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Mitochondrial DNA

- Mitochondria carry about 37 genes.
- The mitochondrial genes are responsible for the construction of several important enzymes involved in cellular respiration.
- Mitochondrial genes are inherited from the mother only.

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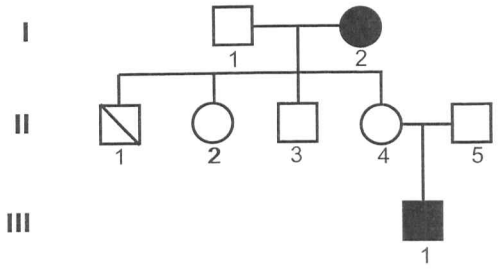
Impact of faulty mitochondrial genes

- General - Small stature and poor appetite
- Central nervous system - Developmental delay/intellectual disability, progressive neurological deterioration (dementia such as the late-onset form of Alzheimer disease), seizures, stroke-like episodes (often reversible), difficulty swallowing, visual difficulties and deafness
- Skeletal and muscle - Floppiness, weakness and exercise intolerance
- Heart - Heart failure (cardiomyopathy) and cardiac rhythm conditions
- Kidney - Problems in kidney function

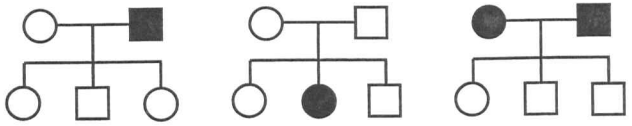
The Australasian Genetics Resource Book

Working with pedigrees

1. Explain what all the symbols used in the following pedigree represent and how *trait x* is inherited.(individuals with *trait x* are shaded.)



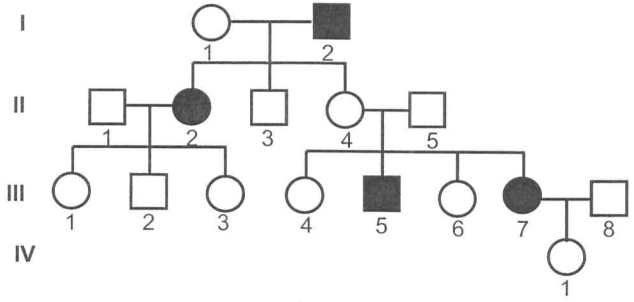
2.



a) In which of the above pedigrees can you be certain of the dominance of the trait?

b) Work out the genotype of the individuals in the above pedigrees.

3. The following pedigree shows the occurrence of polydactyly (an extra finger) in a family. Individuals with polydactyly are shaded black.



a) What is the relationship between a) I 1 and IV 1, b) III 1 and III 3, c) III 3 and III 4?

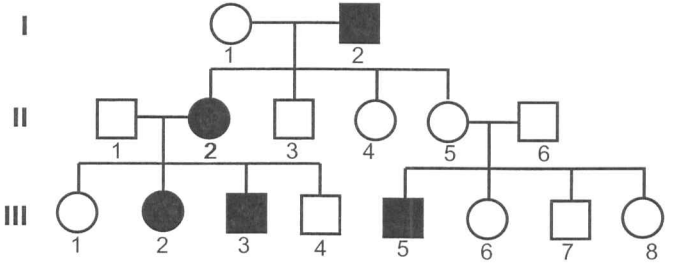
b) Which males in this pedigree display polydactyly?

c) Which individuals in this pedigree must be carriers of the gene responsible for polydactyly?

d) Which individuals in this pedigree have a genotype that can be determined with certainty?

e) Is polydactyly a dominant or a recessive trait?

4. The pedigree below shows the occurrence of an inherited trait in a family. Individuals with the trait are shaded.



a) Is this trait dominant or recessive?

b) Explain your answer to a) nominating specific individuals from the pedigree.

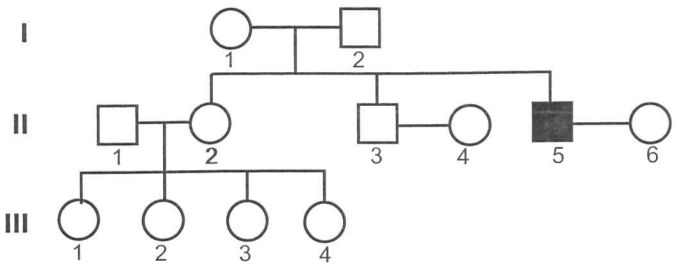
c) Is the mode of inheritance most likely to be autosomal or X-linked?

d) Explain your answer to c) nominating specific individuals from the pedigree.

e) If individual II 5 and II 6 had another child, what is the probability that this child would show the trait?

f) If individual II 5 and II 6 then had two more children, what is the combined probability that they would both show the trait?

