1995

Difficulties Experienced by Year 10 Students When Solving Monohybrid Autosomal and X-Linked Genetic Problems

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DIFFICULTIES EXPERIENCED BY YEAR 10 STUDENTS WHEN SOLVING MONOHYBRID AUTOSOMAL AND X-LINKED GENETIC PROBLEMS

By

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A Thesis Submitted in Partial Fufilment of the Requirements for the Award of Bachelor of Education with Honours at the Faculty of Education, Edith Cowan University

Date of Submission: 10 April 1995

USE OF THESIS

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

ABSTRACT

Genetics is an important aspect of secondary education as it improves students' understanding of the inheritance of genetic diseases which are present in many families. Genetics also provides opportunities for students to learn important problem solving skills.

Students experience difficulties with problem solving in genetics as they tend to rote learn algorithmic methods and not have a meaningful understanding of the concepts of meiosis, gametes and fertilisation which underpin genetics problems. Following instruction in the area of genetics, think-aloud protocols were collected from 20 Year 10 students, while solving four genetic problems. The students were also interviewed to probe their understanding of concepts related to the genetic problems. This enabled the researcher to explore how well students understood the concepts in relation to the algorithms they used to solve the four problems.

This research found that students are more successful in autosomal and forwards working problems than in X-linkage and backwards working problems. There is a low level of meaningful problem solving and a poor understanding of the terms genotype, phenotype, meiosis, gametes and fertilisation. Students also have difficulties understanding why X-linked characteristics are inherited in the way they are, and the difference in structure of the X and Y chromosomes.

DECLARATION

I certify that this thesis does not incorporate, without acknowledgement, any material submitted for a degree or diploma in any institution of higher education and that, to the best of my knowledge and belief, it does not contain any material previously published or written by another person where due reference is made in the text.

Christina Williams

ACKNOWLEDGEMENTS

I acknowledge, with many thanks, the support, advice and neverending encouragement provided by my supervisor, Dr Mark Hackling. His constant encouragement and willingness to help contributed much to the completion of this study.

I also acknowledge, with appreciation, my boyfriend Rod and my family for their support through the times when I thought this thesis would never be completed. A special thank you to my sister Alison is needed here as her expertise with computers was invaluable.

Finally, I wish to acknowledge the 20 students who were willing to be interviewed. Their input into this study was invaluable. My professional colleagues also must be thanked for their cooperation and input.

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CHAPTER 1 INTRODUCTION

Background to the Study

Genetics is a difficult subject that involves many difficult concepts. To understand genetics in a meaningful way, the student needs to be encouraged to link new knowledge to the previous concepts that have already been learnt (Ausubel, Novak & Hanesian, 1978).

One of the concepts commonly taught first in the subject of genetics is that of meiosis which is the process by which the male and female gametes are produced. Students are then taught about fertilisation which produces the zygote containing a combination of the father and mother's genetic material. It is usually later in the course that the students learn to solve genetic problems using the Punnett square method. Students usually will easily master the algorithm of solving these problems, and will be able to successfully do so, but no conscious link is made between these problems and the earlier taught concepts of meiosis, gametes and the process of fertilisation (Stewart, 1983).

The Problem

Studies have revealed that many students have an apparent lack of understanding of meiosis, gametes and fertilisation and the role they play in autosomal and X-linked inheritance (Stewart, 1982). Students also rote learn algorithmic methods of solving genetics problems which enable them to produce correct solutions even though the students are unable to explain the steps of their solution in terms of the underlying conceptual knowledge (Stewart, 1983). Stewart (1985) argues that:

> Procedural knowledge of how to execute a problem solution and conceptual knowledge of concepts, laws and theories which provide meaning or context to

the procedures are both necessary for a meaningful solution to any problem (p. 731)

Significance of the Study

This study will contribute to an understanding of the difficulties that students experience in solving genetic problems. This can lead to more effective instruction in genetics. An improved understanding of genetics may help students understand the advice given by genetic counsellors later in their lives.

This study adds to the literature already written in this area, as it is the first study conducted in Australia involving Year 10 students. Previous studies have been completed at the upper secondary and tertiary levels, this study should therefore benefit secondary teachers, and help them to implement more effective measures for teaching genetics in the classroom.

Purpose and Research Questions

The purpose of this study was to trace the procedural steps taken by Year 10 students while they solved genetic problems, and then probe their understanding of the conceptual basis of what they have done. More specifically, this study addressed the following research questions.

(1) What procedural steps are taken by Year 10 students in solving monohybrid cross problems involving autosomal and X-linked traits?

(2) Can students explain their problem solving steps in terms of genes, chromosomes, meiosis, gametes and fertilisation?

(3) What are the main conceptual and procedural barriers to the production of meaningful solutions to these problems?

CHAPTER 2 LITERATURE REVIEW

Theoretical Framework

Most studies of problem solving today are conducted within the information processing paradigm (Larkin & Rainard, 1984). A typical problem solving task would involve a student in analysing a problem situation, using existing knowledge to create a mental model of the problem, selecting one of a number of possible steps towards its solution, implementing these steps, and evaluating the resulting solution. At each step, the solver is using some form of information processing. Bourne and Ekstrand (1985, p. 205) note:

> A problem solver first analyses the task or problem. The purpose of this analysis is to build on internal memory representation of the problem. This representation is called a 'problem space'. All efforts to solve a problem are conducted within the solver's problem space.

Memory models describe how information is stored in long term memory, and how this current knowledge organisation is continuallly interacting with new knowledge during learning and problem solving (Stewart, 1985).

