do not fit well within the problem-focus/emotion-focus dichotomy as there are elements of both major categories (in general use) represented within their problem-focus factor. In addition, one of the 'acceptance' and one of the 'distraction' items from the COPE scale did not contribute load on to any of the factors (Hastings, *et al.*, 2005b), and it is not immediately clear why religious coping, which is generally viewed as assisting adaptation (Khamis, 2007; Norizan & Shamsuddin, 2010), and denial coping, which tends to lower adaptation (Carver, *et al.*, 1989), should load to a single factor.

The operational definitions and placements of the coping categories are, therefore, subject to some degree of variability across studies. While some consider 'positive re-interpretation and growth' as linked to problem-focused strategies (Carver, *et al.*, 1989; Seltzer, *et al.*, 1995), others prefer to place the concept within the emotion-focused category (Lazarus & Folkman, 1984). Other studies have found that seeking social support for instrumental reasons co-occurs with seeking social support for emotional reasons. Similarly, the concepts of acceptance and restraint may also contain both emotion-focused and problem-focused aspects (Carver, *et al.*, 1989). As a result, there is considerable variation in the use of the concepts in theoretical models, which is evidenced by the repetition of terms within the discussion of various coping strategies.

It is clear that many of the aforementioned strategies can be regarded as either promoting or inhibiting adaptation, depending on the appraisal of each individual. For example, focus/venting may either play a role in reducing perceived stress, or the act of dwelling on the situation may accentuate the stress (Epel, *et al.*, 2009).

3.4 Chapter summary

Stress and coping theory is beset by variability in both the models and the definitions of specific concepts. There is evidence to suggest that a complex interplay of resources, perceptions and coping all influence the effects of stressors. However, the directions and extent of these effects are not yet conclusively determined. The trend is for problem-focused coping to increase adaptation and for emotion-focused coping to

either decrease or have no effect on adaptation. Using the family carers of people with AS or PWS in Western Australia as informants, the present research project aims to determine the coping strategies used by family carers, and also analyse the applicability of the Double ABCX model to the study findings.

Chapter 4 Subjects and methods

4.1 Introduction

Interactions between characteristics of people with AS or PWS, their life experiences, and the effects of these on the lives of their family carers are open to exploration. There are many potential research questions arising from the material covered in the literature review (Chapters 2 & 3). There appears little doubt that a number of factors related to people with IDD, such as communication abilities and behavioural problems, interact in many ways with personal, family, and societal resources and attitudes to affect the stress experienced by family carers. The variety of reports indicating either modifying or mediating effects for an extensive number of factors made it difficult to discern a clear path of investigation for the present study. Chief areas of concern include the effect on carers of physical and/or cognitive deficiencies of their offspring, and the effect of specific coping styles on the perceived stress of carers. In addition, the effects of behavioural issues specific to people with PWS (such as those associated with hyperphagia) and AS (hyperactivity, for example) may, in turn, be modified by various parental resources and attitudes. Of necessity, a small sub-section of these possibilities has been selected for the study.

This chapter will present the research questions and hypotheses, the overarching methodology of the study, a discussion of ethical issues and processes, and a description of the recruitment processes. These will be followed by discussion of the choice and suitability of the research instruments and, finally, a report on the focus groups conducted to identify any problematic aspects of the instruments.

4.2 Research questions and hypotheses

4.2.1 Research question 1

How much care is required during different life stages (infancy, adolescence and adulthood) for persons with AS or PWS?

4.2.1.1 Hypotheses

Based on earlier research, it is expected that:

- a) infants with either syndrome will be cared for in the family home and require specific developmental support services

- b) adolescents with AS or PWS will primarily live in the home setting, but require specific special education services
- c) adults with AS or PWS will increasingly work in supported employment and live outside the family home, primarily as a consequence of age-related health problems affecting their family caregivers and their ability to care full-time for their children

4.2.2 *Research question 2*

What stressors (aA in the Double ABCX model) commonly affect the family carers of people with AS or PWS?

4.2.2.1 *Hypotheses*

It is expected that caregivers of individuals with AS or PWS will be affected by a variety of external and internal stressors, including:

- a) the amount of time required to provide adequate care,
- b) the age of the person with AS or PWS
- c) the extent of care-giving responsibilities for other individuals
- d) their own physical health

4.2.3 *Research question 3*

What resources and supports (bB in the Double ABCX model) are utilised by the family carers of people with AS or PWS?

4.2.3.1 *Hypotheses*

Based on the literature reviewed earlier, it is expected that:

- a) family carers will belong to either a syndrome-specific support group or a generic carer support group
- b) family carers will report that formal support organisations provide practical and emotional assistance
- c) family carers of older persons with AS or PWS will use more respite and/or out-of-home accommodation than the family carers of younger individuals with the disorders

4.2.4 *Research question 4*

How much satisfaction (cC in the Double ABCX model) do family carers of people with AS or PWS get from the role?

4.2.4.1 Hypothesis

Carers will report a mixture of positive and negative feeling associated with their role

4.2.5 *Research question 5*

What coping modes are commonly used by the family carers of people with AS or PWS?

4.2.5.1 Hypothesis

A range of both problem-focused coping strategies and emotion-focused approaches will be reported by family carers

4.2.6 *Research question 6*

How much stress (xX in the Double ABCX model) is reported by the family carers of people with AS or PWS?

4.2.6.1 Hypotheses

After consideration of the literature, it is predicted that

- a) individuals caring for people with greater functional limitations, more behavioural problems and/or no speech will report higher stress levels
- b) stress will be moderated by use of problem-focused coping mechanisms
- c) stress will be moderated by greater use of supports and resources

4.3 *Methodological overview*

The major focus of the project was basic research into the specific stress and coping issues associated with having a child with AS or PWS in Western Australia (WA), with the intention of informing local policies and practices of service providers and support workers, and assisting family members in the caring role. The cross-sectional study was both descriptive, by investigating the lives of people with the disorders and of their family carers, and explanatory, by assessing the effects of the disorders on family carers using the Double ABCX model (Neuman, 2006) as a guide.

Survey instruments either already exist or could have been designed to provide suitable quantitative data to address each of the research questions proposed in 4.2. Within stress and coping research, many of the existing instruments are designed to use closed questions and thus supply quantitative data (Mazzola, *et al.*, 2011), e.g., the Coping Inventory for Stressful Situations requires a response on a Likert-scale to a number of items, such as ‘Work to understand the situation’ (Endler & Parker, 1990; 1994; Dąbrowska & Pisula, 2010). However, it was felt that some of the research questions for this study, particularly Question 5, could well be extended by the use of open-ended questions and a qualitative analysis approach. The stress and coping literature includes descriptions of numerous specific coping strategies (however poorly defined, see 3.3), but there remains the possibility that using only closed-end scales could fail to gather either the complete range of coping mechanisms used by this group of people (Mazzola, *et al.*, 2011), or capture the entire spectrum of stressor events.

It was thus felt that a mixed methods approach was appropriate for this particular study and it was anticipated that this would present a more comprehensive view of factors affecting the lives of family carers of people with AS and PWS in WA.

The overall data collection procedure was sequential, with an initial round of surveys based on a group of volunteer family carers from within a sample frame that included carers of all people identified with AS and PWS in WA. This process was then followed by interviews and further quantitative data collection using a combination criterion/convenience sampling scheme (Onwuegbuzie & Collins, 2007). The criteria were that the family carer had responded to the initial survey invitation, regardless of whether they had been eligible to complete the whole range of survey instruments, and that they were willing to be interviewed. The sample was neither random nor stratified according to the population, but rather selected by convenience.

The original intent of the research project was to correlate the numerical results of the quantitative data with the qualitative data; however the small number of participants precluded meaningful statistical analysis. Some experts believe a minimum of 62 participants is required to provide sufficient statistical power in a study examining correlations between factors (Onwuegbuzie & Collins, 2007). Therefore the results from both methods of data collection were used to clarify and illustrate one another in a complementary style (Bryman, 2006; Onwuegbuzie & Collins, 2007), with

the major emphasis being placed on the qualitative component. Detailed discussion of of data collection and analysis is included in Chapters 5 and 6 as appropriate.

4.4 Ethics approval processes

4.4.1 Approvals from Human Research Ethics Committees

The timelines involved in obtaining ethics approval for the project from the various stake-holders are summarized in Table 4.1.

The initial application for ethics approval was made to the Human Research Ethics Committee of Edith Cowan University (ECU HREC) on 5th December 2005. This Committee granted approval on 8th February 2006 for the project to be initiated, subject to the conduct and reporting of Discussion Groups to assess the survey instruments for effectiveness, suitability, and use of appropriate wording (see Chapter 4.7; Appendix V). Once this process had been completed, full approval was granted to begin the project on 28th August 2006.

In order to access the records of Genetic Services WA an application was submitted to the Princess Margaret Hospital for Children (PMH) HREC on 19th December 2006. At the same time an application for ethics approval to access linked health data through the Data Linkage Unit (DLU) was submitted to the Confidentiality in Health Information Committee (CHIC) of the Western Australian Department of Health.

Following extended correspondence with the secretary of the PMH HREC, it was determined that they were in fact not the correct organisation to be handling this application for ethics approval. Subsequently, an application was prepared and sent to the King Edward Memorial Hospital for Women (KEMH) HREC on 12th April 2007. After further amendments and clarifications, the KEMH HREC granted approval for the project to proceed on 24th July 2007.

A number of separate concerns were raised by CHIC in early February 2007 and were comprehensively addressed by letter on 20th February 2007. The Committee then signified that it would grant approval for the project once the HREC from PMH had also given approval. The final approval to proceed was received from CHIC on 20th August 2007.

The project was also assessed by staff from the Disability Services Commission of WA (DSC) and approved by the Director-General, Dr Ron Chalmers on 5th October 2007.

Table 4.1 Timeline for approvals from the relevant Human Research Ethics Committee and Data Custodians

Group	Dec 2005	Jan-Jun 2006	July-Dec 2006	Jan-Jun 2007	July-Dec 2007	Jan-Jun 2008	July-Dec 2008
ECU	Application	Conditional approval	Full approval			Amendment accepted	Small number accepted
KEMH			Application		Full approval	Amendment accepted	Small number accepted
CHIC			Application		Full approval	Amendment accepted	Small number accepted
DSC				Notified			
DLU					Application		Small number accepted
Data custodians						Application	Small number accepted

On approaching the Department of Health WA HREC with the ethical approvals from ECU and KEMH, further alterations were requested, i.e., the addition of a consent form specifically to authorise access to the linked health data; and proxy consent only to be provided by the legal guardian or holder of an enduring power-of-attorney for any adult with IDD adjudged incapable of giving self-consent. Once these changes had been made, the amended protocols were sent to all Ethics Committees involved on 7th December 2007. Due approval of the amendments was received from all concerned by 19th March 2008.

A further set of amendments was required once it became evident that the total number of participants for the project would not exceed 30 individuals. The Department of Health WA indicated that in light of these small numbers and the increased possibility of identification, express permission to access linked data would be required from each of the HRECs and the custodians of the health datasets for which access was desired. Requests for this access were forwarded to the relevant bodies on 14th June, 2008. The last of the permissions was received on 14th November 2008. In addition, the Department of Health WA stipulated that access to linked data from Federal health databases would not be permitted. Given these restrictions to data access, and the extensive delays in starting the project imposed by these various requirements, it was deemed both impracticable and of little scientific merit to continue with the application for access to linked health data. The health services usage data for all Western Australian residents are accessible to researchers in de-identified form. The Data Linkage Unit coordinates the linkage and dissemination of these data. For further information, go to the website: <http://www.sph.uwa.edu.au/research/wadl>

4.4.2 Ethical issues encountered

4.4.2.1 Consent

Foremost among the ethical considerations of involving people with IDD in research is the question of informed or proxy consent. The problem can be divided into two main issues: 1) who makes the assessment of capacity of individuals with IDD to consent and how is that done; and 2) who has the legal right to provide proxy consent on behalf of a person with IDD?

4.4.2.1.1 Assessment of capacity

A review of the various aspects of gaining informed consent from people with IDD to participate in research projects concluded that there is 'no clear indication of the threshold of performance' indicative of the capacity to provide such consent (Iacono & Murray, 2003). Government Acts, whether Commonwealth or state, often fail to provide guidelines on the competency determination, thus leaving the question open to interpretation. Generally, the person should have a preference between options, and be capable of communicating their choice. These two conditions often cannot be met if the person under consideration has IDD, and more especially in individuals with particularly low levels of function and/or limited communication abilities. The current study wished to include with severe levels of IDD, which translates to people who are typically non-verbal, have multiple physical disabilities (including sensory impairments), and extremely low levels of intellectual function. It is very difficult, and sometimes impossible, for these persons to seek information on their own behalf, understand written or verbal information, or make their concerns known.

Another problematic area is the identity of the person who makes an assessment of the individual with IDD to determine their competency to provide informed consent. It is generally suggested that either the person judging the capability be well known to the affected individual, often a parental carer, or that testing be conducted by the researcher (Iacono, 2006b). But privacy constraints in operation for this project meant that the researcher could not approach most potential participants directly and therefore had no opportunity to judge competence. The candidate was thus reliant on the opinion of family members or direct care staff as to the capacity of the individuals with AS and PWS. There is some concern that, given the position of power held by carers, this might jeopardise the voluntary nature of any decision made by the individual with IDD who is adjudged capable.

If the researcher has the skills necessary to conduct an assessment of competence, or the resources to employ someone else to perform this task, there still remains the issue of proxy consent for the assessment to take place. As discussed in the *Journal of Intellectual & Developmental Disability* (Iacono, 2006b; Iacono, 2006a; McVilly & Dalton, 2006; Ramcharan, 2006), the problem only becomes moved back a step, rather than advancing towards a resolution (Iacono, 2006a).

4.4.2.1.2 Proxy consent

In Australia, a proxy decision may be made on behalf of an adult with IDD who is adjudged incompetent to provide consent. Section 2.2.12 of the National Statement on Ethical Conduct in Human Research (National Health and Medical Research Council, 2007a) requires that a person giving consent on behalf of an incapable adult be '*...a person or statutory body exercising lawful authority*'. However, the definition of the person with lawful authority varies between States. The Information Privacy Bill currently before the Western Australian Parliament defines an 'authorised representative' as a person who:

- i. is a guardian of the individual appointed under law; or*
- ii. has parental responsibility for the person; or*
- iii. is otherwise empowered under law to perform any functions or duties as an agent of or in the best interests of the individual.*

In Western Australia, a majority of adults with IDD do not have a legal guardian appointed. In practice, parents and other carers may be recognised by Centrelink (Commonwealth Welfare Agency) and other service or support organisations as being responsible for decisions about an individual's care, accommodation, and finances. However, they are not legally entitled to provide proxy consent for research participation; a position similarly reported from the USA by Aman and Handen (2006).

The State Administrative Tribunal (formerly the Guardianship and Administration Board) is the body entrusted in Western Australia to grant guardianship to parents, other family members or carers, or to the Public Advocate. Parents of adults with IDD are rarely granted guardianship however, as it is considered they already have the legal right to make proxy decisions about medical treatment, accommodation, and financial matters under s119 of the Guardianship and Administration Act 1990 (WA). According to the then Minister for Health (Hansard 6th June, 1990 [1914] and [1915]), the intent of the Act was to provide for family members to become legal guardians of relatives with diminished capability. In practice, the principle of 'least restrictive alternative' has been invoked to deny family members legal guardianship, e.g., [\[2006\] WASAT 287](#). In addition, the current policy of The Disability Services Commission of Western Australia, the statutory body responsible for service provision to people with IDD, is that legal guardians cannot be paid carers, and therefore the person who, in the

absence of family members, is generally best known to the affected individual may find it difficult be appointed as guardian (see [\[2007\] WASAT 282](#); [\[2007\] WASAT 80](#)).

Ramcharan (2006) recommended that members of the affected person's support circle (family, friends, carers) were best placed to make a decision on behalf of the individual with IDD regarding the advisability of research project involvement, and these persons also were considered to be the most likely to express unease about any breach of ethical procedures. Similarly, Dalton and M^cVilly (2004) and Freedman (2001) suggested that members of an affected person's circle may have greater insight into their best interests than a legal guardian, such as the Public Advocate, who may lack regular contact with the affected individual. The current privacy legislation in Western Australia, however, does not allow the support circle to sanction research participation, as only a legally appointed individual (or statutory body) may provide such consent for an adult without the capacity to self-consent.

In the Australian state of Victoria, Iacono and Murray (2003) needed to obtain proxy consent to allow people with IDD to participate in their study which involved data collection from third parties and a series of blood tests from the individuals with IDD. An application was submitted to the relevant board for limited guardianship to be granted to the Office of the Public Advocate, with the sole purpose of making a decision about each individual's participation in the project. The process, however, consumed time and financial resources that may not be readily available to other researchers. In contrast, another study of adults with IDD in Victoria, which was based on data supplied by staff and family members (Bigby, 2008a) reported that, in the absence of a legal guardian, the next of kin provided consent on behalf of the participants. The Victorian Guardianship and Administration Act 1986 specifically includes a section devoted to Medical Research Procedures (Part 4A, Division 6) and contains a clear definition of the person(s) responsible for providing proxy informed consent (Part 4A, Division 1, Sect 37), which includes a primary caregiver or nearest relative.

4.4.2.2 *Privacy and data security*

The data collected for this project are potentially identifiable. Therefore, once the interviews were completed, the names were replaced by a unique code designating parent/offspring pairs. The code was held by Dr Angus Stewart, Senior Lecturer, School of Exercise, Biomedical and Health Sciences, Edith Cowan University. All hard

copies of data were stored in a locked cabinet within a locked room (Building 19:387) at Edith Cowan University, Joondalup Campus, and electronic records were kept on a password-protected computer at the same location.

All documents, tapes and electronic records will be kept in secure storage at the above location for a minimum of five years after the completion of the study, as required by the governing ethics bodies. If appropriate, at the expiration of this time the records will be disposed of according to the Edith Cowan University policy on the destruction of confidential material.

It is recognised that the small numbers of people participating in the study and the limited total population size of Western Australia may make identification of individuals possible. It is therefore important to avoid the use of identifying information such as birth date, place of residence, or school in all written or verbal reports. Direct quotations from the interviews do not include any identifying features.

A carer-version summary of the results of this study and the implications of these results will be provided to the Angelman Association and the Prader-Willi Association. The Disability Services Commission and Genetic Services WA will also be provided with a report of the results, with the option to disseminate a carer version to their clients if deemed appropriate.

4.5 Recruitment processes

4.5.1 Identification

4.5.1.1 The Disability Services Commission of Western Australia

The Disability Services Commission (DSC) has been the primary State Government agency responsible for providing services to Western Australians with IDD since 1952. DSC is responsible for enhancing the opportunities, inclusiveness and quality of life for individuals with disabilities through direct services and by funding non-government service providers. During the 2008-2009 financial year DSC provided services for 20,540 persons in the state of Western Australia (Disability Services Commission, 2009), which at that time had a total population of 2.26 million (Australian Bureau of Statistics, 2009a).

Any individual registered with DSC (previously Irrabeena) since 1953 has an electronic record stored in the client database. Clients are referred to DSC for an

eligibility assessment from a variety of sources around the state, including hospitals, specialist physicians and general practitioners. Referrals also may be made by family members on behalf of people with IDD.

Demographic data recorded electronically on the DSC database include gender, date of birth, contact details, type of residence (e.g., complete residential care, group home or family home), marital status, employment status and ethnicity. Diagnostic variables that are recorded include the clinical diagnosis, level of intellectual impairment, and additional co-morbidities such as congenital malformations, sensory disabilities, and any associated physical disability. Information is also included if a genetic aetiology is known or suspected, and whether any psychiatric condition exists. This last field is limited in its interpretation and only indicates whether a person has a behavioural, neurotic or psychotic component to their presentation. The electronic and paper records for individuals are sometimes incomplete due to varied contact of the client with DSC-based health care workers, and the age of the client. For example, although older clients potentially have more opportunity for points of contact, they are less likely to have retrospective data transferred to the database when the electronic system was introduced in 1986. The quality of data recorded has also been affected by changes in information collection practices since the inception of the database (Megahey, 1996).

A number of studies have reported that a significant proportion of people with IDD have no specified aetiology for their condition, even as adults (Hou, *et al.*, 1998; Strømme, 2000; van Buggenhout, *et al.*, 2001; Verri, *et al.*, 2004). These late or unconfirmed diagnoses can lead to inappropriate therapeutic regimes, poor management programs, and sub-optimal genetic counselling for the family. The records at DSC similarly include a substantial number of people with no aetiological diagnosis, and Petterson *et al.* (2005) reported that 52% of DSC records studied involving children contained no definite diagnosis. Thus, from the lower than expected numbers of people with AS or PWS identified from the DSC database in a previous study (Thomson, 2005), it is at least possible that undiagnosed cases remain in the record system.

4.5.1.2 *Genetics Services WA*

Genetic Services WA provides services in Western Australia to individuals with genetic disorders or those at risk of carrying genetic disorders, and their families. A

referral from a Medical Practitioner is needed to book a consultation at GSWA, although the initial impetus for investigation may come from an individual or couple if they have reason to be concerned about hereditary disorders.

A range of diagnostic, counselling, and predictive services are available through GSWA for those affected by, or at risk of, genetic and congenital disorders. Details of the specific molecular and/or cytogenetic tests conducted, and the results of those tests, are maintained on the GSWA database along with contact details and other demographic data.

4.5.1.3 Angelman Syndrome Association (WA)

The Angelman Syndrome Association WA is a branch of the Angelman Syndrome Association of Australia, with a vice-president and local committee. The National Association holds a biennial conference to allow members to meet and discuss areas of interest and to learn about the latest research into the syndrome. The most recent conference was held in October 2009 in Perth, Western Australia.

4.5.1.4 Prader-Willi Association (WA)

The national Prader-Willi Syndrome Association has been operating for a number of years and became officially incorporated in 2000. It comprises representatives from all Australian states in which there is an affiliated PWS group. While the State Associations provide support and services for members in their local area, the National Association is involved in issues of national relevance. These include such matters as the development and dissemination of educational material about the condition, liaison with the Federal Government on funding for pharmaceutical treatments and the promotion of ethical research into management of the condition. The Prader-Willi Association (WA) is a major source of information and support for families with a child or adult with PWS in Western Australia.

4.5.2 Recruitment

The majority of the study cohort was initially recruited through direct contact with the organisers of the two family support groups. The candidate is not involved in the conduct of either of these two organisations; however, she joined the Angelman Syndrome Association as an Associate Member in order to present a paper based on the findings of the present study at the National Angelman Syndrome Conference in 2009.

In an attempt to increase the number of participants the records of both DSC and GSWA were examined by staff members of these organisations to identify diagnosed cases of either syndrome. A member of staff at DSC identified from the database all individuals with a clinical or genetic diagnosis of Angelman syndrome or Prader-Willi syndrome using the Terminology and Classification of Mental Retardation codes 6759 and 6715, respectively (Heber, 1959). Similarly, a staff member from GSWA prepared a list containing the name, date of birth and address for all individuals diagnosed with AS or PWS through that service. This was delivered to the liaison at DSC, along with a similar list from the code-holder at Edith Cowan University comprising those individuals from the parent support groups who had already volunteered to take part. After ensuring there was no duplication of the contacts, information sheets (Appendix II) and invitation letters (Appendix III) were sent by the staff member to all the people with AS or PWS who were registered with DSC or to their family caregiver (n = 108). The 'Invitation to participate' from the Head of GSWA (Appendix IV) was posted by the contact within the organisation to the previously identified clients of that service who were not registered at DSC (n = 11).

People who expressed an interest in taking part in the study were asked to contact the investigator. Those who responded were asked a number of preliminary questions:

- a) Does the person you care for have Angelman syndrome or Prader-Willi syndrome?
- b) Is he/she:
 - i. Less than 18 years old
 - ii. 18 years or older?
 - iii. Capable of providing informed consent to participate in the study?
- c) What is your postal address?

Carers were asked if, in their opinion, the person with AS or PWS was capable of informed consent. Best practice would require the supply of guidelines to potential participants to direct their assessment of the capacity of the individual for whom they were caring to provide informed consent.

Study information packs comprising the relevant consent forms (Appendix V), questionnaires and the specific Clinical Data sheet (Appendix VI), and a reply-paid envelope, were posted to each respondent. On return of the completed questionnaires,

carers who consented to be interviewed were contacted to arrange a convenient time and venue.

4.6 Study Instruments

4.6.1 Instruments

The packages sent to all volunteer carers contained a number of items listed in Table 4.2. The complete questionnaires are presented in Appendix VI.

Table 4.2 Contents of the packages posted to participants and the data collected

Item	Data collected from the item
Consent forms	Informed consent and/or proxy consent
Demographic questionnaire	Year of birth, residence, education, employment
Carer Information questionnaire	Demographic information, carer duties, attitudes, general health
Food-related Problems Questionnaire	Presence or absence of behavioural traits related to food
Clinical presentation sheet	Relevant clinical history of the person with IDD
Reply-paid envelope	

The questionnaires were chosen to supply data attributable to the factors of the Double ABCX model, as discussed in Section 3.2.1. Characteristics of the individual with AS or PWS, such as poor verbal ability or physical limitations, that have been previously reported to act as stressors for family carers (Raina, *et al.*, 2004; Norizan & Shamsuddin, 2010), represent the ‘a’ factor. Caregiver characteristics that are potential stressors, e.g., personal health problems, were also assessed (A factor), along with any resources (bB factors) and perceptions (cC), such as the use of respite care and satisfaction with the caring role, which might affect the stress reactions of the carer (xX).

4.6.1.1 Demographic Data questionnaire

The questionnaire (Appendix VI) included questions on the year of birth, diagnosis, postcode of residence and other basic demographic information on the person with IDD. In addition, information with regard to the living arrangements, and education/employment of the individual with AS or PWS, the family structure, and parental education/employment was included.

4.6.1.2 *Carer Information questionnaire*

This included:

- a) Five questions relating to the amount of direct care needed by the individual, and the amount and types of out-of-home services utilised: these questions do not form a recognised scale, but simply asked for factual information.
- b) Four questions aimed at providing a subjective view of the health of the carer, adapted from Lawton *et al.* (1982): this measure has been reported to have good psychometric properties (Pruchno & Patrick, 1999). In their study of 251 mothers of people with IDD or mental health problems the coefficient alpha values were 0.75 and 0.79 (Pruchno & Patrick, 1999). Another good, but longer, measure that could have been used is the MOS 12-item Short Form Health Survey (SF-12) (Ware, *et al.*, 1993), which has been used extensively in research (Bayer, *et al.*, 2007; Llewellyn, *et al.*, 2010b). However, similarly to Minnes *et al.*, (2007), a shorter option was chosen to keep the study instruments to a manageable size.
- c) Six items providing an assessment of carer satisfaction, adapted from Lawton *et al.* (1982): this measure also has good psychometric properties (Pruchno & Patrick, 1999; Heller & Caldwell, 2006), i.e., coefficient alpha values between 0.67 and 0.88. An alternate measure, the Positive Contributions Scale, was considered (at 50 items) too long for the present project, although it has been used successfully in other studies (Hastings, *et al.*, 2005a). Similarly, the 15-item Caregiving Satisfaction Scale (Kramer, 1993) was also slightly longer than required for the present study.

4.6.1.3 *Food-related Problems Questionnaire (FRPQ)*

A significant aspect of the Prader-Willi behavioural profile is hyperphagia, an insatiable appetite. This measure, the only one at the time specifically designed to measure hyperphagia in people with PWS, was chosen for the study as it was hypothesised that aspects of the food obsession might act as stressors for the family carer. The questionnaire was slightly adapted from the original scale for use with adults with PWS with permission from Russell & Oliver (2003) (Appendix VII). In the present study the questionnaire was applied, for the first time, to individuals with AS, as well as those with PWS, with the aim of identifying any behaviour specific to PWS or common to both of these imprinting syndromes.

The FRPQ is a 16-item questionnaire scored on a 7-point Likert scale (0 = 'never' to 6 = 'always'), with four of the items reverse-rated. Five questions relating to communication also have the option for a '7' response (equating to 'does not apply') to be entered in the absence of verbal communication (Russell & Oliver, 2003). The lack of verbal ability generally associated with AS meant that a majority of the AS group had one or more of those items marked as 'does not apply'. In addition, the young age of some of the children meant that a number of questions, e.g., 'Is it necessary to lock away food?' were less likely to be relevant.

The items from the scale were divided into three subscales; preoccupation with food, impairment of satiety, and composite negative behaviour. The latter subscale was further differentiated into; takes/stores food, eats inedible items, and responds inappropriately to unavailability of food. The instrument has been reported to have good psychometric properties with reliability scores of between 0.67 and 0.86, and Cronbach's alpha of 0.87 (Russell & Oliver, 2003). It also discriminated between people with PWS and individuals with IDD from other causes (Russell & Oliver, 2003), and has been used in other studies (Hinton, *et al.*, 2010).

Another Hyperphagia Questionnaire was developed (Dykens, *et al.*, 2007) after the commencement of the current study. It includes a measure of the strength of the hyperphagic behaviour that is not present in the FRPQ, but, when first published, had not been tested thoroughly for psychometric properties. Further evaluation of both the FRPQ and the Hyperphagia Questionnaire, especially comparing the results of both scales has the potential to provide a more authoritative measure of this particular behaviour in people with PWS and others.

4.6.1.4 *Clinical presentation sheets*

These were included to provide further information regarding any specific physical, mental and health limitations which might possibly act as stressors for their family carer/s. They were syndrome-specific and adapted from similar data sheets used internally at DSC. The diagnostic features included on the sheets were those contained in the consensus diagnostic criteria for each disorder (Holm, *et al.*, 1993; Williams, *et al.*, 1995; Williams, *et al.*, 2006). Carers were also asked to include the current height and weight of the person for whom they care (Appendix VI).

4.6.2 Interview protocol

The Family Stress and Coping Interview (FSCI) (Nachshen, *et al.*, 2003) was adapted for this study (Appendix VIII). The modifications, which were approved by the originators of the interview (Appendix VII), were minor and designed to make the interview more specific to the carers of people with PWS or AS, and to incorporate the suggestions of members of the Discussion Groups held prior to ethics approval being granted (Chapter 4.2.2).

The original FSCI was reported to have internal consistency (Cronbach's alpha = 0.89), and both face and discriminant validity (Nachshen, *et al.*, 2003), making it suitable for use as a reliable measure of family stress. The alteration of one item for this study may have affected the psychometric properties. Further studies using a larger number of participants would be needed to assess the reliability and validity of this specific form of the FSCI.

The FSCI total score has been used previously as a measure of the 'C' or perceptions and attitudes component of the ABCX model (Nachshen, *et al.*, 2003; Nachshen & Minnes, 2005; Minnes, *et al.*, 2007). However, in the present study the stress score was taken to represent the outcomes or 'X' component, in keeping with the study conducted by Lopes *et al.* (2008).

The FSCI is a semi-structured interview comprising a Likert scale assessment of stress level, and a directed discussion about the role of caring for an individual with IDD in 24 different situations. The open-ended nature of the discussion assists researchers to identify the coping mechanism/s used by individuals to deal with stress, and to compare the effectiveness of different coping styles (Nachshen, *et al.*, 2003; Nachshen & Minnes, 2005). Stress ratings are given on a scale of 0 (not stressful) to 4 (extremely stressful), thus giving a possible total range of 0-96. At the conclusion of the FSCI interview, additional questions were prepared for this project that expressly cover membership of a syndrome-specific support group and any perceived benefits of such membership.

4.7 *Discussion groups to refine the study instruments*

4.7.1 *Purpose*

As a preliminary exercise to ensure the full study met the requirements of the Human Research Ethics Committee of Edith Cowan University (see Chapter 4.4), two Discussion Groups were established. The Groups were conducted to ensure that the study protocols were effective, non-objectionable and comprehensive, and that consumer perspectives were included in the main study. The specific aims of these Discussion Groups are summarized in Table 4.3.

