

# Pedigree Analysis

## Key ideas

**Symbols:** square = male, circle = female, shaded = affected, dot = carrier; horizontal line = couple, vertical line = their children. Generations use Roman numerals (I, II, III); people are numbered left to right.

**Autosomal recessive:** can skip generations; two unaffected parents can have an affected child.

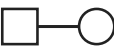
**Autosomal dominant:** every affected person has an affected parent; two affected parents can have an unaffected child.

**X-linked recessive:** mostly males; affected sons usually have carrier mothers; an affected daughter must have an affected father.

## A Pedigree symbols

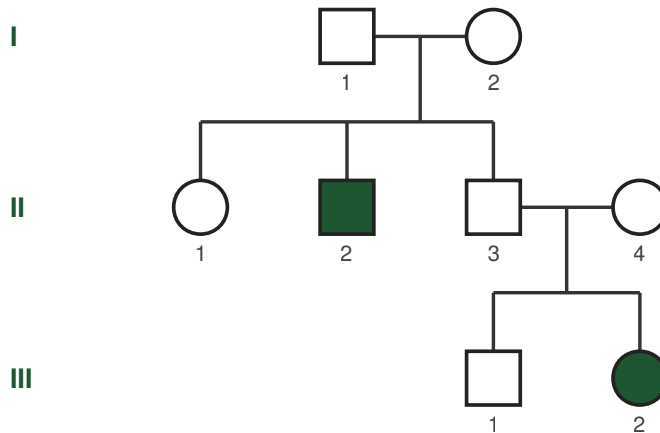
3 marks

Name each symbol (½ mark each).

|                                                                                         |                                                                                         |                                                                                         |                                                                                         |                                                                                           |                                                                                           |
|-----------------------------------------------------------------------------------------|-----------------------------------------------------------------------------------------|-----------------------------------------------------------------------------------------|-----------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------|
| 1.<br> | 2.<br> | 3.<br> | 4.<br> | 5.<br> | 6.<br> |
| _____                                                                                   | _____                                                                                   | _____                                                                                   | _____                                                                                   | _____                                                                                     | _____                                                                                     |

## B Pedigree 1: albinism

6 marks



7. Is this trait dominant or recessive? Give the evidence from the pedigree. (2 marks)

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8. Explain why this trait *cannot* be X-linked recessive.

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9. Give the genotypes of I-1, I-2 and II-2 (use A and a).

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10. II-1 is unaffected. What is the probability that she is a carrier (Aa)?

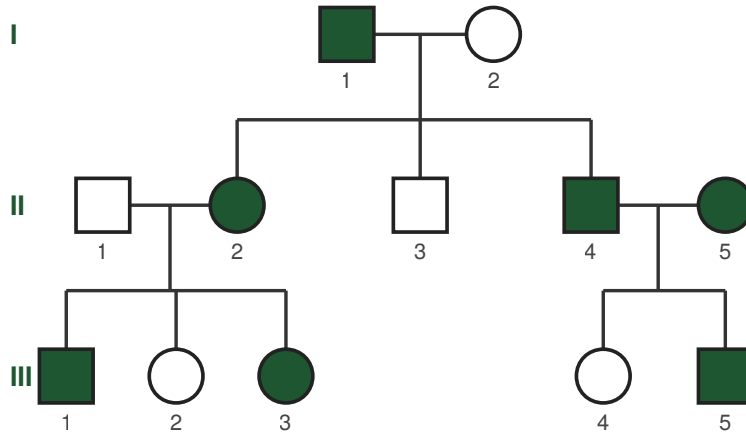
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11. What is the probability that the next child of II-3 and II-4 will be affected?

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### C Pedigree 2: Huntington's disease

6 marks



12. Is this trait dominant or recessive? Give the evidence from the pedigree. (2 marks)

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13. Explain why this trait *cannot* be X-linked dominant.

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14. Give the genotypes of I-2 and II-2.

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15. What is the probability that the next child of II-1 and II-2 will be affected?

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16. What is the probability that the next child of II-4 and II-5 will be affected?

