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## Identifying the difficulties experienced by year 10 high school students when attempting to solve genetic pedigree problems

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**IDENTIFYING THE DIFFICULTIES EXPERIENCED BY YEAR 10  
HIGH SCHOOL STUDENTS WHEN ATTEMPTING TO SOLVE  
GENETIC PEDIGREE PROBLEMS**

**By**

**Richard Hamilton-Brown BA Ed (Secondary)**

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

**Date of Submission: 30th March 1995**

## USE OF THESIS

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

## ABSTRACT

Teachers rate genetics as one of the most difficult biology topics for high school students to understand (Finley, Stewart & Yarroch, 1982). It has been observed that some students are able to solve genetics problems using routine algorithmic methods, without understanding the basic underlying concepts (Stewart, 1982). The aim of this study was to identify the procedural errors and conceptual misunderstandings that the students had when attempting to solve genetic pedigree problems.

Genetic pedigrees are diagrammatic representations of the members and ancestral relationships in a family (Cummings, 1991). Two male and two female students were selected from each of four Year 10 classes. They were required to solve three genetic pedigree problems, identifying the most likely mode of inheritance for each problem. The interviews were tape-recorded and transcribed, and these were followed up with a debriefing session in which each subject's knowledge of the conceptual basis of these problems was probed. The results showed that many students made similar errors in procedure and many lacked the conceptual knowledge to produce meaningful solutions.

The most common procedural error was the failure to falsify all the possible hypotheses, which resulted in students failing to provide complete and conclusive solutions. Other procedural errors included the incorrect use of genotypes, the failure to identify and correctly interpret critical patterns, and the misinterpretation of non-critical patterns. The conceptual misunderstandings included the lack of knowledge regarding the meanings of dominant and recessive, and the mechanisms of x-linkage.

The recommendations for improved teaching of this topic focus on making procedural steps more explicit, and making the link between the procedural steps and underlying conceptual knowledge clearer.

## **DECLARATION**

I certify that this thesis does not incorporate, without acknowledgment, any material previously submitted for a degree or diploma in any institute 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.

Richard Hamilton-Brown

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# **CHAPTER 1**

## **Introduction**

### **Problem**

Teachers rate genetics as one of the most difficult biology topics for high school students to understand (Finley, Stewart & Yarroch, 1982). It has been observed that some students are able to solve routine genetics problems without actually understanding the underlying concepts. This results from the routine application of algorithmic methods such as the Punnett square (Stewart, 1982).

Genetic pedigrees are diagrammatic representations of the members and ancestral relationships in a family (Cummings, 1991). Students are required to determine the mode of inheritance of a trait, from a given pedigree. Genetic pedigree problems are difficult to solve even at the tertiary level. Genetics experts solve pedigree problems by testing inheritance hypotheses using genotypes or patterns of inheritance and rigorously falsifying alternative hypotheses. Tertiary students have been shown to make errors in using genotypes and often misinterpret patterns of inheritance. They also fail to falsify all alternative hypotheses (Hackling & Lawrence, 1988). There have been no studies of pedigree problem solving by secondary students reported in the literature.

In this study, the pedigree problem solving behaviours of secondary students will be observed. Analysis of their problem solving is anticipated to reveal both procedural mistakes and conceptual misunderstandings which are barriers to successful problem solving.

## **Rationale and Significance**

Students continue to be criticised for their lack of problem solving ability (Stewart, 1982). A proficiency at problem-solving is an important key competency expected of students (Mayer, 1992). It is anticipated that the introduction of a national curriculum will enable problem solving practices to be implemented across the curriculum (Ramsey, 1991).

An understanding of genetic pedigrees is important to those students who will need genetic counselling. Some citizens may have to ask questions and make informed decisions involving hereditary diseases which affect them or members of their family. Students will be more capable of dealing with issues such as these, if problem solving is made explicit and meaningful at the high school level where genetics is first introduced as a subject. A complete understanding of the basic genetics concepts introduced at this early stage would help students to build on that existing knowledge, to master more difficult topics such as genetic problem solving or ethical issues in genetics (Thomson & Stewart, 1985).

An understanding of the procedural steps used by students to solve pedigree problems, as well as the conceptual knowledge that they use to warrant these procedures can be used as the basis to plan improved instruction in genetics problem solving. It is anticipated that the findings of this research will allow these recommendations to be made.

Pedigree problems provide a suitable context in which to study students' generation and testing of hypotheses (Smith, 1988). This study will therefore also attempt to provide insights into students' hypothesis testing behaviour, which is an important aspect of scientific inquiry.

## **Purpose and Research Questions**

The purpose of this study is to identify the difficulties experienced by Year 10 science students when attempting to solve genetic pedigree problems. More specifically, this study will address the following research questions:

1. What procedural steps are taken by students as they solve genetic pedigree problems ?
2. What understanding do students have of the conceptual basis of genetic pedigree problems ?
3. What conceptual and procedural difficulties prevent students producing meaningful solutions to pedigree problems ?

## **Definition of Terms**

**Procedural difficulties** are those associated with knowing how to use a problem solving strategy.

**Conceptual difficulties** are those associated with knowing why certain strategies are being used.

**Meaningful solutions** are defined as obtained answers to problems using the correct procedural knowledge to solve the problem and the appropriate conceptual knowledge to justify the procedures used.

(Woolfolk, 1990).

## **CHAPTER 2**

### **Literature Review**

#### **Theoretical Framework**

The collection of data on students' information processing is an integral component of research on problem solving. This includes modelling the procedural steps that students take during problem solving, the reasons they are taken and the errors associated with these steps (Larkin & Rainard, 1984). Stewart (1982) reported that students require two types of knowledge to successfully solve problems. The first is procedural knowledge, which is the knowledge of the possible strategies which can be used to solve problems. The second is conceptual knowledge, which is the knowledge of when and why to implement these problem solving strategies. Hence, knowing what strategies to use and why to use them, leads to meaningful problem solving. Students who only possess the appropriate procedural knowledge tend to simply follow routine, algorithmic problem solving methods.

The conceptual knowledge related to the problem, governs what 'problem space' will be constructed by the problem solver (Stewart, 1982). This refers to the encoding of the relevant features of the problem, allowing an internal representation of the problem to be constructed (Best, 1986). External stimuli are received through sensory receptors into the sensory information storage. These are then encoded into short term memory and some of this information is subsequently retained in the long term memory. To retrieve this information from long term memory, it is necessary to have recognisable patterns presented, which are associated with the previously stored information. Executive control processes facilitate the retrieval of information from the long term to short term memory (Atkinson, Atkinson & Hilgard, 1983). Information

selected from the problem statement and long term memory is used to construct the problem representation in short term memory.

Genetic pedigrees are a diagrammatic representation of inheritance patterns within an extended family. Recognisable patterns in the pedigree activate the appropriate schema in long term memory, bringing it into short term memory. The schema comprises all of the conceptual and procedural knowledge associated with a particular mode of inheritance. Once activated, the conceptual knowledge is used to generate a representation of the problem. The representation is used in selecting appropriate procedural routines for solving the problem.

Smith and Good's (1984) model displayed in Figure 1, outlines the sequence of operations necessary for meaningful problem solving.

**1. Problem Analysis:**

Determining what the problem is asking.

**2. Pattern Recognition:**

Identifying clues which will point them in the right direction.

**3. Schema Activation:**

Retrieving all the relevant conceptual knowledge from the long term memory.

**4. Problem Representation:**

Deciding on what procedure to carry out and why.

**5. Selecting and Implementing Solution Processes:**

Using the chosen problem solving technique.

**6. Checking Solution Adequacy:**

Evaluation of the answers.

Figure 1. A model of the problem solving process (Smith & Good, 1984).