Table 4.3 Aims of the Discussion Groups conducted prior to commencement of the study

-
1. To refine the survey instruments to reflect the common language of the disability sector.
 2. To identify any survey items covered previously by other researchers and remove or modify them appropriately.
 3. To identify any issues of concern to the carers of individuals with IDD which were not adequately represented in the protocol.
 4. To assemble links between a range of organisations involved in the provision of services to individuals with IDD and the support of such people and their family carers.
-

4.7.2 *Methods*

Various organisations, including Carers WA, the Genetic Support Council of WA, identitywa (formerly Catholic Care), and Mosaic Carer Support Group, were approached for assistance in contacting the primary carers of individuals with intellectual disabilities. Anyone caring for a person with PWS or AS was excluded from the pool of possible participants as these individuals would be approached to participate in the main study. Carers WA, Mosaic Carer Support Group and identitywa each included a request for volunteers in their regular newsletter. To facilitate the recruitment process the Genetic Support Council was given a number of packs, each containing:

- a) An information letter for potential participants (Appendix IX)
- b) A consent form (Appendix IX)
- c) A reply-paid envelope

Packs were sent to representatives of various groups affiliated with the Genetic Support Council, i.e., the Fragile-X Support Group, Turner Syndrome Association, for dissemination to their members. Only persons living in the Perth metropolitan area were to be invited, owing to time and distance constraints which made visits to rural or remote areas impractical. Carers were requested to return the completed consent form in the reply paid envelope if they wished to participate in the Discussion Groups.

Unfortunately, this process produced no responses from any of the three support organisations or from members of the groups contacted via the Genetic Support Council. In response, an alternative approach was adopted which involved personal visits by the candidate to Support Group meetings. The two groups contacted were Riverton Commonlink, an initiative of identitywa, and Gosnells Linking Together, for members of Carers WA. A short talk was delivered on the aims of the project, followed by a request for volunteers.

This initiative resulted in the recruitment of 12 carers representing a range of ages and backgrounds, and including two fathers of people with IDD. The age ranges of their offspring with IDD ranged from 7-56 years. The potential participants were contacted by telephone to arrange suitable meeting times and venues and the draft protocols were forwarded to them for evaluation prior to the event: these included all of the survey instruments and the FSCI. All participants signed an informed consent form before commencement of the group discussions.

The first Discussion Group meeting was held on the 1st August 2006 from 1.00-2.15pm at 12 Station Street, Gosnells, WA 6110. This was attended by three mothers of individuals with IDD. The second Discussion Group, on the 3rd August 2006 between 11.00am and 12.30pm, was at 8 Yeeda Street, Riverton, WA 6148. Two mothers and a father participated in this discussion.

The other six individuals who had volunteered were unable to attend either of these sessions and were held in reserve in the event that it was necessary to convene further groups. However, the completed group discussions provided detailed and thorough feedback, and therefore no further groups were organised. All volunteers were thanked for their participation.

The list of recommended amendments elicited from the Group Discussion participants, and the rationale for each change is presented in Table 4.4.

Table 4.4 Amendments and rationales resulting from the Discussion Groups

Site	Amendment	Rationale
Page 1	Insertion: ‘those items that do not apply may be left blank’	It was felt that single parents may not feel happy about including data on their ex-partners, and that some of the questions, e.g., employment were not applicable to many people with IDD.
Page 1	Additional space was created for the diagnosis	It was mentioned that some individuals have more than one problem diagnosed.
Page 2	Insertion: ‘(base this on an average week)’	Although all participants agreed that a ‘typical week’ is very difficult to define, they felt that this insertion was more indicative of the information sought.
Page 2	Insertion: ‘Note; this information, along with all other research data will NOT be seen by any agency or service provider’	It was pointed out that some disadvantages could arise to carers if it was known that the family made use of respite care, so it was deemed advisable to stress the confidentiality of data.
Page 3	Insertion: ‘Note: this information is included to determine whether having a child or adult with IDD affects the earning capacity of parents’	Several participants felt that these questions could be considered intrusive without some explanation of the underlying purpose.
Page 6	Replacement of ‘...if the person is non-verbal’ with ‘...if the person cannot speak’	The language was simplified for greater clarity.
Page 13	An extra question was added to the interview schedule: ‘Dealing with service organisations or government agencies	Many participants indicated service and government organisations were a major source of stress for themselves and the person for whom they cared. It was therefore deemed appropriate to add this question to the interview schedule.

4.7.4 *Discussion*

Although the participants were aware that the purpose of the Discussion Groups was to examine the suitability and applicability of the study protocols, it was quite difficult to keep the conversations away from the personal experiences and problems of each individual. However, the most significant and relevant additions to the interview schedule arose from these personal stories contributed by the participants.

There were few suggestions to alter the wording or language of the questionnaires or interview items (Appendix VII). Discussion Group members had a good grasp of the sense of questions, even if, as was the case with the syndrome-specific clinical profile sheets, they were not familiar with the specific characteristic in question. It may be that having received the protocols in advance the volunteers had time to contemplate the meaning of each item.

The amended questionnaires were then returned to the ECU HREC for approval (Table 4.1). Once approval was granted, applications were made to other HRECs and the Disability Services Commission as detailed in Chapter 4.4.1.

Chapter 5 Survey results

5.1 Methods

5.1.1 Data collection

Quantitative data pertaining to both the people with AS or PWS and to their family carer, were collected by means of postal questionnaires (Table 5.1: Appendix VI) completed by the primary carer of each person with AS or PWS, or in conjunction with the individual themselves if they were competent to do so. Participants were encouraged to contact the candidate if they had any queries or wished to discuss the questions contained in the surveys.

Table 5.1 Contents of the packages posted to participants and the data collected

Item	Data collected from the item
Consent forms	Informed consent and/or proxy consent
Demographic questionnaire	Year of birth, residence, education, employment
Carer Information questionnaire	Demographic information, carer duties, attitudes, general health
Food-related Problems Questionnaire	Presence or absence of behavioural traits related to food
Clinical presentation sheet: syndrome-specific	Relevant clinical history of the person with AS or PWS
Reply-paid envelope	

As previously stated (4.5.2), a total of 119 information and invitation packs were posted by staff from DSC and GSWA to all identified people with AS and PWS. People who responded to the candidate had their details taken, including the diagnosis of the affected individual, to allow the appropriate material to be posted to them (Table 4.2). All of the carers who received a survey package (16 containing all of the questionnaires, 6 with carer questionnaires only) returned completed questionnaires. In addition, a quantitative measure of perceived stress was obtained from the FSCI at the time of the personal interview.

5.1.2 Data analysis

The quantitative data were analysed primarily by means of descriptive statistics using the statistical software package SPSS for Windows, Release 17.0, IBM, Chicago,

USA, with the findings presented in the form of summary statistics. Student t-tests were used to assess the variance of means for some items; however the distributions were far from normal for a majority of variables when tested by the Shapiro-Wilks Test of Normality. As the number of participants was small, the significance level for all t-tests was set at <0.01 to increase the power, even though that also increased the possibility of Type II error.

As stated previously (4.3), correlations between aA, bB and cC factors of the Double ABCX model and the xX factor were not tested: a minimum of 62 participants is suggested to be necessary for these tests to have sufficient power to provide a significant result (Onwuegbuzie & Collins, 2007).

5.2 *Participants*

Twenty-one families and the manager of a group home, all of whom were caring for an individual with AS (n = 14) or PWS (n = 8), volunteered to participate in the study. Non-respondents could not be checked for residence in WA, death status, or ability to read and speak English, any of which would indicate ineligibility to participate. It was therefore impossible to calculate a meaningful response fraction for this study.

Nine of the people with AS and three people with PWS were under the age of 18 years, therefore a parent for each child provided consent for their participation. Proxy consent was provided by the legal guardians of two of the adults with AS and one with PWS. One adult with PWS provided self-consent to participate. However, the remaining six adults were adjudged incapable of self-consent and as they did not have a legal guardian to provide consent on their behalf they had to be excluded from the study group (Figure 5.1). Sixteen individuals with AS or PWS were finally included in the survey phase: 11 with AS and five with PWS. The six family carer/s of the adults with AS or PWS who were excluded were given the opportunity to complete the sections of the study that did not pertain directly to their offspring, i.e., family demographics, carer health, carer satisfaction, FSCI.

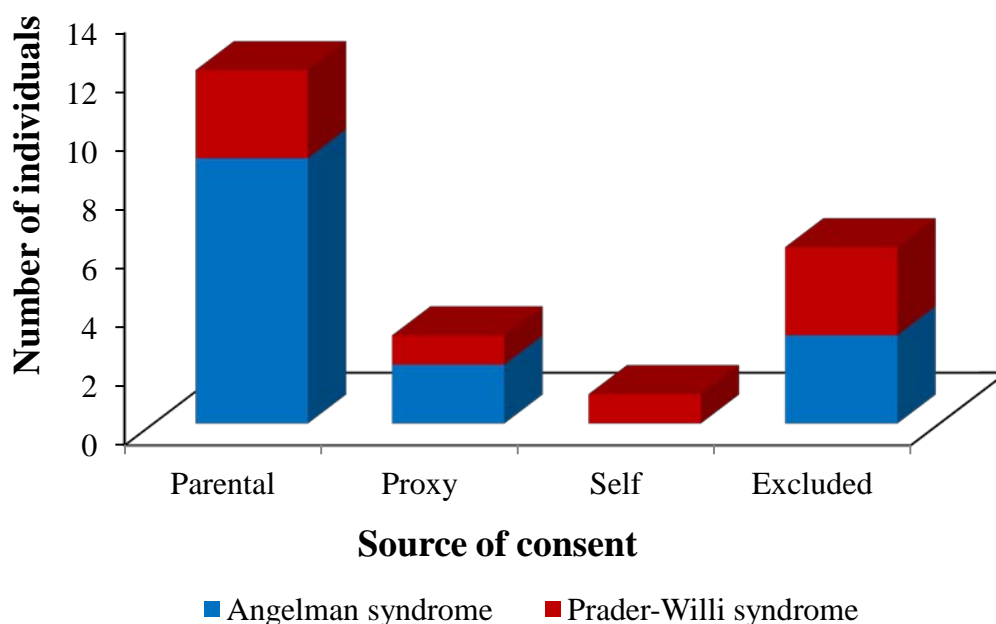


Figure 5.1 Sources of informed or proxy consent allowing data collection regarding the people with AS or PWS

Owing to the consent/proxy consent rules, there were varying numbers of respondents to different sections of the study. The number of informants providing data for each section of the project is listed in Table 5.1. Throughout this chapter the number of respondents will be included where necessary as a denominator to ensure clarity of the relevant sub-groupings.

Table 5.2 Number of people participating in various elements of data collection

	Angelman syndrome	Prader-Willi syndrome	Total
Demographics of cases	11	5	16
Clinical profile	11	5	16
FRPQ	11	5	16
Family demographics	10	7	17
Mothers' age group	12	7	19
Carer Health	11	7	18
Carer Satisfaction	11	7	18
FSCI	13	6	19

5.3 Demographic profiles

5.3.1 Individuals with Angelman or Prader-Willi syndrome

At the census date of 31st December 2008, the mean age of the 11 people with AS was 13.55 years (median 8.00 years; range 4-41 years), and the mean age of the five individuals with PWS was 16.81 years (median 17.10 years; range 9-28 years). See Figure 5.2 for the age group distribution.

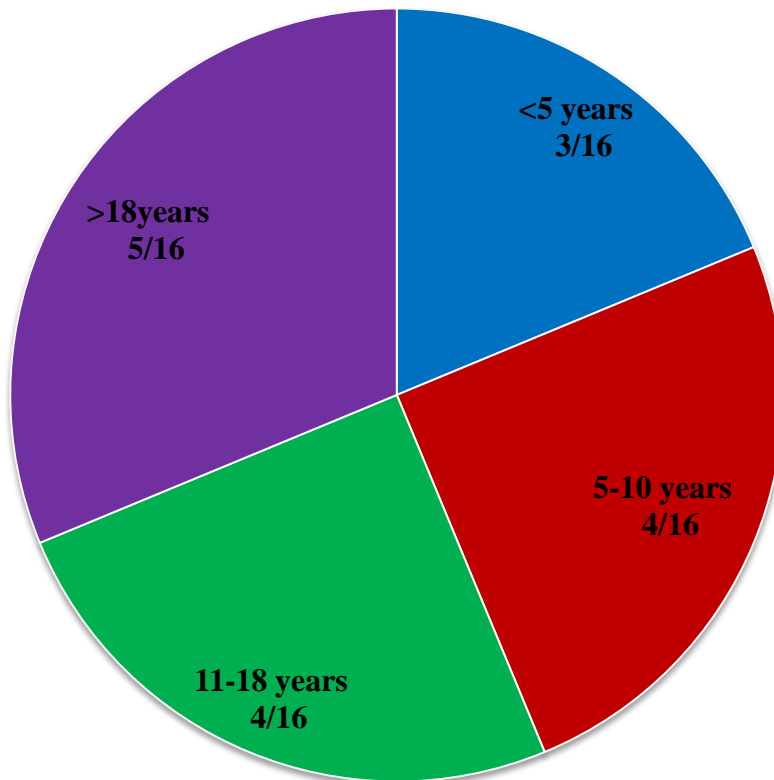


Figure 5.2 Age groups of people with Angelman syndrome (n = 11) or Prader-Willi syndrome (n = 5) at 31st December 2008

As expected, most of the people with AS or PWS were born in WA and lived in a family home, predominantly in the Perth metropolitan area (Table 5.2). Angelman syndrome has not been reported to show a gender bias so it is possible that the high proportion of females with AS was an artefact of the small number of people in the study. Many of the people with AS or PWS had not gone on to secondary schooling, partly due to their young age, and only two had completed secondary schooling at the census date.

Table 5.3 Demographic details of individuals with Angelman syndrome (n = 11) or Prader-Willi syndrome (n = 5)

	Angelman syndrome (number/total)	Prader-Willi syndrome (number/total)
Born in WA	8/11	4/5
Born in Australia, excluding WA	2/11	1/5
Born out of Australia	1/11	0
Female	8/11	2/5
Family home	10/11	5/5
Metropolitan area	10/11	5/5
Secondary education	0	2/5

5.3.2 Family carers

In addition to the 15 family carers of the individuals included in the study, all six of the remaining family carers who had volunteered to participate completed relevant sections of the surveys and/or the FSCI. Two family carers were not available for interview and family demographic data were not supplied by the adoptive parent (see Table 5.1). The mean age of the 19 mothers at 31st December 2008 was 44.5 years (range; 32-58 years: see Figure 5.3 for age group distribution). As the numbers were small, all comparisons throughout the remainder of the study were made between mothers less than 50 years old and those 50 years and older. It is acknowledged that the common cut-off date for 'older' age groups is 55yr (Seltzer, *et al.*, 1995; Chou, *et al.*, 2010; Llewellyn, *et al.*, 2010b); however 'older caregivers' have included individuals less than 50yr (Dillenburger & McKerr, 2011) and other studies have defined 'ageing' parents as 50yr and greater (Minnes & Woodford, 2004).

Two fathers participated in the interview in conjunction with their wives. As there were so few couples interviewed, no comparisons were attempted.

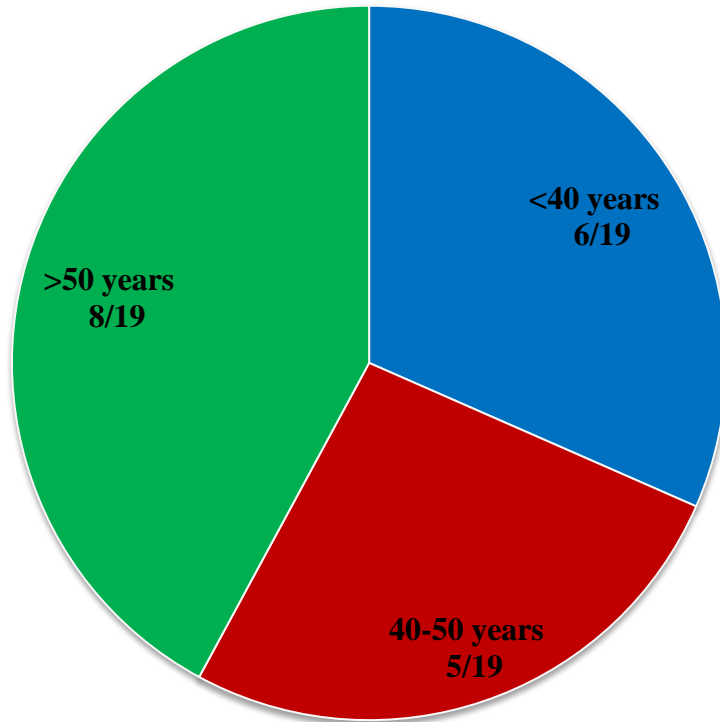


Figure 5.3 Age groups of the mothers of people with Angelman syndrome or Prader-Willi syndrome at 31st December 2008

A majority of families (12/17) comprised a married/de facto couple and one or more children. Two sets of parents were divorced and three people were single or widowed. All but two of the respondents (one foster/adoptive mother, one group home manager) were the natural parents of an individual with AS or PWS.

As can be seen in Table 5.3, there were a higher proportion of parents with a tertiary qualification (59.4%) than was found in the general population of Western Australia (49.8%) in the latter half of the 20th Century (Australian Bureau of Statistics, 2009b). In addition, the percentage of mothers in full-time or part-time employment (76%) was greater than the numbers reported by others in mothers of people with IDD: 64% from Sweden (Olsson & Hwang, 2006), and 67% from Ireland (MacDonald, *et al.*, 2010).

Table 5.4 Educational attainment and current employment of the parents of people with AS or PWS

	Mothers n = 17	Fathers n = 15
Education:		
Tertiary	9	10
Secondary	8	4
Primary	0	1
Employment:		
Full-time	6	12
Part-time/casual	7	1
None	3	0
N/A or not recorded	1	2

5.4 *Stressors (aA factors in the Double ABCX model)*

5.4.1 *Characteristics of people with AS or PWS ('a' factor in the Double ABCX model)*

5.4.1.1 *Care needs*

The collated data indicated that individuals with AS required an average of 52.4 hours of direct care per week while people with PWS required an average of 19.4 hours per week of direct care. The youngest children (<5 years: all with AS) were said to require 70-97 hours care per week*, and the slightly older children (5-10 years) 7.5-52.5 hours. Non-numerical answers to the direct care question included 'constant vigilance' and 'nights/w'end, and if carers don't attend'. On the other hand, some of the older carers did not record any direct care information in the survey, yet referred in the interview to changing nappies, feeding, showering, and dressing their offspring. These discrepancies in the forms of answers provided made it impossible to obtain an unambiguous general picture of the care needs of individuals with the disorders at various life-stages. However, previous research has suggested that the time expended caring for a child with IDD is considerably greater than that required for a typically-developing child (Curran, *et al.*, 2001), and the level of care needed by an older child or adult with IDD is also high (McGrother, *et al.*, 1996).

*Note: '24/7' was coded as 97 hours

5.4.1.2 Clinical signs

Most of the diagnostic clinical features were exhibited by the majority of the people with AS (Figure 5.4). However, very few could walk and many families reported the absence of the typical Angelman facial features, or displayed tongue thrusting behaviour. Among the nine persons for whom data were available, eight were within a healthy weight range and one was overweight for their height.

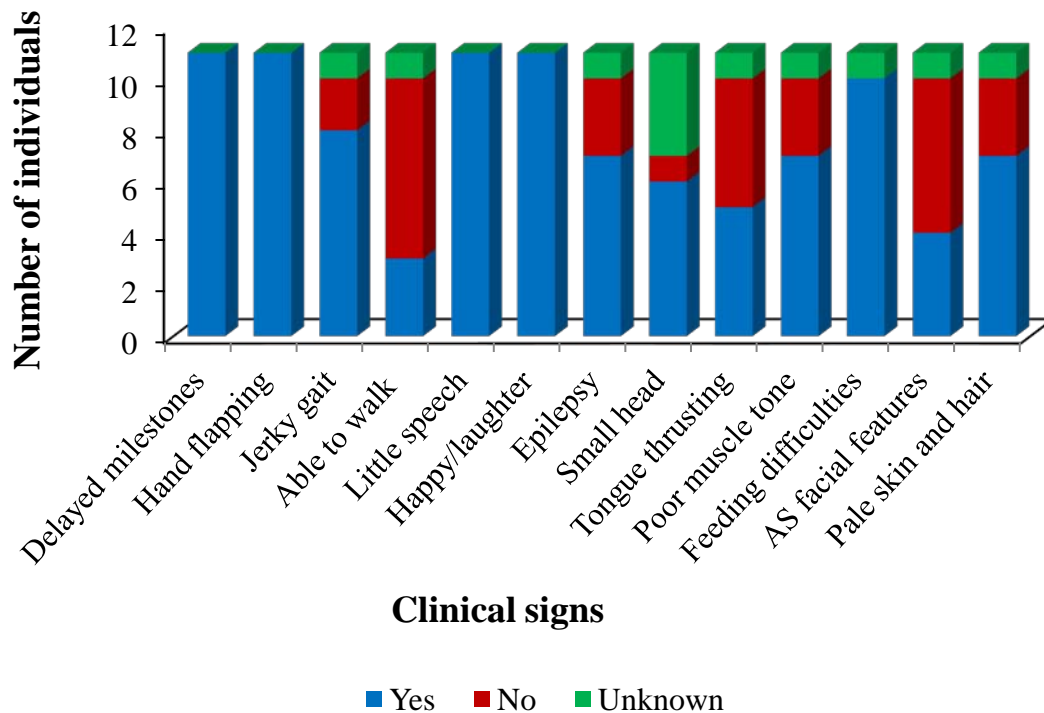


Figure 5.4 Frequency of clinical characteristics of Angelman syndrome (n = 11)

Specific features commonly exhibited by their offspring, such as an inability to walk (8/11), a lack of speech (11/11), and the presence of epilepsy (7/11), are suggested by other researchers as potential stressors for the family carers of individuals with AS (Frey, *et al.*, 1989; van den Borne, *et al.*, 1999).

The major clinical features of PWS were reported to be present in most of the five people within this group (Figure 5.5). One person was recorded as being treated with growth hormone therapy, and was within the normal range of body weight for age and height. The remaining three people with PWS for whom data were available were either overweight or obese.

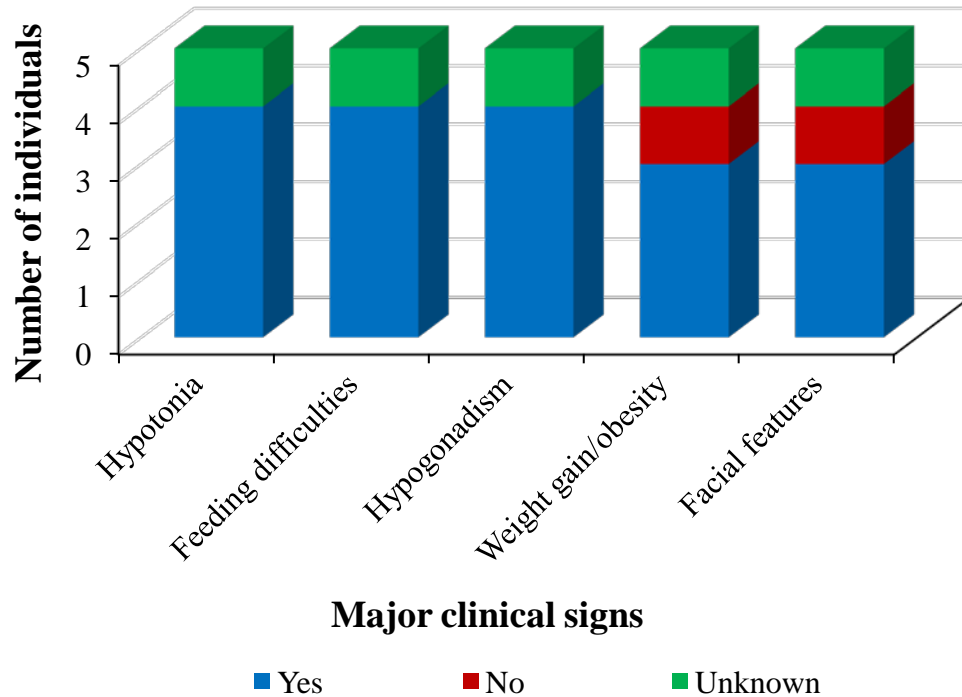


Figure 5.5 Frequency of major clinical signs of Prader-Willi syndrome (n = 5)

Among the minor clinical features of PWS, fat on trunk and thighs, and small hands and feet were reported present in most of the participants. However, only one person was considered to be short-statured compared with other members of the family, and a single individual was reported to engage in skin-picking behaviour (Figure 5.6).

The specific features of PWS that have the potential to act as stressors for family carers are: feeding difficulties and failure to thrive in infancy; weight gain leading to obesity; the typical behavioural problems associated with the syndrome; and sleep disturbances (Cotton & Richdale, 2006; Ho & Dimitropoulos, 2010). All of these traits were reported to occur in three or more of the five people with PWS (Figure 5.5, Figure 5.6).

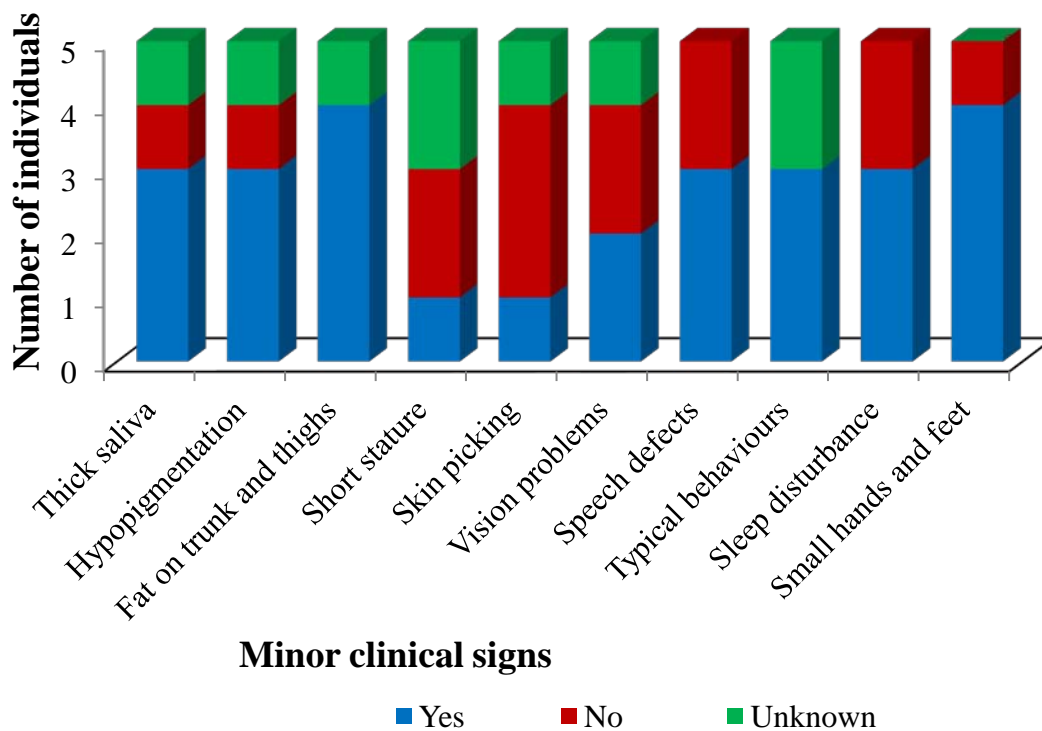


Figure 5.6 Frequency of minor clinical signs of Prader-Willi syndrome (n = 5)

5.4.1.3 Food-Related Problems Questionnaire

The total scores for the 11 people with AS (mean 29.3: range 12-40) were lower on this scale compared to the total for the five people with PWS (mean 36.0: range 20-53). The student t-test was not significant at the 0.01 level ($t = -2.419$, $df (16)$, $p = 0.028$).

However, as can be seen in Figure 5.7, closer examination of the sub-scales reveals that the relationships between the two groups are more complex than suggested by a simple dichotomous comparison.

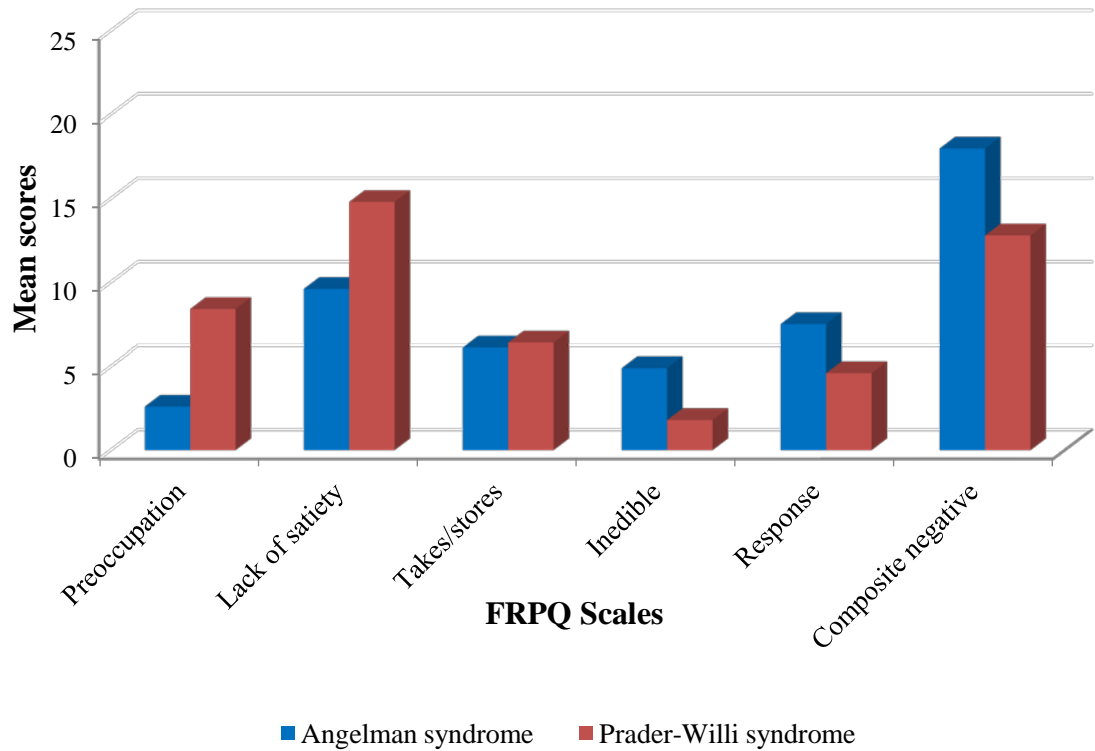


Figure 5.7 Comparison of mean scores between the AS group (n = 11) and the PWS group (n = 5) on sub-scales of the Food-Related Problems Questionnaire

5.4.1.3.1 Preoccupation with food

This scale consists of three questions relating to a preoccupation with food with a maximum possible score of 18. As the scale involves questions that depend largely upon verbal ability, the mean score for the five people with PWS was higher (8.4: 1 N/A) than that of the AS (n = 11) group (2.6: 21 N/A), although it failed to reach significance on the student t-test ($p = 0.0414$) when allowance was made for the non-verbal individuals.

5.4.1.3.2 Impairment of satiety

The five questions in this scale refer to the lack of satiety, ‘feeling full’, often found in people with PWS (maximum possible score = 30). Three of these questions require verbal ability and two do not. The mean score for the PWS group was 14.8 and that of the AS group was 9.6 (Figure 5.7), again non-significant ($p = 0.075$).

