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## Medical conditions in children with Down syndrome: A literature review ; Changes over time in medical conditions and service use by children with Down syndrome

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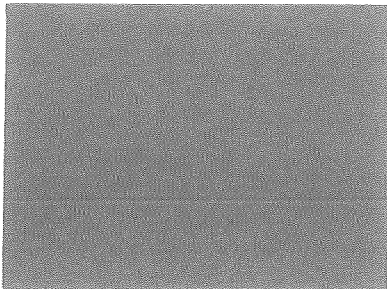
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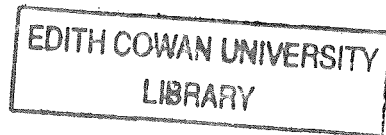
**Medical conditions in children with Down syndrome:**

**a literature review**

**AND**

**Changes over time in medical conditions and service use by children  
with Down syndrome**

Kelly Thomas



A report submitted in Partial Fulfilment of the Requirements  
for the Award of Bachelor of Science (Occupational Therapy)(Honours),

Faculty of Computing, Health and Science

Edith Cowan University.

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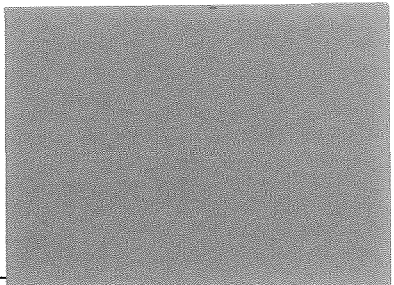
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## **Medical conditions in children with Down syndrome: a literature review**

**Background:** Children with Down syndrome experience an array of medical conditions and health care problems. Although there have been vast improvements in the medical management of these conditions, they still significantly impact on quality of life for families and children with Down syndrome. However, despite the magnitude of this problem limited literature has described the overall health status of children with Down syndrome.

**Aim:** The purpose of this review was to describe the medical conditions commonly experienced by children with Down syndrome and to consider the clinical implications of this knowledge.

**Methods:** Databases Medline, CINHALL and PsychINFO were electronically searched to identify relevant articles from 1990 to 2009. Inclusion criteria were children aged 18 years or younger with a diagnosis of Down syndrome, who had one or more medical co-morbidity. Articles were excluded at the title or abstract level if they were not peer reviewed, English articles, or did not meet the inclusion criteria. A narrative review of this research was possible.

**Results:** Common medical conditions experienced by children with Down syndrome include: cardiac and gastrointestinal disorders, musculoskeletal disorders, respiratory problems, eye and visual defects, ear and hearing impairments, frequent episodic illnesses and infections, and a greater risk of leukaemia and thyroid conditions. Prevalence estimates for these conditions varied depending on the diagnostic criteria or study methodologies.

**Conclusion:** Knowledge of the high levels of co-morbidities experienced by children with Down syndrome will lead to the development of more effective interventions for teachers and health care professionals, and also assist in the provision and resource allocation of disability services.

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Dr Helen Leonard and Ms Jenny Bourke

Submission date: August, 2009

## **Medical conditions in children with Down syndrome: a literature review**

### **Introduction**

Down syndrome is a chromosomal birth disorder and the single most common cause of intellectual disability in Western Australia [1]. The worldwide prevalence of Down syndrome varies and may be determined by accessibility to prenatal screening [2], socio-cultural attitudes, religious beliefs and political legislation regarding termination of pregnancy [3]. In Western Australia since 1980, the number of children born with Down syndrome has remained relatively stable at approximately 1 per 1000 births, however the overall cases of Down syndrome conceptions (live births, still births and terminations of pregnancy) has more than doubled, from 1.2 per 1000 conceptions in 1980-84, to 2.6 per 1000 in 2006 [4]. This change in the epidemiology of Down syndrome has been largely the result of increasing maternal age [5] and the widespread utilisation of early prenatal screening [6].

It is well documented that Down syndrome is strongly correlated with maternal age [7]. Due to the changing roles of women in westernised societies, major socio-demographic shifts have resulted in increasing maternal age. Within the Australian context, women's participation in the workforce has increased from 40% in 1979 to 55% in 2006 [8, 9] and their educational attainment is now similar to that of men [10]. As a result, the postponement of childbirth has contributed to the overall increase in Down syndrome conceptions [4].

The large increase in the reporting of Down syndrome conceptions is attributed to the accessibility and utilisation of antenatal screening programs. In Australia, since 1999, the rate of women's participation in antenatal screening programs has increased 2.5 fold to 44% [11] and the proportion of Down syndrome cases diagnosed prenatally for women under 35 has increased from 3% to 60% [12]. As a result, foetuses which would usually have spontaneously miscarried, are now being detected, contributing to an increase in the reporting of Down syndrome conceptions [12, 13]. However, a corresponding increase in

terminations of Down syndrome pregnancies has resulted in a relatively stable prevalence of Down syndrome live births [14]. Rates of terminations are however influenced by countries' differing legislation regarding termination of pregnancy and their societies socio-cultural beliefs [3].

Developed countries have seen dramatic improvements in infant survival and overall life expectancy for people with Down syndrome. In Australia over the past 40 years, the average life expectancy of people with Down syndrome has increased from 18 to 60 years of age [14]. Of particular significance is the improvement in infant survival whereby 91% of infants are expected to survive to one year, and 85% to ten years [15]. These rates are comparable to other developed countries [16, 17]. Internationally, there are ethnic disparities associated with infant survival with American research reporting black maternal race having a seven -fold increase in the risk of infant mortality, but only among infants without cardiac defects [16]. Within Australia, survival of Aboriginal children with Down syndrome is also significantly reduced compared to non-Aboriginal children [15].

Several factors have contributed to improvements in the survival rates for children with Down syndrome. The most important of these has been surgical advances in the management of congenital heart disease [17]. In addition, improved accessibility to antibiotics and vaccinations have contributed to a decline in morbidity and improved life expectancy of people with Down syndrome [14]. Changes in community and health professionals' attitudes have also influenced health care accessibility and survival [14], whereby people with Down syndrome are now afforded a higher degree of equality in the health care system [18]. However, ethical debates still persist in relation to implementing policies of equality when accounting for limited resources [18].

Children with Down syndrome experience an array of medical conditions and health care problems which have been widely documented. Although there have been vast improvements in the medical management of these conditions leading to increased rates of infant and child survival [13], these comorbidities still impact on quality of life for

families and children with Down syndrome. However, despite the magnitude of this problem limited literature has described the overall health status of children with Down syndrome. Therefore the objective of this narrative literature review is to describe the medical conditions commonly experienced by children with Down syndrome and to consider the clinical implications of this knowledge.

## **Methods**

Databases Medline, CINHALL and PsychINFO were electronically searched to identify relevant articles. The data bases were searched from 1990 to 2009 so as to only include the most recent literature in this review. The databases were also restricted to include only peer reviewed published articles, English articles and those which included children younger than 18 years. The main search terms were: Down syndrome, medical conditions, cardiac, gastrointestinal, respiratory, ophthalmic, ear, hearing, thyroid, diabetes, musculoskeletal, cancer and leukaemia. These terms were truncated and exploded to include all relevant disorders and terminology under these headings. Search terms were also adjusted to match the specific database being searched. Articles were included if they focused on children aged 18 years or younger with a diagnosis of Down syndrome, and one or more medical co-morbidities.

The intention of this review was to provide an overview of the most current and relevant literature and therefore focused on more recent literature discerning important results. Articles were excluded at the title or abstract level if they did not meet the inclusion criteria. Conference proceedings were not searched. Formal quality appraisal was not performed however within epidemiological studies this is not commonly undertaken [19]. Furthermore, no meta-analysis was performed in this literature review due to significant differences between cohorts (population, school or hospital based cohorts) and the different methods of collecting prevalence estimates.

## **Results**

A review of the literature supports the conclusion that children born with Down syndrome commonly experience a variety of additional birth defects and other health related problems. Medical conditions commonly associated with Down syndrome include; cardiac and gastrointestinal disorders, musculoskeletal problems, respiratory problems, eye and visual defects, ear and hearing defects, and frequent episodic illnesses and infections. In addition, research has also highlighted the increased risk that children with Down syndrome have of developing leukaemia and thyroid conditions.

### **Congenital heart defects**

Cardiac defects are reported to be the most common occurring birth defect associated with Down syndrome, with prevalence estimates ranging between 40-50% [16, 20]. The most frequently occurring congenital heart defects include atrioventricular septal defect (AVSD), atrial septal defect (ASD), ventricular septal defect (VSD), tetralogy of Fallot [21], and patent ductus arteriosus (PDA) [22]. Congenital cardiac defects have been a major cause of mortality for children with Down syndrome [2, 17]. Even with advances in corrective surgery for cardiac malformations, which have resulted in significant improvements in survival and quality of life among children with Down syndrome, cardiac disease still remains a major predictor of mortality [15].

### **Gastrointestinal disorders**

Children with Down syndrome experience a higher prevalence of gastrointestinal disorders than their peers [23, 24] with congenital gastrointestinal defects occurring in approximately 6.7% of infants with Down syndrome [25]. Commonly experienced gastrointestinal disorders include duodenal atresia, congenital malformations of the colon and rectum, Hirschsprung disease [23], gastro-oesophageal reflux [26] and coeliac disease [27, 28]. In addition, constipation, although not a serious condition, is relatively common among these children and often involves prolonged management, placing increased demands on family and carers [29].

Although there was considerable literature pertaining to prevalence and screening for coeliac disease, [30-33], research on serious congenital gastrointestinal defects is less comprehensive. This could be attributed to increases in corrective surgery undertaken in infancy resulting in fewer cases of gastrointestinal disorders present in childhood. Hirschsprung's disease however is an exception with a higher representation within the literature. The long term outcomes of Hirschsprung's disease have been researched and indicate that incontinence or soiling is frequent among children with Down syndrome and Hirschsprung's disease [34], although studies have also demonstrated no statistical significance between outcomes in children with or without Down syndrome and Hirschsprung's disease [35, 36]. Results in all studies should however be viewed with caution due to the small sample sizes. Further population based research is needed regarding congenital gastrointestinal disorders by children with Down syndrome and the outcomes and ongoing implications of the disorders for the children, families, and the health care system.

### **Respiratory problems**

Respiratory conditions including pneumonia, bronchitis and obstructive sleep apnoea are considered a major cause of morbidity for children with Down syndrome. Respiratory infections are the leading cause for hospital admissions among children with Down syndrome beyond the neonatal period, especially for those with congenital heart disease [37] and are shown to be a major cause of death [3]. However, due to the varied means of data attainment, discrepancies in the literature exist as to whether respiratory or cardiac defects contribute most to mortality in children with Down syndrome.

Obstructive sleep apnoea, which involves complete or partial upper airway obstruction during sleep, is reported to occur in approximately 60% of children with Down syndrome [38, 39]. The higher prevalence of this condition can be attributed to common physiological and anatomical characteristics observed in children with Down syndrome including midface hypoplasia, large adenoids or tonsils, narrow nasopharynx and eustachian tubes, systemic hypotonia [27] and inner ear dysplasia [40]. It is reported that parents substantially underestimate the prevalence and severity of obstructive sleep

apnoea assuming that irregular breathing during sleep is normal for children with Down syndrome [38]. As obstructive sleep apnoea may be associated with lower IQ performance [41] and behaviour problems [42], parents need to be informed of the prevalence of sleep apnoea and its associated implications.