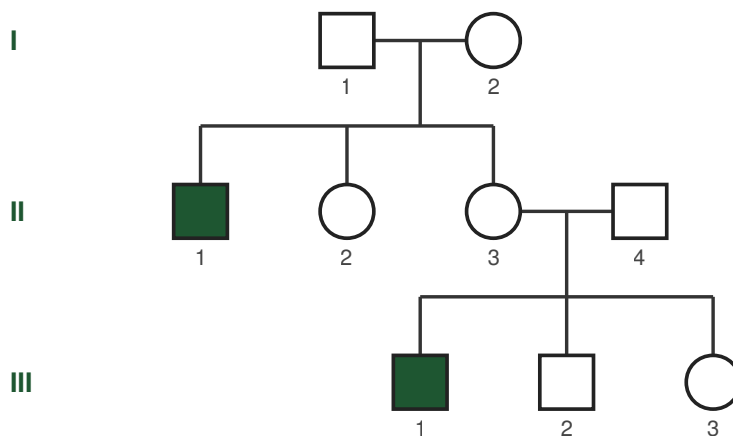
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### D Pedigree 3: haemophilia

5 marks



17. What is the most likely inheritance pattern? Give two clues. (2 marks)

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18. Give the genotypes of I-2 and II-3 (use  $X^H$  and  $X^h$ ).

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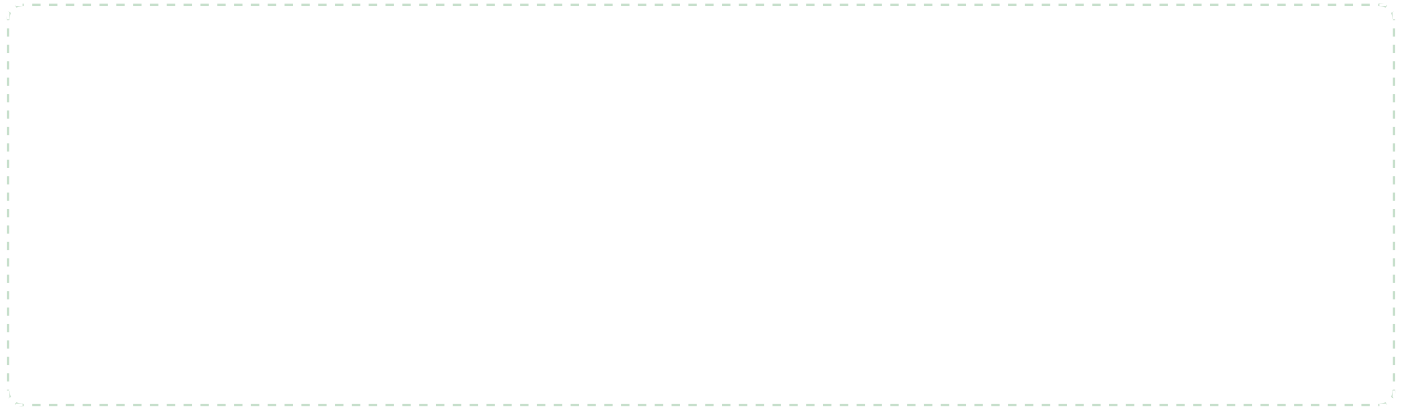
19. II-2 is unaffected. What is the probability that she is a carrier?

20. What is the probability that the next *son* of II-3 and II-4 will be affected?

**E Draw a pedigree**

2 marks

21. Draw the pedigree. Two unaffected parents have three children: an affected son, an unaffected son and an unaffected daughter. The daughter has a child with an unaffected man, and they have one affected daughter. Number the generations and individuals. (2 marks)



**F Spot the mistake**

2 marks

22. A student says: "The trait skips generation II, so it must be dominant." Explain the error and correct it.

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23. A student says: "Only males are affected in this family, so the trait must be on the Y chromosome." Explain the error and correct it.

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# Pedigree Analysis

**ANSWER KEY****Key ideas**

**Symbols:** square = male, circle = female, shaded = affected, dot = carrier; horizontal line = couple, vertical line = their children. Generations use Roman numerals (I, II, III); people are numbered left to right.

