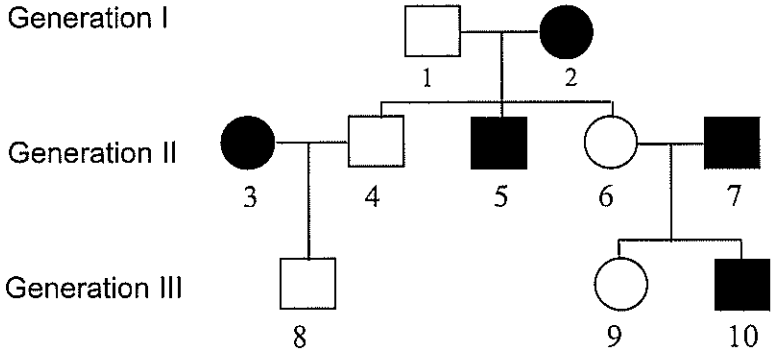
HUMAN BIOLOGICAL SCIENCE
MORE PEDIGREE CHARTS

1. The following diagram is a pedigree of albinism. A square represents a man and a circle a woman. White represents normal while shaded individuals are albino.



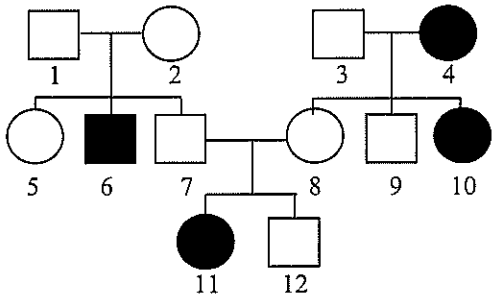
a) If individuals 3 and 4 had10 children, how many of them would be expected to be albinos? Why?

b) In Generation III, both individuals 8 and 9 were normal. Were they homozygous or heterozygous for A?

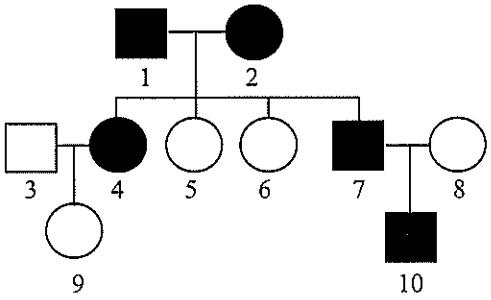
c) What genotypes would you expect among the offspring of the marriage between 8 and 9 and in what proportion?

2. The following diagrams show the pedigrees for the appearance of some human hereditary characteristics (shaded symbols). In each case determine if the inherited trait dominant or recessive or if this cannot be decided from the pedigree given?

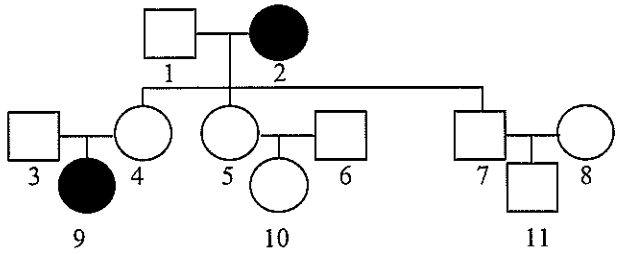
A.



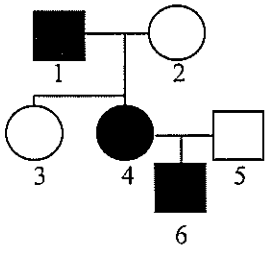
B.



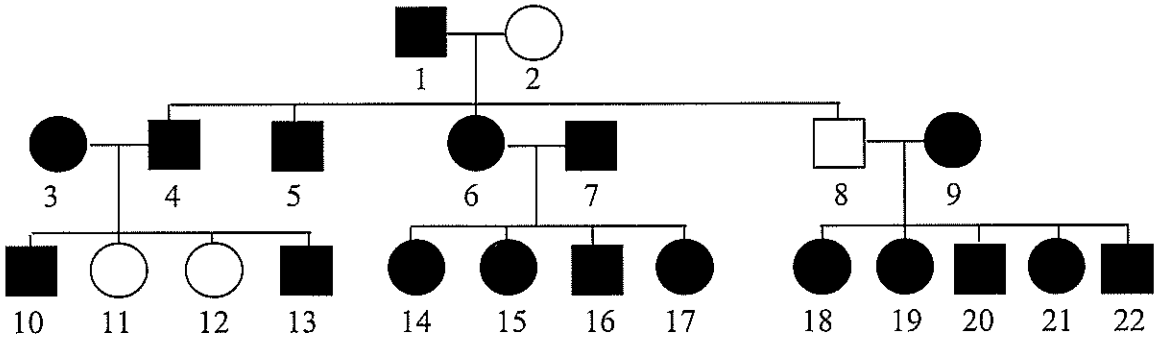
C.