### **Ophthalmic defects**

Ophthalmological problems are over represented in the Down syndrome population and if not corrected can result in substantial secondary disability [26]. Common ophthalmological disorders include refractive errors, strabismus, nystagmus and congenital cataracts [43, 44]. Hyperopia and myopia occur in approximately 43% of children with Down syndrome [44] and contribute to their fivefold increased likelihood of wearing glasses compared to their peers [29]. In addition, as children with Down syndrome frequently present with poor accommodation [45], bifocal spectacles should be considered for those who under-accommodate [46]. Strabismus, occurring in approximately 42% of children with Down syndrome, also contributes to vision impairment [47], and is positively associated with learning difficulties [48]. As sensory defects contribute substantially to disability, vigilant monitoring is required for children with Down syndrome to optimise opportunities for learning and development, and to increase quality of life.

### **Ear and hearing defects**

Children with Down syndrome have a high incidence of hearing impairment [28, 49]. Hearing loss is reported to be prevalent in over 50% of children with Down syndrome [26] and is defined as conductive, sensorineural or mixed [28]. While conductive hearing loss arises frequently in children with Down syndrome due to episodes of otitis media or middle ear dysfunction, sensorineural hearing impairments become increasingly prevalent with age [50]. It is recommended that children with Down syndrome undergo regular audiological screening for early identification of hearing impairments to minimise or correct hearing loss and reduce secondary speech problems [51].

Anatomical malformations, systemic hypotonia and an increased susceptibility to infections result in a high incidence of otitis media among children with Down syndrome



[52] and can lead to glue ear and transient conductive hearing loss. Although grommet insertion is disproportionately higher among children with Down syndrome compared to the general population [53], a systematic review concluded that this procedure is only minimally effective in reversing this loss [54]. Conversely, results of a longitudinal study have suggested that aggressive evaluation and treatment of chronic otitis media significantly improves hearing levels in children with Down syndrome [55]. Clearly more research is needed to guide clinical interventions in this area.

### **Musculoskeletal problems**

Approximately 20% of children with Down syndrome experience musculoskeletal disorders [56] due to a predisposition to hypotonia, lax ligaments and skeletal dysplasias [26]. Commonly occurring musculoskeletal conditions include: patellar instability, metatarsus primus, pes planus (flat foot), scoliosis and atlantoaxial instability [57]. Minor podiatric anomalies including pes planus, foot pronation and metatarsus primus, are often neglected due to a greater focus on the more severe pathologies associated with Down syndrome [58]. There is therefore a need to ensure early identification of minor musculoskeletal impairments is provided to prevent subsequent biomechanical and postural problems.

Atlantoaxial instability, reported to occur in approximately 10% to 20% of people with Down syndrome [59], has been considered to be particularly dangerous due to the potential risk of compression and damage to the spinal cord. Atlantoaxial instability is primarily attributed to laxity of the transverse ligament which results in instability of the spine at C1-C2 [59], or as a result of hypoplasia or malformations of the odontoid [57]. Previously, fear of the potentially catastrophic consequences of atlantoaxial instability led to recommendations for routine screening and the restriction of certain sporting activities [26]. These recommendations have now been revised such that routine screening is mainly indicated for children with Down syndrome who are symptomatic or who participate in contact sports [60].

### **Endocrine**

Endocrine anomalies, especially thyroid dysfunction [61] and diabetes [62], are reported to be more common among people with Down syndrome than the general population. Estimates of thyroid dysfunction among people with Down syndrome have varied from approximately 8.5% [61] to 38% [63]. These estimates vary in relation to population characteristics (age, size, race), diagnostic criteria and variations in operational definitions used in studies. Hypothyroidism can be congenital or acquired and has a greater prevalence than hyperthyroidism with approximately 10% of school aged children with Down syndrome having this condition [26]. As the prevalence of hypothyroidism increases with age, and is similar in its presentation to Down syndrome itself, early annual biochemical screening has been previously recommended to identify thyroid disease [60]. In America, rates of medically treated thyroid disease in people with Down syndrome increased by 73% after the release of the American Academy of Pediatrics Health Supervision Guidelines [64]. Although these rates are based on a study with limited generalisability due to its sample selection, the benefits of increased physician awareness and subsequent rates of screening can be seen. However, there is disagreement over the frequency of screening for thyroid dysfunction with some authors concluding that annual screening is not justified within the first two decades of life [61].

Down syndrome is also associated with diabetes and is reported to have a prevalence rate between 1.4 and 10.6% for Type 1 [65]. The increased prevalence for Type 1 diabetes may be genetically linked to the mutation of chromosome 21 which may be associated with the early onset and high frequency of autoimmune diseases [66]. As diabetes is relatively common among people with Down syndrome in comparison to the general population, further research should examine the substantial secondary health concerns experienced by children with diabetes, and the increased carer responsibility regarding monitoring of dietary requirements and medication management.

### **Leukaemia**

Although the overall risk of cancer for people with Down syndrome is equal to the general population, the risk of developing leukaemia is significantly higher [67, 68].

Children with Down syndrome have a nineteen fold increased risk of developing leukaemia than their normally developing peers [69, 70], with the majority of cases being diagnosed before the age of five [68]. This increased risk is apparent in data from the Western Australian Cancer Registry with 53% of all cancers recorded in people with Down syndrome resulting from childhood leukaemia [3]. Although survival outcomes following treatment for leukaemia for children with Down syndrome are similar to the general population, treatment is often poorly tolerated [28]. This may be associated with the high levels of comorbidity experienced by children with Down syndrome which complicate treatment and management regimes.

Interestingly, people with Down syndrome in all age groups have a decreased risk of solid tumours [67, 68, 71]. Possible reasons for this decrease may be related to reduced exposure to environmental carcinogens or directly related to the tumour –suppressor genes found on chromosome 21 [72]. It has been hypothesised that as people with Down syndrome have three copies of the tumour-suppressor genes due to their extra chromosome 21, their cells are less likely to lose all three functional copies of the tumour-suppressor genes [72] which may therefore provide greater protection against tumours.

### **General infections/viruses**

Structural abnormalities and an immature immune system result in children with Down syndrome being predisposed to recurrent infections and episodic illnesses. Commonly occurring conditions include upper-respiratory infections [28, 73] and otitis media [28, 55]. Poorer immune functioning, leading to greater susceptibility to infection, is further aggravated by frequent attendance at health care appointments, and comorbidity of cardiac and pulmonary diseases [73].

A recent US population-based study demonstrated that children with Down syndrome had a higher prevalence of ear infections, recent colds/flu, stomach illnesses and diarrhoea than did children without Down syndrome [24]. A major contributor to this increased susceptibility to illnesses is the array of immune dysfunctions affecting people

with Down syndrome [74], including severely diminished expansion of T and B lymphocytes [70]. Although the majority of the episodic illnesses common in children with Down syndrome are not life threatening, their prevalent and recurrent nature may add significantly to health care utilisation, increase the financial and emotional impact on carers, and contribute to an overall decreased quality of life.

## **Discussion**

As outlined, Down syndrome is associated with many co-morbid conditions, health problems and an increased risk of mortality compared to the general population. Improvements in the medical management of co-morbidities and surgical advances have reduced morbidity among children with Down syndrome, however congenital cardiac defects, along with respiratory problems are still considered a major cause of mortality [3]. Children with Down syndrome are predisposed to an array of health conditions and impairments, transcending the sensory, endocrine, musculoskeletal and haematological systems. While the estimated prevalences for these conditions vary between studies, this is likely the result of diverse sample characteristics, data collection methods, coding terminologies and study methodologies (population-based versus clinical or hospital based). Overall, it is evident that high levels of co-morbidity significantly impact on the quality of life for children with Down syndrome and their families.

Parents of children with Down syndrome may experience an emotional and financial impact from the medical co-morbidities experienced by their child. Parents have reported that their child's increased needs had diminished their finances to the extent that recreational activities and material items were often unaffordable [75]. Having a child with Down syndrome has also been associated with limited parental time for other children, reduced family socialisation [75] and poor maternal health [76]. Parents therefore need a clear understanding of the medical co-morbidities associated with Down syndrome to assist in making informed decisions regarding possible terminations of pregnancy or in making future plans for their child and family.

Despite the widespread acknowledgement of the co-morbidities experienced by children with Down syndrome and their health effect, limited literature is available on medical utilisation and health expenditures. A Western Australian study [29] presented an overview of service utilisation for children with Down syndrome. However, as the role of the Disability Services Commission, which coordinated medical services for people with an intellectual disability, has since changed, these data may no longer reflect the actual service use of children with Down syndrome in Western Australia. For children under five with Down syndrome, medical costs were 12 to 13 times higher than in the general population [77]. Also, children under five with an intellectual disability were more likely to be admitted to hospital and for longer periods of time than their peers [78]. However, limited research has examined health service utilisation for children over five with Down syndrome or how the need for medical care changes with age. This knowledge would result in a better understanding of the service needs for this population and assist in provision and resource allocation of disability and medical services.

This literature review has implications for screening and subsequent management of co-morbidities. Many of the co-morbidities experienced by children with Down syndrome, if left untreated, contribute to substantial secondary disability, resulting in unnecessary hardship and further disadvantage. Sensory defects are a primary example. As vision and hearing contribute to learning, language development and socialisation [48, 79, 80], the lack of regular screening and subsequent management of impairments not only affects the sensory systems involved but a child's chances of academic success and socialisation. Furthermore, as some children with Down syndrome are reported to have as few as one reciprocal friend [81, 82], and experience limited peer interactions outside school [81], the importance of minimising disability and limiting secondary disability is highlighted. Guidelines for the screening of medical conditions associated with Down syndrome have been published [27, 51, 60]. However, to what extent these are actually being implemented within schools and the health care system remains unknown. Further research is needed to provide an understanding of the current screening policies and guidelines being implemented and their effectiveness in identifying all the health conditions experienced by children with Down syndrome.

Finally, this literature review has important implications for health care professionals and education providers. A clear understanding of the co-morbidities experienced by children with Down syndrome provides a base for health care providers to plan and deliver safe and effective intervention programs suitable for each child's physical capacities and limitations. Knowledge of medical conditions is also valuable to health care professionals who are in a position to provide prospective parents with current information to facilitate informed decision making and future plans. In addition, as teachers play a valuable role in children's development, an understanding of the medical conditions and their functional impact on children with Down syndrome may facilitate the implementation of classroom strategies which minimise disability and foster academic achievement and socialisation with peers.

This literature review has synthesised the most current and relevant literature relating to the medical conditions experienced by children with Down syndrome, however its limitations must be acknowledged. As this was not a systematic review the methodological quality of the research was not formally evaluated. Instead, this narrative literature review was undertaken to summarise findings and provide an overview of the common medical conditions experienced by children with Down syndrome. Furthermore, a meta-analysis was not performed due to the differences between cohorts (population, school or hospital based cohorts) and the varied methods of collecting prevalence estimates.

## **Conclusion**

This review of the literature supports the conclusion that many children with Down syndrome experience additional birth defects and health related problems which impact on their quality of life. Medical conditions commonly associated with Down syndrome include; cardiac and gastrointestinal disorders, musculoskeletal disorders, respiratory problems, eye and visual defects, ear and hearing impairments, and frequent episodic illnesses and infections. In addition, children with Down syndrome have an increased risk of developing leukaemia and thyroid conditions. Although considerable research has documented these conditions, there is limited research examining the impact of levels of

co-morbidity on everyday functioning. Understanding the impact of high levels of co-morbidity would lead to the development of more effective interventions for teachers and health care professionals, and also assist in the provision and resource allocation of disability services.