5.4.1.3.3 Composite negative behaviour

This scale comprises three sub-scale scores, with a combined maximum possible score of 48. The group of people with AS scored slightly higher on average than the PWS group (18.0 and 12.8 respectively: $p = 319$, n.s.). The biggest difference between the two groups was on the 'Eats inedible items' sub-scale in which the AS mean was more than double that of the people with PWS (Figure 5.7).

a) Takes/stores food

The first of the sub-scales consists of three questions relating to taking and storing food and to the necessity of locking food away (maximum = 18). Although verbal ability is not necessary, some degree of mobility and autonomy is required. Members of the AS group scored slightly less than the PWS group (6.1 and 6.4 respectively: $p = 0.931$, n.s.), possibly reflecting the reduced mobility of younger children with AS.

b) Eats inedible items

These two questions relate to eating non-food items and to eating unsuitable food such as raw or frozen foodstuffs (maximum score = 12). It is possible that the difference in the mean scores (AS = 4.9: PWS = 1.8: $p = 0.120$, n.s.) was due to the lower average age of the AS group, as it is not unknown for younger children to eat inappropriate substances.

c) Responds in inappropriate way

The final three questions involve negative responses to the denial of food, a delay at mealtime and changes to the menu (maximum score = 18). As these are behavioural responses, none of the items require verbal ability. On average the people with AS scored slightly higher than those with PWS (7.5 and 4.6 respectively: $p = 0.160$, n.s.).

5.4.2 *Carer health (A factor in the Double ABCX model)*

The three questions of this scale yield a maximum score of 12. Within the study group the mean carer health score was 7.9. The data shown in Figure 5.9 suggest that most respondents enjoyed good health (16/18) which had not deteriorated over the past three years (15/18) and sometimes restricted their activities (17/18). Many (11/18) considered that their health status was equivalent to or better than that of peers who were not responsible for caring for a child or adult with an inherited disorder.

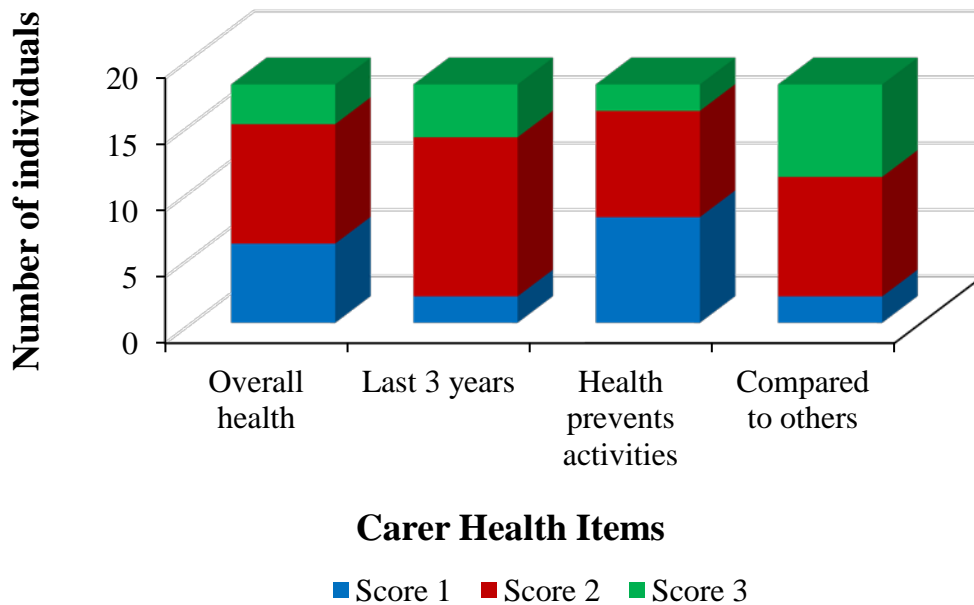


Figure 5.8 Frequency of responses to individual Carer Health Items (n = 18)*

*Note: higher scores reflect poorer self-rated health status.

Although there was a slight trend towards better health ratings for the 11 mothers less than 50yr, the mean carer health scores did not differ significantly across the age groups (Figure 5.10: $t = -1.134$, $df (16)$, $p = 0.274$). The slight worsening of the self-reported health among the seven older carers may be reflective of age-related limitations, rather than indicative of physical strain caused by the caregiving role.

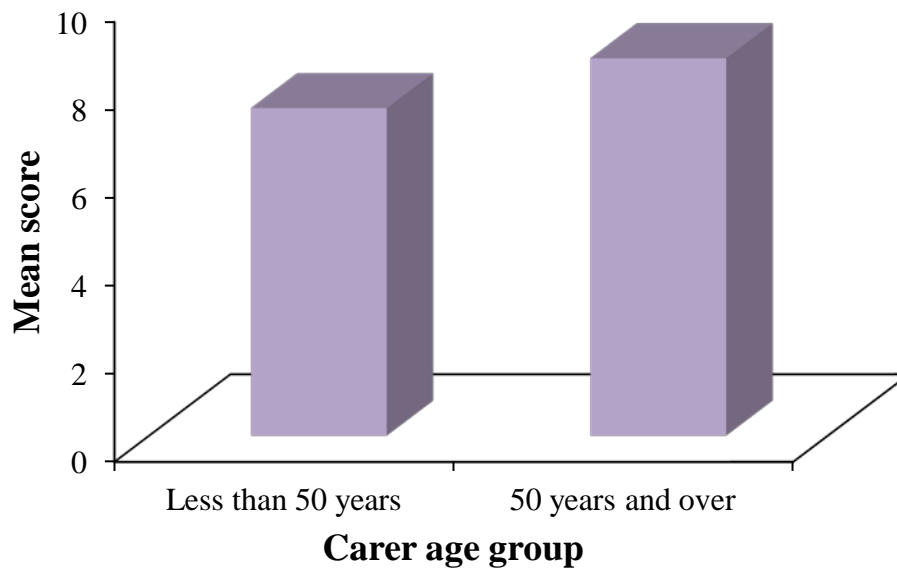


Figure 5.9 Mean Carer Health scores compared by maternal age group (<50yr = 11: 50yr and over = 7)

5.5 *Resources and supports (bB factors of the Double ABCX model)*

5.5.1 *Services and activities utilized*

Among the eight people with AS attending school, work, or a day centre, an average of 17.1 hours per week was spent at that venue, compared to 25.9 hours for four people with PWS. In addition, four people with PWS spent an average of 5.9 hours per week engaged in leisure activities, and the equivalent time for those with AS (n = 5) was 4.6 hours; possibly a reflection of the higher ages of the individuals with PWS. Nine of fifteen families used respite care for periods ranging from one day outing per week plus four nights per fortnight to two nights every six months. Use of respite services for their affected offspring has been described previously as a ‘godsend’ (Caldwell, 2007), and many of the families in the present study were utilising the services available in WA.

5.6 *Perceptions (cC factors of the Double ABCX model)*

5.6.1 *Carer Satisfaction Survey*

The mean satisfaction score across the six questions for the entire group of 18 respondents was 20.6 (maximum possible = 30), indicating that most parents felt

satisfied in their role sometimes or often (Table 5.4). Almost all (17/18) carers reported feeling pleasure when their offspring with AS or PWS felt pleasure, and that they derived enjoyment from caring for their offspring (16/18), similar proportions to those reported to feel positive about caring in a study from the Netherlands (Al-Janabi, *et al.*, 2010). There was less agreement on the item which asked if caring improved the carer's self-esteem: slightly more than half of the respondents (10/18) rated this item as never or occasionally, while a third (6/18) replied often or always (Table 5.4).

Table 5.5 Number of responses to individual items of the Carer Satisfaction Scale (n = 18)

	Never (1 pt)	Occasionally (2 pts)	Sometimes (3 pts)	Often (4 pts)	Always (5 pts)
Gives a feeling of satisfaction	1	2	5	4	6
Enjoy being with child	0	2	2	6	8
Feel closer to child	2	2	3	4	7
Improves self-esteem	6	4	2	5	1
Feel pleasure in child's pleasure	0	1	0	5	12
Adds meaning to life	4	3	4	3	4

5.7 *Outcomes (xX factors of the Double ABCX model)*

5.7.1 *Family Stress and Coping Scale (quantitative)*

5.7.1.1 *Total scores*

The scores on the Family Stress and Coping Interview were highly variable and ranged from a low of 1 to a high of 64, with a mean of 38.2. Mothers caring for a person with AS tended to report lower stress levels (mean 32.6 range 1-53) than those caring for a person with PWS (mean 50.3: range 31-64). The student t-test was not significant at the 0.01 level ($t = -2.528$, $df (17)$, $p = 0.022$). It was not clear if this tendency was related to the mean age difference between the individuals with AS and those with PWS, or to real differences in the stress involved in the caring role.

There was a trend for stress ratings to be greater for the mothers caring for an adult with AS or PWS when compared with the mothers of the other children's age groups (Figure 5.10). The very small number of individuals in each child age group

meant that no statistical test could be conducted to test this trend. Mothers who were 50yr and greater (n = 7) scored higher overall stress ratings than mothers <50yr (n = 11) (t = -3.302, df (16), p = 0.005). Among the older mothers and the mothers caring for an adult, groups which largely overlap, transitions and future planning, including guardianship issues, are hypothesised to be matters of importance both now and in the near future.

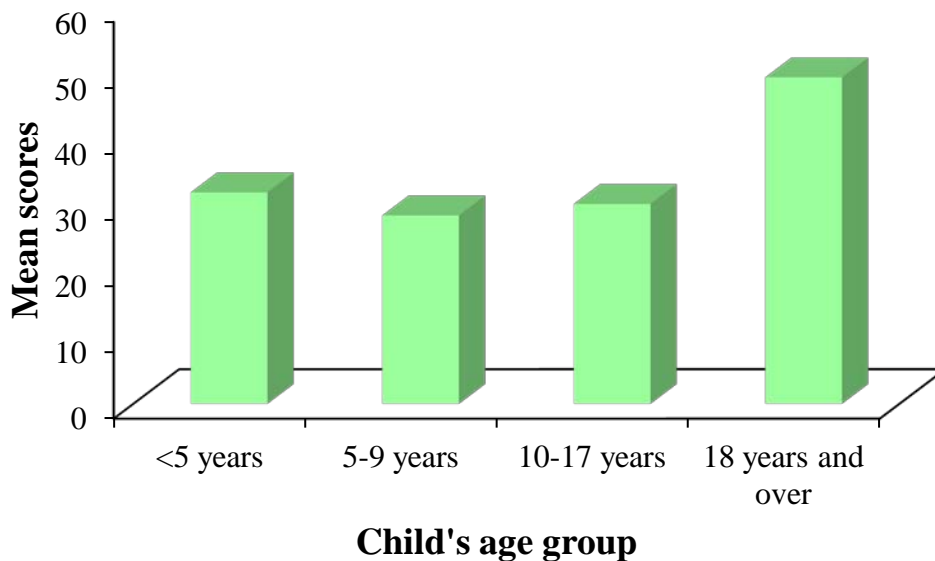


Figure 5.10 Mean scores on the Family Stress and Coping Interview compared by child age group (<5yr = 3, 5-10yr = 4, 10-18yr = 4, 18yr⁺ = 5)

5.7.1.2 Individual item scores

A mean exceeding two was calculated for 12 items of the FSCI, as shown in Figure 5.11. With the exception of the initial diagnosis, the carers of people with AS reported lower mean stress levels for these items compared to the carers of people with PWS. For many of the mothers of a child with AS, the relatively short time since diagnosis may explain their higher stress levels with respect to this item.

There was a substantial differential between the mothers of the AS and PWS groups regarding the issue of work placements or employment for their offspring: the individuals with PWS were considerably older and had higher levels of intellectual function than members of the AS group, and therefore were more likely to be looking for supported or sheltered employment (Figure 5.11).

Dealing with teachers and the education system was another area in which the mothers of people with PWS experienced greater stress than the carers of people with AS (Figure 5.11). Due to differences in cognitive functioning, individuals with PWS tended to be integrated into the general education system, albeit with education support, whereas those with AS mostly attended special schools and centres that could cater more specifically for the child's cognitive and verbal limitations, and behavioural problems.

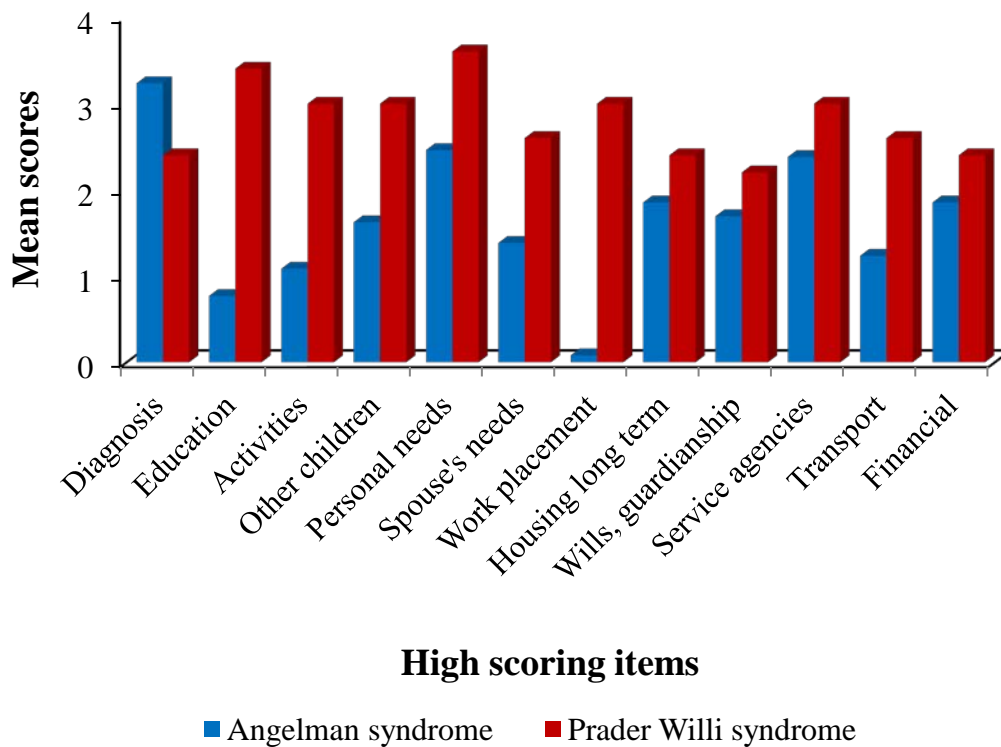


Figure 5.11 Comparison between the mothers caring for a person with Angelman syndrome (n = 11) and mothers caring for a person with Prader-Willi syndrome (n = 8) for the high scoring FSCI items

The remaining 12 items from the FSCI garnered mean scores less than two (Figure 5.12). Within this sub-set, the mothers of people with AS reported deciding on the best level of integration for their child, and day-to-day assistance with care of their child to be considerably more stressful than did the carers of individuals with PWS. By comparison, dealing with their child's sexuality was more stressful for the mothers of people with PWS (Figure 5.12). These results appeared to principally reflect the different life stages currently experienced by the participants.

The least stressful situation for members of both groups was their feelings about the cause of their child’s condition (Figure 5.12). Most of the mothers (12/17) were fully aware of the genetic causality of these disorders, and were untroubled by any feelings of guilt or distress.

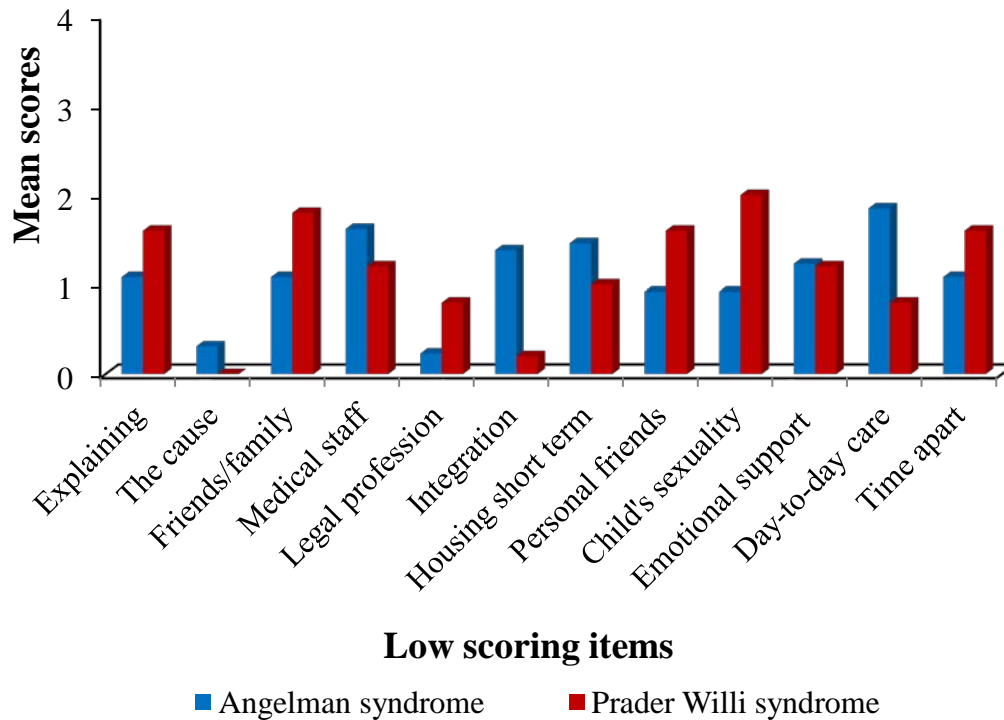


Figure 5.12 Comparison between the mothers caring for a person with Angelman syndrome and mothers caring for a person with Prader-Willi syndrome for the low scoring FSCI items

Very little mean stress was reported by the study group in dealing with legal professionals. However, most carers (15/19) had not had this situation arise in relation to their offspring with AS or PWS and therefore rated the stress as 0.

When the ratings were compared by mother’s age group, 11 items produced a mean score of two or greater. In each of these 11 situations the eight mothers over 50 years old reported higher stress levels than their younger counterparts (n = 11). The greatest discrepancies between the two age groups were shown in the areas of meeting their spouse’s needs, maintaining personal friendships, and wills, trusts and guardianship issues (Figure 5.13), all of which the mothers less than 50 years old found either less difficult or irrelevant.

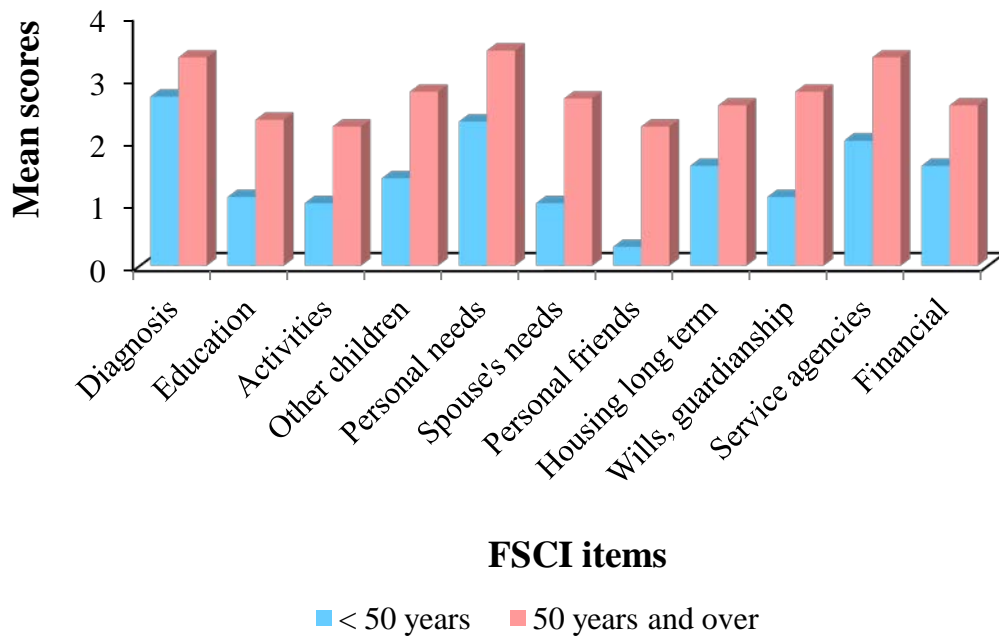


Figure 5.13 Comparison between mothers <50yr (n = 11) and >50yr (n = 8) on high scoring FSCI items

Within the 13 lower scoring items, the mothers less than 50yr reported slightly higher stress levels in their feelings about the cause of the condition, dealing with the medical profession, and in deciding on levels of integration (Figure 5.14). These issues are likely to be more relevant among those with younger offspring. However, mothers under the age of 50yr reported less stress than their older counterparts attached to dealing with friends, family and neighbours, work placement and employment for their offspring, and emotional and social support for their offspring with AS or PWS, all issues more likely to arise as children make the transition into adulthood.

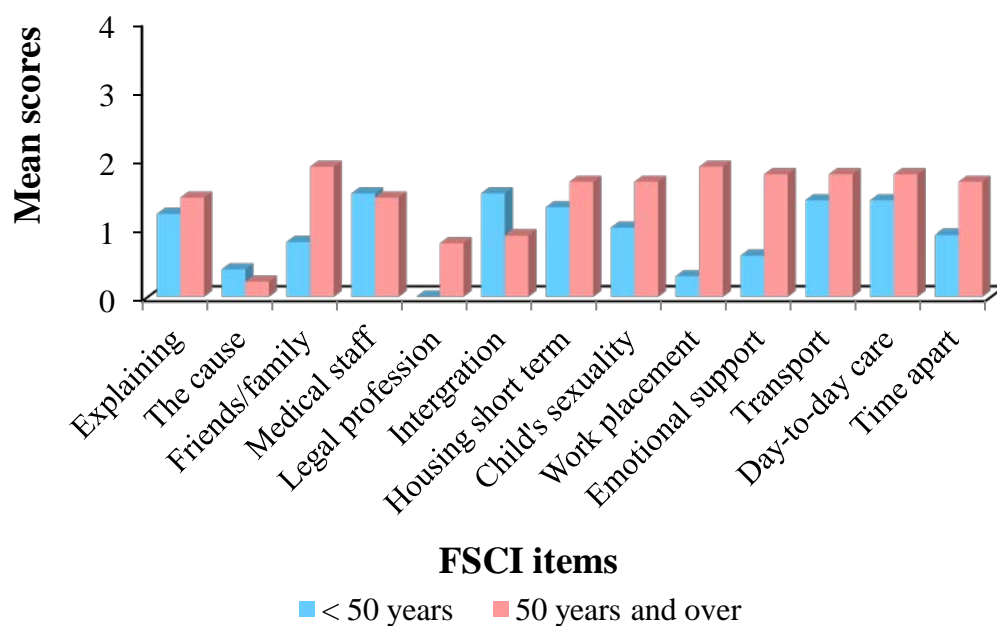


Figure 5.14 Comparison between mothers <50yr (n = 11) and >50yr (n = 5) on low scoring FSCI items

5.8 *Support group membership*

Thirteen of the 19 families involved in the interview were members of either the Angelman Syndrome Association of WA (11/13), or the Prader-Willi Syndrome Association (WA) (2/6). Another individual was a member of Carers Australia, an umbrella organisation supporting family carers of all ages. Seven of the 13 family carers who were members of the Angelman Syndrome Association of WA or the Prader-Willi Syndrome Association (WA) felt that there were benefits in being a member of such a group; four said they had not received any advantage; and the remaining two persons anticipated benefit in the future.

Five of the six carers who were not members of a support group were mothers over 50 years old with adult offspring with AS/PWS. Two of these carers had no previous knowledge of the groups, but two others had been involved with them in the past and felt that they no longer fitted in.

5.9 *Chapter Summary*

The individuals cared for by the participants in this study each showed many of the typical signs of their particular disorder. They required substantial amounts of care from both family carers and formal services, and many were in receipt of respite care.

The family carers who participated were generally healthy and reasonably content with their role. However, the majority reported considerable amounts of stress associated with caring. In the next chapter, attention is given to the specific events that cause stress, and the coping mechanisms engaged by the participants in an effort to control or reduce that stress.

Chapter 6 Family Stress and Coping Interviews

6.1 *Brief orientation*

The key concepts of stress and coping theory that underpin the study were discussed in detail within Chapter 3. The assumptions are that various aspects of the caring role will act as stressors (aA), that the effects of these stressors will be modified to varying degrees by a variety of resources, supports (bB), perceptions and attitudes (cC), and by the employment of various coping mechanisms (bB). Further, that family carers will exhibit adaptation to stress (xX) along a continuum from ‘maladaptation’ to ‘bonadaptation’ (Kramer, 1993). Identification of these factors can be achieved by using quantitative survey methods (see Chapter 5) however much more information can be obtained by utilisation of qualitative data collection and analysis. This can lead to a fuller description of the various components of the Double ABCX model and be used as a basis for assessing interactions between the factors.

The qualitative section of the research design was based on the directed content analysis of text from the Family Stress and Coping Interview (Appendix VIII) with a view to obtaining an understanding of the stress factors acting on the family carers of the people with PWS and AS (Research question 2) and the of coping methods used to combat these stressors (Research question 5). The chapter includes the methods, a description of the study participants, the analytic approach used, and a question-by-question report of the findings.

6.2 *Methods*

This sub-section of the study comprised the Family Stress and Coping interview with some additional questions (Appendix VIII). All participants chose to be interviewed in their own home, at a time suited to them. Prior to commencement of each interview the participants were reminded that they could withdraw at any time and they were asked for verbal confirmation of their consent to participate. Each of the interview situations was read out and the participants were asked to rate the stress related to that situation on a scale of 0 (no stress) to 4 (extreme stress). They were then asked to elaborate on their reply, with the occasional interpolation of a probe question, such as ‘What successes have you had in dealing with this issue?’ (see Appendix VIII). Participants were asked at the conclusion to talk about any issues that had not been

covered in the interview. Additional notes were made during and after each interview concerning the conduct of the interview and the personal impressions of the interviewer based on the interview content.

Each interview was tape-recorded and transcribed verbatim: three by the candidate and 16 by a professional typist employed for this specific task. All identifying names or places were removed at this time so that all transcripts were only identified by the unique code number. Verbatim transcription assists with the maintenance of both authenticity (does this represent what was actually said?) and confirmability (can other researchers see or hear the same material?), two concepts that have been associated with validity in qualitative data analysis (Dellinger & Leech, 2007; Koro-Ljungberg, 2010). As the texts were de-identified at this point, there was no attempt to get participants to review the interview transcripts or the analysis.

Although the concept of data saturation, the point where no new information arises from subsequent interviews, is considered to be an important check on data completeness, it is more often connected with the grounded theory of data analysis (Bowen, 2008). It depends mainly on a pattern of theoretical sampling and the constant comparison method of data analysis, rather than the convenience sampling and directed content analysis used for the current study.

6.3 *Participants*

All eligible carers consented to an interview and the interviews were conducted by the candidate at a time and venue suited to each individual. Two families were not interviewed owing to difficulties arranging a suitable time within the data collection phase of the study, so that a total of 21 family carers (19 mothers only and two mother/father dyads) were interviewed, all of them in their own home. No further participants for the interviews were sought, partly owing to the confidentiality conditions which prohibited the candidate from making direct contact with potential participants and requesting their consent.

Most of the families interviewed (13/19) contained an individual with AS, and generally the people with AS or PWS lived in the family home (14/16), similarly to the proportions participating in the survey section of the study. More than half of the mothers interviewed (10/18) were aged <50 yr, but there were four more mothers 50yr or greater who took part in the interview when compared to the survey section of the

study. All four of these additional participants cared for an adult with AS or PWS. The educational profile and employment status of the interview participants was similar to that of the survey participants.

6.4 Data analysis

Analysis of the interview transcripts was conducted as a directed content analysis (CA). The aim of directed CA is to expand or refine extant theory (Hsieh & Shannon, 2005).

“Existing theory or research can help focus the research question. It can provide predictions about the variables of interest or about the relationships among variables, thus helping to determine the initial coding scheme or relationships between codes” (Hsieh & Shannon, 2005)

The Double ABCX model of stress and coping theory was used for the present study, providing both an overarching coding plan and specific coding variables that were tentatively formulated prior to data analysis. The decision to employ a deductive rather than inductive analytical approach (Neuman, 2006) was in some measure forced upon the candidate by the circumstance of presenting a comprehensive literature review within the study research proposal. Some authorities advise that deduction is an acceptable approach to content analysis (Potter & Levine-Donnerstein, 1999; Flick, 2009); in fact these authors suggest that the development of a coding scheme can be faced with more confidence, especially by novice investigators, when there is a theoretical basis for the codes.

Close correspondence between theory and the operationalization of the coding scheme has been viewed as a test for the validity of qualitative data (Potter & Levine-Donnerstein, 1999; Hsieh & Shannon, 2005). Potter and Levine-Donnerstein regard the development of a quality coding scheme as one of the keys to face, predictive and construct validity. Face validity relies on clearly defined, logical categories and can be assessed by judging the coherence of the operationalization of the concepts as supplied by theory. The same authors consider that theory helps to indicate relationships between concepts which can then be tested to determine predictive validity (Potter & Levine-Donnerstein, 1999). Construct validation can also be guided by theory to test for convergence and discrimination, i.e., is the standard related to certain concepts (convergence) and not related to others (discrimination) (Potter & Levine-Donnerstein, 1999).

The literature on stress and coping was examined for common themes and concepts that could be used as the initial coding categories. A number of concepts drawn from the Coping Orientations to Problems Experienced Scale (Carver, *et al.*, 1989) and from the Family Crisis-Orientated Coping Scale (Woodford, 1998) were defined and placed within either the problem-focused or emotion-focused top order categories in order to aid identification of the common coping strategies used by participants (Research question 5).

The text from each interview was read through once to verify the accuracy of the transcription, and then at least once more to allow thorough immersion in the text. Then each transcript was read slowly, phrase by phrase and line by line. Sections of text that appeared to be related to a specific type of coping were assigned to one or more of the previously-defined sub-categories. In the event of a seemingly important piece of text failing to correspond to any of the codes, it was coded initially as a miscellaneous item. Subsequent discussion between research colleagues helped to identify core concepts behind the strategies and confirmed the appropriate placement of codes within the coding matrix. Ultimately, one new coding item (Advocacy) was added to the original list within the Perceptions and Attitudes higher order category: it appears in Table 6.1b.

Separate sub-coded items were identified that corresponded to specific types of problems expressed by the carers (Research question 2). These emerged from the interview data and are defined in Table 6.2. These items were helpful in indicating the most common sources of stress in greater detail, regardless of the actual situation or event that was being considered at that point of the interview. Stressors from this table that were implied within an actual question, e.g., Planning in Question 18 'Planning for wills, trusts and guardianships', were not coded within that item.

After several of the interview transcripts had been coded by the candidate, another colleague was enlisted to independently check the material for consistency and reliability of coding. Two of the interview transcripts were analysed separately by another researcher with a background in disability research and qualitative analyses. Differences in the use of the coding model were discussed, and consensus agreements were made. Minor changes were made to the coding sub-categories and the remaining pre-analysed transcripts were revised to adhere to the same coding categories before all of the other transcripts were coded.

