16. Pedigree Analysis

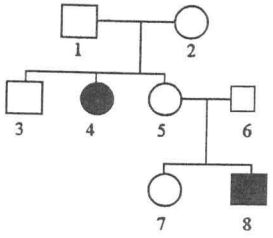
General Objectives

- **Use pedigrees to predict the inheritance of human characteristics**

Content

Pedigree construction and symbols used. Sex determination. Autosomal, X-linked, dominant, recessive, multiple alleles, co-dominance and polygenic modes of inheritance. Examples including Huntington disease, PKU, Duchenne muscular dystrophy, and skin colour (dihybrid crosses not required). Also ABO blood groups and the existence of other blood grouping systems.

Question **1** refers to the pedigree below showing the inheritance of a recessive characteristic in a family.

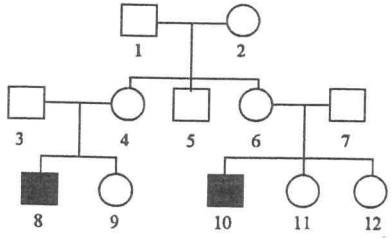


1. 1997 / 15

Which one of the following alternatives lists individuals in this pedigree who are definitely heterozygous for the recessive characteristic?

(a) 1, 2 and 7.
(b) 3, 6 and 7.
(c) 1, 3 and 6.
(d) 1, 5 and 6.

Question **2** refers to the pedigree below of Duchenne type muscular dystrophy which is inherited as a sex-linked characteristic.



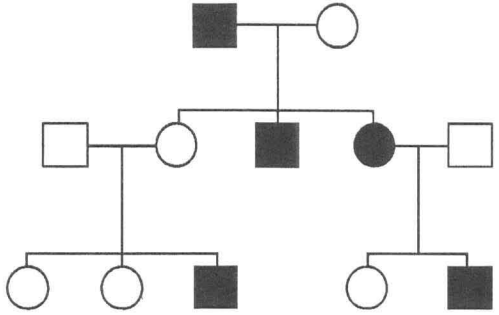
2. 1997 / 16

What is the probability that individual 9 is a carrier of the gene?

(a) 0
(b) 0.25
(c) 0.5
(d) 1

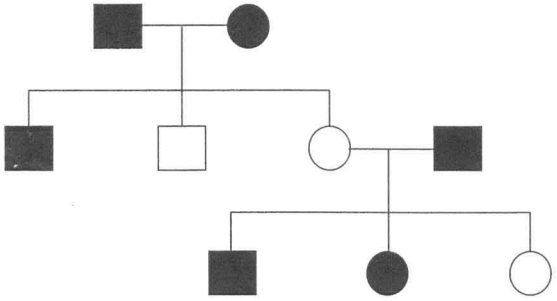
3. 1997 / 17

It can be concluded that the shaded characteristic is determined by



(a) an autosomal dominant gene.
(b) an autosomal recessive gene.
(c) a sex-linked recessive gene.
(d) a co-dominant gene.

4. 1998 / 14

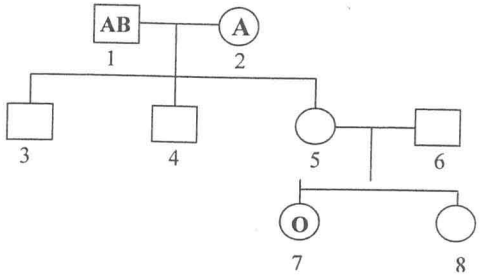


It can be concluded that the shaded characteristic is determined by

(a) an autosomal recessive gene.
(b) an autosomal dominant gene.
(c) an X-linked recessive gene.
(d) an X-linked dominant gene.

5. 1998 / 16

The question refers to the pedigree chart below, which shows the blood groups of three family members (1, 2 and 7).

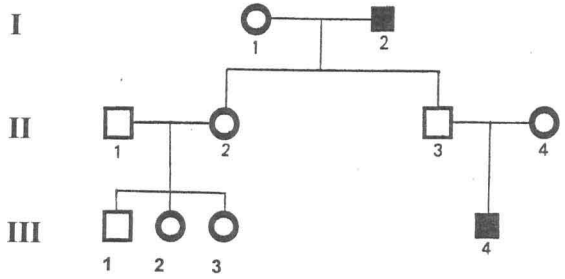


Regarding the individuals in the pedigree above,

(a) individual 2 must have the genotype I^AI^A
(b) individual 3 could be blood group A, AB or O.
(c) individual 5 could be blood group B or O.
(d) individual 6 must have the allele *i*.

6. 1996 / 49

Question 6 refers to the pedigree below indicating haemophilia, an X-linked genetic disorder.



(a) What is the genotype of individuals

I2? _____

II2? _____

II4? _____ (3)

(b) Which female/s are definite carrier/s?

_____ (1)

(c) If II3 and II4 had another child, what is the probability of it being a haemophiliac?

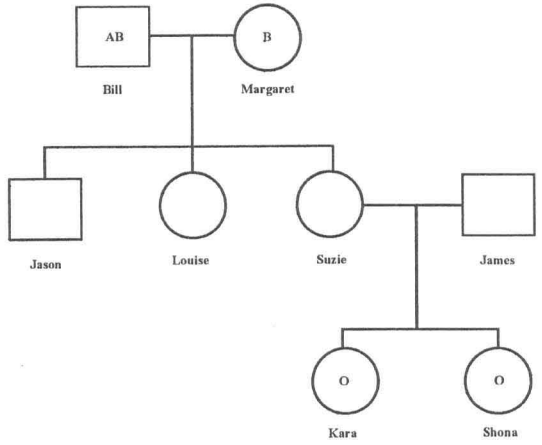
_____ (1)

(d) If II1 and II2 had another son what is the probability that he will be a haemophiliac?