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# Appendix A

# Down Syndrome Research and Practice Submissions Requirements

Further policies and guidelines

- [Content guidelines](#)
- [Standards](#)
- [Publication policies](#)
- [Editorial process](#)
- [Structure and style](#)

All submissions to *Down Syndrome Research and Practice* should meet the following requirements.

## Cover letter

Please include a cover letter explaining why the manuscript is suitable for publication in *Down Syndrome Research and Practice*.

## Prior publication

All authors are asked to indicate that they have not submitted a similar manuscript for publication elsewhere. If related work has been submitted elsewhere, then a copy must be included with the article submitted to *Down Syndrome Research and Practice*.

## Listed authors

All authors must consent to the submission of the manuscript. All authors will be contacted to confirm their approval. The involvement of any professional medical writer in publication must be declared (see the [European Medical Writers Association \(EMWA\) guidelines \[PDF\]](#) on the role of medical writers in developing peer-reviewed publications). Please refer to [section II.A. of the Uniform Requirements for Manuscripts Submitted to Biomedical Journals: Writing and Editing for Biomedical Publication](#) for guidance about recognising authorship.

## Confirmation of ethics approval

All research involving humans and animals must have been approved by the authors' institutional review board or equivalent committee, and that board must be named by the authors. In the case of human participants, informed consent must have been obtained, and all clinical investigation must have been conducted according to the principles expressed in the [Declaration of Helsinki](#). Authors should submit a statement from the ethics committee or institutional review board indicating approval of the research.

## Informed consent to research

We encourage authors to submit a sample of their participant consent form and may sometimes require a copy. Authors must confirm that informed consent was properly obtained from participants.

For further information, see sections 20-27 of the [Declaration of Helsinki](#) and [section 8.02 of the APA's Ethical Principles of Psychologists and Code of Conduct](#).

## Patient privacy and informed consent for publication

Please refer to [section II.E. of the Uniform Requirements of the International Committee of Medical Journal Editors](#). Complete anonymity is difficult to achieve, and informed consent for publication should be obtained if there is any doubt. When informed consent has been obtained it should be indicated in the published article.

## Competing interests

All authors will be asked to declare whether they have any financial, personal or professional interests that could be construed to have influenced their paper. Reviewers are also asked to declare any interests that might interfere with their objective assessment of a manuscript. Any relevant competing interests of authors must be available to editors and reviewers during the review process and will be stated in published articles.

## Epidemiological Studies

Reports of epidemiological studies should use the [STROBE initiative](#) as a guide.

## Registration of clinical trials

*Down Syndrome Research and Practice* supports the position of the International Committee of Medical Journal Editors (ICMJE) on trial registration. All trials initiated after 1 July 2005 must be registered prospectively in a publicly accessible registry (i.e., before patient recruitment has begun), or they will not be considered for publication. For trials initiated before 1 July 2005, all trials must be registered before submission. The trial's registration number must be provided at the time of submission. See the [ICMJE's Frequently Asked Questions \[PDF\]](#) for further information.

## CONSORT reporting guidelines for clinical trials

For reports of clinical trials, authors must submit original protocols as supporting information to allow editors and reviewers to assess manuscripts fully. Any deviation from the protocol must be explained. Authors of clinical trials must adhere to the [CONSORT](#) reporting guidelines appropriate to their trial design. All reports of the results of randomised clinical trials should include the [CONSORT](#) flow diagram, and authors should complete and submit the checklist contained within the [CONSORT](#) statement.

## Systematic reviews and meta-analyses of randomised controlled trials

Reports of meta-analyses of randomized controlled studies should use the [QUOROM statement](#) as a guide, and should include a copy of the [QUOROM checklist \[PDF\]](#).



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# Down Syndrome Research and Practice

## Structure and Style

Further policies and guidelines

- [Content guidelines](#)
- [Standards](#)
- [Publication policies](#)
- [Editorial process](#)
- [Submission requirements](#)

The following describes the structure and style expected of submissions to *Down Syndrome Research and Practice*.

Further guidance about the structure, style and presentation of papers for publication can be found in -

- [APA Guide to Preparing Manuscripts for Journal Publication](#)
- [Section IV of the Uniform Requirements for Manuscripts Submitted to Biomedical Journals: Writing and Editing for Biomedical Publication](#)
- [Publication manual of the American Psychological Association \(5th edition\)](#)
- [Scientific style and format - The Council of Science Editors' Manual for Authors, Editors, and Publishers \(7th edition\)](#)

## Readability

*Down Syndrome Research and Practice* is an international journal covering many disciplines and many subjects, and is read by people from many cultural, scientific and socioeconomic backgrounds.

Contributions should therefore be written clearly and simply so that they are accessible to the broadest range of readers, including those for whom English is not their first language.

## Format

Contributions should be double-spaced and written in English (spellings as in the [Oxford English Dictionary](#)).

Contributions should be organised in the sequence: title, text, methods, references, Supplementary Information (if any), acknowledgements, author contributions, author information (containing data deposition statement, interest declaration and corresponding author line), tables and figure legends.

Hard copies of manuscripts are not required. Text files can be submitted in the following formats: Word, WordPerfect, Rich Text Format (RTF) and Portable Document Format (PDF). Graphics files can be submitted in the following formats: Portable Document Format (PDF), Encapsulated PostScript (EPS),

Excel, Portable Network Graphics (PNG), Adobe Illustrator, Joint Photographic Experts Group Interchange Format (JPEG), Photoshop, PowerPoint, or Tagged Image File Format (TIFF).

## References

Only published or accepted papers and books should be included in the reference list. Meetings abstracts, conference talks, or papers that have been submitted but not yet accepted should not be cited. Limited citation of unpublished work should be included in the body of the text only. All personal communications should be supported by a letter from the relevant authors.

From 2007 (volume 12), *Down Syndrome Research and Practice* uses the numbered citation (citation-sequence) method. References are listed and numbered in the order that they appear in the text. In the text, citations should be indicated by the reference number in ([square]) brackets. Multiple citations within a single set of brackets should be separated by commas. Where there are more than three sequential citations, they should be given as a range.

To assist electronic linking, references must follow the following format. The format is based on the International Committee of Medical Journal Editors Uniform Requirements for Manuscripts Submitted to Biomedical Journals: Sample References, which are maintained by the [US National Library of Medicine Bibliographic Services Division](#). This standard is sometimes known as the [Vancouver system](#) and broadly conforms to [ISO-690](#). The differences from this format are:

1. All authors should be listed in the reference list (as per [NLM's standard](#)).
2. Journal titles should not be abbreviated. Full journal titles are more accessible to a wide readership.
3. Journal and book titles should be italicised (but not journal article or book chapter titles).

Wherever possible, please provide DOIs with references. Some examples follow:

### *Journal article*

1. Anupama R, Hills RK, Stiller C, Gibson BE, Graaf SSN, Hann IM, O'Marcaigh A, Wheatley K, Webb DKH. Treatment for myeloid leukaemia of Down syndrome: population-based experience in the UK and results from the Medical Research Council AML 10 and AML 12 trials. *British Journal of Haematology*. 2006;132(5):576-583. doi:10.1111/j.1365-2141.2005.05906.x.

### *Book*

2. Bird GA, Buckley SJ. *Number skills for individuals with Down syndrome - An overview*. Portsmouth, UK: Down Syndrome Education International; 2001.

### *Book chapter*

3. Buckley SJ, Bird GA, Byrne A. The practical and theoretical significance of teaching literacy skills to children with Down's syndrome. In: Rondal JA and Perera J, editors. *Down's syndrome: psychological, psychobiological and socio-educational perspectives*. London: Whurr; 1996. p. 119-128.

## Acknowledgments

People who contributed to the work, but do not fit the criteria for authors should be listed in the Acknowledgments, along with their contributions. You must also ensure that anyone named in the acknowledgments agrees to being so named.

## Funding

The sources of funding that have supported the work should be described in a section titled 'Funding'. Please also describe the role of the study sponsor(s), if any, in study design; collection, analysis, and interpretation of data; writing of the paper; and decision to submit it for publication.

## Author contributions

A detailed list of the contributions of each of the authors should be included.

## Competing interests

Competing interests associated with any of the authors must be detailed. If authors declare that no competing interests exist, then this should be stated in the contribution.

## Abbreviations

Please keep abbreviations to a minimum. A list of definitions for all non-standard abbreviations should be provided. Non-standard abbreviations should not be used unless they appear at least three times in the text.

## Nomenclature

The correct and established nomenclature should be used wherever possible. In particular:

- We strongly encourage the use of SI units. If you do not use these exclusively, please provide the SI value in parentheses after each value.
- Species names should be italicized (e.g., *Homo sapiens*).
- Genes, mutations, genotypes, and alleles should be indicated in italics. Use the recommended name by consulting the appropriate genetic nomenclature database, e.g., HUGO for human genes. It is sometimes advisable to indicate the synonyms for the gene the first time it appears in the text.
- The Recommended International Non-Proprietary Name (rINN) of drugs should be provided.

## Accession numbers

All appropriate datasets, images, and information should be deposited in appropriate public resources. Please provide the relevant accession numbers (and version numbers, if appropriate). In addition, as much as possible, please provide accession numbers or identifiers for all entities such as genes, proteins, mutants, diseases, etc., for which there is an entry in a public database. Providing accession numbers allows linking to and from established databases and integrates your article with a broader collection of scientific information. Please list all accession numbers directly after the Supporting Information section.

## Supplementary information

This is online-only, peer-reviewed material that is essential background to the contribution (for example, large data sets, methods, calculations), but which is too large or impractical, or of interest only to a few specialists, to justify inclusion in the printed version of the paper.

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**Changes over time in medical conditions and service use by children  
with Down syndrome**

Kelly Thomas

A report submitted in Partial Fulfilment of the Requirements  
for the Award of Bachelor of Science (Occupational Therapy)(Honours),  
Faculty of Computing, Health and Science  
Edith Cowan University.

September, 2009

I declare that this written assignment is my  
own work and does not include:

- i. Material from published sources used  
without proper acknowledgment; or
- ii. Material copied from the work of other students

Signature:

Date:

  
18/12/2009

## **Changes over time in medical conditions and service use by children with Down syndrome**

**Background:** Children with Down syndrome commonly experience a variety of medical conditions and health care problems. Although vast improvements in the medical management of these conditions has occurred, changes in the overall health status of children with Down syndrome over time, is yet to be explored. Furthermore, research is limited which examines service utilisation for children with Down syndrome or the impact of socio-demographic factors on access to services.

**Aim:** To compare the prevalences of parent reported medical conditions, and use of services, by school aged children with Down syndrome in Western Australia between 1997 and 2004. In addition this study aims to describe the impact of socio-demographic factors on medical service utilisation in 2004.

**Methods:** This study involved the comparison of two cross sectional surveys completed by parents of children with Down syndrome in 1997 and 2004. The surveys collected information of family demographics, medical conditions, health issues and service utilisation. Frequency distributions were used to describe medical conditions in 2004 and formal tests of association were based on chi-square tests. Regression analyses were used to compare medical conditions and service use between the 1997 and 2004 cohorts.