Whilst undergoing coding of the remaining documents, some further difficulties were encountered, suggesting the codes needed to be modified slightly. Following consultation with an academic support worker at Edith Cowan University the existing coding scheme was rearranged, and the major and minor categories redefined. These rearrangements are summarised in Table 6.1a, 6.1b. The decision was made to divide the coping strategies into ‘Perceptions and attitudes’ and ‘Resources and supports’, as it was considered that these two categories were more applicable to the Double ABCX model upon which the research was based than were the Problem-focus and Emotion-focus categories more commonly used in stress and coping research. As mentioned previously (Chapter 3.1.4.3), there are many variations in the definitions and the placement of coping strategies within the problem/emotion dichotomy, and it appeared that the allocation of the coping strategies into either perceptions or resources presented a clearer and more useful interpretation of the data. Not only did this scheme better represent the Double ABCX model on which the study was based, but it allowed greater flexibility in placing specific codes within the major categories which would otherwise have seemed ambiguous if fitted into the problem/emotion coping plan. For example, Emotional support has been placed in both the Emotion-focused coping category (Carver, et al., 1989), and in the Problem-focused coping category (Schnider, *et al.*, 2007; Glass, *et al.*, 2009). For the present study, Emotional support formed part of the Resources and supports section.

Table 6.1a Definitions of coping strategies associated with Resources and Supports (bB Factor in the Double ABCX model))

Concept (number of references)	Definition	Examples
Effort (116)	Concentrate time and effort; put aside personal and family needs; do what has to be done	‘We just carry on, going and going...’ ‘...we couldn’t do it as a family because it was too stressful with [offspring with PWS]’
Emotional (54)	Gain emotional support, talk about feelings of distress; gain sympathy and understanding	‘...we felt I think we needed to let out to others as well...’ ‘...bombarded with accolades or sympathy or whatever it is’
Instrumental (105)	Tangible support from family, friends, paid staff or professionals; mobilization of agency or professional help	‘My Dad’s here, he helps me...’ ‘...since all the therapy services are in place, she goes to this group and that group’
Knowledge (82)	Find out more; talk to others in a similar situation to obtain information or knowledge; seek advice from family, friends or professionals	‘...passing on strategies, information, knowledge, whatever we can, share with each other...’ ‘...my Local Co-ordinator is...giving me some information...’
Planning (82)	Work on a strategy; make a plan of action; think about what needs to be done; wait for the right time to act	‘...we’ve got to choose which one’s more important ...’ ‘...preparing yourself for what’s going to happen’
Practical (84)	Practical solutions; physical actions; day-to-day activities; routine and structure	‘...we have to put her on the toilet for quite some time before bed...’ ‘Had...a giant-sized cot that fitted around her bed. I had that made’

Table 6.1b Definitions of coping strategies associated with Perceptions and Attitudes (cC Factor in the Double ABCX model)

Concept (number of references)	Definition	Examples
Advocacy (65)	Work for policy, legislative or community attitude change; stand up for the rights of the child; outreach	‘I like...explaining to people what he has so that more word gets out about the Syndrome’ ‘We have put a lot of effort in writing and campaigning...’
Detachment (42)	Give up trying; admit inability to deal with the situation; take mind off the situation by use of distractions	‘...just go out and leave it, forget about it for a little while and enjoy myself...’ ‘I go to work, ‘cause I have to work for my sanity’
Positive outlook (58)	Look for the good in a situation; learn from a situation; personal growth; make light of the situation	‘...she brings huge joys as well...’ ‘...it’s given me great life lessons in choosing who I want in my life...’
Reinterpretation (133)	Adjust goals; redefine the problem; acknowledge things cannot be changed; accept the situation	‘We decided against integration for everybody’s sake really’ ‘I don’t think you ever fully accept it but you get used to it, don’t you?’
Spiritual (4)	Gain comfort or strength from spiritual or religious belief	‘When I start to focus on the Lord and what my daughter’s needs are, then I cope and I can stay above it all’

Table 6.2 Definitions of generalized stressors independent of the interview items (A Factor in the Double ABCX model)

Concept (number of references)	Definition	Examples
Anger (66)	Expressions of anger; frustration	'...you're very tired and you're angry and it's our kid' '...you do get frustrated with asking, being asked the same questions...'
Funding (51)	Issues of funding availability; financial difficulties	'...especially the way funding is, it's quite hard...to get the funding for it' '...we keep putting in for the funding...'
Label (32)	Feeling stigmatized; labelling	'They say labels don't matter, but they do; you feel like you've got a direction' 'I feel labelled largely...'
Paperwork (20)	Dealing with bureaucracy, forms, and paperwork	'...it's all just a waste of paperwork and time and money for everyone...' '...a duplicate addressed to me, in a separate envelope with separate postage, what a waste...'
Time (57)	Time pressures; not having enough time; taking too much time	'...just not enough hours in the day...' '...an hour and a half morning and an hour and a half afternoon...'

6.5 *Results by question*

6.5.1 *Method of reporting*

Data from the Family Stress and Coping Interview are herein presented as responses to each of the 24 items commencing with the most stressful and progressing to the least stressful. Additional questions relating to membership of a support group are also included. In the tables that precede the commentary, responses to the 24 items (in the order in which they appear in the interview) have been classified in three different ways: according to the five identified generalised stressors (Table 6.3a & b); the six identified coping strategies associated with resources and supports (Table 6.4); and the five identified coping strategies associated with perceptions and attitudes (Table 6.5). Direct illustrative quotes from the respondents have been included throughout to preclude misinterpretation. Relevance to the results of the surveys and the implications of such links will be discussed in depth in Chapter 7.

In reporting the findings the number of participant families giving a particular score or coding item is stated as a fraction, e.g., 14/19. A number in curved brackets after a coding category represents the number of references within the relevant text for that item, e.g., Knowledge (18). This information is also contained in Tables 6.3a & b, 6.4 and 6.5). The information in curved brackets after each direct quotation is the stress score allocated by the respondent for that item followed by the coding category/ies, e.g., (3: Planning).

Throughout this section a stress rating score of 0 (not stressful) or 1 (a little stressful) is referred to as 'not stressful', and a score of 2 (somewhat stressful) or more (considerably, extremely stressful) as 'stressful'. Items that were not applicable in any instance are scored as zero (0). While logic may dictate that each category should retain its classification, in practical terms, and in light of the highly non-normal distribution of the responses, this dichotomy seemed more appropriate.

Several formatting conventions have been followed when presenting direct quotations from the interviews. The format '...' (three dots) is used when one or more words have been omitted from the respondent's quotation, and '....' (four dots) is used to indicate an exclusion between sentences. Square brackets [] are used to

signify that either the transcriber was uncertain of the word or words used by the respondent, or had replaced a specific name, e.g., a hospital, school or individual, with a generic term to ensure the anonymity of participants. Confidentiality was protected by the use of ‘child’, ‘other child’ etc.

Table 6.3a References to Stressors and stress reactions (aA factor) from family carers (n = 19) by individual interview item

Item description	Anger	Funding	Label	Paperwork	Time
Initial diagnosis	6		14		2
Explaining to others	2		10		
Feelings about cause	2				
Dealing with friends/family	2		1		1
Dealing with health professionals	12		2		3
Dealing with legal professionals	1			3	1
Dealing with teachers/education	5	1		2	5
Finding opportunities for friends/activities			1	1	1
Deciding on integration level			1		3
Accommodation: immediate		13		2	1
Meeting needs of other children					4
Meeting personal needs	1	2		1	14
Meeting needs of spouse					6
Maintaining personal friendships	1				5

Table 6.3b References to Stressors and stress reactions (aA factor) from family carers (n = 19) by individual interview item

Item description	Anger	Funding	Label	Paperwork	Time
Dealing with offspring's sexuality	2				
Employment for offspring	5	5		3	
Accommodation: future		6			
Planning for wills, guardianship		3		2	
Dealing with service or government agencies	21	7	3	17	3
Planning emotional and social support	1	1			1
Transportation		5		1	3
Day-to-day assistance with care		2			3
Time apart from offspring	1	2			6
Financial and insurance		17		3	
Any other comments	3	3	3	1	2

Table 6.4 References to Resources and supports (bB factor) from family carers (n = 19) by individual interview item

Item description	Effort	Emotional	Instrumental	Knowledge	Planning	Practical
Initial diagnosis	5	8	8	21	5	2
Explaining to others	1	1	1	6		5
Feelings about cause	2			6		1
Dealing with friends/family	6	10	4	2	3	4
Dealing with health professionals	7		7	13	4	5
Dealing with legal professionals				1	1	1
Dealing with teachers/education	7	1	7	2	4	10
Finding opportunities for friends/activities	7	3	6	2	4	4
Deciding on integration level	5		3		5	
Accommodation: immediate	3	2	1	3	10	3
Meeting needs of other children	8	1	4		4	4
Meeting personal needs	19	2	6		4	2
Meeting needs of spouse	6	2	6		4	1
Maintaining personal friendships	7	18	2	1		
Dealing with offspring's sexuality	4	1	4	3	2	3
Employment for offspring	4	1	4	1	8	2
Accommodation: future	1		2	1	12	2
Planning for wills, guardianship	4		4	3	13	4
Dealing with service or government agencies	4	1	9	4	1	7
Planning emotional and social support	1	1	1	2	6	2
Transportation	2		3	2	6	4
Day-to-day assistance with care	3	1	7		4	5
Time apart from offspring	4		15		8	5
Financial and insurance	6				5	4
Any other comments	8	23		28	3	10

Table 6.5 References to Perceptions and attitudes (cC factor) from family carers (n = 19) by individual interview item

Item description	Advocacy	Detachment	Positive outlook	Reinterpretation	Spiritual
Initial diagnosis		3	7	21	1
Explaining to others	7		4	16	
Feelings about cause	1	1	1	7	
Dealing with friends/family			5	7	
Dealing with health professionals	9	1	4	5	
Dealing with legal professionals	1				
Dealing with teachers/education	7		5	8	
Finding opportunities for friends/activities		2	6	15	
Deciding on integration level					
Accommodation: immediate	4	3	3	9	
Meeting needs of other children			1	1	1
Meeting personal needs		4	1	3	
Meeting needs of spouse		5	1		1
Maintaining personal friendships	1	1	3	4	
Dealing with offspring's sexuality	3			1	
Employment for offspring	3	3	2	3	
Accommodation: future	4	2	1	3	
Planning for wills, guardianship	6	4			
Dealing with service or government agencies	15	5	1	6	
Planning emotional and social support		2	2	1	
Transportation			2	1	
Day-to-day assistance with care	1	2	2	2	
Time apart from offspring	2	4		4	
Financial and insurance			5		
Any other comments	11	2	3	6	5

6.5.2 The initial diagnosis of your child as having Angelman/Prader-Willi syndrome

6.5.2.1 Introduction

This was the single most stressful situation covered by the interview schedule (mean = 3.00: maximum possible score = 4.00). Seven of the eight mothers 50yr and greater, and six of the ten mothers <50yr found this item considerably or extremely stressful. Reinterpretation (24) and Knowledge (22) were the coping strategies expressed most frequently by the family carers in this situation.

6.5.2.2 Generalised stressors

The main stress categories to appear in this item were Label (14) and Anger (8); however some of the references to labelling were (partially) positive while others expressed a more pejorative view of the term.

‘So the actually ‘having a label’ was good, um, but the actual label was fairly devastating’ (4: Label)

‘...you feel labelled, you feel like you live in another world.’ (4: Label)

Anger was typically expressed as dissatisfaction with the process of disclosure, perceived lack of support, or the general lack of knowledge available at the time of diagnosis. Interestingly, these references came in equal parts from family carers of all ages, and therefore were unlikely to reflect changes in procedure across specific eras.

‘...when we got the diagnosis, the paediatrician was really off-hand and she gave us no back-up support systems to speak to.’ (4: Anger)

‘...the information that was out there was outdated and any documentation of cases of Prader-Willi were of severe cases and so all of the documentation we read was extremely negative.’ (4: Anger)

6.5.2.3 Stressful

Most carers (15/19) found this situation stressful. It was apparent that feelings of distress caused by the diagnosis persisted in many carers, often for years. Respondents who reported improved perceptions of the diagnosis, although it did not reduced their stress score, attributed this to a number of factors including time, acceptance, and knowledge.

‘...I guess...over time it just becomes easier...You become more accepting of it, you get used to the idea. We’re talking to other people as well, in the same situations.’ (4: Reinterpretation, Knowledge)

‘...I guess once we got some knowledge, that became...a bit of a weapon against the stress...’ (4: Knowledge)

Some carers recalled feelings of disappointment or changed expectations encountered during the diagnostic process, while a few people viewed the diagnosis as a relief from either uncertainty or a ‘worse’ possible scenario.

‘...just the realisation that what you perceive as normal is not or no longer will apply, and that your dreams and hopes for your child’s future...have all been turned upside down...’ (4: Reinterpretation).

‘...initially we needed to know what the outcomes are going to be with regards to [child’s] diagnosis and it was just such a relief to be able to know the knowledge and the background of it, and that I think settles you a bit more.’ (4: Knowledge)

There was one reference to grief within this item and the same respondent elaborated on this theme at the end of the interview. This concept was, however, not categorised as there were insufficient carers who discussed the process.

‘...I think like everybody you go through a grief process and whilst you deal day-to-day...’ (4: Effort)

‘...grief is a normal process and it is extremely stressful...it puts pressure on your relationship because you both grieve differently and at different times sometimes, and that can be difficult....grief affects everybody differently. Some people grieve for life. Some people grieve and accept and move on quicker than the other person....in our case it’s a pretty much of an ongoing grief because...you feel labelled every day you walk into the school yourself...’

A few carers referred within this item to various positive aspects of caring for a child with AS or PWS, even within the context of high stress levels. This was a reflection of the moderately high scores given by respondents to the Carer Satisfaction Scale (Chapter 5.5.1), and supportive of data from Wales (Hastings, *et al.*, 2002; Hastings, *et al.*, 2005a; Kenny & McGilloway, 2007) indicating that reframing or reinterpretation may be associated with positive perceptions of the child with IDD.

‘That’s something that’s so important to convey, especially to the new mums, is the possibility of the joy and the good things that can come into it...’ (4: Positive outlook)

6.5.2.4 *Not stressful*

The family carers who rated this item as not stressful (4/19) comprised a foster parent and three carers under the age of 50. They mentioned similar coping strategies to the people who had rated the stress as high.

'It was a bit of a shock but basically I just got used to it, it's all you could do really, there's not much else to do....I had family around as well, so that helped. Yeah, family was the main...' (0: Reinterpretation; Emotional support)

'...a panel of experts...said...that he was going to do basically nothing at all. That was devastating. And then we got a phone call...saying that [child] had Angelman syndrome and we were over the moon....It was definitely a relief.' (0)

'I use all my kids as a learning process so that's how I cope....I looked into it, I even got a book on it and tried to work out what it was and the doctors at [hospital]...helped me to understand what it was and how to try to counteract it.' (0: Positive outlook; Knowledge; Instrumental)

6.5.3 *Meeting your own personal needs*

6.5.3.1 *Introduction*

The mean score for this item was 2.84. Carers 50yr or greater (7/8) in the current study were as likely as the carers <50yr (8/10) to score this item as stressful. This was higher than the results of Minnes and Woodford (2004) who reported 55% of older carers rated this item as stressful. It has been suggested that the stress experienced by the older group corresponds to a lack of 'normality', in that they are still responsible for the care of their child with IDD at an age when their own contemporaries are getting time to themselves (Gill & Renwick, 2007).

Effort (19), in the form of putting aside one's needs to concentrate on the child's needs, was the most common coping strategy. A dominant theme within this item was the issue of babysitters or respite care. Similarly to reports from Canada and the UK (Gill & Renwick, 2007; Wodehouse & McGill, 2009) most carers experienced considerable difficulty accessing respite as, and when, needed.

'So if you had a special event coming up, to get a sitter for it....And then we had a sitter coming in. That was stressful in itself as well.' (4: Instrumental)

6.5.3.2 *Generalised stressors*

Time (14) was the general stressor most often mentioned in regard to this item. A few family carers also talked about tiredness or exhaustion resulting from disrupted and insufficient sleep.

'That's right up there because there is no time. You're just down at the bottom when you can fit yourself in.' (4: Time, Effort)

'I didn't have time, didn't have the energy. I was so tired. I mean, you know what Angels are like – they don't sleep.' (4: Time)

6.5.3.3 Stressful

This situation was scored as stressful by 16/19 respondents. The coding concept of Effort was most often expressed here as putting aside personal and family needs in order to concentrate on the needs of the child with AS or PWS.

'...our life has basically revolved around [child], [husband] and I's, so everything's just been put on hold and that's it.' (4: Effort)

'...I didn't have any personal needs until lately because...my whole life was centred around the children.' (4: Effort)

However, there were individuals who spoke positively about this aspect of their lives, despite rating the situation as highly stressful.

'I'm pretty lucky in that I've got [husband], we will bounce off each other and we can work in together.' (4: Positive outlook, Instrumental, Emotional)

Although some carers seemed to find time to meet their own needs, there was still a measure of stress associated with finding and organising suitable care for their child.

'...[husband] and I go out together and do separate things, sometimes together, 4 hours a week....[however] it falls to me to organise it...' (2: Practical)

'...we're looking at respite, occasional respite. We've put our name down for that at [respite centre].' (2: Instrumental, Planning)

'...I book in massages for myself, I go for walks...we have the [sitter service] and at the moment we've increased the hours and we're getting the maximum.' (2: Planning, Instrumental)

6.5.3.4 Not stressful

For a minority (3/19) of carers there was little or no stress associated with this situation. Within these families, members of the extended family, friends, or paid staff would care for the individual with AS or PWS to allow the carer some personal time.

'Mum or my sister-in-law will come into play, look after [child] for us, or if it happens to coincide with one of his respite weekends.' (1: Instrumental)

One carer seemed to feel that meeting her own personal needs did not necessitate time apart from her child.

‘...I’m lucky – I’m probably the perfect person to have her – I’ve never been a social butterfly....if I had the choice of going out at night or staying home reading my book, I would stay home and read a book. I like reading, I like the garden, I like going to the beach...’ (0: Positive outlook, Reinterpretation)

6.5.4 Dealing with service organisations/government agencies

6.5.4.1 Introduction

This situation was rated as stressful (mean = 2.63) by both younger (7/10) and older (7/8) carers. The coping strategy used most often was Instrumental (8) even though it was coded within this item only if the source of support was other than a service organisation or government agency.

Several of the family carers expressed attitudes and described actions related to the concept of Advocacy (14). Some of these references related to educating professionals, while others referred to lending support to others in a similar situation.

*‘I used to teach the foster parents, I also used to teach the LAC’s....I get involved with the organisations and I become part of them so that’s my way of coping because you see things from the inside’ (2: Advocacy)**

**Note: LAC = Local Area Co-ordinator, a DSC staff member.*

‘...we felt so strongly about it that we started up our own body of care for people...’ (4: Advocacy)

6.5.4.2 Generalised stressors

The generalised stressor predominating within this item was Anger (21). A number of people spoke of the needless duplication of letters, forms and assessments ‘...some of them are so bogged down in paperwork,’ (4), and the resultant cost ‘...what a waste of money, sending out all this paperwork’ (3).

Another major issue was the lack of a specific category for permanent intellectual disability in many official files. This topic was a source of frustration for a number of carers.

‘...every election he gets a letter asking him why he hasn’t enrolled, and every time I ring them up and say ‘because he’s disabled’ and they say, ‘oh, thank you for letting us know.’ And then the next election he’ll get another letter – because they have no facility to mark that on the roll, so merely the fact that he’s turned 18 and he hasn’t enrolled, we keep getting letters after every...local...state ...federal government election.’ (4: Anger)

‘...every 6 months sometimes, up to 3 years, you’ll get a letter from some department saying about our son, and you say, ‘Well, he’s permanently disabled.’ ‘We don’t have that section. We’ll just mark it off and we’ll contact

you when it comes up again.’ And that’s frustrating. And it’s all just a waste of paperwork and time and money for everyone.’ (4: Anger, Time)

‘...the very strict criteria that you have to fit into a bracket for funding....aide time at the school demands that a box is ticked....they ask for IQ tests and I don’t want him to have an IQ test...it creates all these issues just to get a box ticked....the way the services are structured, to have to fit into a box to get funding, is extremely stressful and it labels you...I don’t get funding from DSC because I refuse for him to have an IQ test....it’s extremely stressful. I’m trying to go the inclusive way and here’s an organisation that’s making you feel labelled.’ (4: Label, Funding, Anger, Advocacy)

A perceived lack of continuity in staffing and inconsistency of service provision was also apparent in a number of interviews. This was similar to a report from the UK (Wodehouse & McGill, 2009).

‘...they’re just inconsistent, the changeover in staff, you’re having to re-explain everything continuously....it just makes you so angry because you’re having these people in your home, they’re coming in for three times and then they’re off’ (4: Anger)

‘...there’s one agency that just [unclear], our daughter has had 10 different people...’ (3)

‘...the high turnover of staff. There’s always somebody new, somebody leaving. You just get to know someone and feel like you’re really progressing with the therapy, and then they leave...there seems to be a lapse between the next one starting...’ (3)

6.5.4.3 Stressful

More than half of the respondents (15/19) rated this item as stressful. There was a great deal of frustration and anger expressed by these carers. Advocacy and taking practical steps to improve the situation were among the coping strategies utilised.

‘...when I went over to Canada I looked at the Prader-Willi Association over there and their group homes.’ (4: Advocacy, Planning)

‘...we looked after several intellectually disabled people ...over the period of time...a lot of people came through our care....And our daughter [with AS] benefited greatly from that...we had people who were right alongside to support us and help us.’ (4: Instrumental, Emotional, Advocacy)

‘I’ve printed off all this stuff, highlighted everything, and then you get new staff in and it doesn’t get passed on...’ (4: Practical, Planning)

‘But you do start getting resentful towards them. You do start thinking, ‘um, I’m not going to do too much in case you are going to leave’...so you’re putting yourself on guard...’ (4: Reinterpretation, Anger)

The carers also mentioned a number of other coping strategies, including gaining knowledge: ‘...[I] got involved with [agency] and made sure I knew what was going on.’ (2: Knowledge, Advocacy); and maintaining a positive outlook.

‘Other people squawk and moan and I say nasty things about DSC but, they have done well. We have been very fortunate. Again, it’s the way perhaps I’ve gone about it.’ (2: Positive outlook)

6.5.4.4 *Not stressful*

The four individuals who rated this item as minimally stressful all used strategies from the Perceptions and attitudes category: three said they felt fortunate to receive the services they did: ‘I think we’ve been very lucky in our relationship with all agencies.’ (0: Positive outlook), and two carers spoke of adjusting their expectations to match the availability of services:

‘I have built a relationship with the occupational therapist and physiotherapist....I don’t expect them to come over all the time, because they’ve got a huge workload....I take what they can give me.’ (0: Reinterpretation, Positive outlook)

‘...it sort of happened without me insisting. Because she’s so high functioning that I don’t...feel the need for her to access extra therapy groups outside school.’ (0: Detachment)

6.5.5 *Meeting the needs of your (other) children*

6.5.5.1 *Introduction*

This was another stressful situation (mean = 2.05) for both older (6/8) and younger (5/10) carers. In three of the 19 families their offspring with AS or PWS had been an only child and therefore this item did not apply. The coping strategy most often described was concentrating one’s efforts (8).

6.5.5.2 *Generalised stressors*

Time pressures and constraints (4) were the major stressors within this item.

‘Just lack of energy [mother upset] and lack of time.’ (4: Time)

‘I really realise that I’m not giving enough to [older daughter] but it’s hard to get time with her alone.’ (3: Time, Effort)

6.5.5.3 *Stressful*

Many of the 12 carers in this category had difficulty in giving time and attention to their other children, and experienced feelings of guilt at putting the needs of the other children aside to care for their sibling.

‘Not being able to sort of take her where she wanted to go...or even when she had things on at school, that I couldn’t get anybody to look after him...’ (4: Effort)

‘...the [other] children missed out, not a lot, they didn’t miss out a lot...’ (3)

‘Just sort of balancing between doing extra therapy for him [child with AS] and running around with the other ones.’ (2: Effort, Planning)

Even having supports in place to allow time with the other children did not necessarily mean that the associated stress was eliminated, and Planning was an important strategy:

‘...[staff] would come in extra between 4 and 6 so that I have time with the children coming home from school....I’ve always had that support....I couldn’t have had any of the kids if God hadn’t supported me (4: Planning, Instrumental, Spiritual)

‘...we just have to bear in mind what toys [other child] can play with and what friends she can have over, if they might respond negatively. If [other child] has a play-date, and [child with AS] comes in and bashes all the toys up or shouts....It just limits what ways [other child] can play, what toys she can have....if we borrow books from the library for [other child], we have to hang them on a hook above arms reach for her sister...you’re constantly being a bit of a prison warder.’ (2: Planning, Practical)

6.5.5.4 *Not stressful*

The seven carers who rated this item as not stressful included the three single-offspring families for whom it did not apply. There was no dominant coping strategy evident in these particular responses, with a range of approaches described.

‘They’d come in from school, wash their hands, change their clothes and they’d take over and I wouldn’t have anything to do with their sister anymore.’ (0: Instrumental)

‘I always make a point of doing things with [other child] in that time...’ (0: Planning)

‘We’re lucky that we’ve actually got quite placid children, my older boy who’s very placid, and [baby] who’s very, very good.’ (0: Positive outlook)

6.5.6 Long-term accommodation planning for your child

6.5.6.1 Introduction

The mean score for this item was 2.05. Within the current study, a greater proportion of the carers 50yr and greater (5/8) found this stressful compared with the carers <50yr (4/10). This is not as high a proportion as the 83% of carers 50yo and greater reported in Minnes and Woodford (2004) who found this situation stressful. Reinterpretation (6) was the dominant coping strategy.

6.5.6.2 General stressors

Funding (10) issues were the greatest concern expressed within this item. Many people accepted that there would be delays in the availability of funding, and that their child with AS or PWS was unlikely to be able to earn a living wage.

'I've put money aside...they take most of the money off them in the homes, I did find that out when I was secretary, so I want to make sure she's got enough money to do a holiday if she can.' (4: Planning, Funding)

'...so it's all...financially very stressful....he's never going to have a good income' (3: Funding)

6.5.6.3 Stressful

Many of these ten carers expressed a wish to keep their child with AS or PWS in the family home for as long as possible, because, as one mother put it '...no-one can look after him like I can.' (3: Advocacy). People also recognised the general insufficiency of supported accommodation, and the difficulty in gaining access to funding.

'...we want her at home for as long as possible. We're looking for something very unusual...We want to choose who the staff are, because she has so many needs and we're very particular about how her needs are met...' (4: Practical, Effort)

'Because, like all parents of people with disability, we know that there is 'x' amount of housing and 'x' plus thousands the number of people who need it. And so we've, so far adopted the 'head in the sand' approach and not looked into it at all.' (4: Detachment, Funding)

6.5.6.4 Not stressful

Two families with a young child with AS had not considered this issue at all. Among the other seven carers who found the item less stressful, some families had

discussed possibilities for the future with similar families, whilst another carer credited inside knowledge of the system with helping her to manage more effectively.

'We've...talked about it with another couple who have a child with Angelman syndrome...a year younger than [child], and we'd like to eventually put them into shared care, shared accommodation.' (0: Instrumental)

'...it's crisis-driven and you fill in extensive paperwork...you could wait several years before you get something....I knew how to play the game. So when she was 10, I put in the first funding submission for accommodation support...' (0: Funding, Paperwork, Practical)

6.5.7 Dealing with financial and insurance issues

6.5.7.1 Introduction

There was a moderate amount of stress associated with financial matters (mean = 2.05) with older carers (6/8) more likely than younger carers (5/10) to rate this item as stressful. Effort (6), Positive outlook and Planning (5 each) were the most common coping strategies.

6.5.7.2 General stressors

Family carers spoke of issues including accessing the funds, and the actual amount of financial support.

'...the Centrelink forms and stuff like that...' (3: Paperwork)

'...just the therapies and the medications and the nappies and the extra stuff that he has, does not outweigh what you do get from the government...' (4)

6.5.7.3 Stressful

Almost half of the respondents (9/19) rated the stress for this item as considerable or extreme, and a further two carers rated it as somewhat stressful. The costs of supplying necessary items and facilities for their child were a matter of concern for a number of families. Several carers referred to the expense of the nappies needed for incontinent adults with AS (i.e., at 2008/9 prices, \$A95 for two weeks supply).

'Yeah, even nappies, the day-to-day living expenses...and trying to live off one income...you can't really go back to work. And then it's all the other costs....you feel bad because you're putting pressure on your husband...' (4)

'And...[day centre], even though he's funded for 3 days, I still pay a dollar an hour...that's \$80 or something a month....And realistically, his pension doesn't cover anywhere near the outlay for him....he's on five types of medication...' (3)

Some family carers oversaw financial matters on behalf of their offspring, even though not all of them were legal guardians.

'I asked for his monthly accounts...it took me a while...but they send me now monthly accounts and I check off everything that is being spent. Anything that looks a bit strange I will...ask the accountant what it is.' (3: Advocacy)

Other carers expended effort and planning in an attempt to establish stability in their financial situation:

'...we put in an application form which we thought can help us get some stability with carers....it's been a long time coming....We just carry on, going and going, and make our changes as we go along...' (2: Paperwork, Planning, Effort, Reinterpretation)

6.5.7.4 *Not stressful*

Seven of the 19 carers did not find financial matters particularly stressful. This may be yet another reflection of the relatively high education and employment levels of the study participants. They spoke generally of feeling fortunate and of utilising sound money management practices.

'Financially we're pretty well set, so we're fortunate. I'm sure it could be a lot more stressful' (1: Positive attitude)

'I run a tight budget and I do all my own budgeting. We were doing very well...' (0: Practical)

Another carer had a relaxed attitude:

'If I pay a bill late, I pay a bill late, that's basically it. Not a stress.' (0: Reinterpretation)

The family carers of one high functioning child were looking toward a future when their child would be living more independently.

'...we need to teach him about money and so we have to put extra effort into....we need to concentrate a bit more on teaching him about finances because we need him to be independent and knowledgeable about that.' (1: Effort, Planning)

6.5.8 *Planning for wills, trusts and guardianships*

6.5.8.1 *Introduction*

This was another slightly stressful situation (mean = 1.89) with all eight of the older carers and 4/10 younger carers finding it stressful. The rate for the older carers was much higher the 50% reported by Minnes and Woodford (2004).

A number of different coping strategies were evident: Advocacy (8), Effort, Practical, Detachment and Instrumental (4 each). Stress involved not only the processes of applying for guardianship, but also being sure that the right person would be available to take that responsibility:

'...[name]...[applied for guardianship] and she said it was the most stressful and invasive process that she had ever experienced...' (3: Knowledge)

'We had looked into...our daughter being able to buy her own house so we had to get an order for that, and that was much more involved and tedious than what we thought....There was a lot of red tape...' (2: Advocacy, Paperwork)

'...just deciding who you think would do the best job and cope with our child and who's willing.' (2)

6.5.8.2 *General stressors*

Funding issues (3) and Paperwork (2) were the major stressors. There was also some concern voiced regarding the adequacy of the Guardianship and Administration Act 1990 (WA).

'...you're getting the guardianship law saying you don't need it [guardianship] until there's a conflict, and I don't want to wait until there's a conflict...' (4).

6.5.8.3 *Stressful*

Some of the 13 carers who rated this item as stressful found the guardianship and trust procedures to be incomprehensible. However, all carers expressed the desire to be included in the decision-making process when it came to their child.

'...just explain it to people in layman's terms and have them in somewhere...preferably in their own home rather than having to go into an office...' (4: Anger)

'We thought it was something that we didn't have to worry about. And now we find out we do....you find out more from other parents....And most of them are quite open in discussing what they've gone through and the shortcuts that can help you out, which road not to go down.' (2: Knowledge)

'I handle all her finances....it gets very, very complicated with family trusts....just going onto the website...was confusing. I had to go back to our solicitor and say, 'I don't know whether I have to apply for administration or whether I have to apply for guardianship.' And she said, 'Look, I'll make some enquiries and I'll let you know what you have to do'.' (3: Advocacy, Instrumental)

'We did it [made a will] a long time ago...just recently...I had contact with a gentleman that's put a submission into some board over in Canberra in relation to adjusting the [rules?] for trust funds for people with disabilities....it's just something that you feel compelled to support and have input into to

enable...trust funds to be set up that aren't taxed as heavily...' (2: Advocacy, Effort)

One carer felt it necessary to apply for guardianship so that the staff in her offspring's group home would have to include her in the decision-making process.