**Autosomal recessive:** can skip generations; two unaffected parents can have an affected child.

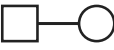
**Autosomal dominant:** every affected person has an affected parent; two affected parents can have an unaffected child.

**X-linked recessive:** mostly males; affected sons usually have carrier mothers; an affected daughter must have an affected father.

**A Pedigree symbols**

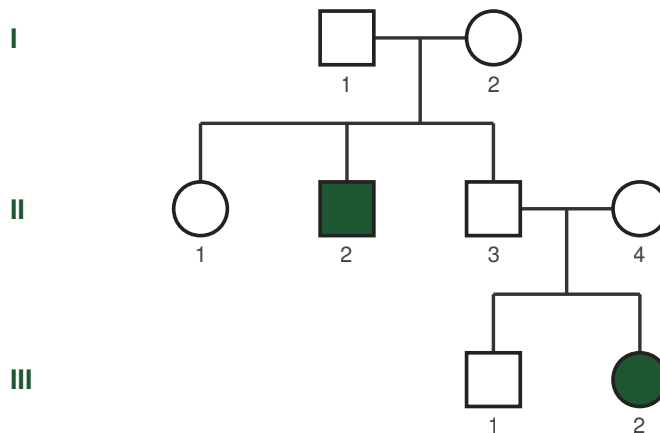
3 marks

Name each symbol (½ mark each).

|                                                                                         |                                                                                         |                                                                                         |                                                                                         |                                                                                           |                                                                                           |
|-----------------------------------------------------------------------------------------|-----------------------------------------------------------------------------------------|-----------------------------------------------------------------------------------------|-----------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------|
| 1.<br> | 2.<br> | 3.<br> | 4.<br> | 5.<br> | 6.<br> |
| Affected male                                                                           | Unaffected female                                                                       | Mating (couple) line                                                                    | Affected female                                                                         | Unaffected male                                                                           | Carrier female                                                                            |

**B Pedigree 1: albinism**

6 marks



7. Is this trait dominant or recessive? Give the evidence from the pedigree. (2 marks)

**Recessive.** I-1 and I-2 are both unaffected but have an affected son (II-2), so they must carry a hidden allele.

8. Explain why this trait *cannot* be X-linked recessive.

III-2 is an affected daughter with an unaffected father (II-3). An X-linked recessive daughter must get an affected X from her father.

9. Give the genotypes of I-1, I-2 and II-2 (use A and a).

I-1 Aa, I-2 Aa, II-2 aa

10. II-1 is unaffected. What is the probability that she is a carrier (Aa)?

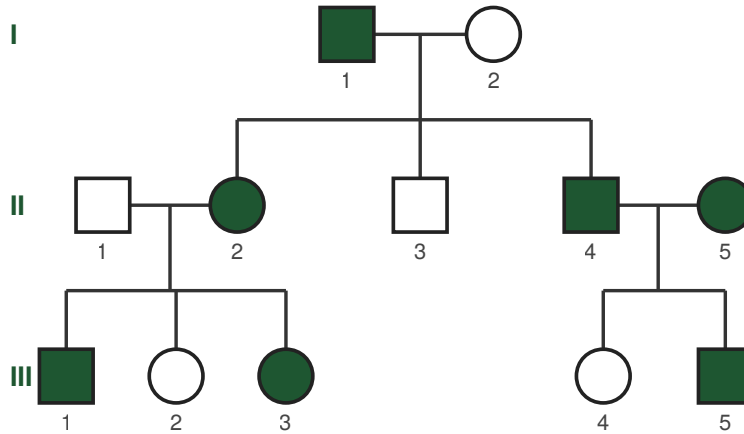
Of the unaffected outcomes (AA, Aa, Aa), two are carriers: 2/3