**Results:** Children with Down syndrome in 2004 had significantly greater odds of having a bowel condition than children in 1997 (OR 1.68, 95% CI 1.16 - 2.45). In 2004 children had approximately 50% less likelihood of having a current problem due to their cardiac condition (OR 0.52, 95% CI 0.28-1.00). An overall reduction in the incidence rates of episodic illnesses and infections was seen in 2004. The use of GP services (IRR=0.91, 95% CI 0.83, -1.00) and combined medical specialist visits (IRR=0.92, 95% CI 0.84 - 1.01) were reduced in 2004. Overnight hospital admissions (IRR 0.60, 95% CI 0.37 - 0.96) and length of stay (IRR 0.33 95% CI 0.24 - 0.44) were also reduced in 2004.

**Conclusion:** The health status of children with Down syndrome has varied over time. Most importantly, children in 2004 experienced a significant reduction in current cardiac problems and episodic illnesses that may contribute to improvements in overall health and impact quality of life. In addition, children with Down syndrome are using less medical services in 2004. These findings have implications for parents, health professionals and in the provision and resource allocation of disability services in Western Australia.

Author: Kelly Thomas  
Supervisors: Dr Sonya Girdler,  
Dr Helen Leonard and Ms Jenny Bourke  
Submission date: September, 2009

## **Changes over time in medical conditions and service use by children with Down syndrome**

### **Introduction**

Down syndrome is a chromosomal birth disorder and the single most common cause of intellectual disability [1], affecting 1 per 1000 births. It is well documented that Down syndrome is strongly correlated with maternal age [2]. Major demographic shifts in westernised societies have resulted in increasing maternal age due to the changing roles of women leading to the postponement of pregnancy. As a result, the number of overall Down syndrome pregnancies has increased [2]. However, with the greater utilisation and accessibility of antenatal screening programs [4], and increases in terminations of pregnancies [5], the prevalence of Down syndrome births has remained relatively stable.

Children with Down syndrome experience an array of medical conditions and health care problems which have been widely documented. There is a predisposition for children with Down syndrome to develop medical co-morbidities [6] which may present ongoing health issues and contribute to an increased risk of developing recurrent episodic illness and infections. However, improvements in the medical management of these conditions has lead to increased rates of infant and child survival and decreased morbidity [7]. These medical improvements include, surgical advances and management of congenital heart defects [8], improved accessibility to antibiotics and vaccinations, and changes in community and health professionals' attitudes towards the treatment methods for people with Down syndrome [9]. With these improvements in health management, it is expected that further improvements in the health of children with Down syndrome will be seen over time.

Although data are limited, there is a growing body of research which suggests that children with Down syndrome have higher rates of health utilisation than their peers [10-12]. However, to our knowledge, no literature has examined changes in service utilisation over time, especially within the Down syndrome population. Furthermore, since 2002, Western Australia has seen changes in the provision of disability medical specialist



services with these services no longer being provided by the Disability Services Commission (DSC). As a result the service utilisation of children with Down syndrome, and how these have changed over time, is currently unknown. Furthermore, the impact of socio-demographic factors on accessibility of medical services for children with Down syndrome is also unexplored. As families of children with an intellectual disability are shown to be more frequently economically disadvantaged [13], it is reasonable to assume that inequalities exist in access and utilisation of specialist medical services which are not fully government subsidised. An understanding of the socio-demographic disparities which exist for families of children with Down syndrome is important for effective policy development and equality in resource allocation.

Although the benefits of improved medical management have been described in terms of survival [8,14], limited literature has examined the reduction of co-morbidities, or the general health and frequency of illnesses in children with Down syndrome over time. With the improvements of medical care, and more effective treatments early in life, it is expected in this study that children with Down syndrome would experience less episodic illnesses and ongoing problems associated with their medical conditions over time. Furthermore, we predicted that with these reductions in illnesses and ongoing problems due to medical conditions, coupled with the restructuring of the specialist services out into the community, a decline in service utilisation may be evident. Finally, we aim to describe the impact of socio-demographic factors on medical service utilisation and examine whether they impacted on specialist medical services which are not fully government subsidised.

## **Methodology**

### **Methods**

This study involved the comparison of two cross sectional surveys conducted in 1997 and 2004 using a Western Australian population data source of children with Down syndrome. Figure 1 illustrates the study process. In 1997 cases were principally ascertained directly through the Disability Services Commission (DSC), a government body coordinating medical and therapy services in Western Australia for people with an

intellectual disability. Families with a child with Down syndrome born between 1980 and 1991, living in Western Australia were identified and mailed a questionnaire. The total response fraction obtained was 79.5% ( $n = 210/264$ ).

In 2004 a second survey was conducted with cases identified through the Intellectual Disability Exploring Answers (IDEA) database, a population data source which uses multiple sources of ascertainment including the Disability Services Commission and the Department of Education and Training [15]. Five hundred families, with a child with Down syndrome were aged 25 years and younger and registered with the Disability Service Commission, were invited to participate in the study. In 2004, the total response fraction was 73% ( $n = 363/500$ ). The present analysis restricted data to subjects who were school aged in 2004 ( $n=208$ ) to ensure a comparable age sample to the 1997 cohort.

### **Instruments**

The 1997 and 2004 questionnaires sent to families of children with Down syndrome obtained similar information on family demographic characteristics, medical conditions and health related conditions experienced by their child. Questions relating to medical conditions asked if the child with Down syndrome had ever had a certain medical condition, and if so, was the condition considered a current problem or requiring ongoing medical treatment. In addition, the questionnaire gathered information on health service utilisation including type and frequency of health care professional visits over the previous 12 months. Both questionnaires were pilot tested to ensure their capacity to elicit relevant and important information.

### **Data coding and management**

In coding the data children were categorised into three age groups (5-9 years; 10-13 years; 14-17 years). Socio-demographic predictors used in data analysis for the 2004 cohort included parental education, father's occupational skill level, maternal work status, family income and place of residence (See Table I). Participants' postcodes were used to categorise place of residence. Father's occupational skill level was based on the Australian and New Zealand Standard Classification of Occupations [16], however categories were recoded so that the lowest skill level would become the baseline. Data

was entered into FileMaker Pro version 8.0, and variables consistently categorised across the two cohorts. Data were then exported into STATA version 10 and the 1997 and 2004 data were linked for statistical analysis by matching on DSC record numbers.

Ethics approval for the 1996 study was granted by the Ethics Committee at Princess Margaret Hospital for Children, and by the Disability Services Commission. Ethical approval for the 2004 study was obtained through the Ethics Committee of the Women's and Children's Health Services in Western Australia. Approval to link the two data sets was obtained directly from the Disability Services Commission. Ethical approval for the current study was granted by the Edith Cowan Human Research Ethics Committee.

### **Data analysis**

The prevalences of medical conditions in 2004 were described as frequency distributions. Formal tests of association between medical conditions and child characteristic factors of age or gender, were based on chi-square tests. P-values below 0.05 were considered statistically significant, however the clinical meaning of differences in variables was also considered. The 2004 medical services, hospital admissions and episodic illnesses data were annualised in order to provide an estimate comparable to the 1997 data. Logistic regression was used to compare the prevalence of medical conditions between 1997 and 2004, and Poisson regression to compare rates of episodic illnesses and medical service use. Two outliers where the respondents had clearly misinterpreted the question about the use of medical services were excluded. To account for overlap of children within the 1997 and 2004 cohorts, regression analyses were conducted within a generalised estimating equation framework [1]. Analysis of the impact of socio-demographic factors on service use in 2004 involved multivariate regression models. Hospitalisation rates for the general population for school aged 5-17 years in 1997 and 2004 were accessed [18], and incidence rate ratios calculated in order to make comparisons between the general population and children with Down syndrome.

## Results

### Sample characteristics

The sample characteristics of the 1997 cohort have previously been described in detail [19]. Table I outlines the socio-demographic characteristics of families in 2004. The mean age of the 2004 study subjects was 11.7 years (range 5.02 -17.98) and there were 90 females and 118 males. All children whose parents responded 202/202 (100%) lived in their family home and 135/149 (90.6%) lived with both parents. Mothers provided the majority of day-to-day care needs in 200/208 (96.2%) cases.

### Medical conditions

#### *Medical conditions in the 2004 cohort*

Specific medical conditions in school aged children with Down syndrome in the 1997 cohort have been previously described [19]. The medical conditions in the 2004 cohort are shown in Table II. Cardiac conditions were present in 95 (55.7%) children, with the most common cardiac conditions being ventricular septal defects (N=33, 16%) and atrial septal defects 25 (12%). Congenital bowel conditions were reported in 66 (31.7%) children, with duodenal atresia occurring in 9 (4.3%) cases, while constipation was present in 20% (N=41) of children with Down syndrome. Over half of the children were reported to have had an ear condition 123 (59.1%). Glue ear occurred most frequently (86, 41.3%) while hearing loss was reported in 36 (17.3%) children. Over two thirds of children presented with visual defects (140, 67.3%), the most frequent of these were short sightedness (52, 25%) and strabismus (45, 21.6%), while parents of 45 (21.6%) children reported their child was long sighted. Twenty-six (12.5%) parents reported their child to have had a thyroid disorder, with hypothyroidism occurring in 22 (22.6%) of children and hyperthyroidism reported in three (1.4%) children. No statistically significant differences were apparent across age groups or gender for all these medical conditions.

Seventy-four (35.6%) children were reported to have ever had a musculoskeletal condition. Flat feet occurred most frequently (N= 47, 22.6%) and were more common among girls ( $\chi^2 = 6.6$ ,  $P = 0.01$ ). Twenty-eight (59.6%) of these children wore orthotics

and use was again higher among females ( $\chi^2=5.65$ ,  $P=0.02$ ). Fifty eight (27.9%) children had had X-rays of the cervical spine and in 4.3% ( $N=9$ ) a diagnosis of atlantoaxial instability had been made. Forty-eight (65%) parents reported that their child currently experienced a problem associated with their musculoskeletal condition and this was most commonly flatfeet (29/48, 60.4%).

Two thirds (126/208, 60%) of children had ever had a respiratory condition. Nearly 20% of children were reported to have sleep apnoea and 42 (20.2%) had asthma. No association was found between respiratory conditions and gender, however sleep apnoea was more prevalent in the 10-19 year old age group ( $\chi^2=7.89$ ,  $P=0.02$ ). Only 15% of parents reported that their child had current or continued respiratory problems and no statistically significant difference was apparent across age groups ( $\chi^2=3.92$ ,  $P=0.14$ ).

#### ***Medical conditions in 2004 compared with 1997.***

Table III outlines the likelihood of medical conditions and episodic illnesses in children with Down syndrome in 2004 compared with 1997, including if they have ever had the medical condition, and if they experience current problems associated with the condition. School-aged children with Down syndrome in 2004 had greater odds (OR 1.68; 95% CI 1.16 - 2.45) of having any bowel condition than children in 1997 even after adjusting for age (OR 1.69; 95% CI 1.13 - 2.52), and gender (OR 1.65; 95% CI 1.13 - 2.37). Specifically, children in 2004 had higher odds of having a congenital bowel condition, mainly duodenal atresia (OR 1.17; 95% CI 0.60 - 2.28) as well as constipation (OR 1.20; 95% CI 0.77 - 1.86). In 2004, children had a slightly higher likelihood of ever having a cardiac (OR 1.11; 95% CI 0.90 - 1.39), ear (OR 1.13; OR 0.18 - 1.56) or thyroid condition (OR 1.08; 95% CI 0.69 - 1.70), and hearing loss had a 13% increased likelihood (OR 1.13; 95% CI 0.68 - 1.90). In 2004 there was a reduced prevalence of overall eye conditions (OR 0.79; 95% CI 0.55 - 1.15), however long sightedness was increased (OR 1.54; 95% CI 1.03 - 2.31) as was the likelihood of wearing glasses (OR 1.18; 95% CI 0.83 - 1.66).