'...you're getting the guardianship law saying you don't need it until there's a conflict, and I don't want to wait until there's a conflict because it could be too late. And because they have such adverse drug reactions....I don't want them to take him to a doctor and put him on medication because that's what has caused his respiratory arrest...' (4: Advocacy, Effort)

For other family carers the fact that they hadn't prepared a will was stressful.

'I haven't done anything about it; it stresses me thinking that I do have to do something.' (3: Detachment)

'No, no, we haven't, go on, put huge stress! Am I the only person you've interviewed that hasn't got a will?' (4)

6.5.8.4 *Not stressful*

Three carers said that they had not considered these issues at all, and all six who gave this a no-stress rating were <50yr. Two families in this group had completed a will.

'I did a will last year...I know I need to review it because when I did it, we didn't have the permanent accommodation.' (0)

'We've done the wills and the guardianship in case my husband and I are both killed....we did that before we knew of her diagnosis. Once we did know her diagnosis we did go back and say 'Are you still OK?'....But later on it will [be stressful] when we have to look at power of attorney and that sort of thing.' (0: Practical)

6.5.9 *Meeting the needs of your spouse or partner*

6.5.9.1 *Introduction*

Carers found this to be a mildly stressful item (mean = 1.79). More of the older carers (2/8) rated this situation highly stressful than did their younger counterparts (3/10). Instrumental and Effort (6 each) were the most frequently expressed coping strategies.

'I have always taken [husband] and myself away and got people in to look after the kids....I've tried to sort it so that all the kids go at the same time and then I have time with my husband....that's very important time with your husband.' (4: Planning, Instrumental)

'I don't think his needs ever factored into my reckoning at all. He had to get on and look after himself; I didn't have the energy to look after him as well.' (3: Effort)

6.5.9.2 *General stressors*

As with meeting their own personal needs (Section 6.2.2), family carers reported that Time (6) was the single biggest difficulty.

'...it would be nice to have time for 'us' together as well. That is a huge thing.' (2: Time).

6.5.9.3 *Stressful*

Slightly more than half of respondents (10/19) rated this item as stressful. A variety of strategies were described, often by the same individual. Most people seemed to recognise the importance of both parents feeling supported in the caring role.

'...we try to make sure that we each get what we need...individually as well as together.' (2: Planning, Effort)

'...we bounce off each other pretty well...We quite often have separate weekends away. We've got a holiday house...we're able to just take off any time we want to....He might go away with a group of boys...I can do the same thing, just to get some time away....you come back with your batteries recharged.' (2: Detachment, Positive outlook)

'Just determination, I think....my husband understood that we couldn't, I couldn't look after him totally on my own. And I realised that my husband couldn't look after him totally on his own, so it was really our son keeping us together...his care needs...' (4: Reinterpretation, Effort)

'...we've been to psychologists, both of us...we're still having counselling...' (4: Instrumental)

'We have a routine with the children....we love each other unconditionally....My husband tends to do a lot of things around the house so he keeps himself busy....We have a very strong spiritual belief as well...' (3: Practical, Emotional, Detachment, Spiritual)

6.5.9.4 *Not stressful*

Four family carers did not find this question applicable. A common theme voiced by the remaining five respondents was tiredness and lack of time, even though the issue was only 'a little' stressful.

'...we'll get a sitter and go out....if we're talking about intimacy, I guess you're too tired for it, so depends what you call his needs and my needs. I'd rather he played golf than had sex, because that doesn't involve me having to do anything (laugh).' (1: Instrumental, Positive outlook)

‘...I keep going, and the kids are in bed and that’s the only chance you get to do extra things...for myself, and by the time you sit down to watch telly together...you end up falling asleep on the lounge. Crash out. So it’s probably not enough hours in the day.’ (1: Time, Effort)

6.5.10 Dealing with your child’s teachers and the education system

6.5.10.1 Introduction

This was a slightly stressful situation (mean = 1.68), with higher scores being given by both older (5/8) and younger (4/10) carers. No child attended a mainstream school without either having an educational aide or being part of a Special Education Unit. Practical coping (10) was the most common strategy, followed by Reinterpretation (8). Almost every carer made some positive remark about one or more of their child’s teachers.

‘I love his teachers...because we sent him to special needs schools, they’re all there because they want to be there, they enjoy working with children who are disabled....I build quite good relationships with them.’ (0: Positive outlook)

‘...they’re caring...they show a lot of love to those kids, which is so good.’ (3: Positive outlook)

6.5.10.2 General stressors

Anger and Time (5 each) were the most common stressors. Much of the anger and frustration related to difficulties encountered when trying to get their child integrated into the system.

‘...inclusion is something that is just not being done to fit the legislative act in Western Australia...it’s a huge issue, huge, huge issue.’ (4: Anger)

Time pressures related to the amount of preparation necessary before school, or by the time required to get to a distant location.

‘It was stressful getting her ready because she had to...go on special transport, she had a bus pick her up, and we had to be ready, and that was stressful.’ (0: Time)

6.5.10.3 Stressful

Most of these carers were keen advocates for inclusion, including in education. Among the problems discussed were the options available given the child’s capabilities:

‘...she couldn’t go...into the actually learning class so I had to bring her full-time back to the handicapped school, and I just don’t believe in handicapped schools.’ (4: Reinterpretation)

‘...with the conductive education, they’re fantastic but they do have certain expectations and they just don’t all apply...it’s just not always what’s relevant for mine is relevant for another, and sometimes I think they really do make them fit that mould to a certain extent...’ (2: Reinterpretation)

Carers also discussed some practical issues related to rural living, and the processes required to acquire funding for teaching aide time.

‘I used to travel...from [country town]...all the way to the [town] host family, pick him up on Friday and have to drive all the way back to the farm for the weekend, so he could be with his family. And then take him all the way back again on Sunday.’ (4: Practical)

‘You [had] some teachers that were so caring and wonderful, that knew your child and different behaviours, and how to handle them, then you’ve got others phone you, ‘such and such is happening’ and ‘what do I do now?’, these are professionals here, so that to me was quite stressful, having to be by the phone all day when he was at school.’ (2: Effort)

‘...dealing with the bureaucrats in the education department. Just trying to have her go through...in the inclusion model...you had to get all sorts of letters from doctors....we had to...tell them all the dreadful things about her...how incontinent she was and how incompetent she was so that we could get the minimum aide time...just to help her function within the environment.’ (3: Paperwork, Instrumental)

6.5.10.4 Not stressful

Of the nine carers who rated this item as low stress, eight had placed their child into Special Education facilities. The other child was attending the Special Education Centre at a mainstream school with aide support.

‘I think I was prepared to approach people if I was unhappy...if you’ve got the right attitude, you can sort things through...’ (0: Effort)

‘I’m very cut and dried and practical, I had no huge aspirations. I didn’t feel I needed to be hounding them to be doing an inappropriate...program that was never going to work.’ (0: Reinterpretation)

However, there were some difficulties attributable to the limited availability of suitable schools in WA at this time.

‘...his teachers are great...they run a programme called School for Parents, and I go there one morning a week, and I stay with him. They’re teaching us how to teach him, and that’s fantastic [but] it’s the only one in WA...it takes an hour to get there...in the peak traffic. (0: Practical, Positive outlook, Time)

6.5.11 Creating/finding opportunities for your child to make friends and participate in activities

6.5.11.1 Introduction

This was a mildly stressful situation (mean = 1.58). Almost two-thirds of carers aged 50yo and greater rated this as stressful, slightly higher than the 56% reported in a Canadian study (Minnes & Woodford, 2004). The most prominent coping strategies were Effort (7) followed by Reinterpretation, Positive, and Instrumental (6 each). Interestingly, four carers spoke of birthday party invitations in relation to this item: two 'stressful' and two 'not stressful'. This may relate to the reported difficulty many children with IDD have in making friends either at school or in community settings (Gray, 2002).

6.5.11.2 General stressors

There were no dominant stressors mentioned by respondents.

'Even though you're ringing somebody who's doing a disabled program, just getting on the phone and saying, 'I've got this kid with a disability'...that's stressful. (2: Label)

6.5.11.3 Stressful

Eight of 19 carers rated this item as stressful. The level of integration and inclusion did not appear to influence the carer stress rating.

'...it is stressful because you can't...send her anywhere by herself so you always have to be there. I've tried going dancing...when she was younger. I did horse-riding with her. It is all hands-on and it is stressful.' (4: Practical, Effort)

'He has no friends at school. He's not had any invites [carer upset]. In the street he has friends who play with him....He swims every Saturday but that's just one-on-one...he has Cubs every week...but he's not making friends there.' (4: Instrumental)

Some carers worked within the limitations of their child with regard to the type of activities they promoted:

'I tried so hard to get her into things. Not into generic programs, that was just not going to be. I tried very hard to get her into a lot of programs.' (2: Effort, Reinterpretation)

'...we have to limit it to what works for her....I don't try too hard to make her new friends, she's got her friends at school...we do family things on the weekend...' (2: Reinterpretation)

6.5.11.4 *Not stressful*

More than half of all respondents (11/19) felt this item was not stressful. A majority of these (8/11) were <50yr. Carers appeared to feel under less pressure to organise activities and friends for young children, especially in the years before school age. Both formal and informal groups were mentioned as forming the basis of a child's network.

'...now that she's in...more structured groups...since all the therapy services are in place, she goes to this group and that group. But in the beginning it was 'what do we do with her?', but I've got plenty going on now.' (1: Instrumental)

'...really lucky...we started at playgroup when he was about eight weeks so they've grown up [together]...I don't think I would like to join a [new] group...I would feel too odd. But because...they've seen him grow, and they've all been so fantastic, it's been easy.' (0: Positive outlook, Emotional)

Some carers felt that the extent of their child's limitations made it very unlikely that they would be able to make friends and participate in activities.

'It just doesn't happen. It's pie in the sky. People who think it's going to happen are obviously not dealing with the situation...[service provider] making themselves feel good...' (0: Reinterpretation)

6.5.12 Transportation

6.5.12.1 *Introduction*

Transportation was a slightly stressful item (mean = 1.58), with nine carers rating it as stressful (5/8 older and 4/10 younger carers). Planning (6) and Practical (4) were the two most common coping strategies.

6.5.12.2 *General stressors*

Funding (5) and Time (3) were the major sources of stress.

'Just the logistics of getting her to school, her sister to day-care, and me to work....It would take me an hour and a half morning and an hour and a half afternoon to do the drop offs and get to work.' (0: Effort, Time)

'...we have to fiddle and juggle with funds, and we're allowed a little bit of petrol in one lot of funding...but it doesn't cover the week. If the carers use their own car, they need to be compensated...we've got to pay them....that's stressful for me, because I do the paperwork....they specify it's respite only but don't take into account that...for respite she needs a car...' (3: Funding, Paperwork)

6.5.12.3 Stressful

Among the nine carers who found this item stressful the problems included the need for modifications to the car to accommodate a wheelchair, public transport issues, and their own physical limitations.

'...it cost me \$100 to get her to [respite] and \$100 to get her back....this carer...said to me, 'There's an organisation,' which I didn't know about....I only pay \$6 now – \$6 for a maxi cab...life's going to be a lot easier...even when I get the car...I can't drive that far...' (4: Funding, Knowledge, Instrumental)

'She gets a school bus....We were lucky...we get the school bus to [supported accommodation] as well, a different school bus. If...that stopped, then that would become a problem...you'd just have to book, and she'd have to travel on a taxi...' (2: Positive outlook, Planning)

There was also an element of disappointment that things were not 'normal' given their child's lack of mobility.

'...when I take [older child] to school and I've got two kids in the car, and just for that five minute drop-off, I've got to get two kids out, and he could be, he should be walking, and other Mums that have got kids the same age as me are carrying a baby and their three-year-old is walking behind them, and I've got to get a pram out.' (2: Effort)

'It's...having to get her in and out of the car...if we just go up to the shop, we...put her in the car and then put her wheelchair in, we go to the shop, we take that out, we take her out, get back in the car, do the exact same thing again. Then come home and do the exact same thing and get her out of the car.' (4)

6.5.12.4 Not stressful

More than half of all respondents (10/19) experienced little or no stress when dealing with transportation issues.

'Mum gave us some money to put towards a car....I think it might have been [a stress], if she hadn't been so generous.' (0: Instrumental, Positive outlook)

A number of carers used wheelchairs, prams, or harness to suit the capabilities of their child and the specific situation.

'Just beginning to think about having to deal with wheelchairs....But it's not really an issue yet.' (1: Planning)

'[when] we go to...big outings...we will actually hire a wheelchair.' (0: Practical)

'She's mobile so that's good, she's helping. She helps a little bit...we've just got a safety harness for her, because she's wriggling around...and she's outgrown her seat, the seat she's in for a while...' (1: Practical)

'...we have a four-wheel-drive, he has no problems getting in and out of that. If we...go to a shopping centre or...into town...it's more for our convenience we put him in a wheelchair.' (0: Practical)

6.5.13 Day-to-day assistance with care of your child

6.5.13.1 Introduction

This was a slightly stressful item (mean = 1.58). Five older and two younger carers received no assistance with day-to-day care, either because their child was not living in the home or was capable of self-care, or because such assistance was not available. Instrumental (7) and Practical (5) were the most common coping strategies.

6.5.13.2 General stressors

Time pressures (3) were the most frequent stressor.

'... that's all got to be done before [unclear] shower and work and do breakfast...' (3: Time)

6.5.13.3 Stressful

More than one half of respondents (10/19) rated this item as stressful. Co-ordination of care assistants, including arranging payment, was a source of considerable stress for some carers.

'We had Silver Chain...Monday to Friday...also...in-home respite from [Association], [agency], [another agency], and we had out-of-home respite....And then we had sitter services as well and...coordinating it, which organisation's coming, how...do you...pay, do you pay this one cash, do you pay that one on account every month?' (4: Instrumental, Effort)

'...she leaves to go to school so early, trying to get everything organised....And just not having the help....It's...difficult finding the staff for that time of the morning. She gets picked up by the school bus...by 7 o'clock...' (3: Effort)

Staff reliability issues were also a source of stress, especially for any carer who was not physically able to undertake all of the care duties:

'It seems like it's harder on your back, physically it's harder....I just hope she becomes a bit more mobile, and she has...it's not been as bad...as she's getting taller, it's easier in you don't have to bend over so much....she's progressing with being able to help with bits and pieces...dressing.' (2: Positive outlook)

'...we've been very lucky to have had...good carers. A lot of them, I've gotten from work...working in the disability sector has been a bonus in some ways....I self-manage the funding money...' (2: Positive outlook, Funding)

'If they don't come, I'm the one who does it all day...because I've got to do it.'
(4: Effort)

6.4.13.4 *Not stressful*

The carers experiencing little or no stress (9/19) may not have required much assistance, especially with younger children. However, as the child grows larger and stronger, there may be a need for increasing amounts of assistance.

'...I find that very easy. My Dad's here, he helps me, and when he does go [on holiday] then I've got a network of friends who can help me out as well.' (0: Instrumental)

'...when it comes to showering, because [child] has the fascination with water, you would need to give some assistance....and maybe just putting her nappy...because she moves so much, so the carers do find it a little difficult to do on their own...' (1: Practical, Effort)

6.5.14 *Dealing with doctors and other health professionals*

6.5.14.1 *Introduction*

This was a slightly stressful situation (mean = 1.47). 37.5% of the carers 50yr and greater in the present study rated this item as stressful, in contrast to two other studies, both of which reported considerably higher percentages: 82% (Jones & Passey, 2004), and 50% (Minnes & Woodford, 2004). Knowledge was the predominant strategy (15), followed by Advocacy (9).

'...we've done a lot of research ourselves in the background...so we knew what we would be dealing with. We've always gone into medical appointments very knowledgeable.' (1: Knowledge, Planning)

'Our first GP, we were the ones who told him what was happening, told him all about it. The GP at the time used to open up his medical books in front of us.' (1: Advocacy)

6.5.14.2 *General stressors*

Anger (12) was the most prevalent stressor for this item. Carers spoke of the frustration of being more knowledgeable than the medical staff, and delays in getting medical appointments.

'...a lot of them either don't know what it is or have very fixed ideas...and there's definitely more than the clinical notes tell you and they don't all fit in the box...it is frustrating when they don't know...' (2: Anger)

'...it's frustrating to have to teach your doctor how to help your kid.' (1: Anger)

'...you don't always get the appointment on that day...he doesn't like doctor's surgeries, he doesn't like hospitals...he gets to a doctor's surgery and he makes a lot of noise because he doesn't want to be there....I basically said, 'If you get me in to see the doctor and out of here, he'll be quiet – he doesn't like being here.' (3: Anger, Practical)

6.5.14.3 Stressful

Eight carers rated this item as stressful. The general problem seemed to be a lack of knowledge on the part of medical practitioners. Some carers spoke of educating the doctors and hospital staff.

'Lack of understanding of Prader-Willi because...it's quite rare. It's getting more common...but back when he was younger, it was so rare....I go armed with my book of Prader-Willi Syndrome [laughs] and give them a copy of it.' (4: Practical, Planning)

Several carers also referred to problems getting appointments, or dealing with their child in a waiting room.

'Definitely stressful, especially if you haven't seen the doctor, or house professional...having to wait for appointments that never seem to happen...' (3: Time)

'...our daughter doesn't like to sit still. So you're in this little confined room with lots of interesting things to do. I...will always ring up and say, "So are you running on time?" Because if they're not I would rather have her [home] and arrive at the last minute.' (3: Planning, Practical)

However, many carers were generally happy with their doctor or specialist:

'I have pretty good experience; it helps working in the profession, doesn't it? You know the system.' (2: Knowledge)

'Just sometimes it's really hard to get appointments and things like that....the doctors are great, bend over backwards to help.' (2: Positive outlook)

6.5.14.4 Not stressful

A majority of carers (11/19) rated this item as little or no stress. Many of them emphasised the importance of educating medical staff, asking questions of practitioners, and preparing prior to a medical appointment.

'...junior residents...who have absolutely no idea about things...you soon get into the education system...telling them what's what....I've got all the new mums doing this as well, the Angelman Syndrome brochure, I had attached to the inside of her [hospital] file.' (1: Practical, Advocacy)

'I write lists if we had a doctor's appointment, a week prior everything I thought of I would write it down. I would come in with my list and so I would ask the questions, I would take notes.' (1: Planning, Knowledge)

‘...I get to know what it’s all about and try to work through it. I believe we should get information and we should use the resources around.’ (0: Knowledge, Instrumental)

‘I find it quite easy, and that’s because we’ve been exposed to a lot of doctors since [child] was seven weeks old. And we just gain lots of information from them....we ask them lots of questions.’ (0: Knowledge)

6.5.15 Making the decision concerning accommodation in the home or in the community

6.5.15.1 Introduction

This was a slightly stressful question (mean = 1.47). A small number of older carers (2/8) found it stressful, compared to 54% in a previous study (Minnes & Woodford, 2004), and all six respondents who found this stressful gave a score of 4 (extremely stressful). Planning (10) and Reinterpretation (9) were the most common coping strategies.

6.5.15.2 General stressors

The most frequent stressor was Funding (13); another area of concern was the availability of supported accommodation.

‘...we started putting in for funding for him...to go into a group home, on his right to be able to move out of home. But the stress came when the funding isn’t there...’ (1: Funding)

6.5.15.3 Stressful

Almost one-third of respondents (6/19) rated this as stressful. It was apparent that some carers had reached or were very close to breaking point in caring for their child.

‘...we feel...burnt out, and we don’t feel we’re being fair on [child]...hanging around with our friends...we’ve just had him placed at one group home...we’ve got another one coming up....a huge stress in trying to get him settled....I’ve learnt that I can trust other people to meet his needs.’ (4: Reinterpretation, Planning)

‘...sometimes I just can’t handle her at that time....I just want to go, ‘take her away and let me get a bit of peace and quiet and relaxation’.’ (4: Detachment, Emotional)

One carer was undecided on the issue of accommodation, while another had made a decision, but still needed to arrange matters in order to achieve that aim.

'...there is a couple of other little boys...the same age...[name], her daughter's in a home with a couple of the Angel children. So do we look at doing something like that?' (4: Knowledge, Planning)

'...had to sacrifice my need to keep him mainstream....do I go with my pride or do I swallow that and get him sorted because he needs to be top of the heap, not bottom of the heap.' (4)

'...she's going to stay in the home for as long as I can live. In my will it's set that she will go to [accommodation]...I need her where she's safe and not exploited, and I've got that all set up...for when I die. I just pray that we go together...' (4: Effort, Planning)

6.5.15.4 *Not stressful*

Six of the carers <50yr had not considered this matter at all and were scored 0. Among the eight carers who both rated this item as low stress and had given the matter some consideration, several understood the difficulties associated with funding, but had taken positive steps to deal with them.

'...the funding...is crisis-driven...in a perfect world...they would say, 'Right, high support needs, when she turns 17 or 18 or whenever the need arises, there will be a place in a group home for her.' That's not the way it works...it's crisis-driven and you fill in extensive paperwork...you could wait several years before you get something....it was still 5 years from start to finish....it was paperwork that had to be filled in because I was looking into the long-term...' (0: Planning, Paperwork, Funding)

'...we'll try and keep [child] with us for as long as possible. At the same time we are applying for funding so if it ever comes up...it's there...I couldn't imagine that happening, not for 10 years anyway.' (0: Funding, Planning)

More than one family struggled to function when there was a person with AS or PWS living at home and two of the older mothers had previously experienced a breakdown.

'...I was not going to have [child] breaking my family up....we knew that we had this dysfunctional family....There was really no choice, for our own sanity, that she went anywhere except into a group home.' (1: Effort, Reinterpretation, Detachment)

6.5.16 *Explaining to other people about your child's disorder*

6.5.16.1 *Introduction*

This was a mildly stressful situation (mean = 1.32). More than half (11/19) carers did not score this issue as stressful. Reinterpretation (16) was the strategy voiced most frequently by the family carers. Carers adjusted the amount of information they

offered to other people depending on the relationship between themselves and the enquirer.

‘...at playgroup recently I had a new person introduced and [child with AS] is going up to touch the baby...you’re just trying to explain...‘My son’s got Angelman Syndrome’...if they’re going to keep going...you try and explain a little more. But if you’re in a shopping centre...keep it brief...’ (2, Effort)

‘Some people are interested and have some sort of knowledge about it. Other people...don’t want to even know about it.’ (2: Reinterpretation)

6.5.16.2 General stressors

The general stressor mentioned most often within this item was Label (10). Two different carers expressed, in very similar fashion, the thought that a diagnosis or label in fact made it easier to explain their child to other people.

‘...it was easier when she had a label than when she didn’t have a label, even though I didn’t think a label made a difference...’ (0: Label, Knowledge)

‘They say labels don’t matter, but they do; you feel like you’ve got a direction. It makes a big difference.’ (1: Label, Reinterpretation)

6.5.16.3 Stressful

Among the eight carers (4/11 <50yr, and 4/8 50yr and greater) who rated this item as stressful, a common reaction was anger or frustration.

‘...you tell them and they still [say] ‘Well, so what does that mean?’ And...having to repeat it all over and over again....some people haven’t heard of it at all.’ (4: Anger)

‘...you always had to keep explaining and you had to make allowances and excuses, and I used to get angry at having to do that....when I first took him to playgroup...the first thing he did was run amok...pushing all the kids over....I was so horrified that I’d just take him home....we didn’t know there was something wrong with him....no-one had really given us any answers to anything...’ (3: Anger, Practical)

Improving their own knowledge and educating other people was important to some carers.

‘...a lot of people couldn’t cope. Some family members couldn’t cope....We keep away from negativity...’ (2: Reinterpretation)

‘...definitely easier....five years ago I didn’t know what the issues were. Now I can put a label to it, and I’ve done all the research so I’m more competent and confident in what I’m saying.’ (2: Label, Knowledge)

‘...me finding out more about it and being more at home with the diagnosis...’ (2: Knowledge, Reinterpretation)

'I do a lot of staff orientation especially for DSC and you say, 'My son has Prader-Willi Syndrome,' and they say, 'What?' (laugh). So you have to go through all the symptoms and everything.' (2: Advocacy, Positive outlook)

6.5.16.4 *Not stressful*

The respondents who felt this to be a low-stress item were keen to inform others about the syndrome. They also adapted their explanations to the demands of a situation and took the opportunity to pass on knowledge to other people.

'...people can be embarrassed about asking, like curious but not wanting to offend us. Whereas I'm thinking, 'hey, the more knowledge that's out there, the better you'll understand my girl'...I'm quite happy to explain.' (1: Advocacy)

'...everything that we've learnt, we pass on to other people...give them as much information as we know.' (1: Knowledge, Advocacy).

'...you learn to shorten it because it's just too draining to go into a lengthy explanation.' (1: Reinterpretation)

6.5.17 *Dealing with friends/family/neighbours on a day-to-day basis*

6.5.17.1 *Introduction*

This was a generally mildly stressful situation (mean = 1.32), in contrast to the study by Jones and Passey (2004) which reported a mean score of 3.02 on this item. Gaining Emotional support (11) was the coping strategy expressed most often by respondents in the present study.

'My neighbours around here...were really very, very good...' (2: Emotional)

6.5.17.2 *General stressors*

There were very few generalised stressors associated with this item.

'And angry, very angry, because whenever I'd leave [family member] with [child]...something would happen, and I was terrified, because she'd leave pills around...I was terrified.' (2: Anger)

6.5.17.3 *Stressful*

Five of the nine individuals who found this stressful were 50yo or greater. The behavioural characteristics of their child seemed to be the greatest source of stress within this item. These carers appeared to have some friends, family or neighbours who did not cope well themselves with the child with AS or PWS.

'...the lack of understanding, [of] insight into the disability. Because he looks pretty normal...the lack of realisation of the effort on our behalf....we do everything right...a lot of time and effort is put in...when they see him

misbehave, they think it's just a really badly behaved child or bad parenting. So I'm always on edge....I feel labelled largely and I don't like that.' (4: Anger, Advocacy, Effort, Label)

'When he was younger, he was cute, and some of the things were tolerated. A lot of the stuff he does now [as] an older child, they aren't as tolerant. Family were a very big support to us in the early days but now....you can become isolated socially....you stick with the same friends, or you tend to go towards families who have kids with disabilities because they're more understanding, more tolerant.' (3: Emotional, Reinterpretation)

Emotional support is especially evident within this group, usually associated with other positive aspects.

'Our friends are pretty good with our daughter, they love her as well, can be very helpful...' (2: Emotional, Instrumental)

Several carers spoke of the capabilities of their child falling behind those of their chronological peers, resulting in exclusion.

'As she gets older...kids are less inclusive....They tend to treat [child with AS] like wallpaper at parties....it's just being defensive of your own kid, not wanting them to be excluded....like she's staying at two, and they're at seven.' (2: Reinterpretation)

Some family carers, however, found dealing with friends, family and neighbours could have positive features.

'...I got through a whole two children of doing tee-ball without ever having to do scoring because I had [child with AS] (laugh).' (2: Positive outlook)

6.5.17.4 Not stressful

More than half of respondents did not find this situation to be stressful. Members of this group were predominantly younger (7/10 were <50yr) than those who reported higher stress levels. They spoke of receiving considerable emotional support, and tended to accept that people without a child with IDD could not fully understand what it was like.

'...they are quite understanding...as much as they can be...' (1: Emotional, Reinterpretation)

'...my family and friends are quite supportive...I surround myself with people who are positive...' (0: Emotional, Positive outlook)

There was also a tendency for these carers to avoid people who did not understand or accept their child, while embracing those who dealt well with the child.

'Keep away from the negative ones.' (0: Reinterpretation)

'If they cause me stress then...you cull them out.' (0: Reinterpretation)

'But the people around here have been amazing, absolutely wonderful...' (0: Positive outlook)

6.5.18 Dealing with your child's sexuality

6.5.18.1 Introduction

This was a slightly stressful situation (mean = 1.32), with seven carers rating it as 2 or higher. There were few references to general stressors, and only a small number to different coping strategies (Instrumental, Effort, 4 each) utilised.

6.5.18.2 Stressful

Most of the carers who rated this item as stressful (4/7) were 50yr or greater, with generally older offspring. They were more likely to be currently dealing with their offspring's sexuality, or to be anticipating the necessity in the near future. The perceived vulnerability of people with AS or PWS was a source of concern for some family carers.

'...part of it is dealing with the period and the second part would be...hearing about children that get sexually abused and she wouldn't be able to tell us.' (4)

'...her periods came on and the doctors couldn't turn it off...she would have been in pain but she wouldn't understand because she's used pads all her life....And to go to court for 4 years and fight everybody [for a hysterectomy]...' (4: Effort, Advocacy)

'...I'm just working out what to do about menstruation....about Depo-Provera shots....I'm not worried about her being a sexual being, just worried about her being preyed on....I'm just concerned about people taking advantage of her affectionate nature.' (2: Planning, Reinterpretation)

'She did have PMT and she suffered a lot...when she was premenstrual she had worse nights....the greatest impact on her life was in the summertime when she couldn't go swimming with the other kids, and it impacted on the whole of the family because the other kids couldn't go swimming at all....I talked to our endocrinologist...' (2: Effort, Instrumental)

6.5.18.3 Not stressful

More than half of the carers (7/12) who rated this as low stress were <50yr, and four carers had not given the matter any consideration at all, possibly a reflection of their child's young age.

'When she first started her periods, I...put her on the pill and ran it straight through back to back, and that was fine....we've had a very bad number of months recently with seizures so I've taken her off [the pill] because the anti-

convulsion medications that she's now on were affecting it, but she's full-time padded....where the girls aren't full-time padded, then there's management issues.' (0: Practical)

'...we've taken that as it comes...sometimes he's masturbating, we say 'go to your room, that's private'....We always feel...there's plenty of other things at the moment than worry about that.' (0: Reinterpretation)

6.5.19 Time apart from your child

6.5.19.1 Introduction

Overall this item was again slightly stressful (mean = 1.26). Three-quarters (75 %) of the respondents 50yr and greater rated the situation as stressful in comparison to the 51% reported in Minnes and Woodford (2004). Instrumental (13) and Planning (7) were the two most common coping strategies.

6.5.19.2 General stressors

Funding issues were mentioned twice.

'...they wanted \$300 a night....It used to be \$25 a night.' (0: Funding)

6.5.19.3 Stressful

Of the nine carers who scored this item as 2 or more, six were 50yr or greater. One source of stress was the behaviour of the child.

'Well, he didn't like respite....I used to...take him to a respite place, and they loved him, and once he was over the hump he settled down a bit....But he hated it, it was just a screaming session from school all the way through so I never used to push it very much so we just took time to spend with the other children individually...' (3: Instrumental, Detachment, Effort)

'...things that happen when they're on respite, like they get bites on them or some other kid's bitten them or pulled their hair, or your kid does the same thing to somebody else. So just going on respite is not a stress-free period at all. You still stress. It just takes the physical pressure off.' (2)

A variety of coping strategies were used by these carers.

'...my husband has her a lot, and I have her a lot. I go to work, 'cause I have to work for my sanity. I enjoy that...it's having the time alone together, both away from her, that's the hard bit.' (2: Instrumental, Detachment)

'...at school it is [stressful]...there's always that 'oh my God, when's it going to happen again' but when he's out with people, generally not, but it's getting harder. Like my parents are getting older and even though they have him sometimes, there's always that fear that he's going to get cross and that's not going to be dealt with effectively and he might try and hit my Mum and my Dad won't tolerate that...that's sort of happened a couple of times...' (2: Instrumental)

6.5.19.4 *Not stressful*

A majority of the carers who did not feel stressed by this item (7/10) were less than 50 years old. Parents of the youngest children tended to treat the issue as normative compared to those with children of an age to be expected to look after themselves.