11. What is the probability that the next child of II-3 and II-4 will be affected?

They already have an affected child, so both are Aa: 1/4

### C Pedigree 2: Huntington's disease

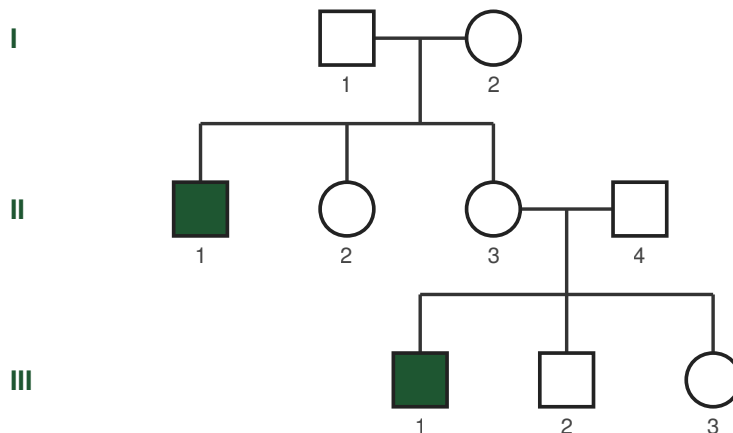
6 marks



12. Is this trait dominant or recessive? Give the evidence from the pedigree. (2 marks)  
**Dominant. II-4 and II-5 are both affected but have an unaffected daughter (III-4), so both must be Aa and the affected allele is dominant.**
13. Explain why this trait *cannot* be X-linked dominant.  
**Affected son II-4 has an unaffected mother (I-2). A son gets his X only from his mother, so the trait cannot be X-linked dominant.**
14. Give the genotypes of I-2 and II-2.  
**I-2 aa (unaffected); II-2 Aa (affected, with an unaffected mother)**
15. What is the probability that the next child of II-1 and II-2 will be affected?  
**aa × Aa: 1/2**
16. What is the probability that the next child of II-4 and II-5 will be affected?  
**Aa × Aa: P(A\_) = 3/4**

### D Pedigree 3: haemophilia

5 marks



17. What is the most likely inheritance pattern? Give two clues. (2 marks)  
**X-linked recessive. Only males are affected, and affected sons (II-1, III-1) have unaffected parents, so the allele passes through carrier mothers.**
18. Give the genotypes of I-2 and II-3 (use  $X^H$  and  $X^h$ ).  
**Both are carriers:  $X^H X^h$  (each has an affected son)**
19. II-2 is unaffected. What is the probability that she is a carrier?  
**Her mother is  $X^H X^h$  and her father  $X^H Y$ : 1/2**

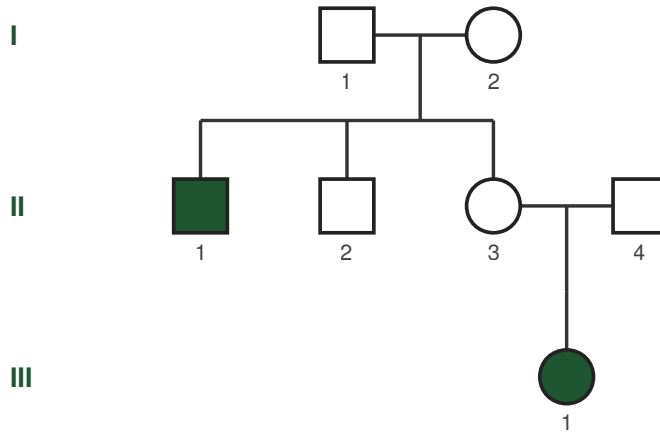
20. What is the probability that the next son of II-3 and II-4 will be affected?

**Sons get the mother's X: 1/2**

**E Draw a pedigree**

2 marks

21. Draw the pedigree. Two unaffected parents have three children: an affected son, an unaffected son and an unaffected daughter. The daughter has a child with an unaffected man, and they have one affected daughter. Number the generations and individuals. (2 marks)



Couple line between the parents, a vertical line down to the three children, and the daughter (II-3) joined to her partner with one affected daughter in generation III.

**F Spot the mistake**

2 marks

22. A student says: "The trait skips generation II, so it must be dominant." Explain the error and correct it.

**Skipping a generation is a sign of a recessive trait: unaffected carriers pass it on without showing it.**

23. A student says: "Only males are affected in this family, so the trait must be on the Y chromosome." Explain the error and correct it.

**A Y-linked trait would pass from every affected father to all his sons. Mostly-male patterns with unaffected fathers point to X-linked recessive inheritance.**