Children in 2004 had approximately 50% less likelihood of having a current problem due to their cardiac condition (OR 0.52; 95% CI 0.28-1.00) which was further reduced when

adjusting for age and gender (OR 0.32; 95% CI 0.15 – 0.68). In 2004, there was an increased likelihood of bowel (OR 1.29; 95% CI 0.77 – 2.18), ear (OR 1.06; 95% CI 0.74– 1.53) and thyroid (OR 1.54; 95% CI 0.89 – 2.66) conditions causing a current problem or requiring ongoing treatment, while eye conditions showed a reduction (OR 0.84; 95% CI 0.60 – 1.18)

An overall reduction in the incidence rates of episodic illnesses and infections among children with Down syndrome was found in 2004 compared to 1997. The greatest reductions were seen for episodes of tonsillitis (IRR 0.56; 95% CI 0.38-0.83) and ear infections (IRR 0.77; 95% CI 0.62 - 0.95).

### **Medical service use**

Table IV illustrates the rate of service use in 2004 compared to 1997. The use of a general practitioner (GP) (IRR 0.91; 95% CI 0.83 -1.00) and the combined medical specialist visits (IRR 0.92; 95% CI 0.84 – 1.01) showed a slight rate reduction in 2004. There was also a reduction in disability specialist services (IRR 0.25; 95% CI 0.15- 0.41) and audiologist use (IRR 0.80; 95% CI 0.61 – 1.04), but an increase for ear, nose and throat (ENT) (IRR 1.12; 95% CI 0.89 – 6.88), podiatry (IRR 0.67; 95% CI 0.50 – 0.91) and ‘other’ specialists (IRR 1.29; 95% CI 0.97 – 1.72). Cardiologists were unable to be compared over the two years due small numbers in cardiologist attendance.

Overnight hospital admissions (IRR 0.60, 95% CI 0.24 – 0.44) and the total nights spent in hospital (IRR 0.33 95% CI 0.24 – 0.44) both showed marked decreased incidence rates in 2004 compared with 1997. This paralleled, but was reduced further than the general population of Western Australian school-age children for these two years (OR 0.88, 95% CI 0.86 - 0.90). With respiratory conditions (IRR 0.28, 95% CI 0.09 – 0.90), there was the greatest reduction in hospital admissions in 2004 with the average nights in hospital decreasing from approximately five in 1997 to 1.6 nights in 2004. However there was an increased rate of day hospital admissions (IRR 1.20, 95% CI 0.81 – 1.76) in 2004 and in both years these were commonly associated with dental procedures. This increase paralleled the increase seen in the general population (OR 1.33 95% CI 1.29 - 1.36).

The frequency of surgical interventions was reported in 1997 and 2004. The odds of having cardiac and ENT surgery were comparable across years (OR 1.08; 95% CI 0.88 – 1.32) and (OR 1.01; 95% CI 0.74 – 1.38), respectively. However, in 2004 there was a reduced likelihood of children having had eye surgery (OR 0.92; 95% CI 0.58 – 1.46), and a 70% increase in the likelihood of bowel surgery (OR 1.71; 95% CI 0.83 – 3.52).

### **Impact of socio-demographic factors on service use in 2004**

Table V outlines the effects of socio-demographic factors on service use for children with Down syndrome. After adjusting for fathers' occupational skill level, family income, maternal work status and place of residence, the rates of GP service use were three times higher for children whose fathers were well educated (IRR 3.33; 95% CI 2.52 – 4.41). All fathers' skill levels above the baseline showed reductions in service use and children had a comparable rate of GP service use for rural and metropolitan areas (IRR 1.00; 95% CI 0.81– 1.24).

After adjusting for the other socio-demographic factors, higher paternal education was associated with increased specialist service use (IRR 1.63; 95% CI 1.30 – 2.05). Children, whose family's annual income ranged from \$20 000 - \$31 999, or \$32 000 – \$51 999, demonstrated the highest rates of service use, and had the strongest effect size within income. Father's skill level had an opposite effect, with the lowest skill level (the baseline) showing the highest rates of specialist service use. Working mothers were associated with reduced use of services (IRR 0.85; 95% CI 0.72 – 0.99) and little difference was found between children living in rural or metropolitan areas (IRR 1.05; 95% CI 0.88 – 1.27).

Within the model for overnight hospital admissions, none of the socio-demographic indicators had statistically significant effects, except for father's occupational skill level of three (IRR; 0.15, 95% CI 0.028- 0.83) and four (IRR; 0.28; 95% CI 0.08 – 0.99) which both demonstrated a reduction in overnight hospital admissions. Hospital day admissions however, demonstrated a strong association between higher maternal education and increased service use (IRR; 5.15, 95% CI 1.71 – 15.55). Significantly reduced hospital

day admissions were seen for fathers who had an occupational skill level of one (IRR 0.23; 95% CI 0.08 – 0.63), three (IRR 0.30; 95% CI 0.12 – 0.81) and four (IRR 0.28; 95% CI 0.12 – 0.66) compared with the baseline which was the lowest skill level. Family income showed no statistically significant associated with hospital day admissions.

## **Discussion**

This study aimed to compare the prevalences of medical conditions and episodic illness in 1997 and 2004 to see if an improvement in the health of children with Down syndrome has occurred over time. In addition, this study compared the use of medical services between these years in order to assess if the improved health of children with Down syndrome, or the disbandment of specialist services at the Disability Services Commission, has led to decreases in service use. Furthermore, the impact of socio-demographic factors on service use was described. The results of this study found a significant reduction in ongoing cardiac conditions, while bowel and thyroid conditions proved to be associated with increased ongoing problems in the later cohort. Eye conditions showed a 20% reduction, while hearing loss had increased by 13% in 2004. Of importance, was that episodic illnesses reported to have an overall reduction in 2004 indicating the improved health and reduction in illnesses not genetically encoded in children with Down syndrome. Service use paralleled these results, showing a reduction in both GP and specialist appointments, while overnight hospital admissions were also significantly reduced. Higher paternal education appeared to have the strongest relationship to increased service use in the majority of analyses, and socio-demographic factors had the strongest impact on specialist and GP service use.

The most important finding in this study was the 50% reduction in parent reported current cardiac problems in 2004 compared with 1997. It is therefore likely that this later cohort may have benefited considerably from the recent improvements in early surgery and as a result have less ongoing comorbidity associated with their heart lesions compared with the earlier cohort. Several studies highlight the changing survival profile of children with Down syndrome [8,14,19] which attribute increased survival mainly to advances in cardiac surgery and the improved medical management of these conditions. As a result, children with Down syndrome are able to experience not only improved survival, but a



significant decline in the ongoing cardiac comorbidities which pose a health risk for children with Down syndrome and affect quality of life.

In 2004, children were reported to have an overall reduction in the number of episodic illnesses and infections. These results would be in line with the improved cardiac status we observed in 2004 as ongoing cardiac conditions often contribute to secondary illnesses [20]. The improved health of children with Down syndrome can also be attributed to greater access to, and effectiveness of, antibiotic treatments and vaccinations within the health care system [9]. In the later cohort, ear infections especially were found to have the highest rate of reduction. Given the similar rate of ENT surgery in both years and the increased use of ENT specialists in 2004, the results indicate a trend towards increased preventative health care which focuses on medical treatment other than surgical intervention. The reduction in episodic illnesses was also reflected in the use of GP services which saw a significant reduction in 2004. Our study is unique as it would appear to be the first to have used population data to address rates of illnesses and infections and service use, in children with Down syndrome over time. However, given the reductions seen in episodic illnesses and infections, our study would support the thesis that the health of children with Down syndrome has improved in recent years.

Hearing impairments in children with Down syndrome are well documented [21,22]. In 2004, a 13% increase in the number of Western Australian school-aged children with hearing loss was found compared with 1997. As hearing is vital to language development [23], and therefore contributes to learning and socialisation, regular screening and subsequent management of impairments is vital in facilitating chances of academic success and socialisation for children with Down syndrome. With the statistically significant reduction in the use of audiologist services, and the increase of hearing loss seen in the later cohort, our findings show some support for children with Down syndrome to undergo regular screening for early identification of hearing impairments [24] in order to minimise disability and limit secondary disability.

In 2004 parents reported higher rates of ongoing problems or the requirement of continued treatment for thyroid and bowel conditions. For thyroid conditions, this might be expected as once hypothyroidism is diagnosed, treatment is ongoing [25].

Furthermore, greater awareness and the possible recognition and identification of thyroid disease [26] in later years may also account for some of the increase in thyroid conditions requiring treatment. In 2004, despite the apparent increase in bowel conditions, the ongoing associated problems were actually lower than what would be expected. Given the increases in bowel surgery in the later cohort, the ongoing problems may be the result of continued constipation. Although parents report thyroid and bowel conditions as a current problem, they are most likely attributable to the necessity of continued treatment rather than the cause of a current medical condition.

A high percentage (60%) of respiratory conditions was reported by parents in 2004. As respiratory infections are considered a leading cause for hospital admissions among children with Down syndrome [27] and are shown to be a major cause of mortality [28], this may be cause for concern. However, parental report showed low prevalences in current respiratory problems, and illnesses such as pneumonia and bronchitis were also reduced. Furthermore, respiratory conditions represented the greatest reduction in overnight hospital admissions and length of stay in 2004. These results demonstrate a possible improvement in the frequency and severity of respiratory conditions in school-aged children with Down syndrome. Sleep apnoea was reported to occur in 20% of children with Down syndrome in the 2004 cohort. These prevalences were lower than other studies [29,30]. This may be due to the smaller sample sizes than in the present study, or that parents are reported to substantially underestimate the prevalence of sleep apnoea assuming that irregular breathing during sleep is normal for children with Down syndrome. As sleep apnoea may be associated with lower IQ performance [31], and behaviour problems [32] parents need to be informed of the prevalence of sleep apnoea and its associated implications.

In 2004, musculoskeletal conditions, the majority of which were 'flat feet', occurred in 35.6% of children with Down syndrome. 'Flat feet' is the most common musculoskeletal condition reported for children with Down syndrome [33] and is temporarily correctable with orthotics, worn by over half of the children with flatfeet in our study sample. Although only a minor musculoskeletal impairment, early identification and correction should be provided to children with Down syndrome to prevent subsequent

biomechanical and postural problems [33]. However, a reduction in podiatry services were reported in 2004 indicating that children may no longer be receiving the level of specialist services they require. Atlantoaxial instability was diagnosed in 4.3% of children by cervical spine x-ray. These results are comparably lower than previous studies [34,35] However, these studies are relatively dated, and since their publication recommendations for routine screening for atlantoaxial instability have been revised such that routine screening is currently only indicated for children with Down syndrome who are symptomatic [36].

As previously mentioned, the use in GP services and combined specialist services were decreased in 2004 compared with the earlier cohort. Given the phasing out of specialist disability medical services at the Disability Services Commission in 2002, this trend is to be expected as specialist services may no longer be as accessible to families. Specialist services previously coordinated by the Disability Services Commission, for example audiologists, have shown reductions in service use, yet the conditions managed by these health professionals have increased, indicating the possibility that specialists services may not be as accessible to this population as they once were in 1997. With regards to the reduction in GP services, the general improvements in health of children with Down syndrome are likely to influence the frequency of GP service attendance. These reductions in GP service use, are a positive indicator that the general health of children with Down syndrome is improving.