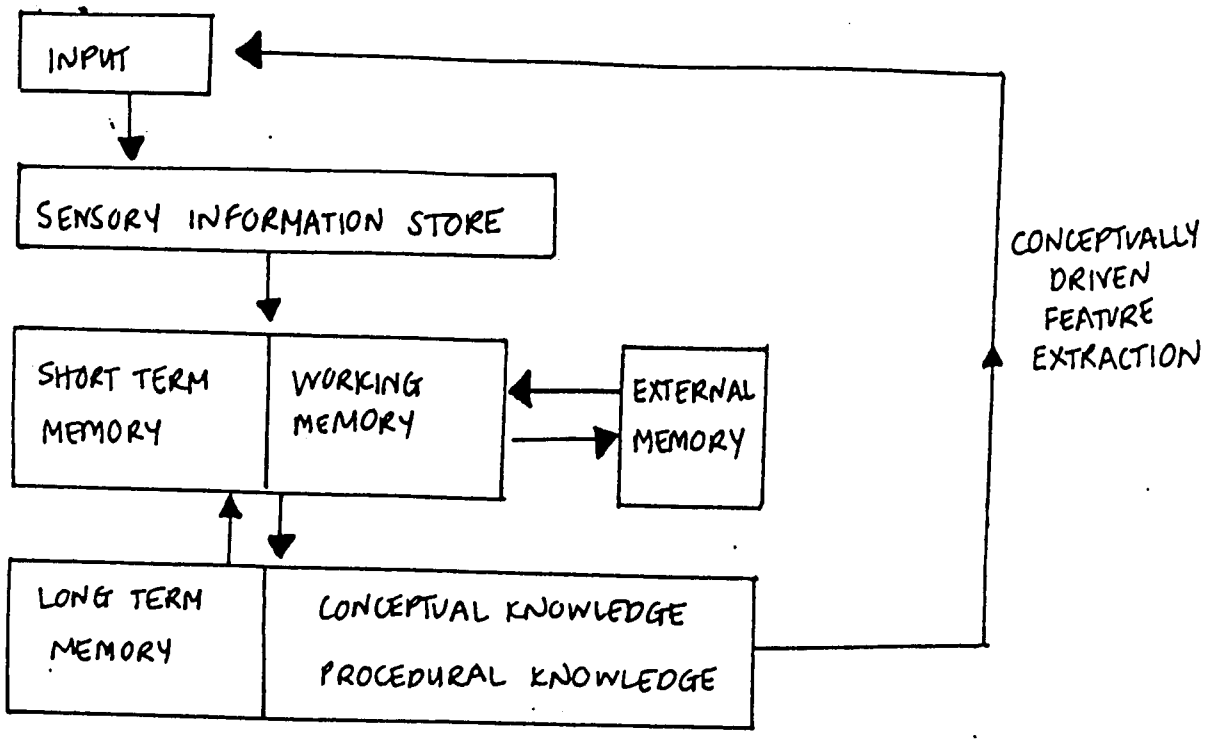

Figure 1. An outline of human memory as proposed by Stewart & Atkin (1985) based on Lindsay and Norman (1977)

Humans are capable of performing complex tasks because of their cognitive abilities, although there are limits to how much information can be held in short term memory. Short term memory serves as a mental 'work space' for solving problems and only has a limited storage capacity of $7^{+}2$ chunks (Atkinson, Atkinson, Smith, Ben & Hilgard, 1990).

To successfully solve a problem, conceptual and procedural knowledge must be retrieved from long term memory and applied to the problem solving process (Chi & Glaser, 1985). Knowledge in memory is highly interrelated.

The importance of the organisation and structure provided by current schemata is emphasised by Chi and Glaser (1985) as efficient organisation will allow relevant knowledge to be retrieved from long term memory. A lack of access to knowledge because of inadequate structuring may be the reason for failure to solve a problem.

Atkinson et al. (1990) found that information is usually encoded according to its meaning. If there are inadequate connections between items to be remembered, memory can be impaired. To improve this, meaningful connections must be added that will provide retrieval paths. The more one elaborates the meaning, the better retrieval from memory will be.

Previous Research Relevant to the Topic

Genetics is often dealt with in a drill and practice nature (Brown, 1990) and this approach often leads to misconceptions of meiosis, and a lack of understanding of how this concept relates to genetic problem solving. Longden (1982) suggests that these difficulties could be related to the separation in teaching time between presentation of meiosis and the introduction of genetic problem solving. As one of Longden's (1982) interviewed students stated:

> I find that...err.. I put meiosis separately on its own and then mitosis and DNA, finally protein synthesis...all as separate categories and don't let them flow into one another so that when I come to do the genetic problems I see it only as a mathematical problem not as part of the whole process. (p. 139)

Hackling and Treagust (1984) reported that Year 10 students fail to understand segregation of chromosomes and genes during meiosis. Most students are aware that the sperm and egg each carry half the genetic information to the offspring, but a common misconception (48% of the students) is that the sperm carry genes for half the features found in the offspring, rather that one of two alleles needed for each feature. The majority of students were not aware of the process by which the gametes were produced. (p. 206)

In earlier research, Hackling (1982) found that few students defined the concepts of meiosis, gametes and fertilisation in terms of their relationship to chromosomes and

genes. He also found the concepts of gametes and fertilisation were far more readily associated with sexual reproduction than with inheritance. Hackling (1982) noted that "...only 6% of students associated inheritance with sexual reproduction" (p. 17), indicating that the behaviour of chromosomes and genes from parent cell through meiosis to gametes, and then to fertilisation is very poorly understood. These concepts seem to bear no relation, in many students, to the inheritance problems they solve. Smith and Simmons (1992) note:

> In many cases, this failure to relate genes and chromosomes appears to be due, not so much to incorrect or missing concepts, but to poorly developed or absent linkages between concepts...Where these functional relationships have not been internalised, students will not recognise and interpret behavious of genes and alleles as consequences of chromosomal events during meiosis. (p. 49)

Stewart (1982) argued that meaningful solutions are ones in which students are able to explain why they have carried out each step in their solution of a genetic problem. To faciliatate a more meaningful understanding of genetics in terms of the relationship between meiosis, genetics, fertilisation and inheritance problems, the teaching in this area must be investigated closely. Hackling (1982) suggests that the relationship must be made much more explicit and be firmly established in the cognitive structure of the student. To do this, the concept of inheritance should be introduced early in the topic so it can act as a subsumer for the less generalised topics such as meiosis, gametes and fertilisation. Cho, Kahle and Nordland (1985) carried out an investigation of the organisation of three textbooks commonly used in the teaching of genetics. Cho et al. (1985, p. 710) reported:

> All three textbooks researched treated meiosis and genetics in separate chapters, with meiosis preceding genetics. Also meiosis was isolated from that of heredity. The topic of heredity was followed by the concepts of chromosomal theory and of genetic material in that order.

The inclusion of meiosis with mitosis may improve student's understanding of cellular reproduction, but this separation from genetics is likely to impede student's ability to relate meiosis to inheritance problems.

Hackling (1983) found that only 10% of students in his research comprehended that gametes carry one chromosome and one gene from each pair. This lack of knowledge affects students's ability to successfully solve monohybrid autosomal and X-linkage problems. Many students who do not understand X-linkage and why males are normally affected whereas females are often carriers of X-linked recessive disorders. Students would benefit from the genes-on-chromosome model (Hackling, 1990) which shows gametes as a separate and distinctive cells which carry one chromosome and one allele from each pair found in the body cells of the parent.