## Previous Studies

There has been little research on secondary students' genetic pedigree problem solving. Smith (1988), Smith and Good (1984), Hackling (1984) and Hackling and Lawrence (1988) have studied pedigree problem solving by tertiary students and expert geneticists. It was anticipated that the findings of these studies would provide a background for this study.

There are four common modes of inheritance which form the basis for hypothesis testing of pedigree problems. These are autosomal dominant (AD), autosomal recessive (AR), x-linked dominant (XD) and x-linked recessive (XR). Inheritance hypotheses are generated in response to the recognition of patterns of inheritance within the pedigree. The hypotheses are then tested either by assigning genotypes to all the individuals in the pedigree or by interpreting patterns of inheritance (Stewart, 1982). These provide evidence to either support or falsify hypotheses. Popper (1959) argued that supporting evidence could not conclusively prove a hypothesis, whereas only falsification could be conclusive. Hence, the falsification of alternative hypotheses becomes an important step in the problem solving procedure.

An analysis of hypothesis testing undertaken by students, revealed that subjects tended to test the most plausible hypothesis first (Hackling, 1984). The degree to which students further tested the hypothesis to completion, depended on their ability to identify other patterns in the problem. The same study identified that more efficient problem solvers falsified alternative hypotheses. Falsifying alternative hypotheses was considered important as it provides additional evidence to support the initial hypothesis. Hackling's findings suggested that differences in problem solving ability were focused on both the procedural and conceptual knowledge of the basis of pedigree problems, with regards to the generation and testing of hypotheses.

Hackling (1984) subsequently identified four sequential steps representative of the approach to pedigree problem solving by experts. The steps included the identification of the most likely mode of inheritance, complete testing of this hypothesis and the falsification of all alternative hypotheses. The final step involved the problem solver's ability to suggest the most likely mode of inheritance from the given data, where it was not possible to eliminate all the alternative hypotheses.

Hackling and Lawrence (1988) studied the differences between expert and novice solutions of genetic pedigree problems. The findings identified that whilst experts did not obtain more correct answers than the novices, they did provide more complete and conclusive answers. The completeness of a solution depended on the extent to which alternative hypotheses were falsified. The study also identified that the experts were more successful at using pattern recognition to generate and test hypotheses, and as a result performed more proficiently on the more difficult problems. These findings and those from Hackling's (1994) study, suggested that experts had a more extensive and accurate conceptual knowledge and systematic procedural knowledge than the novices.

Smith (1988) found that unsuccessful problem solvers tended not to exhibit any logical sequence representative of a problem solving agenda. Their problem solving behaviour differed each time a problem was approached. He suggested that the lack of understanding of the conceptual basis of pedigree problems was responsible for such behaviours. Smith also identified a number of behaviours characteristic of unsuccessful problem solving, such as an inability to identify patterns of inheritance and only considering the first obtained solution. He suggested that these behaviours were caused by a lack of procedural and conceptual knowledge, contributing to difficulties which subsequently prevented meaningful solutions being obtained.

Cavallo and Schafer (1994) stated that relevant prior knowledge was the most important criterion for meaningful learning and for this to take place, the concepts presented to the learner must be potentially meaningful and must allow the learner to integrate the new concept with existing conceptual frameworks as the basis of understanding. Hackling and Treagust (1982) reported that there was a low level of understanding of inheritance concepts among Year 10 students, due to the abstract nature of the concepts involved. Without a sound understanding of concepts such as meiosis and x-linkage, it is likely that students will be limited to using routine algorithmic problem solving methods.

Many of the findings from the previous research are helpful in constructing a framework for identifying the difficulties experienced by secondary school students in pedigree problem solving.

Previous research from Smith (1988), Hackling and Lawrence (1988), Stewart (1982), and Smith and Simmons (1992) have suggested a number of recommendations for the improved teaching of genetic problem solving; all have focused on the procedural and conceptual aspects. Smith and Simmons (1992) suggested that the learners' understanding of genetics concepts and problem solving strategies could be promoted through learning opportunities which enable them to interpret, analyse and experience general and specific genetic concepts, and differing genetic models. In these situations, the learners generate their own questions and hypotheses, and present and receive feedback on arguments in support of the steps they propose. As the learner progresses, his/her performance gets closer to that of expert performance and this is modified by self-assessment.

Throughout the process, the role of the teacher is to provide only the level of support necessary at the time, until the learners' performance improves to the stage where the teachers support and input is no longer needed. The intended result is a learner who is both skilled and independent.

Drawing from the suggestions put forward in previous studies, the recommendations made to improve instruction in genetic problem solving should focus on the enactment and integration of skills. The teacher's role is to diagnose students' problem solving procedures and their conceptual knowledge of the basis of pedigree problems.

### **Methodological Issues**

The most important aspects of the methodology, are the instruments and techniques used to gather data. In this case, a concurrent verbal or think-aloud protocol as described by Ericcson and Simon (1984) will be used to gather data. The verbal explanations are provided concurrently with the generation of written answers. The think-aloud protocol, when encouraged with minimal interruption from the researcher is designed to reveal the sequence of information considered by the subject without altering the cognitive processes used in solving the problem (Larkin & Rainard, 1984).

A debriefing session following the think-aloud-protocol will be used to probe the subjects' knowledge of the conceptual basis for solving genetic pedigree problems. It is anticipated that these data will reveal to what extent the solutions generated by the subjects were meaningful or based on non-meaningful algorithmic methods.

A task analysis was used to determine the appropriate knowledge required by the subjects to solve the pedigree problems and to develop the questions to be used in the debriefing.

## CHAPTER 3

### Method

#### Design

The research design involved a number of subjects solving three genetic pedigree problems with concurrent verbalisation, determining the mode of inheritance for each problem. After all three problems had been solved, a debriefing was conducted in which each subject's knowledge of the conceptual basis of these problems was probed.

#### Sample

The sample was selected from a group of Year 10 students which had completed *Unit 6.2: Biological Change* in which students were taught to solve genetic pedigree problems. A sample size of 16 students was considered sufficient to reveal a number of difficulties experienced by the students at this level. The sample size in this instance would not be large enough to allow generalisations of the results (Gay, 1987), however it was the desired size to comply with the constraints of this study. Four students, two boys and two girls, were selected from four science classes. Each class had been taught genetics by a different teacher. The selection of equal numbers of boys and girls was anticipated to eliminate any aspect of gender bias. The selection of students from different classes was to ensure that a variety of problem solving strategies would be analysed, rather than a single strategy taught by a particular teacher.

It was also necessary to select students with relatively average to high abilities, as less capable students might not have been able to make reasonable attempts at answering these difficult questions. It was anticipated that even the more capable students would demonstrate inadequacies in both the procedural and conceptual domains.

## Instruments

The three pedigree problems (Appendix 1) were ones similar to that which would have been part of the assessment for the *Biological Change* unit, where the subjects were required to determine the mode of inheritance of the trait. The answers to Problems A and B were autosomal dominant and autosomal recessive, respectively. There were two possible modes of inheritance for Problem C, either autosomal recessive or x-linked recessive. An important aspect of this study was to identify whether students could identify that there were two possible modes of inheritance for Problem C, and whether they were able to use the evidence presented in the pedigree to suggest which mode of inheritance was most likely.

A short debriefing at the end of the session with each subject, was required to confirm the strategy they used to solve the problems and the justifications for why they opted to use that particular strategy. A number of questions were used to probe the students' understanding of the conceptual basis of solving pedigree problems (Appendix 2). Answers to these were cross referenced with notes taken and the transcripts to triangulate the data, making sure that any conclusions made about particular subjects could be justified (Cohen & Manion, 1980). This was also done to increase the validity of the study, ensuring that it set out to measure the criteria that were intended (Gay, 1987).

## **Data Analysis**

The think aloud protocols were transcribed. A number of characteristic problem solving features were coded out from the protocol. The problem solving steps taken by the subjects were identified directly from the transcripts to model their solution processes, and to identify where the omission of key steps accounted for any incorrect or incomplete answers. Other factors such as the number and types of hypotheses tested, and how each hypothesis was tested, were also coded from the transcripts. The degree to which the subjects provided complete answers could be determined by identifying the number of hypotheses tested, and whether these were correctly supported or falsified.