In 2004 school-aged children with Down syndrome were reported to have increased day admissions to hospital, while overnight hospital admissions were significantly reduced compared with 1997. Interestingly, these trends in hospitalisations mirror that of the general population of school-aged children. However, young children with Down syndrome demonstrated a greater decrease in overnight hospital admissions compared with the general population, which may be indicative of improvements in health for children with Down syndrome.

The impact of socio-demographic factors on service use were outlined in this study and in places, showed an interesting contrast. Overnight hospital admissions and specialist

services use demonstrated divergent trends. While many socio-demographic factors impact on specialist services, few factors impact on overnight hospital admissions. The limited association of socio-demographic factors on overnight hospital admissions may demonstrate that overnight hospital admissions are not dependent on the socio-economic gradient and are equitable for all children. This contrasts with the findings relating to specialist service use which is greatly impacted on by socio-demographic factors. These results may provide an insight into the level of accessibility in specialist service use for families in Western Australia with a child with Down syndrome. However, some results presented were difficult to explain. For example, higher paternal education was associated with increased GP and specialist service use, while the lowest skill level for fathers also demonstrated increased rates of service use. Little explanation can be offered with regards to these findings, except that perhaps biases existed in the reporting of parents. Future research would be beneficial which analyses the impact of socio-demographic factors on service use for children with Down syndrome based on government Medicare data. This would also provide a comparison between parental report and government data to understand biases which may exist in parental reporting.

In interpreting the findings of this study several limitations should be acknowledged. As the data collection was based on a questionnaire, random recall error may have occurred whereby parents inaccurately recorded answers due to memory recall over time. In cases where there was missing data, results may be affected. Finally, the varied responses given by parents due to the differing terminology they may have used could have lead to possible encoding errors. However, although these limitations exist, this study provides a valuable insight into the changing health of children with Down syndrome and their service needs. To our knowledge, no other studies have described the changing health of children with Down syndrome over time. Furthermore, although limited research has described the increased medical service utilisation for children with Down syndrome [10-12], our study is unique in providing an insight into the changes in medical service use over time, and the impact socio-demographic factors may have on service use, which until now have been unexplored.

The information provided in this study has important implications for parents, health care professionals and education providers. A clear understanding of the co-morbidities experience by children with Down syndrome and the knowledge that a reduction in ongoing cardiac problems and improved health has occurred over time, will assist parents in making informed decisions regarding future plans or possible terminations of pregnancy. This information is also valuable to health care professionals in providing effective programs and interventions suitable for each child's physical capacities and limitations which minimise disability and facilitate development, functioning and increase quality of life for children with Down syndrome. Finally, this research contributes to the knowledge of medical service utilisation by this population and may assist in the planning and provision of disability services in Western Australia.

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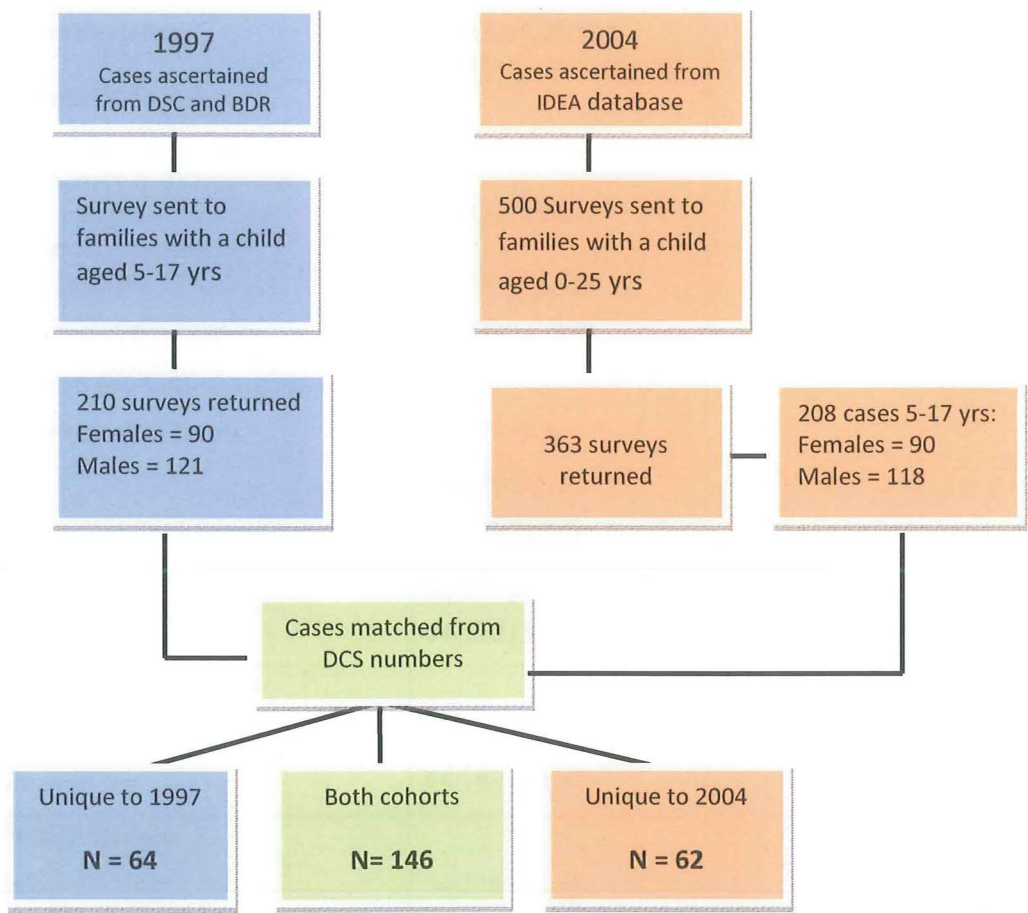
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Figure 1: The study process



**Table I: Demographic characteristics of families in 2004**

| <b>Characteristics</b>                                    | <b>Families, N(%)<br/>within each category</b> |
|---|--|
| <b>Parental Education</b>                                 |  |
| High maternal education (post high school qualifications) | 62/202 (30.7)                                  |
| Low maternal education (secondary education)              | 140/202 (69.3)                                 |
| High paternal education                                   | 44/200 (22.0)                                  |
| Low paternal education                                    | 156/200 (78.0)                                 |
| <b>Fathers occupational skill level</b>                   |  |
| 0 (Lowest skill level)                                    | 15/170 (8.8)                                   |
| 1   | 28/170 (16.5)                                  |
| 2   | 34/170 (20.0)                                  |
| 3   | 29/170 (16.1)                                  |
| 4 (Highest skill level)                                   | 64/170 (37.6)                                  |
| <b>Maternal work status</b>                               |  |
| Working   | 97/195 (49.75)                                 |
| Not working   | 98/195 (50.25)                                 |
| <b>Family income</b>                                      |  |
| Below \$20,000  | 34/208 (16.4)                                  |
| \$20,000 - \$31,999                                       | 20/208 (9.6)                                   |
| \$32,000 – \$51,999                                       | 37/208 (17.8)                                  |
| \$ 52,000 – \$77,999                                      | 34/208 (16.4)                                  |
| \$78,000 onwards  | 44/208 (21.1)                                  |
| Missing data  | 39/208 (18.8)                                  |
| <b>Place of Residence</b>                                 |  |
| Rural   | 164 (78.8)                                     |
| Metropolitan  | 44 (21.2)                                      |

**Table II: Specific medical conditions in children with Down syndrome in 2004.**

| <b>Condition</b>            | <b>Children reported to have the condition N (%)</b> |
|-----------------------------|--|
| <b>Cardiac: any</b>         | 95 (45.7)  |
| Ventricular septal defect   | 33 (15.9)  |
| Atrial septal defect        | 25 (12.0)  |
| Aventricular septal defect  | 16 (7.7)   |
| 'Hole in the heart'         | 11 (5.3)   |
| Patent ductus arteriosis    | 8 (3.9)  |
| Tetralogy of Fallot         | 2 (1.0)  |
| <b>Bowel: any</b>           | 66 (31.7)  |
| Constipation                | 41 (19.7)  |
| Duodenal atresia            | 9 (4.3)  |
| Coeliac disease             | 0 (0.0)  |
| Hirschsprung disease        | 0 (0.0)  |
| <b>Ear or hearing: any</b>  | 123 (59.1)   |
| Glue ear                    | 86 (41.3)  |
| Perforated ear drum         | 18 (8.7)   |
| Hearing loss                | 36 (17.3)  |
| <b>Eye or visual: any</b>   | 140 (67.3)   |
| Short sightedness           | 52 (25.0)  |
| Long sightedness            | 45 (21.6)  |
| Strabismus                  | 49 (23.6)  |
| Astigmatism                 | 18 (8.6)   |
| Nystagmus                   | 9 (4.3)  |
| Cataracts                   | 6 (2.9)  |
| <b>Thyroid disease: any</b> | 26 (12.5)  |
| Hypothyroidism              | 22 (22.6)  |
| Hyperthyroidism             | 3 (1.4)  |
| <b>Musculoskeletal</b>      | 74 (35.6)  |
| Atlantoaxial instability    | 9 (4.33)   |
| Flat feet                   | 47 (22.6)  |
| Scoliosis                   | 5 (2.4)  |
| Perthes                     | 3 (1.44)   |
| <b>Respiratory</b>          | 126 (60.0)   |
| Sleep apnoea                | 39 (19.8)  |
| Asthma                      | 42 (20.2)  |

**Table III: Medical conditions and episodic illnesses in school aged children with Down syndrome in 2004 compared with 1997.**

|  | Condition               | Odds Ratio | 95% Conf Interval        | P value        |
|--|-------------------------|------------|--------------------------|----------------|
| <b>Medical condition: ever</b>                           | Cardiac                 | 1.11       | 0.90 1.39                | 0.32           |
|  | Bowel                   | 1.68       | 1.16 2.45                | 0.01           |
|  | Ear                     | 1.13       | 0.81 1.56                | 0.47           |
|  | Eye                     | 0.79       | 0.55 1.15                | 0.23           |
|  | Thyroid                 | 1.08       | 0.69 1.70                | 0.73           |
| <b>Current problem associated with medical condition</b> | Cardiac                 | 0.52       | 0.28 1.00                | 0.05           |
|  | Bowel                   | 1.29       | 0.77 2.18                | 0.33           |
|  | Ear                     | 1.06       | 0.74 1.53                | 0.73           |
|  | Eye                     | 0.84       | 0.60 1.18                | 0.31           |
|  | Thyroid                 | 1.54       | 0.89 2.66                | 0.12           |
| <b>Frequency of episodic illnesses</b>                   | <b>Episodic illness</b> | <b>IRR</b> | <b>95% Conf Interval</b> | <b>P value</b> |
|  | Cold or flu             | 0.92       | 0.81 1.04                | 0.19           |
|  | Tonsillitis             | 0.56       | 0.38 0.83                | <0.001         |
|  | Pneumonia               | 0.90       | 0.31 2.57                | 0.84           |
|  | Bronchitis              | 0.80       | 0.52 1.23                | 0.32           |
|  | Ear infection           | 0.77       | 0.62 0.95                | 0.01           |

