

Case 13

Marking scheme	Indicative content	Learning outcome
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Question 1a [2]

Valid reason (1)	Recording FH in schematic way (1) Builds pattern of inheritance (1) Aids diagnosis (1)	Describe how to construct a family tree and when and why it may be important to do so
Valid reason (1)	Aids further investigations (1) Calculate probabilities (1)	

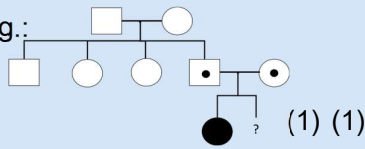
Question 1b [2]

Correctly identified (1)	Mitochondrial (1)	Apply the principles of clinical reasoning in formulating a diagnosis of genetic disease by using a family tree
Appropriate justification (1)	Maternal line only (1)	

Question 1c [1]

Correctly stated (1)	0% (1)	Apply the principles of clinical reasoning in formulating a diagnosis of genetic disease by using a family tree
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Question 2a [2]

Correct tree diagram (2)	E.g.: 	Apply the principles of clinical reasoning in formulating a diagnosis of genetic disease by using a family tree
Correct probability (1)	1 in 2 / 50% (1)	

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Question 3a

[1]

Correctly definition (1)	Process of protecting vulnerable adults or children from abuse or neglect (1)	Describe different ways patients can be vulnerable and the role of healthcare professionals in safeguarding their interests
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Question 3b

[2]

Valid suggestion (1)	Has care/support needs (1) May be experiencing abuse and may be unable to protect themselves from this (1)	Describe different ways patients can be vulnerable and the role of healthcare professionals in safeguarding their interests
Valid suggestion (1)	High risk group e.g. disabled, injured, alone (1)	

Question 3c

[3]

Valid suggestion (1)	Trauma informed practice e.g. minimal touching, seeking permission from patient to proceed (1) Allowing extra time (1) Facilitating communication – e.g. quieter room, visual aids (1) More familiar member of staff (1)	Describe how the consultation may need to be adapted when dealing with vulnerable patients
Valid suggestion (1)		
Valid suggestion (1)		

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Question 4a

[4]

A valid point relating to legal issues (1)	Informed consent – knowledge / risks / benefits to patient / issue of incidental findings (1)	Critically evaluate the ethical and legal issues raised in current and potential developments in human genetics
A second valid point relating to legal issues (1)	Confidentiality – genetic information is sensitive (1) Article 6 of Human Genome and Human Rights (1) Discrimination / Equality Act 2010 / protected characteristics (1)	
A valid point relating to ethical issues (1)	Feelings from patient – guilt? Relief? Fear? (1) Privacy (1)	
A second valid point relating to ethical issues (1)	“Genetic rights” – who owns genes? (1) Mis-leading data – stats, lack of understanding, positive / negative lifestyle changes in response to data (1)	