It was also necessary to determine if subjects interpreted the patterns of inheritance correctly, and if they were able to correctly assign genotypes to the individuals in the pedigree.

The debriefing session, was essential in determining the students' understanding of the conceptual basis of the problems. The students were asked four debriefing questions which were designed to probe their knowledge about genetics concepts. The questions probed their knowledge of the mechanisms for the inheritance of dominant and recessive characteristics as well as autosomal and x-linked traits. The degree of understanding in these areas would determine the degree to which their solutions could be deemed as meaningful.

## CHAPTER 4

### Results

To adequately collect data from the problem solutions and interviews, these had to be transcribed to have a written account of each student's solutions and answers to the debriefing questions. Once this had been achieved, these were used to analyse student performance in terms of the research questions presented in Chapter 1.

This chapter presents the data regarding students' overall success on the problems, the procedural steps taken, their conceptual understanding of the problems, and the procedural and conceptual barriers to meaningful and correct solution of the problems.

#### Overall Success

The students' performance on the three pedigree problems was analysed to obtain a measure of 'overall success'. Two aspects of 'overall success' were identified; the number of correct answers, and the completeness of solutions.

##### *Number of correct answers*

All students made a reasonable attempt to solve each problem and provided an answer. The correct answers, were autosomal dominant (AD) for Problem A, autosomal recessive (AR) for Problem B, and autosomal recessive (AR) or x-linked recessive (XR) for Problem C. The percentages of students who obtained correct answers to the three problems are presented in Table 1.

Table 1. The percentages of students (n=16) who obtained correct answers on Problems A, B and C.

Problem	Correct answer	Percentage
A	AD	100
B	AR	100
C	AR and XR <sup>a</sup>	62.5

Note. <sup>a</sup> For Problem C, students were required to identify that both AR and XR were possible answers.

All of the students were able to produce correct answers for Problems A and B, whilst only 62.5% of the students were able to obtain the correct answer for Problem C. This measure of 'overall success' was not considered the most effective way of representing the students' performance on the pedigree problems, as it was possible to obtain the correct answer based on little evidence or even by guessing. The lower success rate for Problem C, which had two possible answers may indicate that students guessed the most likely mode of inheritance or just accepted the first answer they obtained.

Table 1 reports the percentage of students who obtained the correct answers, irrespective of whether they used evidence to support and falsify inheritance hypotheses. The quality of the solution depends on the extent to which hypotheses have been supported and/or falsified. It was therefore considered necessary to examine the completeness of the students' solutions, analysing the degree to which they supported and falsified hypotheses.

### *Completeness of solutions*

In order to analyse the students' solution in terms of completeness, it was necessary to establish the number of possible inheritance hypotheses that the students could test. In each case, the students were expected to test four hypotheses; these being autosomal dominant, autosomal recessive, x-linked dominant and x-linked recessive. For each problem, the students were expected to cite evidence to support one or more of the hypotheses and subsequently cite evidence to falsify the alternative hypotheses. This procedure would result in the students obtaining a complete solution.

To establish the degree to which the students generated complete solutions, a completeness of solution score was allocated to each student's solution. Since there were four hypotheses to test, the maximum possible score for each problem would be four. The students were allocated one mark for correctly supporting a hypothesis and one mark for correctly falsifying each of the three alternative hypotheses. Figure 2 shows the completeness of solution scores given to S3 for her solutions of Problems A and C.

Problem	Hypothesis	Supported	Falsified	Score
A	AD	/		4
	AR		/	
	XD		/	
	XR		/	
C	AD		/	3
	AR	/		
	XD			
	XR	/		

Figure 2. S3's completeness of solution scores on Problems A and C.

The examples presented in Figure 2 showed that S3 obtained a maximum completeness of solution score of four for Problem A, as she was able to correctly support one hypothesis and falsify the three alternatives. However, she only obtained a score of three for Problem C, as she failed to either support or falsify the x-linked dominant hypothesis. The completeness scores for all subjects are presented in Table 2.

Table 2. Completeness of solution scores for each student's solution of Problems A, B and C.

Student	Problem		
	A	B	C
1	2	2	2
2	1	2	2
3	4	4	3
4	3	4	3
5	3	4	4
6	4	4	3
7	1	2	1
8	4	3	2
9	4	3	2
10	3	3	2
11	3	3	2
12	2	3	2
13	4	4	3
14	3	3	2
15	1	1	1
16	3	3	2
Average	2.7	2.9	2.2

The results shown in Table 2 indicate that a completeness of solution score of four was only obtained on 11 occasions. The most common score was three, which was obtained on 17 occasions and then two, which was obtained on 14 occasions. The average completeness scores show that students were more successful on Problem B, with a score of 2.9. The students were less successful on Problem C, with a score of 2.2.

Since all the students were able to use evidence from the pedigree to support the correct hypotheses on Problems A and B, and most were able to support the correct hypotheses on Problem C, the data indicate that the difficulty resulted from the students' failure to falsify the alternative hypotheses. Table 3 shows the percentage of occasions when students failed to falsify the alternative hypotheses for the three problems.

Table 3. Percentage of occasions on which students failed to falsify alternative inheritance hypotheses.

Hypothesis	Percentage
AD	50
AR	69
XD	52
XR	31

The results from Table 3 show that the most common hypothesis which students failed to falsify most often was the autosomal recessive (AR) hypothesis, which was not falsified 69% of the time. Students only needed to falsify this hypothesis in Problem A, since autosomal recessive (AR) is a correct answer for Problems B and C. The students failed to falsify the x-linked dominant hypothesis (XD) 52% of the time. This hypothesis was not one of the answers to any of the problems and therefore should have been falsified for each solution by each of the students. The autosomal dominant (AD) hypothesis should have been falsified for Problems B and C, but students failed to do so 50% of the time. Finally, students had the most success falsifying the x-linked recessive hypothesis, where they only failed to do so 31% of the time.

## Procedural Steps

By listing the procedural steps taken by students on each problem, it was possible to gain insights into each student's understanding of the problem solving process, the strategies they used, how they supported or falsified hypotheses, their ability to recognise patterns, any misinterpretation of these patterns and the use of genotype nomenclature.

### *Solution processes*

Most of the students used similar strategies for solving the problems, with regards to the ways they started the problem, the patterns they identified and even the procedural errors that were made. Figure 3 shows the solution process used by S3 on Problem A and illustrates a complete and correct solution.

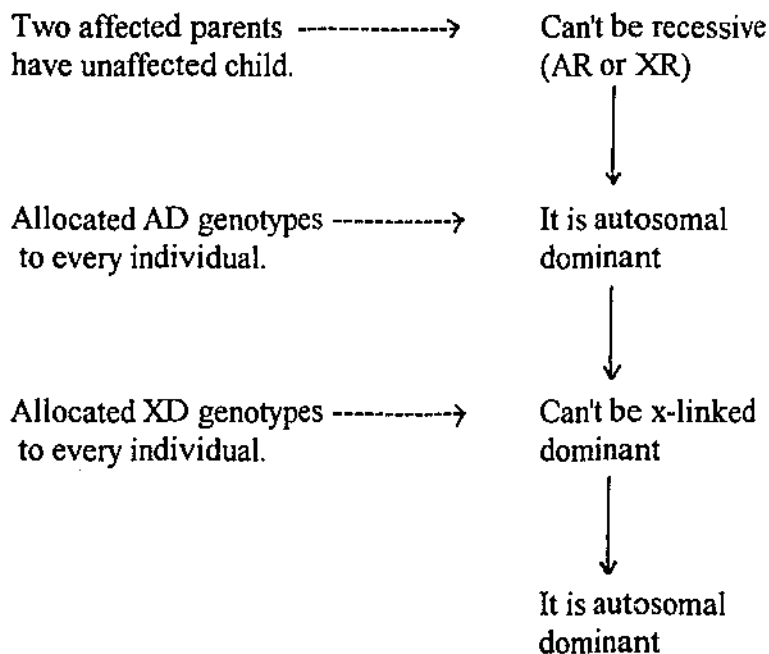


Figure 3. S3's complete and correct solution of Problem A.