**Table IV: The use of medical services in 2004 compared with 1997.**

| Service                          | IRR        | 95% Conf. Interval        | P value        |
|----------------------------------|------------|---------------------------|----------------|
| GP service visits                | 0.91       | 0.83 1.00                 | 0.05           |
| Specialist services combined     | 0.92       | 0.84 1.01                 | 0.09           |
| Paediatrician                    | 0.96       | 0.72 1.28                 | 0.77           |
| Cardiologist                     | -          | -                         | -              |
| ENT specialist                   | 1.12       | 0.89 1.42                 | 0.34           |
| Disability specialist            | 0.11       | 0.05 0.22                 | <0.001         |
| Eye specialist                   | 0.97       | 0.76 1.22                 | 0.77           |
| Audiologist                      | 0.80       | 0.61 1.04                 | 0.09           |
| Podiatrist                       | 0.67       | 0.50 0.91                 | 0.01           |
| Dentist                          | 1.13       | 0.96 1.33                 | 0.14           |
| <b>Hospitalisations</b>          | <b>IRR</b> | <b>95% Conf. Interval</b> | <b>P value</b> |
| Day admissions                   | 1.20       | 0.81 1.76                 | 0.37           |
| Overnight admissions             | 0.60       | 0.37 0.96                 | 0.03           |
| Nights in hospital per admission | 0.33       | 0.24 0.44                 | <0.001         |

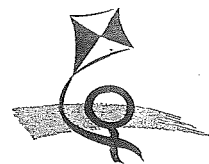
**Table V: The effects of socio-demographic factors on service use for children with Down syndrome in Western Australia in 2004.**

| <b>GP service use</b>                | <b>IRR</b> | <b>95% conf. Interval</b> |       | <b>P value</b> |
|--------------------------------------|------------|---------------------------|-------|----------------|
| High maternal education              | 1.09       | 0.90                      | 1.31  | 0.39           |
| High paternal education              | 3.33       | 2.52                      | 4.41  | <0.001         |
| Father's skill level 1               | 0.21       | 0.17                      | 0.28  | <0.001         |
| Father's skill level 2               | 0.14       | 0.10                      | 0.18  | <0.001         |
| Father's skill level 3               | 0.30       | 0.24                      | 0.38  | <0.001         |
| Father's skill level 4               | 0.16       | 0.13                      | 0.20  | <0.001         |
| \$20,000 - \$31,999                  | 0.88       | 0.60                      | 1.28  | 0.50           |
| \$32,000 - \$51,999                  | 0.86       | 0.63                      | 1.19  | 0.38           |
| \$ 52,000 - \$77,999                 | 1.13       | 0.82                      | 1.55  | 0.45           |
| \$78,000 onwards                     | 0.93       | 0.68                      | 1.26  | 0.64           |
| Missing data                         | 2.48       | 1.86                      | 3.29  | <0.001         |
| Metropolitan area                    | 1.00       | 0.81                      | 1.24  | 0.05           |
| Working mothers                      | 0.62       | 0.53                      | 0.73  | <0.001         |
| <b>Specialist service use</b>        |            |                           |       |                |
| High maternal education              | 1.18       | 0.98                      | 1.42  | 0.09           |
| High paternal education              | 1.63       | 1.30                      | 2.05  | <0.001         |
| Father's skill level 1               | 0.59       | 0.44                      | 0.79  | <0.001         |
| Father's skill level 2               | 0.63       | 0.47                      | 0.84  | 0.002          |
| Father's skill level 3               | 0.66       | 0.49                      | 0.89  | 0.006          |
| Father's skill level 4               | 0.60       | 0.46                      | 0.79  | <0.001         |
| \$20,000 - \$31,999                  | 1.67       | 1.16                      | 2.40  | 0.006          |
| \$32,000 - \$51,999                  | 1.80       | 1.31                      | 2.48  | <0.001         |
| \$ 52,000 - \$77,999                 | 1.21       | 0.86                      | 1.69  | 0.28           |
| \$78,000 onwards                     | 1.20       | 0.87                      | 1.66  | 0.28           |
| Missing data                         | 1.24       | 0.88                      | 1.74  | 0.21           |
| Metropolitan area                    | 1.05       | 0.88                      | 1.27  | 0.58           |
| Working mothers                      | 0.85       | 0.72                      | 0.99  | 0.04           |
| <b>Overnight hospital admissions</b> |            |                           |       |                |
| High maternal education              | 1.28       | 0.48                      | 3.44  | 0.48           |
| High paternal education              | 0.99       | 0.29                      | 3.34  | 0.98           |
| Father's skill level 1               | 0.44       | 0.12                      | 1.65  | 0.22           |
| Father's skill level 2               | 0.32       | 0.08                      | 1.26  | 0.10           |
| Father's skill level 3               | 0.15       | 0.03                      | 0.833 | 0.03           |
| Father's skill level 4               | 0.29       | 0.08                      | 0.99  | 0.49           |
| \$20,000 - \$31,999                  | 0.30       | 0.18                      | 5.04  | 0.40           |
| \$32,000 - \$51,999                  | 0.49       | 0.07                      | 3.72  | 0.49           |
| \$ 52,000 - \$77,999                 | 3.91       | 0.83                      | 18.40 | 0.08           |
| \$78,000 onwards                     | 0.86       | 0.15                      | 5.03  | 0.87           |
| Missing data                         | 2.65       | 0.54                      | 12.95 | 0.22           |
| Metropolitan area                    | 1.39       | 0.56                      | 3.44  | 0.48           |

|                                |      |      |      |       |
|--------------------------------|------|------|------|-------|
| Working mothers                | 0.76 | 0.34 | 1.69 | 0.50  |
| <b>Hospital day admissions</b> |      |      |      |       |
| High maternal education        | 5.15 | 1.71 | 15.6 | 0.004 |
| High paternal education        | 1.90 | 0.83 | 4.38 | 0.13  |
| Father's skill level 1         | 0.23 | 0.08 | 0.63 | 0.004 |
| Father's skill level 2         | 0.49 | 0.21 | 1.17 | 0.12  |
| Father's skill level 3         | 0.30 | 0.11 | 0.81 | 0.02  |
| Father's skill level 4         | 0.28 | 0.12 | 0.66 | 0.004 |
| \$20,000 - \$31,999            | 1.68 | 0.51 | 5.50 | 0.39  |
| \$32,000 – \$51,999            | 2.15 | 0.72 | 6.44 | 0.17  |
| \$ 52,000 – \$77,999           | 1.14 | 0.34 | 3.84 | 0.83  |
| \$78,000 onwards               | 0.66 | 0.19 | 2.31 | 0.52  |
| Missing data                   | 1.04 | 0.31 | 3.44 | 0.95  |
| Metropolitan area              | 0.96 | 0.47 | 1.95 | 0.91  |
| Working mothers                | 0.60 | 0.33 | 1.09 | 0.09  |

# Appendix A





Dr Helen Leonard  
Telethon Institute for Child Health Research  
PO Box 855  
WEST PERTH WA 6872

Dear Dr Leonard

**REGISTRATION NUMBER:** 1003/EP

**TITLE:** The health, functioning and needs of  
children and young adults with Down  
Syndrome in 2004

**REFERENCE NUMBER:** EC04-55.1

**MEETING DATE:** 15 July 2004

The Ethics Committee has recommended approval be given for you to undertake the abovenamed research study. This recommendation has been ratified by the Women's and Children's Health Service.

The Ethics Committee does however wish to be informed immediately of:

- I. any untoward effects experienced by any participant in the trial where those effects in degree or nature were not anticipated by the researchers, and steps taken to deal with these,
- II. substantial changes in the research protocol together with an indication of ethical implications, and
- III. other unforeseen events.

The Ethics Committee has been charged with the responsibility of keeping the progress of all approved research under surveillance. A copy of the final result must be forwarded to the Committee upon completion of the research or if the research is not completed within twelve months you are asked to submit a progress report and annually thereafter. This information should include:

- a) The status of the project (completed/in progress/abandoned/not commenced). In the event that a project does not commence within 12 months of being approved by the Ethics Committee the study must be resubmitted to the Committee for approval.
- b) Compliance with conditions of ethical approval, including security of records and procedures for consent.



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Hospital for Women

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Princess Margaret Hospital  
for Children

Roberts Road  
Subiaco WA 6008

GPO Box D184  
Perth WA 6840

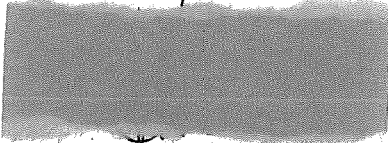
Tel: (08) 9340 8222  
Fax: (08) 9340 8111

- c) Compliance with any special conditions stated by the Ethics Committee as a condition of approval.
- d) Results from the study to date, including outcome.

Please note that approval for studies is for three years and the research should be commenced and completed within that period of time. Projects must be resubmitted if an extension of time is required. In the event that a project does not commence within 12 months of being approved by the Ethics Committee the study must be resubmitted to the Committee for approval.

**Please quote the above registration number on all correspondence.**

Yours sincerely



**Dr Geoff Masters  
Executive Director  
Medical Services**

16 July 2004

cc Professor Carol Bower, Professor Nick de Klerk, Professor Sven Silburn

- **The Ethics Committee is constituted, and operates in accordance with the National Health and Medical Research Council's National Statement on Ethical Conduct in Research Involving Humans**



Department of Health  
Government of Western Australia

Child and Adolescent Health Service

Dr Helen Leonard  
ICHR  
Roberts Road  
SUBIACO WA 6008

Dear Dr Leonard

**REGISTRATION NUMBER:** 1003/EP

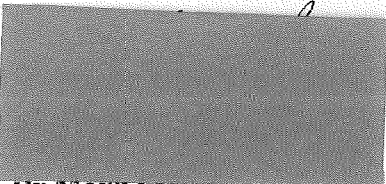
**TITLE:** The health, functioning and needs of children and young adults with Down Syndrome in 2004

**MEETING DATE:** 15 May 2008

The Princess Margaret Hospital for Children Ethics Committee has recommended approval of a 3 year extension to this study. **This extension will expire on 15 July 2010.** This recommendation has been ratified and confirmed by the Child and Adolescent Health Service.

It should be noted that all other aspects of the approval remain unchanged. This is so, in particular, in relation to the progress reports required and regarding any further amendments to the protocols.

**Please quote the above registration number on all correspondence.**



**Dr Mark Salmon**  
**Executive Director**  
**Medical Services**

28 May 2008

- The Ethics Committee is constituted, and operates in accordance with the National Health and Medical Research Council's National Statement on Ethical Conduct in Research Involving Humans



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[www.cahs.health.wa.gov.au](http://www.cahs.health.wa.gov.au)



TO WHOM IT MAY CONCERN

ETHICS COMMITTEE APPROVAL FOR HONOURS STUDENT PROJECT 1997  
CANDIDATE SEONAI LEONARD B.Sc (UWA)

CHIEF SUPERVISORS: DR BEVERLEY PETTERSON BSc. (Hons) PhD.  
DR CAROL BOWER MBBS MSc. PhD FAFPHM

TITLE: A WESTERN AUSTRALIAN DOWN SYNDROME STUDY

This department approves this project and access to client files for the following reasons:

1. The student is under the supervision of Dr Petterson who is currently employed by Disability Services Commission in a research capacity.
2. The student will have signed a standard Public Service Confidentiality Statement which encompasses standard protocols. Working files and the final report will not contain any identifying information.
3. We foresee that this study will provide important practical data for prevention and management of disability in people with Down syndrome and for their families.