When solving problems in a typical high school genetics course, students are commonly exposed to two types of problems. One requires the student to reason from causes to effects (eg. to predict offspring genotype and phenotype from given parental data) and one which asks students to reason from effects to causes (eg. determining parental genotypes from offspring phenotype data) (Stewart, 1988). The first type of problem is often carried out algorithmically with little conceptual knowledge, but the second type of problem requires students to make judgements and decisions about parental genotypes. They have to generate hypotheses and exercise judgement as to it being either accepted or rejected. Stewart (1988) notes

> Since typical textbook problems are of the cause-to-effect type and can be solved algorithmically, there is no necessity to, and thus no guarantee that students will use all the conceptual knowledge that teachers expect. (p. 252)

Stewart (1988) also suggests that students may benefit from observing an expert solve effect-to-cause problems and gain insight into the logic of an experts approach. This will help students develop procedures that are efficient and meaningful.

Methodological Issues

Studies by Stewart (1982; 1985, Hackling and Treagust (1984), and Longden (1982), as well as many other researchers have demonstrated the contribution to understanding that can be gained by analysing recorded interviews with students.

Areas of difficulty in genetic problem solving can easily be identified by the use of think-aloud interviews in which the student talks out loud their thinking as they attempt various problems. The transcribed protocol can be used to identify the procedural steps and conceptual knowledge used in the problem solving task. Larkin and Rainard (1984) comment:

> In a useful protocol, the solver talks steadily, continuously reflecting what he or she is doing. There are only small amounts of neutral speech from the experimenter. There are no constraints that may guide or divert the solver's thinking...Even when a solver has apparently completed his or her work on a problem, it is important that the experimenter does not begin to talk too soon. Often a subject will give a correct answer to it, but not recognise it. By remaining silent after answer is given, one learns how the subject evaluates his or her own answer. (p.250)

When the student has finished solving the problem, the interviewer can conduct a debriefing in which the student's understanding of the conceptual basis of the problem can be probed. Interviews develop a personal relationship between the subject and the interviewer, which will motivate the student to give accurate and complete answers. Questions must be answered, and a guess by the students can easily be identified by the interviewer. Elaboration of any explanation can be asked for, and any other interesting responses can be further investigated. By communicating through talking, the interviewer can gain information directly from the student without being limited by the

student's skills of written expression. Students can clarify the meaning of questions, and the interviewer can clarify the meaning of the student's answers.

CHAPTER 3 METHODOLOGY

Research Design

Following regular instruction, students solved genetic problems, while thinking out aloud. This was followed by a debriefing in which the interviewer asked the subject to justify the steps taken in terms of the related conceptual knowledge.

Subjects

Twenty Year 10 students were selected at random from a high school with a total population of 156 Year 10 students. Subjects were selected using stratified sampling in which two males and two female students were chosen at random from each of the five Year 10 science groups which were all taught by different teachers.

Problems

Students solved four genetic problems with concurrent verbalisation. Two of these problems were forward working, and two required the student to work backwards. Two problems involved autosomal inheritance, and two involved X-linkage. These four problems are as follows:

Problem One

Blue eye colour is determined by a recessive gene. Brown eye colour is dominant. If Mr Jones is a brown eyed heterozygous male and Mrs Jones is a brown eyed heterozygous female, what are the possible genotypes and phenotypes of the offspring?
What is the probability that these two parents can produce a blue eyed child?
Mrs Jones is pregnant. What could be the colour of the baby's eyes?

Problem Two

Colourblindness is a recessive trait which is carried on the X chromosome. If a non-colourblind male has children with a female who has normal vision but is a carrier for colourblindness, what are the possible genotypes and phenotypes of the offspring produced?

What is the probability of these parents producing a son who is colourblind?

Problem Three

Haemophilia is a recessive trait which is carried on the X chromosome.

A man has been diagnosed as having the disorder of haemophilia. If this man's father is a non-haemophiliac, what is the genotype of the mother?

Problem Four

Big ears is inherited as a dominant trait to small ears.

A couple who both have big ears produce one child with small ears, and one child with big ears. What are the possible genotypes of the parents?

Debriefing Questions

After problem solving was completed, the following questions were asked of the students. These questions sought to probe students' understanding of the conceptual basis of the problems.

(1) Can you explain why you separate the genotypes (eg. Bb) into separate components when solving the problems using a Punnett square?

(2) Could you explain what gametes are? On the Punnett square, could you identify the gametes for me?

(3) How are the gametes produced? What is the process called?

(4) Could you explain for me, how this process occurs?

(5) Could you place where this process must occur in reference to the Punnett square?

(6) What is fertilisation? When would fertilisation have to occur in this Punnett square?

(7) You have outlined what the offspring are, and you have used the terms genotype and phenotype. Can you explain these terms for me, in reference to the answers that you have written down.

(8) You have also completed some problems with X-linked inheritance for me. Why are males most often affected if they carry the affected allele, and females are normally carriers of X-linked traits?

(9) Can you tell me of any difference between the X and Y chromosome in appearance? Does this play a part in how X-linked characteristics are inherited?

Data Collection and Analysis

The interview instrument was carefully constructed so that there was a sequence of probes to be used in the debriefing after the four problems had been solved. Early probes did not influence the response to probes later in the sequence. This was ensured through appraisal of the instrument by science educators and pilot studies.

After the interview instrument was developed, pilot testing with 10 Year 10 students was conducted. This helped to identify incorrect sequenceing and ambiguous questions which were identifed and corrected before interviewing the subjects of the study.

The data were analysed in terms of eight problem solving steps. These steps were based on studies conducted by Stewart (1985) and Hackling and Lawrence (1988), and are as follows:

STEP A: Construction of symbolic key to alleles, before commencing the problem solution.

STEP B: Determination of parent genotypes and gamete types.

STEP C: Determination of offspring genotypes and phenotypes.

STEP D: Determination of parental genotypes given offspring genotypes and limited detail of parents.

STEP E: Relationship of meiosis, gametes and fertilisation to parental genotypes and gametes.

STEP F: Understanding of X-linked inheritance, and how genes are carried on the X chromosome and not on the Y chromosome.

STEP G: Understanding of why males mainly show X-linked recessive traits, and females are mainly carriers.

Data regarding steps A, B, C and D were generated as students worked on the problem. Data sources included the taped think aloud protocol and the students written workings on the problem sheet.

Data regarding subgoals E, F and G were generated in the debriefing following students completion of the problem. These data were recorded on tape.

CHAPTER 4 RESULTS

Twenty subjects each solved four genetic problems which involved either autosomal or X-linked inheritance, and working forwards or working backwards.