'I've probably only left him for as much as a night... I haven't been stressed....but I'd probably be more stressed if it was longer...' (0)

'...when he was younger we used to send him to the [unclear], it took us ages to do that, just to get organised. But once I got for him to trust...that someone else can look after him...' (0: Instrumental)

'...we do worry if she's alright when we go out. But then, we worry about the boys as well, but more so we think, 'ooh, I hope the babysitter's coping with her,' rather than, 'I hope she's ok.' (1: Instrumental)

Carers who had access to respite tended to find that setting up a routine and being prepared was beneficial.

'It's fairly routine...for example, next month he goes in [to respite] I'm...having a general anaesthetic...and my husband's going to be [overseas]. So I just rang up [respite] and said can I leave him there two extra days...financially...it's going to cost us....All I have to worry about is packing his bag and organising his meds...' (0: Instrumental, Funding, Practical, Planning)

6.5.20 *Deciding on the best level of integration for your child*

6.5.20.1 *Introduction*

This was a slightly stressful situation (mean = 1.21). More carers aged <50yr (4/10) than those 50yr and greater (1/8) found this situation stressful. The most common coping strategy was Reinterpretation (15).

'...once we had the diagnosis it was easy, because I knew the probable baseline....Once I knew she would never be verbal then I had to go where she'd get the best communication strategies and that would not be in a mainstream school.' (0: Reinterpretation)

'I didn't want to put [child] into [mainstream] environment because he goes into his shell very quickly. So we said right from the word go that he would go to Special Ed....And looking at my other two children, for the best for them as well....not to have their brother in the same school.' (0: Reinterpretation)

6.5.20.2 Stressful

More than a quarter of respondents (6/19) rated this item as stressful. Most stress appeared to be associated with indecision or with discontinuity in the child's education.

'...I really pushed...for younger parents to let their kids do primary school because I [think] it's great....when [child] entered the system, they weren't ready for him at all...they weren't prepared. So it failed and he ended up in Special Needs. But then he started a program at [primary school] and he was integrated there and that worked...' (3: Advocacy, Effort)

'...just worrying about what is the best thing to do?...what is the best school?...do I go special school? do I go private? do I go public?...where we're living, how much of a strain it's going to be getting him to...[distant school] or something like that?...how's it going to work in with the rest of the family life?' (4: Planning)

'We have put a little bit of thought into it. The school that [older child] goes to has a special needs unit...not sure how [child] will go, but we have enrolled him there....I hope...he might end up there. I've spoken to his teacher at [special school] and she said 'once he does learn to walk, feed himself, and all that sort of thing...he won't need the conductive education...anymore'. That could take quite a few years...' (2: Planning, Reinterpretation, Knowledge, Time)

'When we moved...we...looked into the schools....did our homework and researching, calling the child psychologists...and discussed it all...and we made a choice between two schools...' (4: Knowledge, Instrumental, Planning)

6.5.20.3 Not stressful

A majority of carers (13/19) felt that deciding on the level of integration for their offspring involved little or no stress. Acceptance of the limitations of their offspring was most evident, and appears to have greatly influenced any decision.

'...not with the severity of...her physical and her intellectual together...I think with an intellectual disability, then you really do need specialist programs and those smaller classes like...two staff to three to six kids...where you're getting the maximum input and the maximum output that you can...' (1: Reinterpretation)

'We've decided on special needs schools...just because of the facilities...they have a swimming pool, physiotherapy, and they focus on day-to-day living, life skills rather than colouring-in and drawing.' (0: Reinterpretation)

'...we always knew that [child could not go mainstream]. We accepted that a long time ago.' (0: Reinterpretation)

'...the ideal is that he attends the school that's in the community that you live in and that's why...' (0: Advocacy)

6.5.21 Maintaining satisfying friendships for yourself

6.5.21.1 Introduction

This was a slightly stressful situation (mean = 1.21). Five of the six individuals who scored this item as 2 or more were 50yr or greater. Most carers spoke of Emotional support (18) in relation to this item.

'We maintain them pretty well actually. Those who appreciate us for who we are and what we are, I tend to keep those friends....the others merely by the way, acquaintances....I know we do have friends....because they totally understand and they will give us our space, so I think they are all very good friends.' (0: Emotional)

6.5.21.2 General stressors

Time (5) was the major stressor mentioned in regard to this item.

'...finding time, I find at the end of the day, I can't be bothered phoning people....I've got some good friendships, but ones I used to have sort of drifted off...I don't have time to fit in seeing them....I'm sure it gets easier once [child] go[es] to school.' (1: Time, Detachment)

6.5.21.3 Stressful

Many of the six respondents who felt this was stressful indicated that the stress resulted from restrictions on the freedom that would usually be available to carers in later middle-age. Reciprocal visiting was not possible:

'She is first in my life and everybody else comes after. Her care is more important to me than talking to somebody....I don't go to anybody's house. When people come here they do find it difficult.' (4: Effort, Reinterpretation)

'...we find that it's very rare that people drop in here. And we don't maintain friendships because we haven't got the freedom. I think that's quite a big thing.' (4: Detachment)

Two carers emphasised the importance of emotional support in supplying personal friendships.

'...if you've got a support network that...will listen and you can talk things through...get you through for another week....that's a direct impact of having a child with a disability. It isolates you socially.' (2: Emotional, Effort)

'...we've lost lots of friends, we were close to each other, and we all had kids at the same time. We all thought we'd have growing up kids, growing up together....Our son's first twelve months, he was quite normal really. We just saw the milestones not happening....after that we realised there was going to be a problem and he would need more attention and time....I think it's important...someone to talk to or whatever.' (3: Emotional)

6.5.21.4 *Not stressful*

Most of the carers for whom this was a low stress item were <50yr (10/13). When caring for younger children it could be considered to be more normative to have a friendship circle restricted primarily to like families, so that there is less of a feeling of exclusion. A variety of coping strategies were expressed.

'...I'll go out of my way to stay in touch with people....initially I had a core group of friends....I had to get rid of them because they were just too...demanding...I just had to...make a conscious decision that I no longer wanted them in my life...' (0: Effort, Emotional, Detachment)

'I've got a lovely group of friends...the only thing that's a bit hard is getting time to see them...I don't take [child] out with me....I like people to come here...I don't go to restaurants or anything like that...' (0: Positive outlook, Time, Reinterpretation)

'I've still got all my old friends, but I probably don't see them as often, and I've made new friends since having [child] that I'd probably see more. I guess you know where each other's at and more accepting of each other's kids.' (0: Emotional)

6.5.22 *Planning for emotional and social support for your child*

6.5.22.1 *Introduction*

This was a low stress item (mean = 1.16). The carers who felt this was stressful included four who were 50yr or greater, and two who were aged <50yr. However, fewer of the older participants in this study scored this item as stressful (50%) when compared to the 68% of Minnes and Woodford (2004). There were no prominent coping strategies discussed in relation to this item.

6.5.22.2 *Stressful*

Over one third of respondents (7/19) found this situation stressful. For some of them, at least, the future for their child did not encompass emotional and social support.

'You can't buy friendships and if you could, it would be really easy....She doesn't have a friendship circle. She has the people that she lives with...in her family and the people on the day program, and really that's...the extent of her social life....She doesn't get out and about a great deal now.' (3: Reinterpretation)

'... everybody says that their biggest fear is what happens to their children when the mum and dad have gone....I don't even like to think about it....The reality is that...if something happened to me, if I was going to be killed in a car accident, I would hope to hell that she was with me and got killed as well.' (4: Detachment)

'We tried to introduce a few friends....almost like an informal network?....we know it's there if something [unclear] but then again I suppose [group home] that will form a friendship for him there and they can handle the emotional side of things while we're not around.' (2: Effort)

6.5.22.3 *Not stressful*

Finding emotional and social support for their child was not considered to be a problem for many respondents (12/19). For some, the idea of such support being necessary had not entered their thinking, while other carers had considered the matter and taken steps to achieve their aim.

'...I've had carers that have promised...that they will be here for her...if they were here for her after I'm gone, they'd be here while I'm [still] here. And I don't see them....there are people I do keep in touch with. I send Christmas cards, I write letters and keep them involved....that they will be involved with her after I'm gone....one of my foster sons, he absolutely adores her....he will go and see her and he will jump up and down if anything's wrong.' (1: Instrumental, Effort)

'...at the moment he does take care of himself, he's got his own bowling group, he's got his own netball group....So he's got...a good social side....he is not afraid to do anything. I brought him up to have no fear of life.' (0: Positive outlook)

'...I'm sure that my brother and sister-in-law, I'm sure that [name] would keep in touch...her cousin and things like that....I have looked at that but haven't really thought about it...' (1: Detachment)

6.5.23 *Work placements or employment for your child*

6.5.23.1 *Introduction*

This item was not considered stressful (mean = 1.05), with 14 of the 19 respondents reporting no or little stress. Planning (8) was the most common coping strategy.

'She'll get...full post-school options funding....the kids stay at [full-time] school until the year they turn 18....the maximum...funding you'll buy yourself 3 days of day-placement. Kids who are living at home...mum's...worked around the children being at school from Monday to Friday, suddenly...2 days that [child is] at home. Respite tends to...halve when you go from school age to adult....That is why I wanted permanent accommodation set in place before she left school. So now, it'll be the [service provider's] problem.' (0: Planning)

6.5.23.2 *General stressors*

The main stressor identified within this item was Anger (5).

'...I remember the frustrations that I used to have, and that went on for about 6 years, of total frustration...' (4: Anger)

6.5.23.3 Stressful

Most carers who found this stressful were older (4/5), and their children tended to be higher functioning. The principles of inclusion and normalisation appeared to be very important to the carers.

'We went to a workshop [about open employment]...we've got...some tips on how to link [child] in with that...and how to up-skill him....we really didn't have the energy to go, we pushed ourselves to go....we know that just as passionate [about] inclusion as we are, that he will have open employment....for 7 years we've been within a group that believes in inclusion, fights for senior partnership and we know that he will go down that [path]....It's something we're constantly striving for....it's for him...to come out...with...a better quality of life. (3: Advocacy, Knowledge, Effort, Instrumental)

'...I didn't know what the solution was....he didn't have a severe disability but he wasn't normal...he was in limbo....that's even worse than being one or the other. It was extremely, extremely hard, and I used to fight...a typical mum, go off at these people for not doing the right thing...in the end I just gave up.' (4: Detachment, Effort)

'[employment] that's failed completely....he [is] very good at manipulating people, and he worked out...if he played up as soon as he got there, they'd phone me and I'd come and pick him up....we had huge problems trying to keep him in work...' (4: Detachment)

6.5.23.4 Not stressful

Commonly, the carers who found this item to be not stressful were <50yr (10/12). Acceptance was a prominent attitude among these carers.

'We knew...that he just doesn't have the tolerance or patience to sit there...' (0: Reinterpretation)

'He just can't, he doesn't follow instructions....he just can't do it. And we actually don't know how much he understands.' (0: Reinterpretation)

'...the school had the transition going between school and [day centre], so...in the last year of school she was going to [day centre] one day a week and they were getting used to her and she was getting used to them. The transition was very smooth.' (0: Planning, Instrumental)

'I saw someone working at [supermarket] who was very obviously special needs, and I'm thinking 'could my child do that?'. He was just wiping down the fruit and [vegetable] fridges.' (0: Planning)

6.5.24 Dealing with legal professionals

6.5.24.1 Introduction

This was a low scoring item (mean = 0.37), with most carers (15/19) having experienced no dealings with the legal profession in relation to their offspring with AS or PWS.

6.5.24.2 Stressful

One carer who found this situation stressful spoke of feeling abandoned by the service agencies:

'...she started her periods at 6...took me 'til she was 16, four years and a court case...we got permission to do the partial hysterectomy....it's the waiting, it's the longevity of it...you end up on your own because [agency], everybody just backs off when you go to court. Nobody wants to be involved in a court case so you're left there on your own.' (4: Advocacy, Effort, Time)

A perceived lack of empathy concerned another carer:

'...not very flexible, not very understanding – procedures that are more bookwork than caring and organising.' (2: Paperwork)

6.5.24.3 Not stressful

The one carer who found this a little stressful had used her network to gain some insight into the process. All other carers had scored this issue as not applicable (14/15) or not stressful.

'...talking to other parents who have gone through the paperwork, the amount of forms you have to fill in, but...deal with someone like Centrelink and you're filling in lots and lots of forms anyway....' (1: Knowledge, Paperwork)

6.5.25 Your feelings about the cause of your child's condition

6.5.25.1 Introduction

This was another very low stress item (mean = 0.32). Only one member of each age group scored this item as 2 or more. Reinterpretation (8) and Knowledge (7) were the most commonly expressed coping strategies, although it could be speculated that even those who did not articulate acquiring knowledge may have gained peace of mind from knowing that the condition is generally sporadic.

'It's out of my control, it's nothing to do with me, just one of these things that happen.' (0: Reinterpretation)

6.5.25.2 *General stressors*

There were very limited stressors identified in connection with this issue:

'...[relative] has just come back into the family and his wife wanted to know...if it was genetic. And those issues probably anger me more, the ignorance or whatever but upset me a little bit...' (0: Anger)

6.5.25.3 *Stressful*

Two carers found this item somewhat stressful. This seemed to relate to either a lack of knowledge or to other difficulties encountered during the pregnancy.

'Well, probably if...we were fully informed about the actual condition, how it happened, because it's no-one's fault obviously in the end.' (2: Knowledge)

'...we tried so long to get [child] and before I had him, I did...drink a bit of alcohol...like one or two during the pregnancy...by that stage I had done three pregnancies where I did the perfect thing and still had the result of no child at the end... so by this [pregnancy] I was going 'one or two drinks isn't going to hurt – if it's going to happen, it's going to happen'...' (2: Reinterpretation)

6.5.25.4 *Not stressful*

Most respondents (16/19) experienced little or no stress when considering the cause of their child's disorder. The foster mother had no feelings about the cause at all. Carers referred to acceptance and gaining information as important coping strategies.

'...it was spontaneous mutation – I have no control over it whatsoever.' (0: Reinterpretation, Knowledge)

'I just think 'life's like that, this is what's been dealt to us' so we get on with it and do the best that we can.' (0: Reinterpretation, Effort)

'Not once we found out. It's just something that we've accepted and we've got on with life.' (0: Knowledge, Reinterpretation)

In addition, some carers expressed positive (concentrating on what the child can achieve) and forward-thinking (having the freedom to have more children) attitudes:

'We've got this child, we've got to concentrate more on her and seeing what she can do.' (0: Effort, Positive outlook)

'I guess the fact that it wasn't hereditary was a good feeling, and that we could go ahead and have another baby.' (1: Positive outlook)

6.5.26 Supplementary questions

6.5.26.1 Introduction

A majority of families (13/19) were members of either the Angelman Syndrome Association or the Prader-Willi Syndrome Association. Both groups were relatively new when the data were collected and so did not have extensive membership lists.

6.5.26.2 *Have you received any benefits from your membership of the [support organisation]? If so, what were they?*

Most members (9/13) felt that they had either already gained some benefit or anticipated that they would benefit from joining the support group. There were a number of suggestions aimed at improving the outcomes from group membership.

6.5.26.2.1 Benefits received

The most common benefits were related to the concepts of Knowledge and Emotional support. Drawing on the experience and advice of other parents, and sharing feelings with them were very important to these respondents.

'...people who live the same life, who have the same stresses...who are going to have the same outcomes....gives you a sense of belonging, that you're not on your own...these mums [who] have got young kids who are only 3 or 4 or 5 now, just don't know what's in store for them. And I want to be there for them when they get there because their journey has just started...' (Emotional, Knowledge)

'...great bunch of people that you can actually talk to. It's nice to be able to share....going to the meetings...it's a night for me in a way....sitting there and talking and listening to what other people are going through.' (Emotional)

'...the information that other families have is far more than what service providers give you and DSC give you, and you support each other, you understand where each other's coming from...It's actually quite good to realise that other people have the same stresses and their child is entirely different.' (Emotional, Knowledge)

6.5.26.2.2 No benefits received

Some carers looked forward to obtaining future benefit from membership of the group, either through acquiring knowledge or receiving emotional support. Two carers felt that the group was irrelevant or unsupportive and tended to distance themselves from others.

'Not yet. But I'm sure, in a couple of months I will be. We're going to the conference.' (Knowledge)

‘...stuff comes in on the computer, and I read it but a lot of it – to me...it’s not relevant to us at this point in time.’ (Knowledge)

‘...not at this stage. We met...[other member], which was lovely, fantastic, but the distance is a big barrier.’ (Emotional)

‘...they give me nothing back. They don’t give me any psychological support, they don’t give me any social stimulation...over the years...I feel I’ve given a lot when they were having problems...but I don’t get that back...I’m tending to step away a little bit and avoid...because it’s too draining for me.’ (Detachment)

‘...our daughter is one of the oldest and...in the early days when [group] started...we’d go along and if they asked me any questions I’d say it and I could have them all in tears...so we actually stopped going because I felt so [bad?]...they’ve got nothing to...offer us really...’ (Detachment)

6.5.26.3 *Is there anything that would make membership of the group more rewarding or helpful to you?*

One issue was the small number of members in each of the support groups. Some carers felt that they would be able to obtain advice and support more readily from within a localised network; these could be formed once there were sufficient members in any one area.

‘...more helpful to have a local sort of group and for networking with other parents....you learn most of your stuff from other parents...’ (Knowledge)

‘...more friendship.’ (Emotional)

‘...I’m one of the few [locality], all the others are...the other side of town....if I could just have a coffee with a local Mum....there’s a Mum in [country town] with a child who...would be our next mentor....I look at [child] and think that if she lived locally, we’d see her.’ (Emotional, Knowledge)

‘...as far as any other support, I haven’t really gotten any but now that we are...setting up the WA branch...there will be a lot more networking....hoping to be able to ask more questions, learn more things...I’m hoping I can tread in someone else’s footsteps and make my path easier.’ (Knowledge, Emotional)

6.5.26.4 *Other comments*

The additional comments made by the respondents were reflective of both problems and solutions. Two major issues raised were incontinence and sleep disturbance.

‘Continence [laughs]....our daughter is day-time continent – mostly....we have to put her on the toilet for quite some time before bed...if she needs to do a poo...in bed, it goes everywhere....we can never, ever properly relax until she’s asleep....we just can’t let down the guard....We have her in strait-jacket kind of pyjamas...with a zip down the back and a tie at the back so she can’t get out of them....I can’t relax because you always have to know what she’s doing and

where she is. Because if there's a silence [laughs], she's probably up to no good....She can't get out of her room at night because she would...wander....As she gets older...her poor sleep pattern seems to be improving...' (Practical, Positive outlook)

'Sleep's a big one....Broken sleep, years of it.'

'...she used to...take her nappy off....Stitching her pyjama top to the bottom and putting a zipper in to make all-in-one pyjamas. Problem solved....a giant-sized cot that fitted around her bed....Gates to prevent her from going in the kitchen....a quite set routine...it has helped [child] and it has settled her....and at school they have a routine and they're very big on routine....And I think managing just on a day-to-day basis, having a routine.' (Practical)

Feelings of frustration and anger were expressed, and concerns for other family members:

'...frustration...that the funding isn't available to offer the services that the Prader-Willi clinic should have....one of those is psych[iatric] support for the families, for the parents....for [child] to have psych input...that's not offered through the clinic....We have put a lot of effort...campaigning to get [a multidisciplinary clinic] but...the money's not there for it...' (Anger, Funding, Advocacy)

'She's dreadful to her father...and he's a very quiet man....life isn't that easy....he [father] gets quite morose...and I'm always the meat in the sandwich....Sometimes I'm leaving work and I think, 'will I turn right and come home or will I turn left and go down the hill or over the hill and far away.' (Detachment)

'...having to do the medication [at] the same time every day. In the morning and then...at night-time....It...does stress me out...'

'Siblings....it's ongoing to see how our boys'...lives have been impacted and changed by their sister....they've never felt as though home is somewhere they could bring their mates....they see that she gets...100% of our attention and affection and time, and always has, so that must be hard...' (Effort)

There was one clear statement of Spiritual coping and the effect on the individual in question. Similarly to the respondents in Graham *et al.* (2001), this spiritual belief appeared to help this carer to deal effectively with the role.

'I've got peace in here [touches heart] because I've already been through a breakdown...that lasted for 3 years, 3½ years....the answer for me is...Jesus Christ....When I've got my focus on me, I'm not coping, I don't cope. The more I focus on me, the heavier the load becomes. When I start to focus on the Lord and what my daughter's needs are, then I cope and I can stay above it all. But once I get self-orientated, I just go down...' (Effort, Spiritual)

The Advocacy aspect was also apparent within this section of the interview as carers reflected on some of their successful strategies:

'...you have to be upfront with people, and I'm particularly good at that....I don't take any nonsense...we've learnt that you've got to ask questions...and if you don't agree with something, you actually say so.' (Advocacy)

'You just have to deal with things day to day, and be proactive as well....[child] has been doing school for parents since he was two....And I've been willing to travel...to get him, and then he did...kindy at...[location] twice a week.' (Effort, Advocacy)

Some carers spoke in positive terms about their lives with an individual with AS or PWS:

'...I've been lucky...my husband and I...we've got a really good, strong relationship...we've been able to share through it all the highs and the lows....and when you have a bad day, let it all out...to someone with a good ear....[child] used to have his poo parties in the middle of the night and decorate his bedroom and I used to sit there cleaning it all up, tears rolling down the face....It's part of your day and you just put up with it...but sometimes it just gets overwhelming....Lots of tears shed along the way.' (Positive outlook, Emotional, Effort)

'...when I look back to when I had [child]...that's an era when any child that was born...with any kind of disability, would have had the same if not worse experiences than I did....living out on a fairly isolated farm and...[DSC staff], she was the very first person who was supportive....it hasn't been a terrible, terrible experience because I've not allowed it to be....I refused to accept that institutions should even exist....I was very much an advocate of that...' (Positive outlook, Advocacy)

6.6 Chapter summary

As indicated from the above item summaries and illustrative carer statements, there were a number of issues, e.g., the initial diagnosis of their offspring's condition, which were stressful for more than 50% of carers. Importantly, even some situations not faced by many carers, such as 'dealing with legal professionals', often contributed to high stress levels for the few who had faced those particular issues. Other items, such as 'your feelings about the cause of your child's condition', were not stressful for the majority of carers.

A substantial amount of anger and frustration was expressed by the respondents. Many carers also felt that the demands on their time were ceaseless, and that there was insufficient time left to fulfil their own needs. A third common concern was the financial support and provision for their offspring with AS or PWS.

There was a diverse range of coping mechanisms, often referred to concurrently, expressed by respondents. However, there were insufficient participants to attempt to identify any correlation between the use of any specific type of coping strategy and the

level of stress reported for an issue. There were examples of carers utilising both 'resources' and 'perceptions' coping methods to deal with the same problem, indicative of the complexity of the relationships between these different types of coping strategy.

It appears that membership of a support group was perceived as beneficial for many respondents. They reported gaining both practical and emotional support from their peers, and an increased sense of belonging. In particular, the carers <50yr valued interaction with other people living the same experiences as themselves, and felt encouraged by the support from other family carers.

Chapter 7 Discussion

7.1 Introduction

The research project undertaken as part of this thesis was designed to assess family stress levels and the use of coping strategies among the carers of people with an intellectual disability caused by Angelman syndrome or Prader-Willi syndrome and to examine the strength of the Double ABCX stress and coping model to interpret these data. Information was collected using a range of postal questionnaires and by face-to-face interview with family carers. There was widespread variation in the responses given by the family carers who took part in this study with regard to levels of stress and/or satisfaction, degrees of ‘normality’, and the use of coping strategies. However, the relatively high scores on the FSCI Scale indicated that many carers experience considerable stress, and have done so for extended periods of time. This Chapter will examine these results in relation to the original research questions, and also discuss some of the limitations and implications of the study

Throughout this chapter, it must be kept in mind that the limited sample size means that these data cannot be generalised to other people with IDD or, indeed, other individuals with AS and PWS.

7.2 Consideration of the research questions

7.2.1 How much care is required during different life stages (infancy, adolescence and adulthood) for persons with AS or PWS?

As hypothesised, all eleven children and most of the adults with AS or PWS lived in the family home. Two of the four families participating in the interview only mentioned that their adult offspring with AS/PWS was not living in the family home; however these comments could not be incorporated into the survey section of the study. Within the interview texts every family referred to visits to doctors, and a substantial number of the people with AS/PWS had been in hospital (7/19), often more than once. In addition, references were made to a variety of medical specialists: neurologists, paediatricians, endocrinologists, and psychologists among them. It appears that individuals with AS/PWS require considerable amounts of medical care, as previously determined in a comprehensive project using linked hospital data (Thomson, *et al.*, 2006a; b; Thomson, *et al.*, 2007).

There was one adult whose parent spoke of as working, making this particular individual the only member of the AS/PWS group currently employed. It is uncertain if this result reflected the poorer intellectual function of most people with AS, or if there were insufficient supported employment opportunities for people with IDD in WA. A third alternative, supported in some measure by the words of family carers, was that the parents themselves may not feel that employment is appropriate for their offspring:

'We knew that for a long time, for a long time, that he just doesn't have the tolerance or patience to sit there...'

'He just can't, he doesn't follow instructions. He doesn't, yeah, he just can't do it.'

The amount of time carers reportedly spent engaged in direct care of their offspring with AS or PWS was high, with some young children reported as needing 80 hours of active care per week. There were several examples of carers reporting that six to seven hours a day were spent in the active care of a child attending school five days per week. These care activities variously included dressing, feeding, transport, personal hygiene, and home-based therapy.

Based on these findings, it is possible that the wording of the survey question: 'How many hours per week do you, the primary caregiver, spend actively caring for this individual?' may have been perceived as encompassing emotional as well as physical care. The process of supervising the child's activities could also be construed as active care. An alternate phrase, 'personal care assistance' (Banks, 2003; Mona, 2003, p. 213), may have better described the required concept and its use in the current study could have eliminated any differences in interpretation. It is unclear why the pre-study discussion groups did not give any indication of this ambiguity. Additionally, use of a specific scale, such as the Functional Independence Measure for children (Msall, *et al.*, 1994), the Barthel Index, or the Functional Independence Measure (Kidd, *et al.*, 1995) may have more accurately assessed the care needs of the affected individuals and the extent of care duties undertaken by the carer.

Regardless of any possible misinterpretation, it is clear that a substantial amount of time and energy is spent by family carers in the day-to-day care of their offspring, especially younger children, a conclusion which matches the findings of Haveman, *et al.* (1997) and Edwards, *et al.* (2008). However, while there was a tendency for carers >50yr to spend the smallest amount of time on direct care of their adult offspring, they

also tended to report greater levels of stress for certain items on the FSCI Scale. These items especially pertained to time and caring issues: meeting the needs of other offspring, a spouse and/or oneself, maintaining personal friendships, day-to-day assistance with care of the offspring, and time apart from their child or adult with AS/PWS. Two of these problems, meeting personal needs and time apart from their offspring, were also areas of concern for older family carers in a study by Minnes and Woodford (2004). Other studies have reported that a number of supports, notably instrumental support from the family and friends, tend to decrease in frequency as the individual with IDD grows older (MacDonald & Callery, 2007), leaving the carer responsible for more of the care needs of the person.

Parental attitudes to a child's disability are variable over time and across developmental stages. Very young children are rarely expected to exhibit certain behaviours, such as helping others or showing empathy (Beck, *et al.*, 2004), therefore carers may not perceive atypical social skills development in their child until comparisons are made with typically developing children during the early school years (Neece & Baker, 2008). In support of this contention, the carers of very young children in the current study generally reported lower stress levels than did family members who were caring for older individuals, indicating that while this source of stress may be low for parent carers at this developmental age, other sources of stress are present for these carers. The attitudes of others may also change as the individual with AS or PWS reaches adolescence and adulthood:

“When he was younger, he was cute, and some of the things were tolerated. A lot of the stuff he does now, behaviour-wise, as an adult or an older child, they aren't as tolerant.”

It has been suggested that deviation by their offspring from the normative developmental trajectory can cause parents to feel 'labelled' or stigmatised (Gray, 1993; Rosenthal, *et al.*, 2001; Gray, 2002). Carers may become increasingly affected by the unremitting nature of their role, and experience feelings of isolation from mainstream society (Seltzer, *et al.*, 1995). Indeed, several carers in the present study spoke of the amount of effort necessary to achieve the best outcomes for their child, and of feeling disconnected from families with typically developing children. For example, this situation was described as:

‘...that's a direct impact of having a child with a disability. It isolates you socially.’

7.2.2 What stressors (aA in the Double ABCX model) commonly affect the family carers of people with AS or PWS?

Time pressures and time constraints were common features of the interviews, often relating to either the requirements of caring for their offspring with AS/PWS, or to the needs of other family members. As reported earlier (5.4.1.1), the amount of time family members spent caring for a person with AS/PWS varied considerably, although the younger children generally required more care than adolescents and adults with AS/PWS.

Contrary to the report by Murphy *et al.* (2007) and in support of the findings of Chen *et al.* (2001), family carers in the present study reported only minor health problems that were directly associated with their role. Two carers >50yr, however, had undergone an unspecified 'breakdown' at an earlier period, and among the carers >50yr there was mention within the interviews of personal physical limitations as their child matured into adulthood and increased in body size and/or became less mobile. Previous studies have suggested that some degree of health disadvantage is experienced by family carers, perhaps as a consequence of reduced socio-economic standing (McConnell & Llewellyn, 2006), or as an effect of behaviour problems in their offspring (Eisenhower, *et al.*, 2009). Where financial constraints exist, they may incline carers to forgo personal health care in favour of their child with IDD or that of other children in the family (Altman, *et al.*, 1999). Within the present study group, there were references by some carers to personal financial difficulties; however, these factors did not appear to adversely affect carer health ratings.

7.2.3 What resources and supports (bB in the Double ABCX model) are utilised by the family carers of people with AS or PWS?

Most of the families interviewed belonged to a support group, generally a syndrome-specific one. Almost all of these carers felt that the group contributed emotional support, created a sense of belonging, and enabled sharing of both feelings and knowledge with regard to their offspring's condition, as suggested previously (Solomon, *et al.*, 2001; Hale, *et al.*, 2005). In view of the small numbers within WA who are members of the respective support groups, it could be predicted that there will be a wider range of reactions to group membership as the respective Associations grow and develop.

None of the children with AS younger than five years old had used respite care, compared to more than half of the individuals with AS/PWS older than five. There is very little expectation in general society that typically developing young children will be sent to camps, or to spend weekends with some other carer, so it is unlikely that similar parents in this study would do it either: as one mother of a young child said: ‘...I’ve never felt the need to do it yet.’ However, one mother with experience working in the disability sector placed her child in respite at 18months old because:

“...I knew that as soon as she started it would be better and easier for her and me.”

Some carers spoke of using babysitters without recognising that this constituted respite. This may be a reflection of the perceived normality of using babysitters for children up to a certain age, with other family members often taking the babysitter role: ‘My Dad’s here, he helps me...’ Even among the families with an older child or adult with AS/PWS there were some who did not see the need for respite, perhaps relying also on family members, but also some who would have liked some form of respite but did not have it available: ‘It [time away from her] just doesn’t happen.’ Contrary to a previous report (Burton-Smith, *et al.*, 2009) there was no difference in the present study in the use of respite care when compared by total stress score ($\rho=0.4438$).