Dr J Crowhurst  
ASSISTANT DIRECTOR,  
MEDICAL SERVICES

February 18, 1997 sd/8301/M&SS

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app44:178/EP  
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340 8245



8 April 1997

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Ms Seonaid Leonard  
Department of Anatomy & Human Biology  
University of Western Australia  
NEDLANDS WA 6009

Dear Ms Leonard

**REGISTRATION NUMBER:** 178/EP

**TITLE:** A Western Australian Down Syndrome Study

**REFERENCE NUMBER:** EC97-11.5 20 March 1997

The Ethics Committee has recommended and the Board of Management have ratified approval be given for you to undertake the abovenamed research study.

The Ethics Committee does however wish to be informed immediately of:

- I. any untoward effects experienced by any participant in the trial where those effects in degree or nature were not anticipated by the researchers, and steps taken to deal with these,
- II. substantial changes in the research protocol together with an indication of ethical implications, and
- III. other unforeseen events.

The Ethics Committee has been charged with the responsibility of keeping the progress of all approved research under surveillance. A copy of the final result must be forwarded to the Committee upon completion of the research or if the research is not completed within six months you are asked to submit a progress report and annually thereafter. This information should include:

- a) The status of the project (completed/in progress/abandoned/not commenced).
- b) Compliance with conditions of ethical approval, including security of records and procedures for consent.

King Edward Memorial Hospital For Women, Perth, Western Australia  
Princess Margaret Hospital For Children, Perth, Western Australia

# Appendix B

# Down Syndrome Research and Practice Submissions Requirements

Further policies and guidelines

- [Content guidelines](#)
- [Standards](#)
- [Publication policies](#)
- [Editorial process](#)
- [Structure and style](#)

All submissions to *Down Syndrome Research and Practice* should meet the following requirements.

## Cover letter

Please include a cover letter explaining why the manuscript is suitable for publication in *Down Syndrome Research and Practice*.

## Prior publication

All authors are asked to indicate that they have not submitted a similar manuscript for publication elsewhere. If related work has been submitted elsewhere, then a copy must be included with the article submitted to *Down Syndrome Research and Practice*.

## Listed authors

All authors must consent to the submission of the manuscript. All authors will be contacted to confirm their approval. The involvement of any professional medical writer in publication must be declared (see the [European Medical Writers Association \(EMWA\) guidelines \[PDF\]](#) on the role of medical writers in developing peer-reviewed publications). Please refer to [section II.A. of the Uniform Requirements for Manuscripts Submitted to Biomedical Journals: Writing and Editing for Biomedical Publication](#) for guidance about recognising authorship.

## Confirmation of ethics approval

All research involving humans and animals must have been approved by the authors' institutional review board or equivalent committee, and that board must be named by the authors. In the case of human participants, informed consent must have been obtained, and all clinical investigation must have been conducted according to the principles expressed in the [Declaration of Helsinki](#). Authors should submit a statement from the ethics committee or institutional review board indicating approval of the research.

## Informed consent to research

We encourage authors to submit a sample of their participant consent form and may sometimes require a copy. Authors must confirm that informed consent was properly obtained from participants.

For further information, see sections 20-27 of the [Declaration of Helsinki](#) and [section 8.02 of the APA's Ethical Principles of Psychologists and Code of Conduct](#).

## Patient privacy and informed consent for publication

Please refer to [section II.E. of the Uniform Requirements of the International Committee of Medical Journal Editors](#). Complete anonymity is difficult to achieve, and informed consent for publication should be obtained if there is any doubt. When informed consent has been obtained it should be indicated in the published article.

## Competing interests

All authors will be asked to declare whether they have any financial, personal or professional interests that could be construed to have influenced their paper. Reviewers are also asked to declare any interests that might interfere with their objective assessment of a manuscript. Any relevant competing interests of authors must be available to editors and reviewers during the review process and will be stated in published articles.

## Epidemiological Studies

Reports of epidemiological studies should use the [STROBE initiative](#) as a guide.

## Registration of clinical trials

*Down Syndrome Research and Practice* supports the position of the International Committee of Medical Journal Editors (ICMJE) on trial registration. All trials initiated after 1 July 2005 must be registered prospectively in a publicly accessible registry (i.e., before patient recruitment has begun), or they will not be considered for publication. For trials initiated before 1 July 2005, all trials must be registered before submission. The trial's registration number must be provided at the time of submission. See the [ICMJE's Frequently Asked Questions \[PDF\]](#) for further information.

## CONSORT reporting guidelines for clinical trials

For reports of clinical trials, authors must submit original protocols as supporting information to allow editors and reviewers to assess manuscripts fully. Any deviation from the protocol must be explained. Authors of clinical trials must adhere to the [CONSORT](#) reporting guidelines appropriate to their trial design. All reports of the results of randomised clinical trials should include the [CONSORT](#) flow diagram, and authors should complete and submit the checklist contained within the [CONSORT](#) statement.

## Systematic reviews and meta-analyses of randomised controlled trials

Reports of meta-analyses of randomized controlled studies should use the [QUOROM statement](#) as a guide, and should include a copy of the [QUOROM checklist \[PDF\]](#).



## Support better education for children with Down syndrome



**Children with Down syndrome deserve better education.**

We are working to ensure that every child with Down syndrome benefits from the best possible opportunities.

Our work depends on the support of donors around the world.

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# Down Syndrome Research and Practice

## Structure and Style

Further policies and guidelines

- [Content guidelines](#)
- [Standards](#)
- [Publication policies](#)
- [Editorial process](#)
- [Submission requirements](#)

The following describes the structure and style expected of submissions to *Down Syndrome Research and Practice*.

Further guidance about the structure, style and presentation of papers for publication can be found in -

- [APA Guide to Preparing Manuscripts for Journal Publication](#)
- [Section IV of the Uniform Requirements for Manuscripts Submitted to Biomedical Journals: Writing and Editing for Biomedical Publication](#)
- [Publication manual of the American Psychological Association \(5th edition\)](#)
- [Scientific style and format - The Council of Science Editors' Manual for Authors, Editors, and Publishers \(7th edition\)](#)

## Readability

*Down Syndrome Research and Practice* is an international journal covering many disciplines and many subjects, and is read by people from many cultural, scientific and socioeconomic backgrounds.

Contributions should therefore be written clearly and simply so that they are accessible to the broadest range of readers, including those for whom English is not their first language.

## Format

Contributions should be double-spaced and written in English (spellings as in the [Oxford English Dictionary](#)).

Contributions should be organised in the sequence: title, text, methods, references, Supplementary Information (if any), acknowledgements, author contributions, author information (containing data deposition statement, interest declaration and corresponding author line), tables and figure legends.

Hard copies of manuscripts are not required. Text files can be submitted in the following formats: Word, WordPerfect, Rich Text Format (RTF) and Portable Document Format (PDF). Graphics files can be submitted in the following formats: Portable Document Format (PDF), Encapsulated PostScript (EPS),

Excel, Portable Network Graphics (PNG), Adobe Illustrator, Joint Photographic Experts Group Interchange Format (JPEG), Photoshop, PowerPoint, or Tagged Image File Format (TIFF).

## References

Only published or accepted papers and books should be included in the reference list. Meetings abstracts, conference talks, or papers that have been submitted but not yet accepted should not be cited. Limited citation of unpublished work should be included in the body of the text only. All personal communications should be supported by a letter from the relevant authors.

From 2007 (volume 12), *Down Syndrome Research and Practice* uses the numbered citation (citation-sequence) method. References are listed and numbered in the order that they appear in the text. In the text, citations should be indicated by the reference number in ([square]) brackets. Multiple citations within a single set of brackets should be separated by commas. Where there are more than three sequential citations, they should be given as a range.

To assist electronic linking, references must follow the following format. The format is based on the International Committee of Medical Journal Editors Uniform Requirements for Manuscripts Submitted to Biomedical Journals: Sample References, which are maintained by the [US National Library of Medicine Bibliographic Services Division](#). This standard is sometimes known as the [Vancouver system](#) and broadly conforms to [ISO-690](#). The differences from this format are:

1. All authors should be listed in the reference list (as per [NLM's standard](#)).
2. Journal titles should not be abbreviated. Full journal titles are more accessible to a wide readership.
3. Journal and book titles should be italicised (but not journal article or book chapter titles).

Wherever possible, please provide DOIs with references. Some examples follow:

### *Journal article*

1. Anupama R, Hills RK, Stiller C, Gibson BE, Graaf SSN, Hann IM, O'Marcaigh A, Wheatley K, Webb DKH. Treatment for myeloid leukaemia of Down syndrome: population-based experience in the UK and results from the Medical Research Council AML 10 and AML 12 trials. *British Journal of Haematology*. 2006;132(5):576-583. doi:10.1111/j.1365-2141.2005.05906.x.

### *Book*

2. Bird GA, Buckley SJ. *Number skills for individuals with Down syndrome - An overview*. Portsmouth, UK: Down Syndrome Education International; 2001.

### *Book chapter*

3. Buckley SJ, Bird GA, Byrne A. The practical and theoretical significance of teaching literacy skills to children with Down's syndrome. In: Rondal JA and Perera J, editors. *Down's syndrome: psychological, psychobiological and socio-educational perspectives*. London: Whurr; 1996. p. 119-128.

## Acknowledgments

People who contributed to the work, but do not fit the criteria for authors should be listed in the Acknowledgments, along with their contributions. You must also ensure that anyone named in the acknowledgments agrees to being so named.

## Funding

The sources of funding that have supported the work should be described in a section titled 'Funding'. Please also describe the role of the study sponsor(s), if any, in study design; collection, analysis, and interpretation of data; writing of the paper; and decision to submit it for publication.

## Author contributions

A detailed list of the contributions of each of the authors should be included.

## Competing interests

Competing interests associated with any of the authors must be detailed. If authors declare that no competing interests exist, then this should be stated in the contribution.

## Abbreviations

Please keep abbreviations to a minimum. A list of definitions for all non-standard abbreviations should be provided. Non-standard abbreviations should not be used unless they appear at least three times in the text.

## Nomenclature

The correct and established nomenclature should be used wherever possible. In particular:

- We strongly encourage the use of SI units. If you do not use these exclusively, please provide the SI value in parentheses after each value.
- Species names should be italicized (e.g., *Homo sapiens*).
- Genes, mutations, genotypes, and alleles should be indicated in italics. Use the recommended name by consulting the appropriate genetic nomenclature database, e.g., HUGO for human genes. It is sometimes advisable to indicate the synonyms for the gene the first time it appears in the text.
- The Recommended International Non-Proprietary Name (rINN) of drugs should be provided.

## Accession numbers

All appropriate datasets, images, and information should be deposited in appropriate public resources. Please provide the relevant accession numbers (and version numbers, if appropriate). In addition, as much as possible, please provide accession numbers or identifiers for all entities such as genes, proteins, mutants, diseases, etc., for which there is an entry in a public database. Providing accession numbers allows linking to and from established databases and integrates your article with a broader collection of scientific information. Please list all accession numbers directly after the Supporting Information section.

## Supplementary information

This is online-only, peer-reviewed material that is essential background to the contribution (for example, large data sets, methods, calculations), but which is too large or impractical, or of interest only to a few specialists, to justify inclusion in the printed version of the paper.

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