After the subjects had solved the problems, they were questioned about their understanding of the relationships between meiosis, gametes and fertilisation, and the steps taken to solve the problems. Students were further questioned as to their understanding of X-linked inheritance and the way in which alleles for X-linked genetic traits are carried on the sex chromosomes.

Data are presented for overall success on the problems, and for each of the eight problem solving steps for the genetics problems.

Success in Solving Genetic Problems

The problems involved either autosomal or X-linked modes of inheritance and working forwards or working backwards. The overall success of students in solving these problems and obtaining answers is presented in Table 1.

Table 1. Percentage of students (n=20) who achieved correct answers to genetics problems.

Problem	Type of problem		Percent correct answers
One	Autosomal	Forward	90
Two	X-linkage	Forward	70
Three	X-linkage	Backward	25
Four	Autosomal	Backward	75

The majority of students managed to obtain the correct answers for Problem One, Two and Four. The most difficult of the problems was Problem Three which was an X-linkage problem which required students to work backwards and give two alternative answers. Problem One was the easiest, it involved autosomal inheritance, and required students to work forwards. Students were more successful on the autosomal problems than the X-linkage problems. Students were more successful on the forwards working problems than the backwards working problems.

Sample Problem Solutions

Problems One and Two required the students to work forward from information given about parents to determine the phenotypes and genotypes of the offspring. Problems Three and Four required students to work backwards from information given about the offspring to determine the genotype and phenotypes of the parents.

Sample solutions to Problems One (forward working) and Problem Three (backward working) are presented below to illustrate the solution process used for these two types of problems.

Problem One (Autosomal forward)

Blue eye colour is determined by a recessive gene. Brown eye colour is dominant. If Mr Jones is a brown eyed heterozygous male, and Mrs Jones is a brown eyed heterozygous female, what are the possible genotypes and phenotypes of the offspring?

What is the probability that these two parents can produce a blue eyed child?

Mrs Jones is pregnant. What could be the colour of the baby's eyes?

Transcript of S16's Solution of Problem One.

"Blue eyes equals little b and brown eyes equals big B. The male is heterozygous brown, and Mrs Jones is brown eyed heterozygous, so they both will be Bb. (Constructs a Punnett square, see Figure 2). The offspring will be BB, Bb, Bb and bb. Now...if I am to answer the question of what is the chance that these parents will produce a blue eyed child. Well...genotypes will equal one quarter BB, one half Bb and one quarter bb. The phenotypes equal that three quarters will have brown eyes, and one quarter will have blue eyes. So...there will be a 25% chance of a parent producing a blue eyed baby."

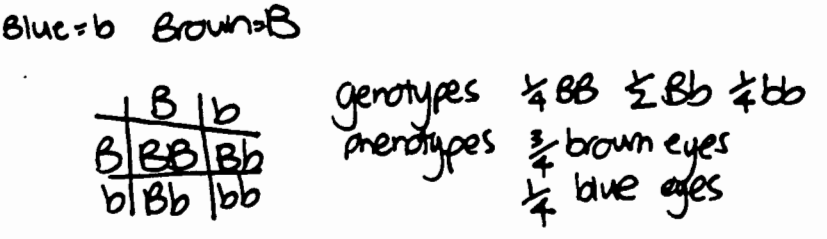

Figure 2. S16's solution of Problem One.

S16 clearly showed an understanding of the terms heterozygous as she identified the parental genotypes correctly. She also correctly identified the genotypes and phenotypes of the offspring.

Problem Three (X-linked backward)

Haemophilia is a recessive trait which is carried on the X chromosome.

A man has been diagnosed as having the disorder of haemophilia. If this man's father is a non-haemophiliac, what is the genotype of the mother?

Transcript of S10's Solution of Problem Three

"I'll just start by doing one of those pedigree things. Now the man has it, and it is a recessive gene. I'll just put little b. The father has the big B gene as he is normal. Now, if I do the Punnett square, the mother can be X^BX^b as this can produce a child with a genotype of X^bY. (Constructs Punnett square, see Figure 3). The mother could also be a haemophiliac X^bX^b as this could also produce a child of X^bY. (Constructs second Punnett square). So...the mother can be X^BX^b or X^bX^b, so there can be two possible genotypes.

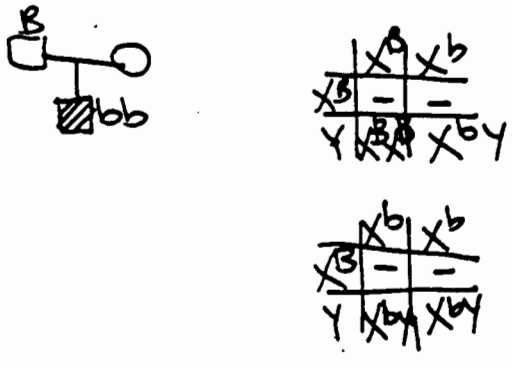

Figure 3. S10's solution of Problem Three.

S10 was one of the 25% of students that correctly solved Problem Three as this student supplied both of the alternative genotypes that the mother could be to produce a son who had the condition of haemophilia.

The students' procedural and conceptual knowledge, as revealed in their solution processes and the debriefing was analysed. The findings are presented in the following sections.

Construction of a symbolic key for the alleles

In genetic problem solving, students are expected to provide a symbolic key to show which letters represent certain alleles. When solving the problems, 30% of students provided a symbolic key and the remaining 70% progressed immediately into solving the problem without formulating any form of symbolic key. Figure 4 shows sample solutions to Problem One that include a symbolic key.

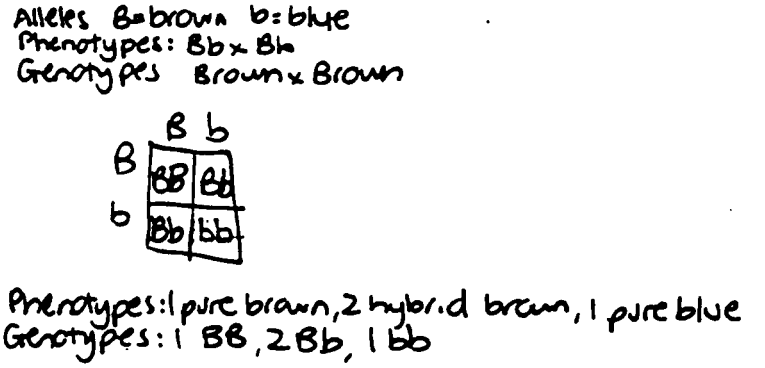

Figure 4. Sample solutions to Problem One illustrating the use of a symbolic key.