Family carers in the current study found maintaining personal friendships relatively stress-free, but respondents identified their current group of friends as predominantly parents who were living under similar situations. Feelings of isolation were rarely expressed, although the carers who currently lived, or had formerly lived, in rural areas spoke of feeling literally disconnected from people with similar experiences. There have been other reports of families caring for a child with IDD functioning poorly when members felt isolated from ‘normal’ society (Rehm & Bradley, 2005). Some carers >50yr in the present study also spoke of feelings of loneliness dating from many years ago, when it seemed to them that few people knew of the problems that they faced and even medical professionals had very little knowledge of the disorders and of the particular needs of the individuals with AS or PWS.

7.2.4 How much satisfaction (cC in the Double ABCX model) do family carers of people with AS or PWS get from the role?

The overall scores for the Carer Satisfaction Scale in this study were moderately high, suggesting that many carers recognized positive facets to their role, similarly to some previous reports (van den Borne, *et al.*, 1999; Hastings, *et al.*, 2002; Rapanaro, *et al.*, 2008). Indeed, some family carers made positive comments about their offspring with AS or PWS during the interviews:

“...she brings huge joys as well...”

“...we’ve got to love her so much.”

There were a small number of comments from the interviews indicative of satisfaction in the caring role.

“...something good did come out of it.”

‘That’s something that’s so important to convey, especially to the new mums, is the possibility of the joy and the good things that can come into it...’

The format of the FSCI may have influenced the expression of positive attitudes: basically the carers were asked to focus on the stresses involved and how they dealt with them, rather than on any satisfactions experienced. Some carers felt that caring did not improve their self-esteem, and more than a third indicated that their role failed to add meaning to their lives. This may demonstrate that a substantial number of family carers in this study were dissatisfied with their life role and circumstances, which could in turn have a bearing on their perceived stress levels.

7.2.5 What coping modes are commonly used by the family carers of people with AS or PWS?

As mentioned in Chapter 3, previous studies have identified a range of factors that predict successful coping in families with a child with IDD (Taanila, *et al.*, 2002a; Jones & Passey, 2004; Paczkowski & Baker, 2007). These characteristics include the use of a variety of coping strategies, adequate personal and couple time for parents, supportive friends and families, and feelings of self-efficacy. Similarly, parents of people with IDD in Western Australia reported that stressful situations could result in a new outlook for themselves, a form of reinterpretation, and strengthen their emotional and instrumental support networks (Rapanaro, *et al.*, 2008). However, other studies have found that the use of coping strategies has minimal effect on carer well-being

(Higgins, *et al.*, 2005; McConkey, *et al.*, 2008). The data in the current study do not support the theory that the use of coping strategies alleviates stress, as items with the highest mean stress level on the FSCI Scale also contained the most coping code instances, indicating a high level of coping strategy usage. However, many carers who provided a rating of 3 or 4 for the item regarding the initial diagnosis actually stated that use of Knowledge and Reinterpretation coping strategies reduced their stress levels. The high stress scores for this item, therefore, may have resulted from the style of the interview, allowing respondents to focus on their past, rather than current, stress levels: *'...you accept it very quickly...you change your whole thinking about it...'*, although long-term high stress associated with the diagnosis of a child with IDD has been reported previously (Todd & Shearn, 1996).

There was no clear pattern of coping strategy use when examined by stress score, offspring's age group, offspring's diagnosis, or carer age group. Similarly, Lopes *et al.* (2008) reported no difference in coping strategy use between people with a typically developing child and those caring for a child with IDD. The initial diagnosis of their child as having AS or PWS drew responses from carers indicating the use of strategies representing both resources and supports (Knowledge, Instrumental support), and attitudes and perceptions (Reinterpretation, Positive outlook). Carers referred to learning about the disorder and of adjusting their expectations for their offspring's future. This may reflect the ambiguity of the situation: the actual condition cannot be changed and therefore attitudinal change is required, but the process of diagnosis can be modified by the use of resources such as information. The receipt of adequate information, whether from professional or personal sources, has previously been identified as crucial to family adaptation to the stress of having a child with IDD (Taanila, *et al.*, 2002a). This gain is encapsulated in the words of one carer:

'...once we found out what it was and being able to read up on the literature and find out more about it...trying to work out what we were going to do with him and where we could take him...'

During the diagnostic period there was considerable use by families of professional and agency assistance, and of family support. Some carers also referred to positive feelings associated with the caring role, and spoke of the personal growth that resulted from learning to cope with adversity; attitudes that have been associated with more effective family function (Taanila, *et al.*, 2002a).

The strategy of Reinterpretation was associated with explaining to other people about the child's disorder. Carers spoke of tailoring their explanation to suit the circumstances; with a short version for casual meetings with other people, and longer and more complex explanations for people who are regularly in contact with the individual with AS/PWS. Reinterpretation was also commonly used when carers were deciding on the best level of integration for their offspring. An acceptance of the capabilities of, and the redefinition of goals for, their offspring featured prominently within this item.

‘...it would not have been appropriate for her to go there...’

When dealing with doctors and other health professionals, many carers depended on Knowledge as a form of support. In some cases, they felt they had a greater knowledge of the disorder than medical staff, and that an integral part of their role was to educate others about the health issues faced by individuals with AS or PWS. Similar attitudes have been reported previously from Canada (Minnes & Steiner, 2009).

There were two interview items in the current study in which Emotional support was especially important to carers: dealing with friends, family and neighbours, and maintaining satisfactory friendships for themselves. Family and friends play a vital role in reducing carers' feelings of isolation, allowing emotional release, and providing some semblance of normality in the lives of family carers (Taanila, *et al.*, 2002a; Ben-Zur, *et al.*, 2005; Guralnick, *et al.*, 2008). Thus families who were isolated from extended family contacts, either by distance or some other cause, tended to depend more strongly on friends and on membership of a support group for emotional and practical assistance.

Another form of emotional support for respondents was provided by membership of a syndrome-specific support group. In common with the findings of others (Rosenthal, *et al.*, 2001; Solomon, *et al.*, 2001; Hale, *et al.*, 2005), many family carers said that being part of the group allowed them to feel connected to their peers, and that they had benefitted from the experience and knowledge of other group members.

‘...gives you a sense of belonging, that you're not on your own, that there are others living that life...’

The item relating to time apart from their child produced varied responses from carers in the current study. Younger carers appeared less likely to desire time apart from their child, while older carers seemed much more inclined to feel the need for

respite, both for their own well-being, and that of their child. These differential attitudes are related to the concept of normality; the families of younger children, even typically developing children, have lesser expectations of being able to spend time apart from their child than do the families of older children and adults. The use of respite (Instrumental support) has been associated with reduced stress in family carers, and can provide greater opportunities for an individual with IDD to engage in community-based activities (Caldwell & Heller, 2003; 2007; MacDonald, *et al.*, 2007; McConkey, *et al.*, 2010). In the current study, carers who had access to respite care recognised the benefits this type of support offered their offspring as well as themselves, as well as articulating the problems associated with gaining access to adequate respite, as also reported from Canada (Doig, *et al.*, 2009).

‘...it’s just respite, recreational I guess respite for him because he just loves being around anybody...’

Making the decision about accommodation in the home or in the community was a Planning issue, as was finding a work placement or employment for the individual with AS/PWS. Unlike older parents in Taiwan (Chou, *et al.*, 2009), many carers in the present study had devoted considerable thought into future plans for their child with AS or PWS, even though many also acknowledged that there was insufficient provision at present for the accommodation of individuals with IDD in the community.

With the exception of Spiritual support, Detachment was used as a coping strategy less often than all others in the current study. Many of the instances of this strategy from the interview texts involved the use of distractions, such as work or recreation, rather than avoidance of the situation. The relatively high employment rate among the mothers in this study may reflect the use of work as a distraction, proving a degree of respite from the caring role. This is consistent with the findings of Grant and Whittell (2000) who reported that a majority of their study group described ‘enjoying personal time’, and ‘taking their mind off a problem’ as two effective coping methods.

There were no instances identified in the interview texts of some of the strategies generally allocated to Emotion-focus coping by other authors: i.e., distancing, wishful thinking, denial, venting, and avoidance. It has previously been noted (Hastings, *et al.*, 2005b; Glass, *et al.*, 2009) that use of avoidance coping, i.e., giving up trying to deal with problems or admitting to one’s inability to cope, can be associated with increased, rather than decreased, distress. Similarly, denial, distancing, and

wishful thinking have been correlated with poorer adaptation (Schnider, *et al.*, 2007; Glass, *et al.*, 2009). The self-selective recruitment method for the current study may have failed to include individuals who commonly use avoidance and/or denial coping methods. If use of these strategies is indeed conducive to high stress (Glass, *et al.*, 2009), then people who adopt them with greatest frequency are likely to be more overwhelmed by their caring role and therefore may decline to participate in a study such as the present research.

According to Grant and Whittell (2000), successful coping depends on a combination of personal growth, a structured routine, a close confidant(e), priority-setting, and the availability of a range of coping strategies. During development of the Family Life Interview, Llewellyn *et al.* (2010a) stressed the adaptive value of a sustained family routine, while noting that ‘frequent, unheralded or uncontrollable change places a strain on family routines’ (Llewellyn, *et al.*, 2010a, p. 53). Family carers of individuals with AS and PWS are often faced with unexpected events, perhaps relating to health or behavioural issues, and there may be little that can be done to control these situations. Probably for this reason, many respondents in the current project found it difficult to maintain their preferred routine. The people who spoke most often of routine or order reported generally lower stress levels than carers who did not mention any use of these techniques.

7.2.6 How much stress (xX in the Double ABCX model) is reported by the family carers of people with AS or PWS?

Overall, the level of stress related to caring for a child or adult with AS/PWS was high, as has been reported in other studies (Hodapp, *et al.*, 1997; Lopes, *et al.*, 2008; Wulffaert, *et al.*, 2010; Griffith, *et al.*, 2011), even when compared to the carers of individuals with IDD with a range of causalities. However, contrary to the carers of people with IDD from various causes surveyed by Nachshen *et al.* (2003) using the FSCI, people in the present study caring for an adult with AS/PWS were likely to report higher stress levels than their younger counterparts.

There were only two individuals with PWS with good speech skills in the study cohort; therefore it was not possible to compare parental stress by that ability. The small number of participants meant that the hypothesis relating to child characteristics could not be assessed.

Throughout the interviews, many family carers expressed anger and frustration toward the topics raised and in particular their recall of personal experience. Previous studies have suggested that chronic stress, such as experienced by family carers of people with IDD, is a common source of anger, and in turn that anger may compound the perceived stress (Neece & Baker, 2008; Benson & Karlof, 2009).

Much of the anger expressed in the present study originated in, and was associated with, dealing with service organisations and government agencies, reminiscent of the parents in another Western Australian study (Rapanaro, *et al.*, 2008). Carers spoke of the frustration involved in having to repeat information and processes owing to the lack of a 'box' for permanent disability (Chapter 6.5.3.2). With respect to therapy services, medical personnel, and agency staff, some carers expressed dissatisfaction with the high turnover rate of staff and the associated inconsistency of service provision (Chapter 6.5.3.2). These experiences were similar to those described by family carers from Canada (Gill & Renwick, 2007).

There was also a perception that some professionals were poorly informed about the disorders, and therefore failed to offer optimum treatment, a concern which has also been identified in studies conducted outside Australia (McGill, *et al.*, 2006a; Wodehouse & McGill, 2009). Some strategies were described as attempts to improve this situation, an example being the attachment by carers of a copy of the relevant support association information booklet to their child's file in the doctor's surgery and the hospital (Chapter 6.5.13.5). In common with some previous reports (e.g., Rehm & Bradley, 2005), most carers (14/19) had opted to act as strong advocates for their offspring as expressed by:

'...I'm inclined to stand up for our son's rights....I will stand up for him....no-one can look after him like I can...'

A substantial proportion of family carers felt that their own personal needs were the most difficult to schedule or meet. In common with earlier reports, finding time was an almost universal problem (Curran, *et al.*, 2001; Brandon, 2007; Murphy, *et al.*, 2007), and several people stated very clearly that their own needs were regarded as being lowest on the list of things that needed to be undertaken (Chapter 6.5.2).

'...there is no time. You're just down at the bottom when you can fit yourself in...'

Younger carers appeared more likely to feel that they had sufficient time to fulfil their own needs, even in light of their child's extensive care requirements. Their expectation of personal time, however, may be lower than that of older parents, and therefore any time when not directly involved in caring for their child may be looked upon by younger carers as an agreeable extra. Another factor involved in meeting personal needs was the fatigue expressed by many carers, especially parents whose offspring had AS. The detrimental effects of sleep deprivation on the mental and physical well-being of carers may be compounded by the necessity to concurrently care for a partner and/or other children (Richdale, *et al.*, 2000; Bayer, *et al.*, 2007).

Issues arising from the financial assistance necessary for the care of their offspring with AS/PWS could be sub-divided into two main categories: the availability of sufficient funding, and the processes involved in accessing the funds available. A number of family carers expressed the opinion that the level of funding allocated by 'the government' was insufficient to meet the needs of the many families requiring financial support.

'...at the end of the day the money's not there for it.'

The disability pension, which was the main source of financial assistance for older people with AS/PWS, did not always adequately cover the outlay required to maintain and care for them in the manner their family wished. However, families were often proud of their ability to use the money they received to best effect, and a number of carers handled all financial matters on behalf of their offspring. Consumer-directed funding may be a better option for these carers, as previous studies have found that family carers generally are good custodians of their child's finances (Caldwell, 2007). This option, known within Australia as individualised funding, is becoming more common. Although the amount of funds available is generally no more than before, the money does not need to be spent within the existing disability support system, and the costs of case management and administration are often much reduced (Laragy & Ottmann, 2011), leaving proportionally more available for purchasing services.

There have been discussions within Australia to introduce a National Disability Insurance Scheme (NDIS) to be funded by all taxpayers to assist people with disabilities and their family carers. The aim would be to '...provide funding for essential care, support, therapy, aids, equipment, home modifications and access to the community, education and training.' (National Disability Insurance Scheme, 2009). An initiative of

this nature has the potential to give the people with disabilities and their carers more control over their future, and to reduce or eliminate the crisis-driven funding model currently in place. Considerable work needs to be undergone before the NDIS becomes a reality, and among the issues to be examined is the level of co-ordination of service provision needed to ensure that adequate measures are in place before its initiation.

Within the current study, and in common with many reports worldwide (Woodford, 1998; Mansell, *et al.*, 2002; Gill & Renwick, 2007; Davys & Haigh, 2008; Eley, *et al.*, 2009b), funding for the provision of residential accommodation was perceived to be inadequate. In 2007/2008 there were 3,493 individuals in Western Australia accessing accommodation support, representing a small increase from 3,453 in 2004/2005 (Disability Services Commission, 2008). Some of the participant families had been waiting for an accommodation place for their child and they felt that insufficient capital funding was made available for supported housing.

‘...waiting for funding and facilities to be found for him, that could be forever and a day....we keep putting in for the funding...’

There was a general assumption that placement would be available only when the family had reached crisis point, a view not confined to Western Australia. The following excerpt is taken from a Four Corners documentary, ‘Breaking Point’, screened by the Australian Broadcasting Commission (ABC) on February 16th, 2010 (Cronau, 2010). Dick Jones is the father of an adult with multiple disabilities and Wendy Carlisle is the interviewer:

WENDY CARLISLE (to Dick Jones): If you acted on your desire, which is for Robbie to be in a home now, in a house somewhere, what would you have to do for that to happen?

DICK JONES: Leave him at the doorstep I suppose of a respite care centre.

WENDY CARLISLE (to Dick Jones): It'd have to come to that?

DICK JONES: Oh I think it'd have to come to that. Or we'd have to die. But that's pretty tragic in one sense, but that we were told sometimes would be the only option you have is when you're in a wooden box. That'll probably be the only time that these types like Rob will get any accommodation.

The extensive processes necessary to access many services also acted as sources of considerable stress for participants in the current study. Similar experiences have been reported elsewhere: e.g., ‘endless mounds of redundant paperwork, meetings, appointments and visits to the doctor to complete forms in order to qualify’ (Caldwell,

2007, p. 556). Respondents in the present study spoke of the frustration of trying to meet the often stringent and complex rules of entitlement for services and funding, experiences that were echoed in the investigations reported by Redmond and Richardson in Ireland (2003) and Doig, McLennan and Urichuk in Canada (2009).

The concept of labelling was briefly discussed in Chapter 6. In some instances the term was applied to the child, and in others the carer was the person who felt labelled. For some parents, the labelling of their child as having AS or PWS was initially very distressing. Carers experienced feelings of grief and disappointment on realising that their child was unlikely to have a 'normal' life (McConkey, *et al.*, 2008).

'...just the realisation that what you perceive as normal is not or no longer will apply, and that your dreams and hopes for your child's future...have all been turned upside down...'

There generally followed a period of readjustment of parental expectations and goals for their child and, ultimately, of acceptance. Indeed, in confirmation of other studies, many carers reported a reduction in perceived stress once a clear diagnosis was provided to them (Lenhard, *et al.*, 2005; Skotko, 2005; Graungaard & Skov, 2007).

Thus the term 'label', in the sense of diagnosis, was generally viewed as a positive force in acting to ameliorate stress. Family carers tended to feel more empowered once a definite causality for their child's disorder had been established, as it allowed them to find out more about the condition and to determine a likely prognosis. One carer stated the value of such knowledge quite explicitly:

'...I guess once we got some knowledge, that became...a bit of a weapon against the stress...'

Information given to family carers at the time of diagnosis was occasionally felt to be inadequate, especially 15-30 years ago, either because so little was known about either AS or PWS at that time, or because the consulting medical professional had no experience of the particular syndrome. The provision of comprehensible and correct information at the time of diagnosis is considered an important component of the adjustment process for family carers (Skotko, 2005; Graungaard & Skov, 2007; Kenny & McGilloway, 2007). It is also of vital importance to convey a balanced picture of possible futures: too often in the past carers have been given worst-case scenarios with positive or hopeful messages omitted (Harnett, *et al.*, 2009).

‘...it was basically...if you’ve got an Angelman Syndrome child they won’t be able to do this, and they won’t...and lots of won’ts, won’ts, won’ts.’

Referred stigma, where the carer or family member feels labelled, has previously been named ‘courtesy’ stigma (Gray, 1993; 2002), and may be ‘enacted’, i.e., rude staring and comments from other people, or ‘felt’, which involves embarrassment or other emotions on the part of the carer (Gray, 2002, p. 737). There were several references to both types of stigma within the interview texts, and this type of ‘labelling’ caused considerable distress to the individuals involved.

‘...there’s girls that I’ve gone all through playgroup with...that now cross the street when they see me coming. And I thought ‘what have I done to upset them?’

Another possible source of carer stress was the food-related behaviours exhibited by children with either syndrome. Given their abnormal responses to food, it was expected that individuals with PWS would score highly on the FRPQ (Russell & Oliver, 2003), but the scores for people with AS, once they had been adjusted for the items which required verbal ability and/or mobility, were also high. Almost all family carers in the current study had to cope with some degree of problem behaviour associated with access to food; with a number of carers remarking on the necessity to keep strict control of their offspring’s eating habits. These types of behavioural problems have been similarly associated with high stress levels in other studies involving the parents of individuals with AS/PWS (Hodapp, *et al.*, 1997; Wulffaert, *et al.*, 2010).

7.3 *Does the Double ABCX model represent the findings of the study?*

Throughout the study it has been clear that the lives of family carers contain many, if not all, of the features of the Double ABCX model. They are regularly exposed to a variety of stressors which they perceive in a variety of ways. A range of supports and resources are utilised and many different coping strategies described. However, it was not feasible to assess the relevance of the Double ABCX model of stress and coping given the present data. The small number of participants precluded complex statistical analysis of the moderating and/or mediating effects of supports, resources, perceptions, attitudes, and coping strategies on the adaptation by family carers to stressful events associated with having offspring with AS or PWS.

It was considered that the small sample size and non-representativeness of the study group precluded any meaningful comparison with people caring for offspring with AS/PWS, offspring with IDD from other causes, or the general population, hence no recommendations have been made to inform policy or to influence practices in the support of family members caring for a individual with AS/PWS in Western Australia.

7.4 Potential limitations of the research and its findings

7.4.1 Problems with the recruitment process

The response rate for the study was lower than anticipated, with fewer than 25% of the families approached volunteering to participate. As was evident from the attempts of recruiting people for the focus groups, described in Section 4.7, recruiting participants via letters and/or newsletters was less effective than a more personal approach. Most of the participants (13/19) in the present study were recruited by means of a personal approach to the local Support Associations. However, the candidate was not permitted to contact people who were not members of the Support Associations in person, and so they had to be approached by means of invitation letters mailed by DSC and GSWA.

As noted by others, the use of gatekeepers or facilitators can result in recruitment being dependent on the interpretation by the gatekeeper of the inclusion and exclusion criteria for the project. In turn, this may lead to disconnection of members of the study team from the recruitment process (Scott, *et al.*, 2006), and at the same time it can inhibit follow-up of non-responders. Perhaps not surprisingly, people with IDD have been identified as a group that is particularly difficult to recruit into research studies (Evenhuis, *et al.*, 2004; Lennox, *et al.*, 2005; Cleaver, *et al.*, 2010). Some of these difficulties relate to consent issues, and others to procedural problems within service organisations (Evenhuis, *et al.*, 2004; Lennox, *et al.*, 2005). A recent review of studies involving individuals with IDD found that higher rates of recruitment were achieved when researchers had personal access to potential participants or to substitute-decision makers (Cleaver, *et al.*, 2010), and if the data collection was non-invasive. However, as mentioned previously (4.4.2.1.1), greater restrictions are being imposed by Human Research Ethics Committees with a view toward preventing invasions of privacy.

The introduction in Australia of the *Privacy Act 1988* has increased the likelihood of HRECs requiring researchers to use the opt-in method of participant recruitment (Trevena, *et al.*, 2006). This method of recruitment, which was adopted for the current study, has been shown to be significantly less productive than the opt-out alternative (Trevena, *et al.*, 2006). According to Junghans *et al.* (2005), opt-out is the more appropriate method of recruitment for studies involving a low risk to participants. Additionally, opt-in recruitment has been associated with varying degrees of selection bias (Edwards, *et al.*, 2002; Boynton, 2004; Hewison & Haines, 2006; Trevena, *et al.*, 2006).

There is a possibility that the final group of participants in the current study was not representative of all families with a child with AS or PWS. The proportion of family carers with tertiary qualifications (59.4%) was substantially greater than that found in the general population in Western Australia at that period in time (49.8%) (Australian Bureau of Statistics, 2009b). It could be argued that aspects of the tertiary education process promote interest and a willingness to participate in research. Comparatively, from a self-selected group of participants in a Canadian study, 69% of parents of a child with IDD ($n = 100$) and 76% of parents of a typically developing child ($n = 100$) had completed college or university education (Nachshen & Minnes, 2005). The participants in the current study may have been more highly motivated and less overwhelmed by the caring role than were individuals who did not respond to the invitations. The mothers who volunteered to participate in the current study were more likely to be in full- or part-time employment than has commonly been reported for similar groups (Olsson & Hwang, 2006; MacDonald, *et al.*, 2010). This elevated employment rate may itself be indicative of and be related to high levels of educational attainment within the study group.

There was under-representation in the study population of people from rural and remote areas of Western Australia, and of people from both indigenous and culturally and linguistically diverse backgrounds. In 2008 more than one quarter of the Western Australian population had been born overseas (Office of Multicultural Interests, 2008), and one quarter of all residents lived outside the Perth metropolitan area (Australian Bureau of Statistics, 2009c). However, all but two individuals with AS or PWS in the current study were of European descent, and only a single participating family lived in rural Western Australia. It is probably significant that the carers of both non-European

families, and of the rural family, specifically referred to the lack of family support available to them.

Another major issue affecting recruitment was the requirement for a legal guardian to act as proxy to consent on behalf of any adult with IDD judged incapable of providing self-consent. Some carers were surprised to be told they were no longer the legal guardian of their older offspring, and one couple had even had the State Administrative Tribunal rule that it was unnecessary for them to be awarded guardianship for their adult child. Iacono and Murray (2003) successfully applied to the State Administrative Tribunal in Victoria, Australia for permission for the Public Advocate or some other person to act as proxy to provide consent on behalf of older individuals with IDD. However, this option was both too time-consuming and too expensive for adoption in the current project, which as part of a PhD study was subject to stringent time constraints.

As previously indicated, the limited number of eligible participants had repercussions for the scientific validity of the study, with insufficient statistical power to analyse correlations between the physical, medical or behavioural features of the people with AS or PWS and the level of stress reported by their family carer. It was also judged impracticable to collect linked health services usage data pertaining to individuals with AS or PWS, because of the potential breach of ethics guidelines that could occur due to the identification of study participants within the quite restricted population of Western Australia.

One intention of this project was to present the views of a specific group of people: the family carers of individuals with AS or PWS. This goal has been affected to some degree by regulations designed to protect personal privacy. The people who were excluded from full participation in this study (6/22) were thus denied the opportunity to provide data leading to a better understanding of the needs of their offspring. Logically, partial and unrepresentative data cannot provide a sufficiently clear picture of the needs of this potentially vulnerable population.

A further consequence of the small number of participants was related to the process of data management. In order to preclude ready identification of the responding individuals by the researcher, the code-holder recruited for the study was unable to release hard copies of completed questionnaires until a sufficient number had been

returned. This significantly slowed the data processing and interview schedules. In terms of the typical duration of post-graduate study and other time-limited projects, these delays are particularly troubling and may jeopardise the continuity of research.

A preferred method of increasing participation rates would have been to send the questionnaires and consent forms directly to all potential participants (Edwards, *et al.*, 2009). However, this option was difficult to organize as specific families required different combinations of consent forms, and pre-set budgetary constraints did not cover the printing and posting in excess of 100 sets of questionnaires. It may have been effective to provide some incentive, such as a Scratch-and-Win (instant lottery) ticket or a small amount of money (e.g., \$2.00AU), to encourage family carers to volunteer to participate in the study, although this would again have been a more expensive option, and its adoption would have required special HREC approval.

7.4.2 Ethics procedures

One major adverse issue in gaining the requisite multiple ethics committee approvals was the sequential nature of the process, the ramifications of which were indicated in Table 4.1. The requirement for approval from one committee before forwarding the application to the next caused the procedure to be incompatible with the time-constraints associated with post-graduate study. In addition, amendments required by later HRECs necessitated ratification by all other committees, requiring a circuitous process that consumed even more time. Concurrent applications, followed by discussion between the HRECs to decide on any issues needing resolution, could have shortened the time required to gain ethics approval, without in any way interfering with or endangering the integrity of the overall process.

The three different ethics committees directly involved in the current study also had varied application forms and information requirements which added considerably to the time spent obtaining their separate approvals. Each HREC has its own annual reporting regime, with different formats and diverse time-frames. The National Ethics Application Form (NEAF) developed in Australia has been specifically designed to eliminate the need for completing applications in a variety of formats, and to facilitate multi-centre research (National Health and Medical Research Council, 2008). Some Australian states are using the NEAF as a tool for ethics assessment of multi-centre

research projects (NHMRC, 2010). However, to date there appears to be very limited acceptance of the NEAF among HRECs in Western Australia.

Internationally, there have also been moves to streamline multi-centre ethics approvals (Lux, *et al.*, 2000; Tully, *et al.*, 2000; Dalton & McVilly, 2004; Mayor, 2005; Blustein, *et al.*, 2007), with varying degrees of success. For example, in the UK, a study already approved by a multi-centre research ethics committee was presented to a number of local research ethics committees. Fewer than half were dealt with at executive level, as is recommended, and only one in ten committees required some alteration or clarification before granting approval. Nonetheless, a number of committees took a minimum of three months to process the applications (Lux, *et al.*, 2000; see also Tully, *et al.*, 2000). Some of these difficulties have since been formally identified, with recommendations for improvement of the multi-centric approval procedure transmitted to the UK National Patient Safety Agency (Mayor, 2005).

7.4.3 Socio-political events

The data collected for this study represent a snapshot in time. Questionnaires were completed and participants interviewed between September 2006 and December 2008. Since that period it seems probable that international economic events, such as the Global Financial Crisis, may have had significant effects on families with limited resources, including many of those caring for a child with IDD. In addition, there were changes of Government in Australia within the relevant time period, at both Federal and state levels. It is not clear what level of importance the two major political parties in Australia place on the support of people with disabilities and their family carers. In Western Australia, for example, seven different Ministers were responsible for Disability Services over the period 2001-2010, with five individuals holding the portfolio over a period of just 14 months. It could be interpreted that Ministerial turnover on this scale is indicative of relatively low priority being placed on the Disability Services portfolio by successive state Governments.

7.5 Questions arising from the study

7.5.1 Progress in research

Over the half century since the initial description of AS and PWS, research dedicated to understanding the causes, clinical profiles and behavioural patterns of these

conditions has continued. Beginning in the 1990s, considerable effort has been devoted to assessing the effects of growth hormone therapy on a number of parameters in people with PWS, including height, body composition, pulmonary function, and quality of life (Höybye, 2007; Myers, *et al.*, 2007; de Lind van Wijngaarden, *et al.*, 2009; Carrel, *et al.*, 2010). These effects can all contribute to improved long-term health outcomes for individuals with PWS and may result in an increase in the life expectancy for this population.

Research findings can provide information to family members of people with PWS regarding issues likely to become problems over the life span, such as an increase in aggressive behaviour during young adulthood (Ho & Dimitropoulos, 2010), a high incidence of musculoskeletal disorders (Shim, *et al.*, 2010), or damaging skin-picking behaviour associated with anxiety or boredom (Morgan, *et al.*, 2010). Carers then have the opportunity to prepare strategies designed to reduce the impact of these problems, prior to their development.

Mouse models of AS are available to researchers, and the neurological deficits in such a mouse have been reported to respond to treatment (van Woerden, *et al.*, 2007). However, it has not yet been shown that these advances can be applied to humans with AS. Augmentative and alternative communication (AAC) systems have been developed which offer hope to the parents of individuals with AS, being a means by which their offspring who lack speech ability can achieve greater autonomy and personal choice (Calculator & Black, 2010). There are also some indications the ketogenic diet may assist in the control of intractable epilepsy (Beniczky, *et al.*, 2010), and this may be a useful option for the carers of people with AS and poorly controlled seizures.

7.5.2 Concerns of carers

There were a number of specific areas in which concern was expressed by family carers. See, for example, Chapter 6.5.22.3 and Chapter 6.5.25.4. Unfortunately, it was not possible to pursue these concerns within the parameters of the current study. They may, however, represent issues affecting them in their daily roles and are potential research pathways to assist family carers to fulfil their role more effectively.

Several families referred to the impact on their other offspring of having a sibling with AS or PWS. This area is largely under-researched; although there has been some discussion on similar issues in respect to other forms of IDD (Orsmond & Seltzer,

2000; Dykens, 2005; Knox & Bigby, 2007; Mulroy, *et al.*, 2007; Canary, 2008; Gold, *et al.*, 2008). It seems probable that specific matters related to the food behaviours of individuals with PWS will affect their brothers or sisters. Siblings may also be reluctant to bring friends or potential partners back to the family home, or they may experience taunting at school as a result of having a sister or brother with AS or PWS.

Family carers also expressed concerns regarding the post-school environment of their offspring with AS or PWS. For more than a decade, researchers have been investigating transitions: from youth to adulthood, from school to employment, and increasingly from middle age to old age, that are experienced by individuals with IDD (Blacher, 2001; Kraemer & Blacher, 2001; Rapanaro, *et al.*, 2008; Docherty & Reid, 2009). It therefore would be appropriate to explore whether any particular issues are faced by people with AS or PWS, and their carers, during the course of these transitions.