5. PKU (phenylketonuria) is an inherited disease. The allele (n) for the disease is recessive to the normal allele (N). The diagram shows how PKU was inherited in a family.



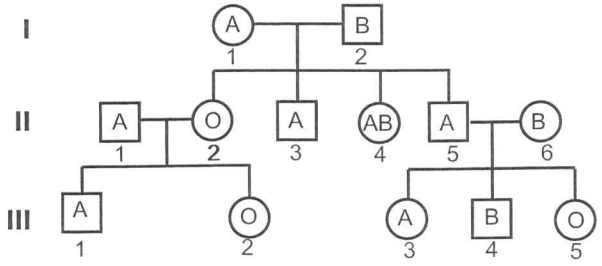
a) Give the genotype of individual I 2 and III 2

b) How many of the children of I 1 and I 2 are homozygous?

c) If II 5 and II 6 have a child, what is the probability that it will have PKU?

d) II 1 and II 2 have four children, all of whom are female. What is the probability that their next child will be female?

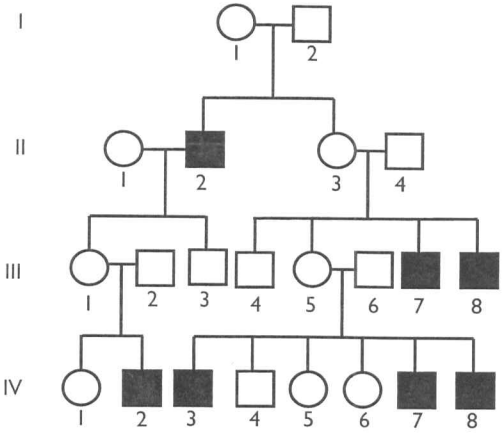
6. The following pedigree shows the blood groups in a family.



a) Predict the genotypes of all the individuals in this pedigree.

b) If individuals II 5 and II 6 were to have another child, what would be the chance of it having blood group AB?

7. The pedigree below shows the occurrence of an inherited trait in four generations of a family. Individuals with the trait are shaded.



a) Is this trait dominant or recessive?

b) Explain your answer to a) nominating specific individuals from the pedigree.

c) Is the most likely mode of inheritance of this trait autosomal or X-linked?

d) Explain your answer to c) nominating specific individuals from the pedigree.

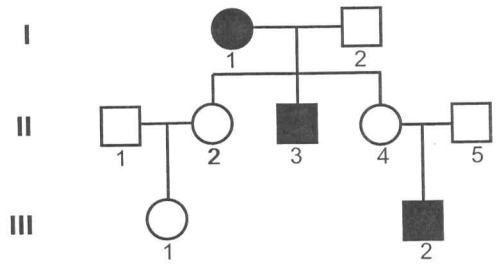
e) Could the occurrence of this trait be explained by any other type of inheritance?

f) Assuming that this trait is X-linked, work out the genotype of all individuals.

g) If this trait was a potentially fatal condition, such as haemophilia, what advice might a genetic counselor give to the children of III 5 and III 6?

8. The following pedigree shows the occurrence of a fairly common, inherited form of colour-blindness in a family. All the colour-blind people are shaded.

[Use "A" to denote the dominant allele and "a" to denote the recessive allele]



a) Is the inheritance of this form of colour-blindness due to a dominant or a recessive gene?

b) Using named individuals, explain why.

c) Could the gene responsible for this form of colour-blindness be X-linked ?

d) Could the gene responsible for this form of colour-blindness be autosomal?

e) Is there any way of telling which is correct?

The next three questions assume that this condition is X-linked.

f) Give the genotypes of individuals I 1, I 2, II 4, II 5 and III 2.

g) If II 4 and 5 have another child, what is the probability that it will be a colour-blind boy?

h) What can be inferred about the phenotype of I 1's parents?