Most students who did not provide a symbolic key for the first problem tended not to provide a key when solving Problems Two, Three and Four.

Identification of parent genotypes and gamete types for forwards working problems

Problems One and Two needed students to formally identify parental genotypes to show full understanding of the problem. Many students omitted this stage, and

immediately progressed into solving the genetic problems using the Punnett square algorithm. Table 2 shows the percentage of students who completed this step before constructing the Punnett square.

Table 2. Percentage of students (n=20) who identified parental genotypes before constructing a Punnett square.

	Identified parental genotypes
Problem One	45
Problem Two	35

Students tended to approach Problem One and Problem Two in a similar fashion, thus if parental genotypes and gametes were omitted in Problem One, the same tended to happen in Problem Two.

Identification of offspring genotypes and phenotypes on forwards working problems

Problems One and Two required students to determine genotypes and phenotypes of the offspring produced in the cross. The success of students in doing this is presented in Table 3.

Table 3. Percentage of students (n=20) that correctly determined offspring genotypes and phenotypes

	Offspring genotypes	Offspring phenotypes
Problem One	90	90
Problem Two	70	70

Students were also questioned about their understanding of the terms genotype and phenotype in the debriefing that was conducted after the problem had been solved. Student responses were coded as no understanding, misconceptions or full understanding. Table 4 shows the percentage of responses in each of these categories.

Table 4. Percentage of students (n=20) with no understanding, misconceptions or full understanding of the terms genotype and phenotype.

	No understanding	Misconceptions	Full understanding
Genotype	15	50	35
Phenotype	10	55	35

Responses from S12, S15 and S20 are presented below to illustrate the difficulties that students experienced in explaining these concepts.

"Genotypes is the letters they get from the parents, and phenotypes is percentage...the chance of them getting the condition." (S12)

"Genotype is the different types of things, and phenotype is the possibility of what happens." (S15)

"I think...phenotype is like the percentage and brown and blue, and genotype is the type of technical term or alleles you give to it." (S20)

Identification of parental genotypes on backwards working problems

Problem Three and Four required students to work backwards and determine the parental genotypes given the genotypes and phenotypes of the offspring.

When solving Problem Three, students were required to state that the mother could be either a carrier of haemopilia (X^BX^b) or affected with the disease (X^bX^b). Twenty-five percent of students recognised that the mother could be either of these two possible genotypes. Fifty percent of students recognised one of the two possibilities, but did not attempt to investigate the other.

Seventy-five percent of students were successful in solving Problem Four in which each parent would have had to be heterozygous for big ears (Bb).

Problems Three and Four did not require students to complete a Punnett square to solve the problem, but 45% of students approached it in this manner. Figure 5 demonstrates how S6 used a Punnett square to find out the possible parental genotypes.

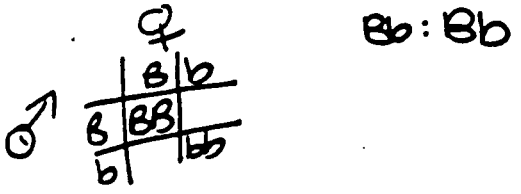

Figure 5 A sample of S6's solution for Problem Three using a Punnett square.

Transcript of S6's solution

"Well...if I have this Punnett square and have two parents, Bb and Bb. It then shows that if I do the solution, you can have a possible offspring who is Bb and also one who is bb. The bb offspring will have small ears and the Bb offspring will have big ears. The parents must be Bb for this to be able to happen."

Forty-eight percent of students approached these problems by working backwards from the offspring genotypes, identifying the alleles that must have come from the mother and the father. Figure 6 shows how S16 used a pedigree type solution process to identify the parental genotypes.

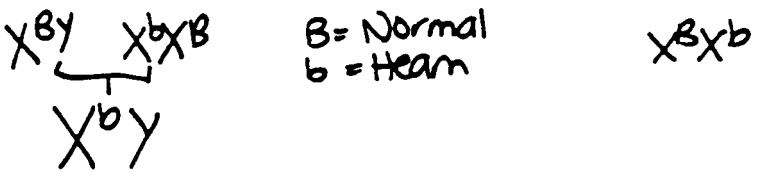

Figure 6. A sample of S16's solution to Problem Three illustrating a 'pedigree' method.

Transcript of S16's solution

"To have a man who is haemophiliac means he must be $X^{b}Y$. Ummm...this means the Y must have come from the dad who is normal $X^{B}Y$ and the X^{b} must have come from the mum. The mother must therefore be a carrier."

Degree of Meaningful Problem Solving

Problem solutions could be separated into three different categories. Problem solutions were either correct and solved in a meaningful fashion, this being with a sound knowledge of meiosis, gametes and fertilisation and how these concepts are related to the inheritance problem. Alternatively, solutions were correct but solved in a non-meaningful fashion, or there was a failure to obtain the correct answer. Table 5 presents these data for the four problems solved.

Table 5. Percentage of students (n=20) who produced correct and meaningful solutions,correct but non-meaningful solutions or failed to solve the problem.

	Correct and meaningful solution	Correct but non-meaningful solution	Failed to solve the problem
Problem One	5	80	10
Problem Two	5	30	30
Problem Three	5	20	75
Problem Four	5	35	25

Certain conceptual and procedural barriers could be identified that contributed to students failing to solve the problems. The common types of errors are presented in the following two tables.

Table 6. Percentage of students (n=20) who made particular types of errors when attempting to solve the autosomal problems.

	Incorrect parental genotypes	Identified as a X-linkedproblem
Problem One	10	0
Problem Two	15	10

Several students incorrectly formulated the parental genotypes and consequently were unable to correctly identify offspring genotypes. Ten percent of students represented autosomal Problem Four as being X-linked.

Table 7. Percentage of students (n=20) who made particular types of errors when solving X-linkage problems.

	Incorrect parent genotypes	Incorrect use of X-linkage	Not identified X-linked	Did not show alternate genotypes
Problem Two	10	10	10	0
Problem Three	0	0	25	50

Students only formulated incorrect parental genotypes with Problem Two where the mother was a carrier (X^BX^b) and the father was normal (X^BY). Fifteen percent of students allocated alleles in a incorrect manner, often attaching them to the Y chromosome, and thus had trouble solving these problems. Several students also had difficulty recognising X-linked problems and solved them as autosomal problems.

Fifty percent of students did not show the alternate genotypes of the mother in Problem Three as being either X^BX^b or X^bX^b which indicates that a large majority of students accepted the first possibility as being the only genotype that the mother could have been.