D.



3. In humans, tongue rolling is determined by the presence of a dominant gene (R), whose recessive allele is represented by (r). The following represents a family tree for a number of individuals.



Squares represent males, and circles represent females. Black squares and circles represent rollers. White squares and circles represent non-rollers.

a. State the genotypes of individuals.

(i) 1
(ii) 3 and 4
(iii) 9

b. Individual 13 and his non-roller wife have a daughter Alice who is a tongue-roller. Individual 21 has a son Tom who is also a roller. But her husband is a non-roller.

i. Make a diagram to show the result of the cross between Alice and Tom.
ii.

iii. State the ratios of the genotypes and phenotypes of offspring in the cross in (i).

iv. Why is it not possible to determine the genotypes of individual 14?

v. If individual 5 marries a non-roller woman, what is the probability that their first child is tongue-roller?

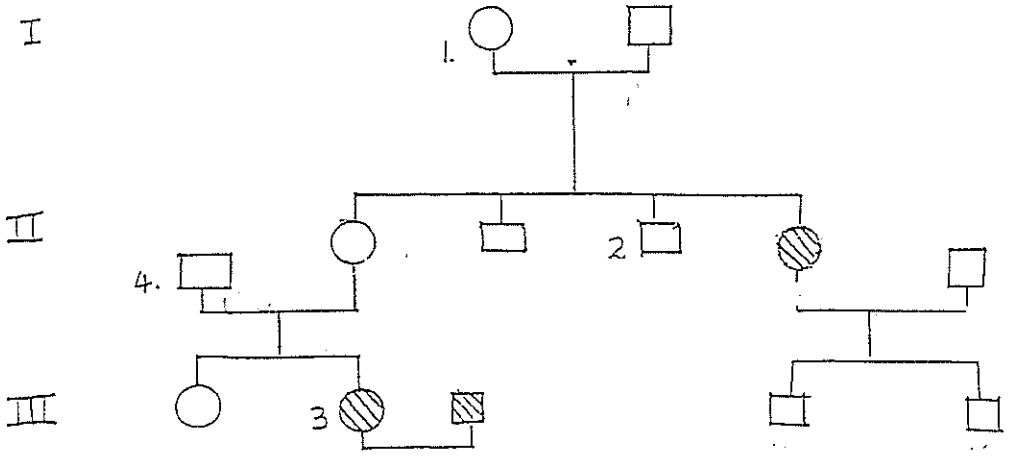
4. A woman who has unusually short fingers marries a man who has fingers of normal length. They have four children, two of each sex. One of their sons and one of their daughters have unusually short fingers.

i. Draw the pedigree.
ii. If a single pair of alleles governs finger length, could the pedigree be explained by autosomal dominant inheritance?
iii. Is the pedigree consistent with it being an example of autosomal recessive inheritance?

H BIOL - GENETICS

SECTION B: Write all your answers to Section B in the spaces provided.

Question 1 is based on the pedigree shown below in which individuals with a common bone disease are indicated by shading.



A Assuming that no new mutation has occurred in the pedigree above, choose from the following list those words which best describe the mode of inheritance of the bone disease; autosomal, X-linked, dominant, recessive and give one conclusive reason why, for each choice.

B What are the genotypes of the following individuals?

I 1

II 2

II 4

III 3

(4 marks)

C Individual III 3 is pregnant. What is the probability that she is carrying a daughter with the bone disease? (Show working)

Question 2

a) Colour blindness is a hereditary condition in which degrees of deficiency in colour perception may occur. The Pedigree Chart in **Figure 11 (below)** shows the inheritance of red- green colour blindness, a common form of the condition.
Red-green colour blindness is an X-linked condition.

i) Is the gene for red—green colour blindness dominant or recessive?

Give one conclusive reason for your answer.