An analysis of the solution displayed in Figure 3, shows that S3 started by using pattern recognition to falsify the autosomal recessive and x-linked recessive hypotheses. The student then allocated the appropriate genotypes to all the individuals in the pedigree to support the autosomal dominant hypothesis. The student subsequently used genotypes to falsify the x-linked dominant hypothesis and concluded that the trait was autosomal dominant. Since one hypothesis was correctly supported and the other three correctly falsified, the student obtained a completeness of solution score of four (See Table 2).

In contrast, some of the students utilised different approaches which neglected pattern recognition and the falsification of alternative hypotheses. Figure 4 shows S12's solution of Problem A, which was typical of that used by most students.

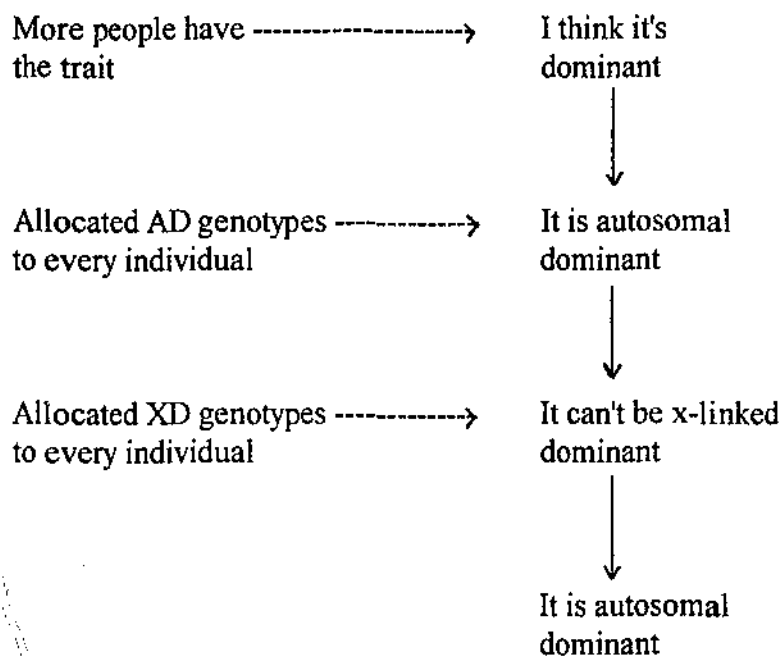


Figure 4. S12's solution of Problem A.

The solution process illustrated in Figure 4, revealed that the student started the problem by identifying a pattern that was not informative and interpreted it inappropriately. The student subsequently supported the autosomal dominant hypothesis, but failed to falsify the autosomal recessive hypothesis. By using genotype allocation again, the student falsified the x-linked dominant hypothesis but failed to falsify the x-linked recessive hypothesis. The student correctly concluded that the trait was autosomal dominant. By correctly supporting one and falsifying another hypothesis, the student obtained a completeness of solution score of two (See Table 2). The inability to falsify alternative hypotheses and use pattern recognition to do so, was typical of those students who did not obtain complete solutions to the problems.

### *Recognition of patterns*

If students were able to identify and correctly interpret the critical patterns in each pedigree, it was expected that they would be able to produce a complete and correct solution to each problem. Critical patterns are ones that either conclusively falsify an inheritance hypothesis or indicate if a particular mode of inheritance is likely. From the analysis of the solution processes used by the students, it was revealed that many students used pattern recognition to start solving the problems. This procedural step was illustrated by the solution process provided by S3 for Problem A (see Figure 3), where she identified that two affected parents had an unaffected child. In this case, this procedural step proved to be most effective, in that the student was able to falsify both the autosomal recessive and x-linked recessive hypotheses in one simple step.

There were two critical patterns in the pedigree of Problem A. The first was that two affected parents had an unaffected child, which indicated that the mode of inheritance of the trait could not be recessive. The second pattern was that an unaffected mother had an affected son, which indicated that the mode of inheritance could not be x-linked dominant. The correct interpretation of both these patterns would reveal to the problem solver that the solution could only be autosomal dominant. Table 4 shows the number of students who were able to identify and correctly interpret these patterns.

Table 4. Number of students (n=16) who identified and correctly interpreted the critical patterns in Problem A.

Pattern	Number of students who identified pattern	Number of students who correctly interpreted pattern
1. Two affected parents had an unaffected child. Can't be recessive (AR or XR). Must be dominant.	7	7
2. Unaffected mother had an affected son. Can't be x-linked dominant.	0	0

The data in Table 4 reveal that only seven students were able to identify the first pattern. All of them were able to deduce that the mode of inheritance was not recessive, hence falsifying the recessive hypotheses. None of the students were able to identify the second pattern to falsify the x-linked dominant hypothesis. Five students did obtain full completeness scores for Problem A (see Table 2). These students falsified the x-linked dominant hypothesis using genotypes.

There were two critical patterns in the pedigree of Problem B. The first was two unaffected parents had an affected child or the trait skipped a generation. This indicated that the mode of inheritance of the trait could not be dominant (AD or XD). The second pattern was that an unaffected father had an affected daughter, which indicated that the mode of inheritance could not be x-linked recessive. Table 5 shows the number of students who were able to identify and correctly interpret these patterns.

Table 5. Number of students (n=16) who identified and correctly interpreted the critical patterns in Problem B.

Pattern	Number of students who identified pattern	Number of students who correctly interpreted pattern
1. Two unaffected parents had an affected child or skipped generation. Can't be dominant (AD or XD). Must be recessive.	12	12
2. Unaffected father had an affected daughter. Can't be x-linked recessive.	0	0

The data in Table 5 reveal that 12 students were able to identify the first pattern and that all of them were also able to correctly interpret it. As a result, they were able to falsify the dominant hypotheses (AD and XD). However, none of the students were able to identify the second pattern to falsify the x-linked recessive hypothesis. Five students obtained full completeness scores for Problem B (see Table 2). These students used genotypes to falsify the x-linked recessive hypothesis.

There were three critical patterns in the pedigree of Problem C. The first was two unaffected parents had an affected child or the trait skipped a generation. This indicated that the mode of inheritance of the trait could not be dominant (AD or XD). The second pattern was that an affected father had an unaffected daughter, which indicated that the mode of inheritance could not be x-linked dominant. The third pattern was that an affected grandfather had an unaffected daughter who in turn had an affected son, which indicated that the mode of inheritance was likely to be x-linked recessive. The correct interpretation of these three patterns would reveal to the problem solver that the mode of inheritance was either autosomal recessive or x-linked recessive, but most likely to be the latter. Table 6 shows the number of students who were able to identify and correctly interpret these patterns.

Table 6. Number of students (n=16) who identified and correctly interpreted the critical patterns in Problem C.

Pattern	Number of students who identified pattern	Number of students who correctly interpreted pattern
1. Two unaffected parents had an affected child or skipped generation. Can't be dominant (AD or XD). Must be recessive.	12	12
2. Affected father had an unaffected daughter. Can't be x-linked dominant.	0	0
3. Affected grandfather had unaffected daughter who had an affected son. Likely to be x-linked recessive.	1	1

The data in Table 6 revealed that 12 students were able to identify and correctly interpret the first pattern. This enabled them to falsify the dominant hypotheses (AD and XD). None of the students were able to identify the second pattern to falsify the x-linked dominant hypothesis. However, this was not considered necessary for those who correctly interpreted the first pattern, which indicated that the trait could not be dominant. One student was able to identify and correctly interpret the third pattern to support the hypothesis that the mode of inheritance was likely to be x-linked recessive. Only one student obtained a full completeness score for Problem C (see Table 2), but other students used genotypes to support the hypothesis that x-linked recessive was the most likely mode of inheritance.