Students understanding of meiosis, gametes and fertilisation and their relationship to the solution process

After the student had finished working on the problems, they were questioned about their understanding of meiosis, gametes and fertilisation. Table 8 presents the range of understandings of the concepts displayed by the students.

Table 8. Percentage of students (n=20) with no understanding, misconceptions, partial understanding or full understanding of the concepts meiosis, gametes and fertilisation.

Concept	No understanding	Misconception	Partial understanding	Full understanding
Meiosis	25	30	40	5
Gametes	5	15	50	30
Fertilisation	0	20	35	45

A large number of students displayed a partial understanding of the concepts or had misconceptions. S7 and S11 displayed misconceptions of the concepts meiosis and gametes.

"Meiosis is the normal cells, and mitosis is the sex cells." (S7)

"Gametes are the genes in the sex cells." (S11)

S6 and S12 displayed partial understanding of the concepts of meiosis and gametes.

"Meiosis occurs in...umm...it has something to do with the sex organs or sex cells." (S6)

"Gametes are the male and female sex cells which are the ovary and the testis." (S12)

When students were asked if they could place the concepts in the context of the processes they used to solve the problem, none of the students could determine where

the process of meiosis took place, 40% could identify the place of gametes, and 50% could determine where the process of fertilisation occurred.

S14 illustrates the difficulty experienced by students in explaining why they separated the alleles in the Punnett square algorithm.

"I separate the genes in the Punnett square because it tells us...like...the different characteristics and which one is recessive and which one is dominant." (S14)

Understanding of X-linked recessive inheritance and how genes are carried on the X chromosome and not on the Y chromosome

Problems Two and Three dealt with the X-linked recessive traits of haemophilia and colourblindness. Students approached the X-linkage problems in two different ways.

Sixty percent of students attempted to solve the X-linkage problems by correctly attaching the recessive or dominant allele to the X chromosomes, and did not allocate an allele to the Y chromosome. Figure 7 shows the work of a student who attempted to solve the problem in this manner.

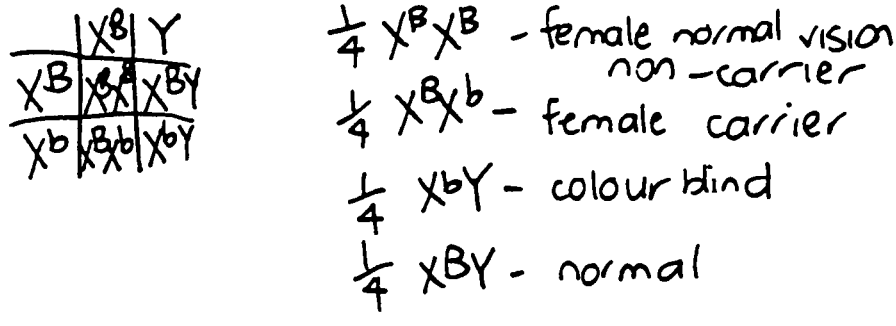

Figure 7. A sample of S10's solution to Problem Two illustrating the allocation of dominant and recessive alleles to the X chromosome.

Twenty percent of students attempted to solve the X-linkage problems by only writing the recessive allele on the X chromosome. If the dominant allele was present on

the X chromosome, it was not shown. Figure 8 demonstrates how students solved X-linkage problems using this method.

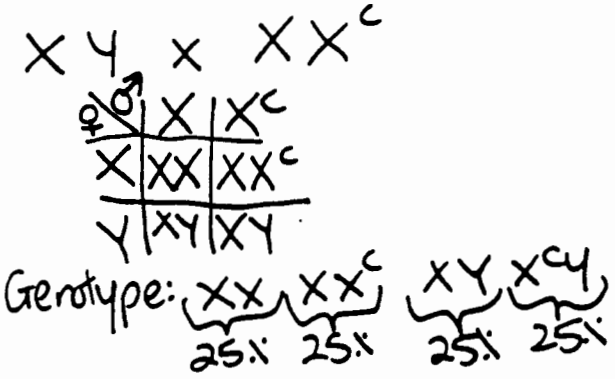

Figure 8. A sample of S12's solution of Problem Two in which only the recessive allele was written on the X chromosome.

Twenty percent of students either attached alleles to both the X and the Y chromosome, or treated the whole of Problem Two as an autosomal problem, displaying an inadequate understanding of X-linked inheritance. Figure 9 shows the work of S1 who did not identify the parents' genotype correctly, and thus showed an inadequate understanding of X-linked inheritance.

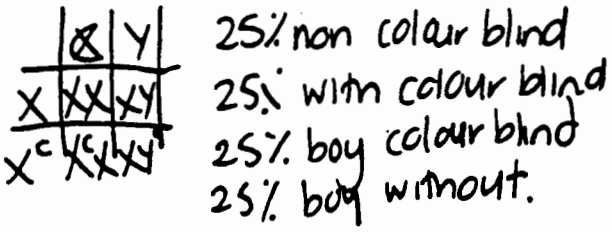

Figure 9. A sample of S1's work showing the incorrect representation of X-linkage.

When questioned as to how the alleles are carried on the X chromosome and not on the Y chromosome, none of the students were able to tell of any significant difference between the X and Y chromosome which could make this possible, and therefore the students were unable to explain why X-linked recessive conditions are more prevalent in males than in females. Following are some student comments that demonstrate the difficulty that students have in explaining the mechanisms of X-linked inheritance.

"Males show the trait because the female is dominant. She carries it, he gets it." (S4)
"The characteristic for the condition only goes to the X and not to the Y." (S13)

"Only the X chromosome can carry it...I don't know if there is any difference between the X and the Y chromosome. Aren't they just the same?" (S14)

Many students were able to solve the X-linkage problems, but the majority of students had no understanding of how the alleles are carried on the X chromosome, and why females are most likely to be carriers of X-linked recessive conditions, and males are more likely to be affected.

CHAPTER 5 DISCUSSION

Analysis of the data revealed that in most instances, students could complete the necessary procedural steps to solve the problems, but few of them did this in a meaningful fashion. Students do complete the Punnett square algorithms in most instances, quite well, but it is the lack of conceptual knowledge linked to these procedures that is the concern.

Analysis of Procedural and Conceptual Knowledge

To solve even a simple problem, a great deal of knowledge is required. What is necessary for a meaningful solution to any problem is the procedural knowledge of how to execute a problem solution, and the conceptual knowledge which provides meaning or context to the procedures (Stewart, 1982). What becomes very noticeable from these studies is the lack of conceptual knowledge of the students. Students tend to solve their Punnett square algorithms in a rote fashion, not demonstrating meaningful understanding.