Few of the people with AS or PWS were engaged in open or supported employment, even though some carers expressed the hope that jobs would be available for their offspring at some point in the future. There is an extensive body of literature discussing employment in people with IDD, but very few of these studies appear to examine the experience of individuals with specific conditions, such as PWS or AS (Fillary & Pernice, 2005; Taanila, *et al.*, 2005; Hensel, *et al.*, 2007; Grant, 2008; Jahoda, *et al.*, 2008; Cramm, *et al.*, 2009).

7.5.3 Alternate methods

Researchers in Western Australia have the opportunity to access comprehensive health services usage data through the assortment of linked data bases that exist in the region (Holman, *et al.*, 1999). A detailed study of carer health utilisation data from state sources, such as the Hospital Morbidity Database and the Cancer Registry, in conjunction with Federal government records from the Pharmaceutical Benefit Scheme database and the Medicare database, would provide a more detailed profile of changes in carer health than was possible in the present study. Although dependent on adequate recruitment, this process could generate relevant and unique data on the health effects of caring for a person with IDD.

It could be argued that the FSCI Scale was not a suitable outcome measure for the present study. Some reports of the use of this instrument (Minnes & Nachshen,

1997; Woodford, 1998; Nachshen, *et al.*, 2003) have indicated that it is a good measure of perceived stress (an attitude or perception), while other groups have used the instrument as an outcome determinant (Lopes, *et al.*, 2008). An alternative approach to the measurement of carer adaptation could have been the adoption of instruments to measure one or more dimension of anxiety, depression or mental health, such as the Questionnaire on Resources and Stress (QRS-F: Friedrich, *et al.*, 1983), the Mental Health Inventory (MHI: Veit & Ware, 1983), the Beck Anxiety Index (Beck, *et al.*, 1988) or the Hospital Anxiety and Depression scale (HAD: Zigmond & Snaith, 1983).

One area that was not considered for the present study was quality of life (QoL). There are numerous reports of the effects on QoL of caring for a person with IDD (Park, *et al.*, 2003; Mactavish, *et al.*, 2007; Hu, *et al.*, 2011; Rillotta, *et al.*, 2011). There are also a number of instruments designed to measure QoL, such as the Family Quality of Life scale (Park, *et al.*, 2003; Hu, *et al.*, 2011). Future research into the families of people with AS and PWS could benefit greatly by including some measure of quality of life

Of necessity, the current study was cross-sectional in design. A longitudinal study, while more time-consuming and costly, could be used as a source of valuable data relating to changes of stress and coping use over time. A larger longitudinal study could encompass a broader subset of people, including people from linguistically and culturally diverse backgrounds, individuals with different conditions causing IDD, and a broader age group of both people with IDD and their family carers. Therefore it would be recommended that future studies of this or similar populations should include more representative sample groups.

7.6 Conclusion

Substantiated claims about the impact on family carers of having offspring with AS or PWS could not be made from the largely unrepresentative data collected from the study participants. The only explicit statement that can be made is that the family carers who participated in this research are under considerable stress, and in many cases, have been so for a number of years. Using the Double ABCX model of stress and coping to form the basis of the present study was of limited benefit, mostly due to the small number of participants. However, the utility of the model has been demonstrated in numerous studies and it will continue to form the basis of much research in this area.

Although the process of more inclusive recruitment for studies of this kind may be difficult to achieve, it is important that researchers continue to strive to improve recruitment methods, and to undertake research projects on behalf of this vulnerable section of society (Lennox, *et al.*, 2005). If the difficulties encountered in the recruitment of people with IDD leads to their exclusion from the research agenda, then they and their carers run the risk of becoming even more marginalized, their voices becoming fainter, and they effectively lose the right to participate in all aspects of society. Inclusion is an ideal for people with IDD: the scientific community has a responsibility to facilitate the realisation of that ideal by promoting rigorous, valid, and relevant social, medical, and health research incorporating the views of people with IDD, their families, and their carers.

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Appendix I List of journals sourced for the study

Acta Paediatrica
American Journal of Human Genetics
American Journal of Medical Genetics
American Journal on Mental Retardation
Annals of the New York Academy of Sciences
Archives of Disease in Childhood
Australian and New Zealand Journal of Public Health
British Journal of Learning Disabilities
British Medical Journal
Child: Care, Health and Development
Clinical Genetics
Current Opinion in Psychiatry
Developmental Medicine and Child Neurology
Disability & Society
European Journal of Human Genetics,
Family Relations
Human Molecular Genetics
Intellectual and Developmental Disabilities
International Journal of Disability, Development and Education
International Journal of Obesity
Journal of Applied Research in Intellectual Disabilities
Journal of Autism and Developmental Disorders
Journal of Child Psychology and Psychiatry
Journal of Clinical Endocrinology & Metabolism
Journal of Disability Policy Studies
Journal of Intellectual and Developmental Disability
Journal of Intellectual Disabilities
Journal of Intellectual Disability Research
Journal of Medical Genetics
Journal of Paediatrics and Child Health
Journal of Pediatrics
Journal of Personality and Social Psychology
Lancet
Mental Retardation
Mental Retardation & Developmental Disability Research
Nature Genetics
NCBI search alerts: Angelman, Prader-Willi, Carer stress
Pediatrics
Research in Developmental Disabilities

Appendix II Participant information sheet



Family- and community-based studies of major syndromes causing intellectual disability

INFORMATION SHEET

Why are we doing the study?

To make information available to service providers about the current and future needs of people with Prader-Willi syndrome or Angelman syndrome, and their family carers.

What will the study tell us?

What is involved in caring for a person with Prader-Willi syndrome or Angelman syndrome, how does it affect your health and well-being, and what supports and ways of coping are helpful? What are the medical needs of people with Prader-Willi syndrome or Angelman syndrome at various ages?

Who is carrying out the study?

Dr Peter Roberts and Ms Allyson Thomson from Edith Cowan University.

What you will be asked to do if you decide to take part?

There are some questionnaires to be completed (Consent Form 1). This should take around one hour.

You will also be asked if you are willing to be interviewed (Consent Form 2). The interview can be face-to-face or over the telephone, and will take 1-1½ hours. The time and place will be chosen to suit you.

Do I have to take part?

No. Involvement in either section of the study will be entirely voluntary on your part.

What about my privacy?

This is most important to us. Once the questionnaires and interviews have been completed, the names of all participants will be replaced by unique numbers. The interview tapes will be maintained under secure storage for up to 5 years before being destroyed. When the results are written down, there will be no names used.

Where is your information kept?

All of the paper records and tapes will be kept in a locked cabinet in a locked room at Edith Cowan University. The computer used is password-protected and also kept in the locked room.

Who has approved the study?

The Human Research Ethics Committee of Edith Cowan University, the Confidentiality in Health Information Committee, and the King Edward Memorial Hospital for Women Ethics Committee.

Who to contact for more information about this study:

If you would like any more information about this study, please do not hesitate to contact one of the research team. They are very happy to answer your questions.

Ms Allyson Thomson

Contact Number: 6304 5623 (wk)

Mobile: 0403 005673

Who to contact if you have any concerns about the organisation or running of the study?

If you have any concerns or complaints regarding this study, you can contact the Research Ethics Officer, Edith Cowan University. Your concerns will be drawn to the attention of the Ethics Committee who is monitoring the study.

Research Ethics Officer
Edith Cowan University
100 Joondalup Drive
JOONDALUP WA 6027
Phone: (08) 6304 2170
Fax: (08) 6304 2661
Email: research.ethics@ecu.edu.au

What to do next if you would like to take part in this research:

If you would like to take part in this research study, please read and sign the appropriate consent form/s provided.

THANK YOU FOR YOUR TIME

Appendix III Invitation letter from Disability Services Commission



Angelman syndrome and Prader-Willi syndrome: care needs and the effects of ageing

This is an invitation to take part in a study into the special needs of individuals with Prader-Willi syndrome or Angelman syndrome as they become older, and of the problems facing the carers of these people. It is now likely that many people with intellectual disability will live almost as long as members of the wider population. This project is aimed at investigating the effects of growing older on people with intellectual disability and their carers/families. We want to be informed on specific health, social and economic aspects of ID and advancing age, so that service providers and policy makers can access information about the future needs of people with ID and their carers.

Phase 1

We will be asking the primary carers of all people in Western Australia with Angelman or Prader-Willi syndromes to complete several questionnaires. The estimated time to complete the questionnaires is 1-1½ hours. The questionnaires are designed to give information on:

1. Family structure, living arrangements, and education or employment of the affected individual, and carer health and satisfaction.
2. Behaviour problems associated with food shown by the affected person.
3. Clinical presentation of the specific syndrome in each individual.

In addition, information on the use of health care services, such as General Practitioners, hospital admissions, and prescription drug usage will be collected on each person with either PWS or AS via electronic data linkage. This linkage is conducted by people in charge of the databases, and information is provided to us with only a unique number for identification. The completed database, which will not contain any names or addresses, will be held on a password-protected computer in the secure area of the Centre for Human Genetics, Edith Cowan University, Joondalup, and all paper materials will be kept in locked storage at the same place.

Phase 2

A representative selection of carers will be asked to agree to an interview. These interviews are intended to find out what stresses are involved in caring for an individual with AS or PWS, and what styles of coping are used to deal with these stresses. There is also a section devoted to attitudes to Support Groups. Interviews are expected to take between 1-1½ hours, and will be audio-taped to ensure accuracy in reporting. The time and venue for these interviews can be chosen to suit the carers.

At the completion of the interviews the tapes will be transcribed and the tapes destroyed. Names will not be included in the transcription, only an identifying number. None of the data will be identified by name in any report or publication, and only the investigators will know the names of any participants. All investigators are bound by a confidentiality agreement, and participants may withdraw from the study at any time, with no reason required. No risks have been identified as consequences of participation in this project.

If you wish to participate in Phase 1 of this project, please sign section 1 of the accompanying Informed Consent Document and return it with the completed questionnaires. If you are willing to participate in Phase 2, please indicate by signing the second section of the Consent Document. At any point you are welcome to contact Allyson Thomson to ask questions and seek further information.

Contact details:

Student: Allyson Thomson
Centre for Human Genetics
Edith Cowan University
100 Joondalup Drive
Perth 6027

Phone: 6304 5684
Fax: 6304 5717

Supervisor: Dr Peter Roberts
Department of Human Biology
Edith Cowan University
100 Joondalup Drive
Perth 6027
Phone: 6304 5455
Fax: 6304 5717

This research project is being undertaken as part of the requirements of a PhD degree at Edith Cowan University.

Faculty: Computing, Health and Science.

School: Exercise, Biomedical and Health Science.

This project has been approved by the Edith Cowan University Human Research Ethics Committee.

If you have any concerns or complaints about the research project and wish to talk to an independent person, you may contact:

Research Ethics Officer
Edith Cowan University
100 Joondalup Drive
JOONDALUP WA 6027
Phone: (08) 6304 2170
Fax: (08) 6304 2661
Email: research.ethics@ecu.edu.au

Appendix IV Invitation letter from Genetic Services WA

Date

Name

Address

Dear

I am writing to ask if you would be willing to be contacted by researchers (Dr Peter Roberts and Ms Allyson Thomson) who are undertaking a project on caring for a person with intellectual disability and wish to recruit suitable former patients from this Clinic. Patients from this Clinic are needed for this study and from our records you/your child would appear to be a potential subject for this study.

The project will be looking at issues related to caring for a person with Angelman syndrome or Prader-Willi syndrome, including sources of stress and methods of coping. The health and support needs of the person cared for will also be assessed. This information can be used to inform service providers of the major areas of need for people with the disorders and their carers.

The people the researchers are looking for to help them with this study need to be:

1. a family member of a person with Angelman syndrome or Prader-Willi syndrome
2. a carer of a person with Angelman syndrome or Prader-Willi syndrome

If you do not wish to hear more about this study, or be contacted further, could you please complete the attached slip and return it in the envelope provided. If we do not hear from you within 3 weeks we will assume you are willing to be contacted and the researcher (Ms Thomson) will contact you shortly after to give you more information regarding the study. Should you wish to have further information about the study before making a decision as to whether or not you wish to be contacted please telephone the researcher (Allyson Thomson: 6304 5623).

Whether or not you participate in this project, any future care you/your child receives at this Hospital will not be affected in any way.

This study has been approved by the King Edward Memorial Hospital for Women Ethics Committee, the Edith Cowan University Human Research Ethics Committee, and the Department of Health Confidentiality in Health Information Committee. The confidentiality of all participants is assured.

Thank you for considering this request.

Yours sincerely

**Director
Clinical Care Unit**

PLEASE TICK ONE BOX, THEN SIGN, DATE AND RETURN THE SLIP IN THE ENVELOPE PROVIDED

Please do not contact me regarding my participation in the 'Family- and community-based studies of major syndromes causing intellectual disability'.

OR

I would like further information on the 'Family- and community-based studies of major syndromes causing intellectual disability' so I can decide whether or not I will participate.

OR

I do not fit the criteria listed in the letter, but I am interested in hearing about future research studies.

Name: (Child's Name):

Signature: Date:

Appendix V Consent forms



Family- and community-based studies of major syndromes causing intellectual disability

Consent Form 1 [to be completed by the carer, questionnaires]

I have read the information letter provided and have had the opportunity to ask questions. Any questions I have asked have been answered to my satisfaction.

I understand that all information provided is treated as strictly confidential and will not be released by the investigator unless required to by law. I have been advised as to which data are being collected and for what purpose.

I am willing to complete the questionnaires that will be provided realising that I may withdraw at any time without given reason and without prejudice.

I agree that research data gathered for the study may be published, provided no name or other identifying information is used.

Participant:.....

Date:.....



Family- and community-based studies of major syndromes causing intellectual disability

Consent Form 2 [to be completed by the guardian or parent if person with Angelman or Prader-Willi syndrome is incompetent or a child]

I have read the information letter provided and have had the opportunity to ask questions. Any questions I have asked have been answered to my satisfaction.

I understand that all information provided is treated as strictly confidential and will not be released by the investigator unless required to by law. I have been advised as to which data are being collected and for what purpose.

I give permission for health data relating to (*person with Angelman or Prader-Willi syndrome*) to be forwarded to the investigator by the Data Linkage Unit from the following health datasets:

- Medicare benefit scheme: 1984-2006
- Pharmaceutical benefit scheme: 1990-2006
- Hospital morbidity database: 1970-2006
- Cancer registry: 1979-2006
- Mental health database: 1966-2006

I agree that research data gathered for the study may be published, provided no name or other identifying information is used.

Parent or guardian of the person with Angelman or Prader-Willi syndrome:.....

Date:.....



Family- and community-based studies of major syndromes causing intellectual disability

Consent Form 3 *[to be completed by the carer, interview]*

I have read the information letter provided and have had the opportunity to ask questions. Any questions I have asked have been answered to my satisfaction.

I understand that all information provided is treated as strictly confidential and will not be released by the investigator unless required to by law. I have been advised as to which data are being collected and for what purpose.

I am willing to participate in an interview, realising that I may withdraw at any time without given reason and without prejudice. I agree to the audio-taping of the interview, with the knowledge that the tapes will be stored securely after they have been typed up, and that the text will not contain any names.

I agree that research data gathered for the study may be published, provided no name or other identifying information is used.

.....

Participant

.....

Date



Family- and community-based studies of major syndromes causing intellectual disability

Consent Form 4 [to be completed by the affected person, if competent, and co-signed by a parent or guardian, if applicable]

I have read the information letter provided and have had the chance to ask questions. I have been happy with the answers to any questions I had.

I understand that everything found out is kept private and will not be spread around unless the law says. I have been told what things will be found out, and why the people want to know them.

I will/will not let a family or staff carer answer some questions about me, and understand that I may stop this at any time and for any reason.

I will/will not let my health information be sent to the investigator by the Data Linkage Unit from these datasets:

- Medicare benefit scheme: 1984-2006
- Pharmaceutical benefit scheme: 1990-2006
- Hospital morbidity database: 1970-2006
- Cancer registry: 1979-2006
- Mental health database: 1966-2006

I agree that the facts collected for the study may be printed, as long as no-one can be identified.

Person with Angelman syndrome or Prader-Willi syndrome (if they are able to give consent):

..... Date:.....

Co-signed by:

Parent (child under 18):.....

Or

Guardian (person 18 or older):.....

Date:.....

Appendix VI Postal questionnaires



Family- and community-based studies of major syndromes causing intellectual disability

Thank you for filling in these questionnaires.

Please answer each question as best you can. Those items that do not apply may be left blank. Please feel free to write comments by any question.

All information will be treated in the strictest confidence.

Please return the completed questionnaires in the enclosed self-addressed envelope.

Questionnaire 1: Demographic Information

Identification number:

*May be completed by family carer or paid staff

Some questions about the person you care for:

First Name:

Diagnosis:

.....
.....

Year of Birth: Gender: M/F Marital status:

.....

Place of Birth: Ethnicity:

Current postcode:.....

Type of residence:Number of co-residents:

Highest level of education completed:

Occupation – current: Previous:.....

Identification number:

*May be completed by family carer or paid staff

Your relationship to the affected person (e.g., Mother, paid carer, foster parent etc.)

.....

Care needs of the affected person: (base this on an average week)

Note: This information, along with all other research data, will NOT be seen by any agency or service provider.

- How many hours per week do you, the primary caregiver, spend actively caring for this individual?
- How many hours per week does the person spend at school/employment/day centre?
- How many hours per week does he or she spend in leisure activities outside the residence?
- Has respite care (inside or outside the home) been used by this person within the last year?
 - How often?
 - For what length of time?

Please describe any out-of-home services this individual has used within the last year (for example: day centre, social club, camp).

.....
.....
.....
.....
.....

Identification number:

*Family carer only

Some questions about the family of the person you care for:

Note: This information is included to determine if having a child with ID affects the earning capacity of parents.

Residential postcode: Father's place of birth:

Father's level of education: (*circle the highest level completed*)

Primary school

High school

Tertiary institution

Current occupation: Full time/part time/casual

Mother's place of birth:

Mother's level of education: (*circle the highest level completed*)

Primary school

High school

Tertiary institution

Current occupation: Full time/part time/casual

Parents' marital status:

Father's age at birth of affected person:

Mother's age at birth of affected person:

Number of other children:Current ages of other children:.....

Family history of intellectual disability: yes/no

Details:

.....
.....
.....
.....

Questionnaire 2: Carer information

Identification number:

*Family carer only

Primary caregiver health status*:

Please circle the best answer with regard to your own health:

1. Overall, my health can be described as

poor *good* *excellent*

2. In the last three years my health has

improved *stayed the same* *worsened*

3. Health problems prevent me from doing the things I want.

not at all *a little* *a great deal*

4. Compared to others my own age, my health is

better *about the same* *not as good*

*Adapted from Lawton et al. (1982)

Identification number:

*Family carer only

Primary caregiver satisfaction*:

Please circle the answer that best describes your feelings about your role as caregiver:

1. Caring for my child gives me a feeling of satisfaction.

never occasionally sometimes often nearly always

2. I enjoy being with my child.

never occasionally sometimes often nearly always

3. I feel closer to my child due to caregiving.

never occasionally sometimes often nearly always

4. Caregiving improves my self-esteem.

never occasionally sometimes often nearly always

5. I feel pleasure when my child feels pleasure.

never occasionally sometimes often nearly always

6. The role of caregiving adds meaning to my life.

never occasionally sometimes often nearly always

*Adapted from Lawton et al. (1982)

Questionnaire 3: Food-related Problems Questionnaire*

Identification number:

*May be completed by family carer or paid staff

Please circle the number you think represents the relative frequency with which the person you care for shows the following behaviours.

(1 is 'never' and 6 is 'always'. 7 is 'does not apply', and is to be used if the person cannot speak).

How often does the person compare the size or content of their meal with others?

1 2 3 4 5 6 7

If given the opportunity, how often would the person 'help themselves' to food which they should not have?

1 2 3 4 5 6

Is the person ever able to accept an explanation given to them if a meal is delayed?

1 2 3 4 5 6

Does the person hide or hoard food?

1 2 3 4 5 6

How often does the person talk about food?

1 2 3 4 5 6 7

If the person was denied food, how often would they respond negatively?

1 2 3 4 5 6

Is it necessary to lock away food to stop the person from taking food?

1 2 3 4 5 6

After a normal sized meal, how often will the person say they still feel hungry?

1 2 3 4 5 6 7

If the person is tired, unwell or upset, how often would this result in them going without food?

1 2 3 4 5 6

If it was available, would the person eat food not suitable for consumption (e.g., frozen food, scraps from the bin)?

1 2 3 4 5 6

If the meal includes an item the person does not like or is not expecting, how often would this result in behaviour difficulties?

1 2 3 4 5 6

Does the person ever eat non-edible items (e.g., dog food, leaves)?

1 2 3 4 5 6

How frequently will the person share food with others?

1 2 3 4 5 6

Does the person ever describe 'feeling full'?

1 2 3 4 5 6 7

Does the person ever associate people and/or places with specific food items or occasions involving food?

1 2 3 4 5 6 7

If given the opportunity, does the person eat more than a standard sized meal?

1 2 3 4 5 6

** Adapted from Russell & Oliver (2003) with kind permission from the authors.*

Angelman syndrome: Clinical data sheet

Identification number:

** May be completed by family carer or paid staff*

Put a 'X' in the box that applies to the person you care for. Please answer these questions as best you can.

	YES	NO	DON'T KNOW
Psychomotor development			
Feeding problems as a baby	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Delayed milestones (sit, walk etc.)	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Able to walk without assistance	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Absent speech or use of less than 6 words	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Able to use sign language	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Intellectual disability	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
IQ (if known) _____			
Loss of previous skills	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Neurological features/movement disorders			
Seizures or fits	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
How often?	<input type="checkbox"/>	< 1 per month	
	<input type="checkbox"/>	1-3 per month	
	<input type="checkbox"/>	1 or more per week	
Poor muscle tone in body	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Strong muscle tone in limbs	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Jerky movements	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Tongue poking out	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Hand flapping	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Behaviour			
Happy disposition	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Easily provoked or inappropriate laughter	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

	YES	NO	DON'T KNOW
Physical features			
Abnormal facial features	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Prominent jaw	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Widely spaced teeth	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Large mouth	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Deep set eyes	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Small head (<25th percentile)	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Fair hair/skin compared to other family members	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

Current height: _____

Current weight: _____

Additional comments or information (including relevant family history)

Prader-Willi syndrome: Clinical data sheet

Identification number:

* May be completed by family carer or paid staff

Put an 'X' in the box that applies to the person you care for. Please answer these questions as best you can.

	YES	NO	DON'T KNOW
Classical features			
Floppy as a baby	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Feeding problems as a baby	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Small penis or undescended testes (males) or Small labia, delayed or irregular menstrual periods (females)	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Rapid weight gain between 1-6 years old	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Characteristic facial features	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Intellectual disability IQ (if known)_____	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Minor features			
Decreased foetal movements	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Typical behaviour problems	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Sleep disturbance/sleep apnoea	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Short stature for family by age 15	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Fair hair/skin compared to family	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Fat concentrated on the trunk and upper legs	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Small hands and feet for height & age	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Narrow hands with straight outside border	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Squint or short sighted	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Thick viscous saliva	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Speech articulation defects	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Skin picking	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

	YES	NO	DON'T KNOW
Supporting features			
High pain tolerance	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Rarely vomits	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Temperature control problems	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Curvature of the spine	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Osteoporosis	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Early puberty	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Good at jigsaws	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

Current height: _____

Current weight: _____

Additional comments or information (including relevant family history)

Appendix VII Alterations to the Family Stress and Coping Interview and Food-Related Problems Questionnaire

Legend: Deletions ~~striketrough~~
 Additions **bold**

Family Stress and Coping Questionnaire

Please rate the following situations on a scale of 1 (not stressful) to 5 (extremely stressful):

1. The diagnosis of _____ as having ~~developmental disability~~ **Angelman/Prader-Willi syndrome**.
2. Explaining to others **people** about _____'s ~~developmental disability~~ **disorder**.
3. Your feelings about the cause of _____'s ~~developmental disability~~ **condition**.
4. Dealing with friends/family/~~people in the neighbourhood~~ **neighbours** on a day-to-day basis.
5. Dealing with doctors and other health professionals.
6. Dealing with legal professionals.
7. Dealing with _____'s teachers and the education system.
8. Creating and/or finding opportunities for _____ to make friends and participate in activities.
9. Deciding on the best level of integration for _____ .
10. Making the decision concerning accommodation in the home or in the community.
11. Meeting the needs of your (other) children.
12. Meeting your own personal needs.
13. Meeting the needs of your spouse.
14. Maintaining satisfying friendships for yourself.
15. Dealing with _____'s sexuality.
16. Work placements or employment for _____ .

17. Long-term planning for accommodation for _____ .
18. Planning for wills, trusts and guardianships.
19. Planning for emotional and social support for _____ .
20. Transportation.
21. Day-to-day assistance with care of _____ .
22. Time apart from _____ .
23. Dealing with financial and insurance issues.
24. **Dealing with service organisations/government agencies.**

Each item will be accompanied by appropriate probes from the following list:

1. What challenges have you faced in ~~this particular issue~~ **relation to this** issue?
2. What successes have you had, and how did you achieve them?
3. How have you tried to cope with this challenge? Why did you choose this strategy?
Has this been helpful?
4. Has the stress related to this issue changed:
 - i. Over the past 5 years? increase no change decrease
 - ii. In the last year? increase no change decrease

If changes in level of stress have occurred, why?

5. What would you like to change that would help make this experience easier for you and for parents of other children with ~~special needs~~ **intellectual disabilities**?

Food-Related Problems Questionnaire modifications

There were two word substitutions to reflect common usage in Australia:

‘ill’ was replaced with ‘unwell’

‘dustbin’ was replaced with ‘bin’

Appendix VIII Family Stress and Coping Interview

*Interview protocol (semi-structured)**

Interviewer: **Date:**

Identification number:

Notes:.....
.....
.....
.....

Introduction

This included a personal introduction, an explanation of the nature and purpose of the study, and a reiteration of the terms of informed consent.

Interview questions

Please rate the following situations on a scale of 1 (not stressful) to 5 (extremely stressful):

1. The initial diagnosis of _____ as having Angelman/Prader-Willi syndrome.
2. Explaining to other people about _____'s disorder.
3. Your feelings about the cause of _____'s condition.
4. Dealing with friends/family/neighbours on a day-to-day basis.
5. Dealing with doctors and other health professionals.
6. Dealing with legal professionals.
7. Dealing with _____'s teachers and the education system.
8. Creating and/or finding opportunities for _____ to make friends and participate in activities.
9. Deciding on the best level of integration for _____ .
10. Making the decision concerning accommodation in the home or in the community.
11. Meeting the needs of your (other) children.
12. Meeting your own personal needs.

13. Meeting the needs of your spouse.
14. Maintaining satisfying friendships for yourself.
15. Dealing with _____'s sexuality.
16. Work placements or employment for _____ .
17. Long-term planning for accommodation for _____ .
18. Planning for wills, trusts and guardianships.
19. Dealing with service organisations/government agencies
20. Planning for emotional and social support for _____ .
21. Transportation.
22. Day-to-day assistance with care of _____ .
23. Time apart from _____ .
24. Dealing with financial and insurance issues.

Each item was accompanied by appropriate probes from the following list:

- b. What challenges have you faced in relation to this issue?
- c. What successes have you had, and how did you achieve them?
- d. How have you tried to cope with this challenge? Why did you choose this strategy?
Has this been helpful?
- e. Has the stress related to this issue changed:
 - i. Over the past 5 years? increase no change decrease
 - ii. In the last year? Increase no change decrease
 - iii. If changes in level of stress have occurred, why?
- f. What would you like to change that would help make this experience easier for you and for parents of other children with intellectual disabilities?

**Adapted from (Nachshen, et al., 2003).*

Additional questions (Support group survey):

1. Are you a member of a Prader-Willi/Angelman Support Association?
2. Have you received any benefits from your membership of the organisation?
If so, what were they?
3. Is there anything that would make membership of the group more rewarding or helpful to you?

At the conclusion of the interview, participants were asked if they have any other concerns, or any suggestions for future questions. It was reiterated that the data will be de-identified, and that no names will be used in any reports or publications.

Appendix IX Information sheet and consent form; Discussion Groups



Angelman syndrome and Prader-Willi syndrome: care needs and the effects of ageing

This is an invitation to take part in a pilot study aimed at assessing items for inclusion in questionnaires and interviews. The developed protocols will form part of the above project investigating the effect of ageing on people with Angelman or Prader-Willi syndrome and on their carers. As intellectual disability is a sensitive area of study, the proposed items need to be considered by people involved in the daily lives of individuals with ID. For this purpose, we are inviting carers (family members or staff) of people with Down syndrome to participate in focus groups and to give suggestions and comments on the proposed protocols. Participants will also be encouraged to offer alternative fields of study which may be of special importance to people in their situation.

With a view to reducing the time required for the discussion groups, the draft protocols will be sent to all participants prior to the meeting. It is anticipated that the discussions will last for 1-1½ hours, and they will be audio-taped to ensure accuracy in reporting. Participants are free to leave the meetings or to withdraw from the pilot study at any time.

Participants in this pilot study will be identified by first names during the focus groups. After the discussion groups have concluded, the tapes will be transcribed and then destroyed. No names will be included in the transcriptions and all subsequent work will be on unidentified material. The end-result will be a set of tools for the future project which will best meet the needs of the researchers and of those who care for people with ID.

If you are interested in participating in these discussion groups, please complete and sign the accompanying Informed Consent Document and return it in the enclosed self-addressed envelope. After selection of a representative sample from the volunteers, the draft protocols will be sent to the participants for consideration. If you have any queries or concerns regarding the procedure, please contact Allyson Thomson.

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This pilot study is being undertaken as part of the requirements of a PhD degree at Edith Cowan University.

Faculty: Computing, Health and Science.

School: Exercise, Biomedical and Health Science.

This study has been approved by the Edith Cowan University Human Research Ethics Committee.

If you have any concerns or complaints about the study and wish to talk to an independent person, you may contact:

Research Ethics Officer
Edith Cowan University
100 Joondalup Drive
JOONDALUP WA 6027
Phone: (08) 6304 2170
Fax: (08) 6304 2661
Email: research.ethics@ecu.edu.au

Consent form



Angelman syndrome and Prader-Willi syndrome: care needs and the effects of ageing

I have read the information letter provided and have had the opportunity to ask questions. Any questions I have asked have been answered to my satisfaction.

I understand that all information provided is treated as strictly confidential and will not be released by the investigator unless required to by law. I have been advised as to which data are being collected and for what purpose.

I agree to participate in a focus group, realising that I may withdraw at any time without given reason and without prejudice. I agree to the audio-taping of the discussion, with the knowledge that the tapes will be destroyed after transcription, and that the transcripts will not include any names.

I agree that research data gathered for the study may be published, provided my name or other identifying information is not used.

.....

Participant

.....

Date

Table IX Timeline for recruitment of discussion group participants

Date	Action
24-2-2006	Approach to Genetic Support Council of WA (GSCWA) requesting access to support groups for recruitment
22-3-2006	Invitation to participate sent to Carers WA for insertion in the newsletter April and May
4-4-2006	Information packs delivered to GSCWA, forwarded to specific support groups
10-5-2006	Asked GSCWA, Carers WA and i.d.entity WA if a personal appeal to carers was possible
24-5-2006	Invitation to participate sent to Mosaic Carer Support Group for distribution to members
1-6-2006	Attended meeting of Commonlink group (i.d.entity WA), successfully recruited four volunteers
12-7-2006	Asked for an invitation to a meeting of Carers WA to address members
20-7-2006	Attended meeting of Linking Together (Carers WA), successfully recruited six volunteers

Appendix X Original Table of Contents

To assist the examiners in visualising the structural changes made to the thesis, the original Table of Contents is presented to facilitate comparisons.

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