### *Misinterpretation of patterns*

The data in Tables 4, 5 and 6 indicate that all the students who identified the critical patterns in the pedigrees were also able to correctly interpret these patterns. However, there were a number of common misinterpretations made with regards to certain other patterns in the pedigrees. Table 7 shows the percentage of students who identified and misinterpreted non-critical patterns present in the three pedigrees.

Table 7. The percentage of students who identified and misinterpreted non-critical patterns in Problems A, B and C.

Problem	Non-critical pattern	Misinterpretation	Percent
A	More people have the trait.	It is dominant.	43.8
B	Less people have the trait.	It is recessive.	25
C	Less people have the trait.	It is recessive.	25
	The trait is rare.	It is x-linked.	18.8
	Only males have the trait.	It is x-linked recessive.	56.3

The data in Table 7 reveal that several of the common misinterpretations of non-critical patterns relate to the frequency of affected individuals in the pedigree. For Problem A, 43.8 % of the students assumed that since there was a relatively large number of affected individuals in the pedigree, then the mode of inheritance of the trait was dominant. They happened to obtain the correct answer in this particular case, but the frequency of affected individuals in the population is not considered to be reliable means of determining the mode of inheritance. The frequency of affected individuals is determined by the frequency of the allele and not its dominance.

The same type of error was evident on Problems B and C, where 25% of the students in each case, assumed that the mode of inheritance was recessive because there was a low frequency of affected individuals in the pedigree. The following quotation illustrates S9's misinterpretation of this pattern.

It looks to be recessive because less people have the trait and most of the people don't have it. If more people had it, then it would be dominant because it's more common. (S9)

For Problem C, the students were expected to select x-linked recessive as the most likely mode of inheritance. The results showed that 18.8 % of them opted for the x-linked hypothesis based on the trait being rare. The data also revealed that 56.3 % of the students selected x-linked recessive as the most likely mode of inheritance because only the males had the trait. The following quotation illustrates S10's misinterpretation of this information.

I think it's probably x-linked recessive. It says that the trait is rare and only the males get the trait, like colour-blindness. (S10)

### *Use of genotypes*

An analysis of the results presented so far, revealed that some students tested, supported and falsified hypotheses using a method other than pattern recognition. In addition to using patterns for testing hypotheses, students also used genotypes. Inheritance hypotheses were tested by allocating genotypes to all the individuals in the pedigree.

Students could have used genotypes to test hypotheses relating to the four common modes of inheritance. The students were expected to use AA or Aa alleles to represent an autosomal dominant genotype and aa alleles to represent an autosomal recessive genotype. To represent an x-linked dominant genotype, students were expected to use the  $X^AX^A$  or  $X^AX^a$  nomenclature to represent females and the  $X^AY$  nomenclature to represent males. To represent an x-linked recessive genotype, students were expected to use the  $X^aX^a$  nomenclature to represent females and the  $X^aY$  nomenclature to represent males. Correct use of genotypes required the correct combination of upper and lower case letters to represent the alleles, but any letter of the alphabet could have been used. Table 8 shows the number of students who correctly used the respective genotype nomenclatures.

Table 8. Number of students (n=16) who correctly used the four categories of genotype nomenclatures.

Genotypes	Number of students who used genotypes	Number of students who used genotypes correctly
AD	16	16
AR	16	16
XD	11	9
XR	11	9

The data in Table 8 show that all of the students used both the autosomal dominant and autosomal recessive genotypes correctly. This meant that they were able to allocate the AA, Aa and aa alleles to the appropriate individuals in the pedigrees. However, when it came to the x-linked traits, only 11 students attempted to use these genotypes and nine of them used it correctly. The other five students did not attempt to test the x-linked hypotheses at all. The two students who used the genotypes incorrectly, allocated alleles to the Y chromosomes of males (ie.  $X^AY^A$ ,  $X^AY^a$  or  $X^aY^a$ ).

## Conceptual Understanding

After students completed their problem solving, a debriefing was conducted to probe students' understanding of the conceptual basis of pedigree problems. The students were asked four questions during the debriefing session, to reveal their understanding about what procedural strategy was required to solve the problem, the differences between dominant and recessive, the mechanisms of x-linked inheritance and the mechanisms of inheriting dominant and recessive alleles.

### *Question 1. How did you decide what to test first ?*

There were only three types of responses to this question. Fifty-six percent of the students indicated that they used pattern recognition as the starting point to solve the problems and subsequently supported and falsified appropriate hypotheses. Twenty-five percent of the students indicated that they used genotype allocation and subsequently tested the hypotheses. The remaining 19% indicated that they analysed the pedigree to identify the number of affected individuals. From there, they suggested that they were able to determine whether the modes of inheritance were dominant or recessive and subsequently tested these hypotheses.

The students who generally relied on the frequency of affected individuals in the pedigrees to solve the problem, failed to understand that the frequency of affected individuals in the pedigree is not a reliable indicator of the mode of inheritance of a trait.

*Question 2. What is meant by dominant and recessive ?*

For this question, the students were required to explain their understanding of the terms dominant and recessive. Brown (1989) defined the term dominant as an allele which is expressed phenotypically in all heterozygotes and recessive as an allele which is only expressed phenotypically in homozygotes, but not expressed in heterozygotes. The students were expected to appropriately describe the occurrences of these alleles in both homozygotes and heterozygotes for their answers to be deemed correct.

Sixty-nine percent of the students were able to demonstrate a satisfactory understanding of the terms dominant and recessive, and all the students were able to describe that dominant genotypes were represented by the alleles AA and Aa, while recessive genes were represented by the alleles aa. Thirty-one percent of the students had misconceptions of dominance. Typical misconceptions are illustrated in the following quotes, which relate to the frequency of alleles in the population.

The gene which shows up the most in the population is dominant. More people will be dominant and if the trait is dominant, more people will have it. (S15)

Recessive genes are not as common as dominant genes, so recessive genes have less chance of occurring in the population. When the trait is rare, it is most likely to be recessive. (S5)

*Question 3. What is meant by x-linked ?*

Cummings (1991) described x-linked genes as those which were only present on the X chromosome and not the Y chromosome. The X chromosome is longer than the Y chromosome, and has a portion which is non-homologous to the Y chromosome. Genes which are carried on this non-homologous portion of the X chromosome do not occur on the Y chromosome, and the mode of inheritance of these genes is described as x-linked. Hence, in males, all x-linked characteristics are inherited from the mother only, since a Y chromosome must be inherited from the father. Since the male only has one copy of the X-linked allele, recessive alleles cannot be masked and X-linked recessive phenotypes occur more often in males than in females.

The students were expected to correctly explain three aspects of x-linkage: x-linked genes are carried on the X chromosomes only; the Y chromosome is shorter than the X chromosome, so it has a missing homologous portion; and, all x-linked characteristics in males are inherited from their mothers. Sixty-nine percent of the students mentioned that the trait was only carried on the X chromosome, 31% of the students mentioned that the Y chromosome was shorter than the X chromosome, and 38% of the students mentioned that all x-linked characteristics in males are inherited from the mother. These results indicate that many students did not have a complete understanding of the x-linkage concept.

According to the answers provided by the students, there appeared to be no specific misconceptions about x-linkage. The results indicated that some of the students understood the concept and the rest did not.