Formulation of a symbolic key to alleles

Seventy percent of students did not construct a symbolic key to the genetic problems they solved. Although a large percentage of these students still solved the genetic problems successfully, they did not formally identify the letters which represent each allele for the genotypes of the individuals. Although this does not normally cause a student to fail to obtain a correct answer, it is a practice which is recommended to help identification of parental genotypes.

It is far easier for students to successfully solve problems, allocating correct genotypes for different individuals if they have a symbolic key to work from. Thomson and Stewart (1985) found that students who did not construct symbolic keys to alleles often displayed a lack of understanding of basic genetic concepts such as dominant, recessive, heterozygous and homozygous.

Identification of parental genotypes

Students found it difficult to explain how the gametes were generated from the parents, and that each gamete could carry one allele for a particular characteristic to the offspring. These findings indicate that students are not linking the algorithm to the process of meiosis which produces the gametes. Longden (1982) stresses:

> Meiosis can be regarded as a major conceptual block for the students which impedes a meaningful understanding of concepts dependent upon it. The distinction between "rote learning" and "meaningful learning" is evident when extracts are studied; the reliance upon a memorised series of stages rather than an appreciation of a process and its function. (p. 138)

With few exceptions, students see the problem solving algorithm as a procedure that could be used to obtain a correct answer, but were unable to relate it to the real worlds of meiosis and fertilisation (Stewart, 1985).

Problems Three and Four required students to identify the parental genotypes from given offspring phenotypes. Twenty-five percent of students were successful in solving Problem Three which involved a X-linked characteristic. This problem proved to be the most difficult as the majority of students only gave one possible genotype for the mother as being X^BX^b or X^bX^b, but did not explore the possibility that the mother could be either of these. Smith (1988) notes:

> Unsuccessful students tend to consider the first option supported to be the acceptable answer while successful students tended to submit the hypothesis to further analysis or to test more hypotheses. (p. 425)

Seventy-five percent of students correctly solved Problem Four which involved autosomal inheritance and working backwards. The success rate of this problem was much higher with the majority of students recognising that each of the parents had to be heterozygous (Bb) for the trait of big ears.

To solve Problems Three and Four required students to work backwards. Students completed these problems in two different ways. Forty-five percent of students used a Punnett square to generate the genotypes of the parents. This was more successful in Problem Four which only had one possible pair of genotypes for the parents. Problem Three had two alternative genotypes for the mother, and a Punnett square did not allow the students to easily see this. Forty-eight percent of students solved the problems using a pedigree type solution process where they worked backwards from the offspring genotypes, determining which alleles came from each parent.

It was noticeablethat many students approached the genetic problems via the Punnett square method before even deciding what sort of genetic problem it was. Stewart (1982) found in his studies that quite often the Punnett square was completed before the justifications for what the students were doing was discussed.

Identification of offspring genotypes and phenotypes

In Problem One and Two, 70% of students were successful in determining genotypes and above 60% were successful in determining phenotypes. When questioned as to their understanding of the terms genotype and phenotype, only 35% of students had

a full understanding and could identify genotype as being a combination of alleles for a characteristic, and phenotype as being the observable traits of the offspring.

About 50% of students displayed misconceptions about these terms, recognising phenotype as being a percentage or possibility and genotypes as just being the letters. This error is largely attributed to the way in which students write down the genotypes and phenotypes from the Punnett square as percentages and ratios (Stewart, 1985).

Degree of Meaningful Understanding

A concern to many educators is the lack of meaningful understanding that students portray when solving Mendelian genetic problems. The application of rote learned algorithms to solve genetics problems eliminates the need for these students to know about the process of meiosis (Hackling, 1982; Stewart, 1982, 1985; Thomson & Stewart, 1982).

On average, only about 30% of students could solve the problems in a meaningful fashion. A larger percentage of students was able to solve the problems, but it was a non-meaningful solution, and when questioned as to their understanding of why they carried out particular procedures, many students could not justify their steps.

Students understanding of meiosis, gametes and fertilisation

Students were questioned after the completion of the genetic problems about their understanding of meiosis, gametes and fertilisation. Twenty-five percent of students had no understanding of meiosis, and only 5% had a sound understanding of the concept. Many of the students did not link the process of meiosis in any way to the Punnett squares they were using to solve the problems. It was found that most students who were able to solve the typical textbook problems did so with very little correct knowledge of meiosis (Slack & Stewart, 1989.).

Similiarly, it was found that the concept of gametes and fertilisation were poorly understood, with many students not able to place these events in the Punnett square. Thirty percent of students fully understood the role of gametes in inheritance, and 45% of students could determine the time of fertilisation when the genes of the two parents united. Based on his research, Hackling(1982) argued that:

> If students are to develop a meaningful understanding of genetic inheritance, the relations between inheritance and sexual reproduction, the role gametes and fertilisation plays in inheritance must be made more explicit and be more firmly established in the cognitive structure. (p. 18)

Understanding of X-linked recessive inheritance and how genes are carried on the X chromosome and not on the Y chromosome

Problem Two dealt with X-linked inheritance where the students had to allocate genes to the mother and the fathers' sex chromosomes. Seventy percent of students succeeded in completing Problem Two. Of these students, 60% solved the X-linkage problems by allocating recessive or dominant alleles to the X chromosome and did not allocate any alleles to the Y chromosome.

By solving the X-linked problems in this way, students can explain with relative ease the parental source of the X and Y chromosome. Alleles that are tied to the X chromosome are easily traced to the mother, in the case of a boy, or to the mother and father (one from each) in the case of a girl (Tolman, 1982).

Twenty percent of students attempted to solve the X-linkage problems by only writing the recessive allele on the X chromosome. These students had no understanding as to how X-linked traits are inherited from the mother and father, as it is very difficult to trace back the alleles.

When asked to explain the difference between the X and Y chromosome, none of the students could describe any difference between them in length or features. This lack of understanding is a concern, as the differences in the length of the X and Y chromosome is one of the basic concepts needed to truly understand X-linked inheritance. If students do not have this knowledge, it is difficult for them to understand why females are more often carriers, and males are affected even if they only carry one X-linked recessive allele.

CHAPTER 6 CONCLUSIONS AND IMPLICATIONS

Limitations of the Study

The findings of this study indicate that there are conceptual and procedural barriers to genetic problem solving by Year 10 students, but due to the small sample space, these findings cannot be generalised to the population of Year 10 students. This study was only implemented at one school with a sample of 20 students selected from four classes taught by four different teachers. The findings are likely to be strongly influenced by the approach to instruction adopted within this science department and these four teachers.