_____ (1)

7. 1997 / 43

Question 7. relates to the pedigree below showing the inheritance within a family of blood groups in the ABO system. Blood groups are shown for **some** of the family members, i.e. Bill, Margaret, Kara and Shona.



In this system three alleles control whether an individual has A, B, AB or O blood group. These alleles are I^A, I^B and I.

(a) Which term is used to describe characteristics determined by more than one pair of alleles?

_____ (1)

(b) What are Suzie's possible genotypes?

_____ (1)

(c) What are Suzie's possible phenotypes?

_____ (1)

(d) Calculate the probability that Jason has the same blood group as his father.

_____ (1)

(e) What is the probability that Jason has blood group O?

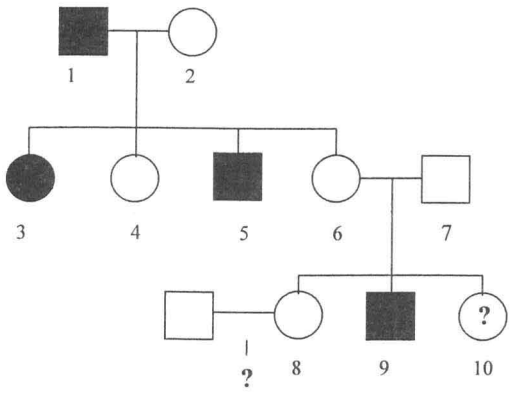
_____ (1)

(f) What is the probability that Jason and Louise both have blood group B?

_____ (1)

8. 1998 / 45

Question **8** relates to the pedigree below. This shows the inheritance, within a family, of the X-linked characteristic known as red-green colour blindness. Shaded individuals possess the characteristic. Individual 10 is a baby and it has not yet been determined if she is affected



(a) Using the symbol *R* to represent the dominant allele and *r* for the recessive allele:

(i) What is the genotype of individual 1?

_____ (1)

(ii) What is the genotype of individual 6?

_____ (1)

(b) What is the probability that newborn daughter 10 will be

(i) red-green colour blind?

_____ (1)

(ii) a carrier of the gene?

_____ (1)

(c) If daughter 8 has a son by her unaffected husband, what is the probability that this son will be red-green colour blind?

_____ (1)

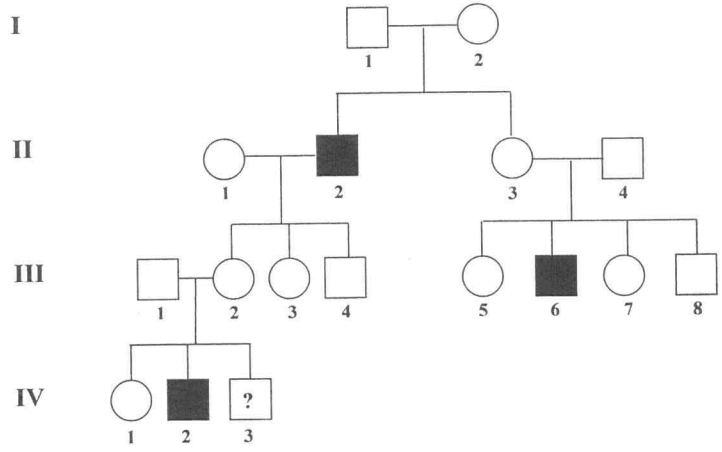
9. 1999 / 43

(b) Draw a correct pedigree from the following information, using the initial letters of the names of the people involved (eg 'N' for Norman). **A freehand drawing is acceptable.** (3)

Alan is married to Beatrice. They have five children born in the following order, Cheryl, Douglas, Edward, Frederick and George. Sadly, Frederick died as a baby. Cheryl is married to Henry and George is married to Isabelle. Cheryl and Henry have two daughters, Julie (eldest) and Kate. Leonard and Martin are brothers. Their mother is Isabelle.

10. 2001 / 46

The pedigree below shows the inheritance, within a family, of a very rare disorder. Individual IV.3 is a newborn baby who has not yet been tested for the disorder.



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(2001 / 46 cont)

(a) Is the disorder inherited as a dominant or recessive trait? _____ (1)

(b) Explain how you arrived at your answer in (a). (1)

_____

_____

(c) Is the trait **more likely** to be autosomal or X-linked?

_____ (1)

(d) Explain how you arrived at your answer in (c). (1)

_____

_____

(e) Using the letters 'A' and 'a' to represent dominant and recessive alleles, respectively, write the full genotype of individual I.2.

_____ (1)

(f) What is the chance that the newborn baby (individual IV.3) has the disorder?

_____ (1)

(g) The ABO blood grouping system displays two phenomena in genetics.
Explain the following terms and give an example using the ABO system.

(i) Multiple alleles (2)

_____

_____

(ii) Co-dominance (2)

_____

_____

(h) Apart from the ABO system, name ONE other blood grouping system.

_____ (1)

EXTENDED ANSWERS

11. 2000 / 55

(b) You have just constructed a family pedigree chart for a particular characteristic. After studying the pedigree, you recognise that the trait shows an autosomal pattern of inheritance. Explain what information the pedigree would contain for a dominant characteristic and what information it would contain if it was recessive. (8)