*Question 4. Explain how you worked out the genotypes in the last generation of offspring?*

The students were expected to describe the mechanism of inheritance of dominant and recessive alleles, in terms of the transmission of alleles from parents to offspring. All of the students provided satisfactory answers to this question. A typical response is illustrated by the following quote:

If both parents are Aa (heterozygous), they can give their children the A or a genes. If the child is dominant, then one parent must give it the A gene. The other parent can give the A or a gene, so the child can become AA or Aa. (S12)

### **Procedural and Conceptual Barriers to Meaningful Solutions**

This section is essentially a summary of the previously reported results which outlines the procedural and conceptual barriers responsible for preventing students from producing meaningful problem solutions. The main procedural difficulty experienced by the students was the inability to provide complete solutions to the problems by supporting one or more hypotheses and falsifying all alternative hypotheses. Students made errors in testing inheritance hypotheses which resulted from: students' inability to recognise critical patterns in the pedigrees and misinterpretation of non-critical patterns in the pedigree.

Other procedural errors that the students made resulted from the confusion they had between frequency and dominance of alleles. This resulted in the students making inaccurate conclusions about inheritance patterns because of their misconception. There were also a number of errors made with allocating incorrect genotypes to test x-linkage hypotheses. Since students revealed no misconceptions about x-linkage, it can only be assumed that they had no knowledge of how to assign genotypes in these situations.

Finally, the debriefing questions and the think-aloud protocols revealed that students lacked complete knowledge about the concept of x-linkage, which possibly accounted for the high incidences of error when it came to testing x-linkage hypotheses using genotypes and patterns of inheritance.

## **CHAPTER 5**

### **Discussion**

Analysis of the obtained data, revealed a number of procedural and conceptual errors which prevented students from providing meaningful solutions to the pedigree problems. Although a large percentage of the students obtained correct solutions to the problems, many of these were considered to be incomplete solutions. Similarly, many students were unable to provide the correct justifications for the problem solving strategies they used. Every student identified the correct answer to Problems A and B, while 62.3% obtained the correct answer to Problem C. The correct solutions to the problems could be obtained using little evidence or simply by guessing, and hence did not provide any insight into the problem solving strategies used or conceptual understandings held by the students.

#### **Procedural knowledge**

Analysis of Tables 2 and 3 revealed that although the students were able to obtain the correct solutions to the problems, the solutions were deemed to be incomplete. Where students were expected to obtain completeness of solution scores of four, for each problem, they obtained average scores of 2.7, 2.9 and 2.2 on Problems A, B and C respectively. This indicated that the students were failing to falsify all of the alternative hypotheses. The data in Table 3 show the frequency with which the various hypotheses were not falsified, indicating that most students did not falsify the autosomal recessive (AR) hypothesis. This result does not necessarily indicate that students were unable to or had difficulty falsifying the AR hypothesis, it is more likely that the students simply omitted this step during their procedure.

Autosomal recessive (AR) appears as possible answers for Problems B and C, but not for Problem A. The results show that 69% of students failed to falsify the AR hypothesis, and this occurred solely for Problem A. It was identified that most students started Problem A by testing the autosomal dominant hypothesis first. When this was supported, they moved on to test the x-linkage hypotheses, and in particular, the XD hypothesis. The step that most students omitted was the testing of the AR hypothesis.

Hackling and Lawrence (1988) reported similar findings where novice problem solvers on many occasions, failed to falsify alternative hypotheses. Expert problem solvers generally falsified more alternative hypotheses, leading to more complete and conclusive solutions. As was the case for Problem A, it was observed that 69% of the pupils sought evidence to confirm the initial hypothesis and failed to consider all of the alternatives.

These findings are consistent with the findings of Mynatt, Doherty and Tweney (1977) who reported that there was a strong tendency for novice problem solvers to seek confirmatory rather than disconfirmatory evidence, and that this bias was a characteristic of human reasoning. They further suggested that the subjects found it difficult to arrive at the correct hypothesis because their initial hypothesis was either totally incorrect or misleading, and the alternative hypotheses were not considered. They further stated that the effects of confirmation bias may not be so disadvantageous if the initial hypothesis was at least partially correct.

Popper (1962) has argued that supporting evidence does not conclusively prove or verify a hypothesis; only falsification of hypotheses can be conclusive. The results showed that systematic errors were made through the solution processes as students failed to realise the relevance of falsifying alternative hypotheses, which would in turn provide additional support to their answers. Consequently, their answers were considered incomplete and inconclusive.

Analysis of the solution processes used by the students was designed to identify the errors made as students carried out their problem solving strategy. Most of the students used either of two strategies to test inheritance hypotheses; pattern recognition or genotype allocation. Of the students who identified the critical patterns in the pedigrees, all were able to correctly interpret these to support and/or falsify the appropriate hypotheses. It was also observed that none of the students who used pattern recognition were able to identify the critical patterns which directly supported or falsified the x-linkage hypotheses. These data shown in Tables 4, 5 and 6, reveal that although the students were able to identify and correctly interpret some of the critical patterns, they had difficulty identifying the critical patterns regarding x-linkage.

Students were also observed to identify and misinterpret non-critical patterns in the pedigrees. Table 7 showed that many students based their problem solving strategies around the frequency with which individuals were affected with the trait in the pedigree. The common misinterpretations made by students were that the trait was dominant if more people in the pedigree were affected and recessive if less people in the pedigree were affected. In these cases, students were obviously unaware that the frequency of affected individuals is determined by the frequency of the allele and not its dominance or recessiveness. The other misinterpretation that some students made, was that the trait in Problem C was most likely to be x-linked because the problem statement indicated that the trait was rare, and only males were affected in the pedigree. Again, these inferences were not scientifically correct and the students used little or no other evidence to further support this claim.

The misinterpretation of these non-critical patterns is therefore based on misconceptions about the incidences of traits in the population.

In addition to using pattern recognition, students also tested inheritance hypotheses by allocating genotypes to each individual in the pedigrees. This procedural strategy appeared to be the most successful until students were required to allocate genotypes, when testing x-linkage hypotheses. The results in Table 8 showed that two of the students used the incorrect genotype nomenclature, preventing them from adequately testing the x-linkage hypotheses for each pedigree.

Hackling (1994) identified similar cases among tertiary subjects and suggested that the knowledge of the locus of genes on X and Y chromosomes was the most essential component of understanding the concept of x-linkage. Students who incorrectly allocated genotypes to the Y chromosome, lacked the understanding of the basic concept of x-linked inheritance and limited themselves in their ability to appropriately test x-linkage hypotheses.

The procedural steps used by the students were generally appropriate. The main feature highlighted by analysing these steps are that the students faced the most difficulty when attempting to falsify alternative hypotheses and testing x-linkage hypotheses.

### **Conceptual understanding**

The four questions posed to the students during the debriefing session, revealed the degree to which the students understood the concepts required to meaningfully solve pedigree problems. When asked how they decided what to test first, most students revealed that they used recognition of patterns or genotype allocation. Unfortunately, 19% of students identified and misinterpreted non-critical patterns as a starting point to their problem solving strategy.

When asked to explain the meanings of the terms dominant and recessive, the students were expected to demonstrate their knowledge of the terms with regards to how the alleles were expressed phenotypically in each individual in the pedigrees. Even though all the students could explain that the dominant genes were represented by the alleles AA and Aa, while recessive genes were represented as aa, 31% of the students were unable to provide satisfactory explanations to the meanings of the terms. Furthermore, these students failed to understand that the frequency of affected individuals in the pedigree is not a reliable indicator of a trait's dominance or recessiveness.

When asked to explain the meaning of x-linkage, the answers provided by the students revealed that many of them did not have a complete understanding of the x-linkage concept. The results indicated that most of the students were unaware that the Y chromosome was shorter than the X chromosome, and as a result could not carry x-linked alleles. Also, the results indicated that some students were unaware that all x-linked characteristics in males are inherited from the mother, since a Y chromosome was inherited from the father. The consequent lack of understanding of the x-linkage concepts may be related to the poor performances on the x-linkage aspects of the three pedigree problems.