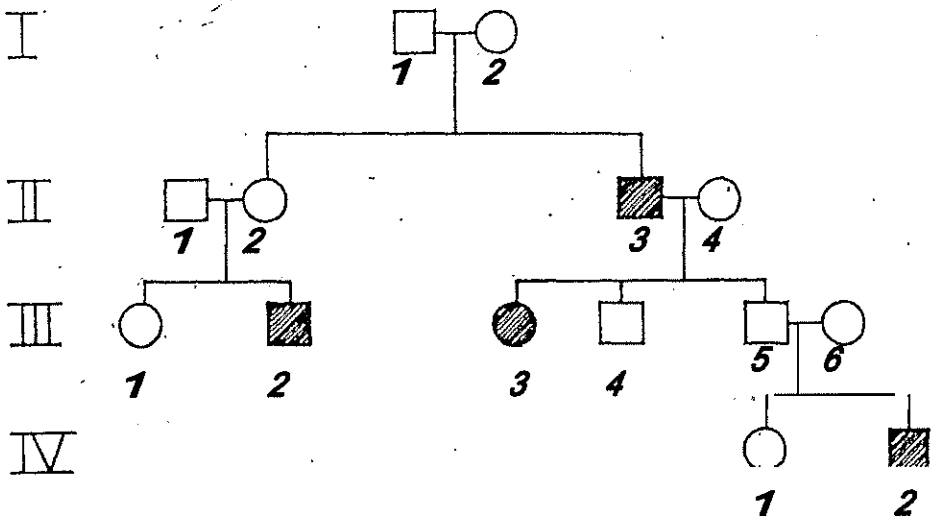


Figure 11
Pedigree Chart

ii) What are the genotypes of the following individuals shown in Figure 11?

I 2

III2

III 4

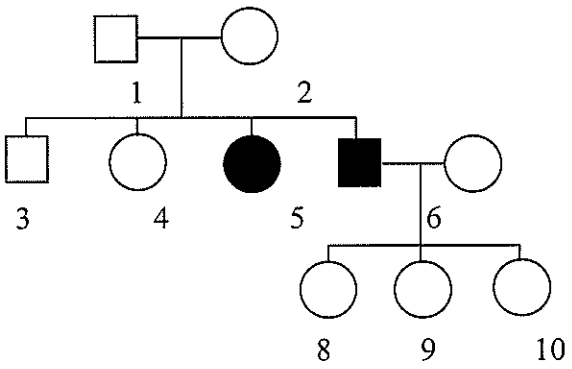
Indicate, in the space below, the meaning of the symbols you are using.

iii) Female IV 1 is pregnant. The father of the child has normal vision. What is the probability that the child will be a red—green colour blind son?: Show working.

WORKSHEET PEDIGREE CHARTS

Study this example showing how to complete a pedigree chart, and then work on the problems that follow.

Albino (*a*) is recessive to normal pigmentation (*A*). Work out the genotype and phenotype of each individual in this pedigree. (Shaded individuals are albino)



STEPS: 1. Begin from the known - what is the genotype of 5.
6.

2. Work back to their parents - what must be the genotype of parents
1.
2.

3. Go forward! - What could be the genotype/s of 3.
4.

4. What must be the genotype of 8.
9.
10.

5. Give the possible genotypes of 7.

QUESTIONS:

6. If individual 8 marries an albino what is the chance of the couple having an albino child?

STEPS: Genotype 8 = Genotype of albino =

Genotypes of offspring?

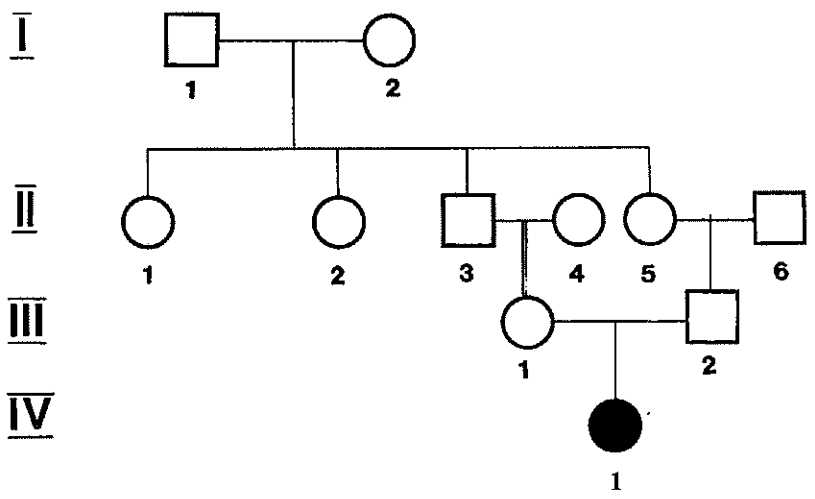
Answer:

7. Which individuals in this pedigree can only be:

a. homozygous (pure breeding)?

b. heterozygous (hybrid)?

HUMAN BIOLOGICAL SCIENCE
PHENYLKETONURIA PEDIGREE



Pedigree in which PKU individuals affected are indicated by shaded symbols.

a) Assuming that no new mutation has occurred in the pedigree, describe the mode of inheritance of P.K.U. Give one conclusive reason for your decision.

2 marks

b) What is the relationship between III.1 and III.2? (apart from husband and wife)

1 mark

c) Identify the genotype of
III.1 IV.1

2 marks

Explain any symbols you use.

d) III.1 is pregnant. What is the probability that her second child will have P.K.U.?