Summary of Findings

Research Question One: What procedural steps are taken by Year 10 students solving monohybrid cross problems involving autosomal and X-linked traits?

When solving forwards working problems involving autosomal and X-linked traits, the majority of students tend to start straight away on the Punnett square without formulating a symbolic key for alleles, and identifying parental genotypes and gametes.

After completing the cross, the students write out the offspring genotypes and phenotypes, often without a meaningful understanding of the terms genotype and phenotype. It appears that students often follow a rote learned algorithm when solving these problems. This is particularly apparent when students solve problems that involve working backwards. So often, it is approached like a forwards working problem using a Punnett square when in fact, a pedigree type solution process is far more appropriate as it enables all possible alternative parental genotypes to be established.

Research Question Two: Can students explain their problem solving steps in terms of genes, chromosomes, meiosis, gametes and fertilisation?

Students have difficulty in explaining their problem solving steps in terms of genes, chromosomes, gametes and fertilisation. When solving the problem, very often no relationship is seen between the separation of the parental genotype in the Punnett square to the segregation of chromosomes and alleles into the gametes by meiosis. Students tend to solve the genetic problems by rote without meaningful understanding.

The terms genotype and phenotype seem to be poorly understood by students and often as they solve the genetic problems, the students will dutifully fill in the genotypes and phenotypes in a rote like manner without fully understanding the terms at all. Phenotype is commonly mistaken for a percentage as they are often expressed as percentages or fractions of the total number of offspring.

Students' performance on X-linkage problems is an area of concern. None of the students involved in the study knew of any difference in appearance of the X and Y chromosome. This also makes it difficult for students to understand how X-linked recessive characteristics predominantly affect males and cause females to be mainly carriers.

Lastly, a significant finding is that most students do not understand meiosis. Meiosis is fundamental to understanding inheritance. The concepts of meiosis, gametes and fertilisation are essential for the understanding of the process of inheritance. Meiosis is a complex and abstract process and is therefore difficult to understand. An additional problem is that meiosis is often taught separately from problem solving and consequently students do not link meiosis with the algorithms used to solve problems in genetics.

Research Question Three: What are the main conceptual and procedural barriers to the production of meaningful solutions to these problems?

The main conceptual barrier to the production of meaningful solutions to genetic problems is the lack of knowledge of meiosis. If the students do not understand meiosis, they don't understand how gametes are produced, and it is then difficult to be sure of when fertilisation takes place. The inability of many students to explain where these processes must take place in genetic algorithms is of great concern, and it really does need to be addressed by science teachers. Instruction, it seems, has been devoted mainly to the algorithm of carrying out the genetic cross using the Punnett square, without devoting enough attention to the processes that underly these genetic crosses. Furthermore, it is noted that many students do not correctly understand the meaning of the terms, genotype and phenotype. As these terms are so commonly used in the solving of genetic problems, it is disturbing that so many students have this lack of understanding.

The majority of students, when interviewed, also seemed to have poor understanding of how the X and Y chromosome are related to X-linked inheritance. If students do not understand the difference in the X and Y chromosome, it is almost impossible for them to have any meaningful understanding of how X-linked traits are inherited.

The procedural barriers which cause students to have difficulty is that many students omit a symbolic key to represent the alleles to the genes in question. They also fail to allocate the genotypes to the parents, but instead work straight into the Punnett square. This has a tendencey to cause students to make silly errors which should be avoided.

Students tended to approach backwards working problems via the Punnett square method, rather than using a pedigree type solution. Although many students do get the

correct answer, if there is alternative genotypes to be noted, a Punnett square makes it easy for them to be overlooked. Using a pedigree method encourages students to note all the alternative parental genotypes.

When solving X-linked problems, students often only allocate the recessive alleles to the X chromosome. This makes it difficult for students to construct a meaningful understanding of X-linked inheritance, as they often do not realise that the dominant allele is also carried by the X chromosome, and the Y chromosome does not carry any allele. In the study, it was also found that none of the students knew of any difference between the X and Y chromosome in appearance, and unless this is understood, it is impossible to have any degree of meaningful understanding of the concepts of X-linked inheritance.

Implications for Teaching

Many of the difficulties students experience in solving genetics problems meaningfully have been attributed to inappropriate sequencing of concepts in genetics textbooks (Cho et al, 1985; Tolman, 1982). As teachers tend to teach from texts, it is no wonder that students end up so frustratingly confused.

Genetics needs to start with broad concepts of inheritance and reproduction which can act as subsumers for concepts of meiosis, followed by sex determination by chromosomes. This should then be followed by X-linked inheritance, as it does teach students to trace the alleles back to the parents. Following this, autosomal inheritance should be introduced, defining terms such as dominant, recessive, homozygous and heterozygous (Tolman, 1982). This order of concepts will enable students to develop a much more meaningful understanding of genetics.

Hackling's (1990) genes-on-chromosome model should be used for instruction for monohybrid autosomal and X-linked problems. This model enables students to

clearly see how each parent contributes one chromosome from each pair with one allele from each pair and also clearly shows when the processes of meiosis and fertilisation take place in the Punnett square algorithm. The model reinforces links between alleles, chromosomes and cells, and shows the different length of the X and Y chromosomes. (See Appendix One)

Concepts such as meiosis need to be made more perceptible to students, and concrete situations can be implemented by teachers to allow students to make a concrete representation of this complex process. This will enable a clearer mental 'picture' of how chromosomes and genes behave during meiosis (Smith, 1990).

Concept maps should also be implemented throughout the topic, focussing on inheritance as a process involving the transmission of genes and chromosomes from parents to offspring through agencies of meiosis, gametes and fertilisation (Hackling, 1982). This will enable the meanings of these terms to be enhanced and allows students to link them to the genetic problems they solve throughout the topic of genetics.

Science teachers also need to ensure they expose students to not only forwards working genetic problems, but also to the backwards working problems. This encourages students to use their conceptual knowledge and exercise their problem solving skills. Teachers should model for students how they solve the problems so that students can employ these procedures that are efficient and meaningful.

When working on genetics problems, group work and talking aloud should be encouraged as students can then have the opportunity to see what procedures others use.

Further Research

A revised genetics curriculum based on an improved instructional sequence, concrete representation of abstract concepts, teacher modelling of problem solving processes, genes-on-chromosomes model, forwards and backwards problems needs to be developed, implemented and evaluated to properly test the recommendation from this and other studies.

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APPENDICES

Appendix One

The genes-on-chromosome model designed by Hackling (1981)