All of the students were able to satisfactorily explain how different alleles were autosomally inherited from the parents to the offspring. This indicated that they had an understanding about the mode of transmission of alleles from one generation to the next, and how genetic traits could be inherited in this manner.

An interesting feature of the results was the contrast in the degree of student understanding with regards to Questions 2 and 4. Although all the students were able to explain the mode of transmission of alleles from one generation to the next (Question 4), 31% were unable to explain the terms; dominant and recessive (Question 2). Hackling and Treagust (1982) suggested that students were able to describe how phenotypic features were inherited as they could easily relate this concept to the inheritance of features within their own families. They further suggested that the students had more success in relating concepts to concrete experiences, and these concepts became more frequently understood rather than concepts such as dominant and recessive characteristics which are more abstract ideas. Dominant and recessive characteristics can only be explained in terms of DNA codes and protein syntheses. These explanations are not included in the Year 10 curriculum.

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 (Stewart, 1982). Since the procedural errors and conceptual misunderstandings have been identified, it is possible to establish how these factors combine to prevent meaningful problem solving. The next chapter identifies these barriers to meaningful problem solving and suggests recommendations for teaching and implications for further research.

## **CHAPTER 6**

### **Conclusion**

The study has identified the procedural steps undertaken by the students during problem solving, students' understanding of the conceptual basis of pedigree problems, and the procedural and conceptual barriers which prevented meaningful problem solving. This chapter is dedicated to identifying the limitations of the study, summarising the findings with regards to the research questions and identifying the implications for teaching and further research.

#### **Limitations of the study**

Some interesting features of problem solving were revealed as a result of this study, but there were a number of limitations which have to be taken into account. Firstly, it would not be appropriate to generalise the findings to the entire Year 10 population, as the subject sample size used was too small. Secondly, since the subjects were selected from only four different classes in one school, it is likely that the problem solving approach used by the students was strongly influenced by the instructional approach used in that school. Finally, lower ability students were not included in this study, as the problems were initially considered too difficult. The results obtained may have been considerably different if the lower ability students were included in the sample as other procedural errors and conceptual misunderstandings may have been revealed.

## Summary of the findings

The results indicated that most of the students either omitted or incorrectly utilised critical steps during their problem solving procedure. The omission of these steps accounted for the incorrect and incomplete solutions to the problems. Among the procedural errors were the failure to falsify all the alternative hypotheses resulting in incomplete solutions, the failure to recognise critical patterns, misinterpretation of non-critical patterns and the incorrect use of genotype nomenclature.

The results showed that many students lacked an understanding of the conceptual basis of pedigree problems. Answers to the debriefing questions revealed that some students did not understand how to start solving pedigree problems and could not justify the procedures which they used. Also, many students could not explain the meanings of the terms dominant and recessive, although they could describe the appropriate genotypic nomenclatures. All of the students could however explain the transmission of inherited alleles from parents to offspring. Finally, many students were unable to explain the meaning and significance of x-linkage.

The failure to correctly use the appropriate procedural steps and the lack of conceptual understanding of the basis of pedigree problems, were the difficulties students experienced which prevented them from producing meaningful solutions to the problems. This was displayed when the lack of knowledge regarding dominant and recessive traits resulted in students misinterpreting non-critical patterns, and also when the lack of knowledge of x-linkage mechanisms resulted in the failure to identify and interpret critical patterns and the incorrect use of genotypes.

## **Implications for teaching**

As a result of this research, some recommendations for teaching have been proposed, based on the procedural errors and conceptual misunderstandings identified, which prevent meaningful problem solving. The fundamental assumption underlying these recommendations is that students will be capable of learning this subject more meaningfully if the instructions are explicitly designed to further this goal (Thomson & Stewart, 1985). The recommendations listed below, focus on making problem solving strategies easier for students, and making a clearer link between the procedural steps and the conceptual knowledge underlying pedigree problems.

- 1. Encourage students to use both pattern recognition and genotype allocation as the basis of their problem solving procedure.*

In this study, students made errors when using only one hypothesis testing procedure, without having another approach to check their answers. Students need to first learn the genotype method of testing inheritance hypotheses and then use this knowledge as a foundation for understanding critical patterns that can be used to test hypotheses. Students need to be familiar with the patterns illustrated in Appendix 4 (Hackling, 1988).

*2. Teach students to use a decision tree for solving pedigree problems.*

A decision tree, similar to that proposed by Hackling (1988), would be beneficial in allowing students to use a particular strategy for all problems, irrespective of the problem's degree of difficulty. Hypothesis testing can be made more systematic by using a decision tree to sequentially test alternative inheritance hypotheses. It would also ensure that students understand the necessity for falsification of alternative hypotheses.

*3. Encourage students to list the justifications for each step they use during the procedure.*

Rather than simply following a routine algorithm, students should list each step in the problem solving process with a justification. This would allow teachers to identify the students understanding of the underlying genetic concepts and the procedural steps used to solve the problem. As a result, the knowledge and strategies of students can be diagnosed and remediation applied where necessary.

*4. Confront misconceptions regarding dominance and frequency of phenotypes.*

Students misconceptions that common traits are dominant and rare traits are recessive must be challenged during instruction. This can be achieved if aspects of dominance and recessiveness are taught in terms of characteristics, not genes.

*5. Explain thoroughly the mechanisms and significance of x-linkage.*

Although x-linkage may be a difficult and abstract concept to understand, a general understanding of the basics, such as x-linked traits only being carried by the X chromosome, the Y chromosome being shorter than the X chromosome and males inheriting x-linked traits from the mothers, should be learnt. The use of Hackling's (1990) genes-on-chromosomes model should be used to illustrate the nature of X and Y chromosomes and the locus of x-linked alleles.

**Implications for research**

Due to the limitations of this study, it would be inappropriate to generalise the findings to the entire population of Year 10 secondary students. Further research with larger samples would help to construct a more complete profile of the procedural and conceptual difficulties experienced by these students when attempting to solve genetic pedigree problems.

Further research should be conducted to test the effectiveness of a revised genetics curriculum based on the recommendations from this study.

Research in these areas are important as they will provide information which will form the basis of curricular and instructional decisions, regarding Year 10 genetics. It is important that students completing compulsory schooling have a sound grasp of genetics as they may face important decisions regarding inherited diseases in their own families.

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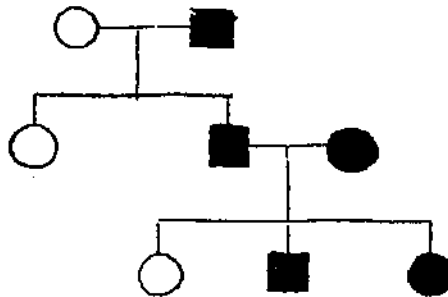
**APPENDIX 1**

The pedigree shows the inheritance of a human characteristic.  
People with the characteristic are shaded in the pedigree.  
Males are represented by squares and females by circles.

Your job is to find out if the characteristic is **autosomal dominant**, **autosomal recessive**, **x-linked dominant** or **x-linked recessive**.  
Provide as complete and conclusive answers as you can.

**Problem A**

This pedigree shows the inheritance of a common trait.



The pedigree shows the inheritance of a human characteristic.

People with the characteristic are shaded in the pedigree.