1 mark

e) P.K.U. originates from gene mutations. Explain the meaning of "gene mutation"

1 mark

f) The symptoms of P.K.U. can be prevented if the defect is detected very early in life, and a diet very low in the amino acid phenylalanine is maintained during development. In which food group is phenylalanine found? 1 mark

g) What problems do you see in providing a diet suitable for young babies with P.K.U., and how could these problems be overcome?

3 marks

Activity 4 Pedigrees

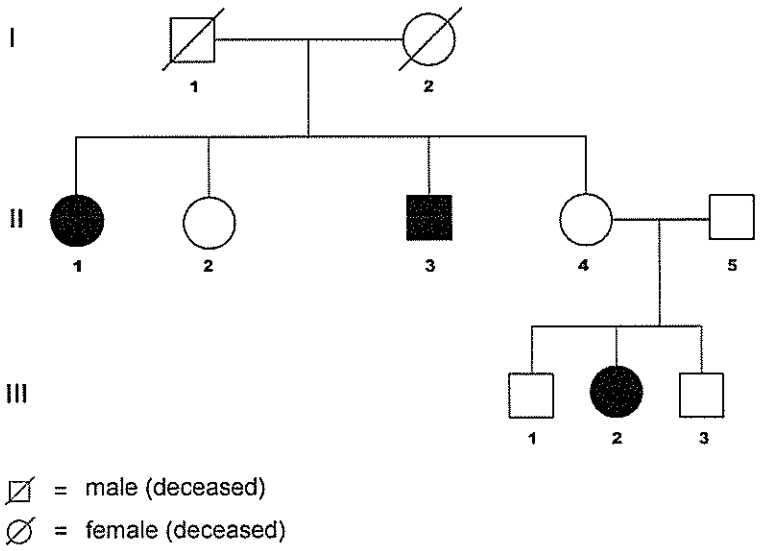
Case Study 1: Cystic fibrosis

Cystic fibrosis is the most common genetic disorder in Australia. About one in every 2 500 babies, male or female, is born with cystic fibrosis. In addition, about 1 in 25 in this population is a carrier of a mutation of this gene.

In people with cystic fibrosis, the mucus lining of the respiratory and gastrointestinal tracts is extremely thick. The sweat glands and the skin are also affected so that the person secretes salty sweat. Individuals with this disease have repeated lung infections and digestive upsets. There is no cure but treatment using pancreatic extracts, antibiotics and physiotherapy can help greatly. CF individuals used to die within the first years of life. As advances in diagnosis and treatments have been made, the life expectancy has lengthened to 31 years for males and 29 years for females.

Examine the pedigree for cystic fibrosis and answer the following questions.

Cystic Fibrosis Pedigree



1. Cystic fibrosis is an autosomal disease. Is the gene dominant or recessive? Explain your reasoning.

2. What are the genotypes of the parents in generation I? Show your working.

3. Parents II4 and II5 are expecting another child. What is the probability that this child will have cystic fibrosis?

4. Given that the first child of parents II4 and II5 is a healthy 10-year-old boy, what is the probability that he is a carrier of the inherited disorder?

Case study 2: Huntington disease

Huntington disease is an inherited disorder which results in lack of control over muscles and progressive mental deterioration. As the disease progresses, walking and speech become more difficult and memory and intellectual functions decline. As the symptoms do not appear until a person is about 30 to 50 years of age, the gene may have been passed on to their offspring.

Consider the following family history of Huntington disease.

Jane (43 years) has just developed some early symptoms of Huntington disease including mood swings, loss of memory and uncontrolled movements. Her father, Don, has Huntington disease and has to be cared for in a nursing home. Her mother, Mary, does not have the disease. Jane's brother, Ian (52), does not have the disease.

Jane's husband, Joe (42) does not have the disease and has no family history of the disease. Jane and Joe have two children, Carol (25) and Max (27). Carol is married to Tim (28) and they have a child Susy (2). Max (should be TIM) has no family history of Huntington disease.

Use this information to answer the following questions.

5. Draw a pedigree to show all the members of Susy's extended family. Remember to shade those who have Huntington disease.

6. Describe the mode of inheritance of Huntington disease. Give evidence from the pedigree.

7. List the possible genotypes of Mary, Don, Jane and Joe. Explain the symbols you are using.

8. What is the probability that Carol will have inherited the disease? Set out the genetics cross in full and show your working.

9. What is the probability that Susy will have inherited the disease? Explain your answer.

10. A genetic test is available to detect the gene for Huntington disease. However, there is no treatment to slow the onset or progression of the disease. There are treatments to reduce some of the symptoms such as antidepressants for depression and medication to reduce the involuntary muscle movements. Consider the decision Carol and Tim face whether to have their child, Susy, tested for Huntington disease. Discuss the advantages and disadvantages of having Susy tested. Consider any ethical issues involved.