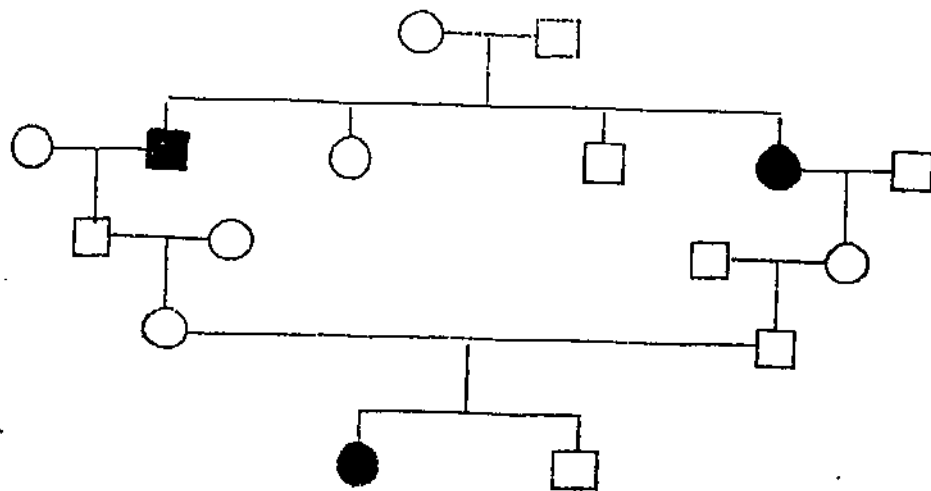
Males are represented by squares and females by circles.

Your job is to find out if the characteristic is autosomal dominant, autosomal recessive, x-linked dominant or x-linked recessive.

Provide as complete and conclusive answers as you can.

### Problem B

This pedigree shows the inheritance of a rare trait.

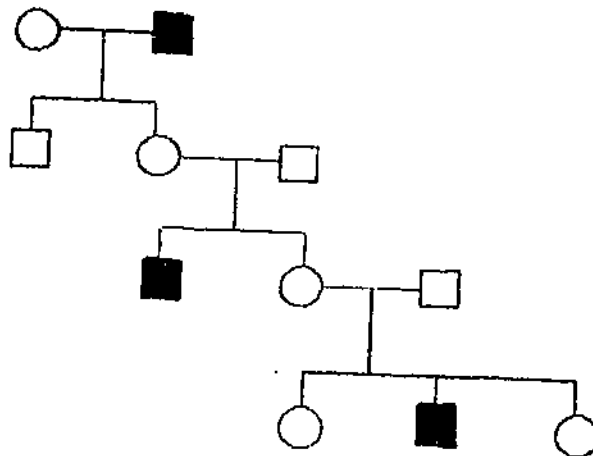


The pedigree shows the inheritance of a human characteristic.  
People with the characteristic are shaded in the pedigree.  
Males are represented by squares and females by circles.

Your job is to find out if the characteristic is autosomal dominant, autosomal recessive, x-linked dominant or x-linked recessive.  
Provide as complete and conclusive answers as you can.

### Problem C

This pedigree shows the inheritance of a rare trait.



## APPENDIX 2

### Debriefing Questions

#### *Standardised Questions:*

1. How did you decide what to test first ?
2. What are meant by dominant and recessive ?
3. What is meant by x-linked ?

#### *Specific Questions:*

4. Explain how you worked out the genotypes of the last generation of offspring.

### **APPENDIX 3**

#### **INTERVIEW PROCEDURE**

**1. Explanation of why interview is being conducted.**

- Part of a research project.
- Aim is to identify difficulties students have.
- Provide teachers with suggestions for how to teach this more effectively.
- Enable students to better understand and solve problems.

**2. Explanation of think-aloud protocol.**

- Subjects to think out loud through each step of procedure as they solve problems.
- Enables researcher to understand what subjects are thinking.
- To be tape recorded as not to miss vital information.

**3. Instruct subject to solve simple mathematical problem.**

- Encourage the use of think-aloud protocol.
- Model procedure of first problem, then get subjects to attempt next two.

**4. Introduce 1st problem and explain instructions to subjects.**

- Genetic pedigree problem to be solved according to instructions.
- Remind subjects to use same procedure as before.
- Subjects work on problem.

**5. Present 2nd problem.**

- Subjects work on problem.

**6. Present 3rd problem.**

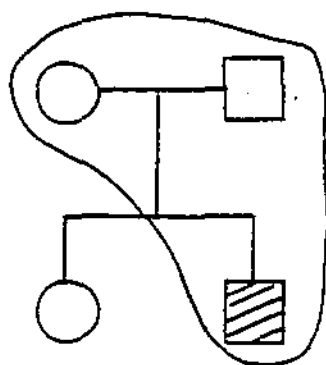
- Subjects work on problem.

**7. Conduct debriefing session when subjects have completed all three problems.**

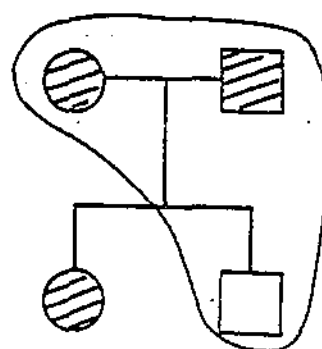
- Ask debriefing questions.

## APPENDIX 4

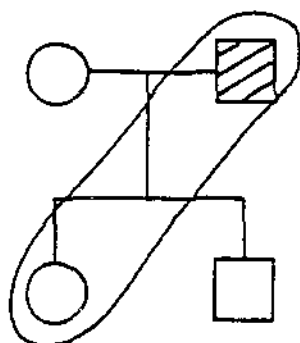
## PATTERNS OF INHERITANCE



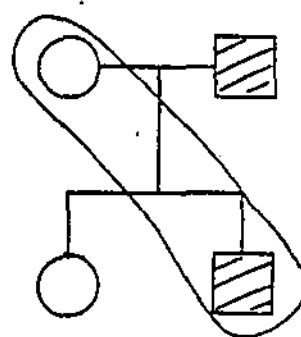
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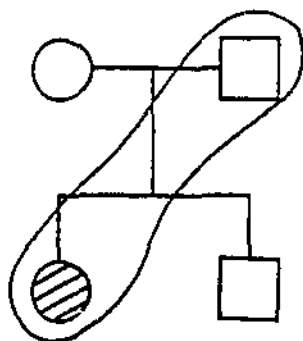
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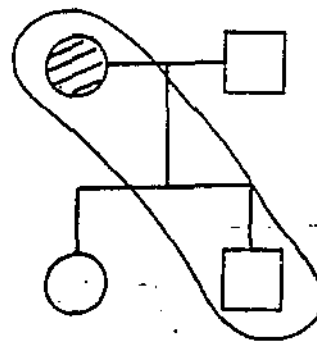
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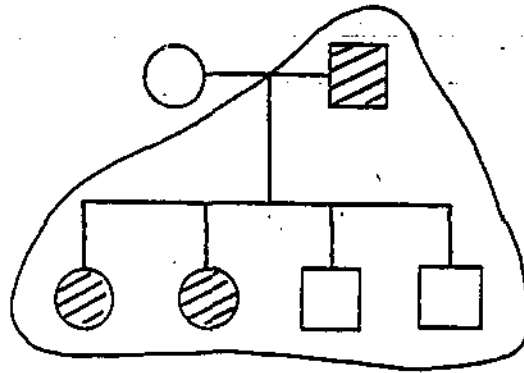
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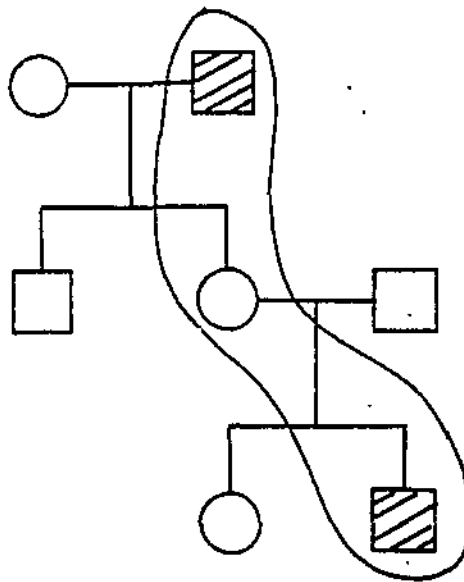
CAN'T BE XR



CAN'T BE XR



LIKELY TO BE XD



LIKELY TO BE XR