Solutions

Marking: **M1** a correct method, **A1** the correct answer, **B1** an independent mark.

Each answer shows the steps, then the **result**.

2.1

autosomal dominant

affected people in every generation

an affected father passes it to his son, which rules out X-linked

2.2

Aa × Aa gives AA : Aa : aa in the ratio 1 : 2 : 1

risk **1/4** for each child

2.3

healthy outcomes: AA, Aa, aA, so 3 equally likely

carrier chance **2/3**

2.4

each son: 1/2 affected

each daughter: 1/2 carrier

no daughter is affected, because she gets a normal X from her father

3.1

(a) **1/2**

(b) $\frac{1}{2} \times \frac{1}{2} = \mathbf{1/4}$

(c) some people with the variant never show the disease, for example people with 36–39 repeats in Huntington disease

(d) there is no cure, so a positive result can cause great distress; effects on family, work and insurance, with time to decide and support afterwards

3.2

(a) the variant is new (de novo) in the child

(b) low, close to the population risk; a small extra risk remains because a parent may carry it in some sperm or eggs (gonadal mosaicism)

(c) **1/2**, because the child now carries one copy of a dominant variant

3.3

(a) **1/4**

(b) $\frac{3}{4} \times \frac{3}{4} = \mathbf{9/16}$

(c) 3 healthy outcomes, 2 of them carriers; **2/3**

(d) $\frac{2}{3} \times \frac{1}{25} \times \frac{1}{4} = \mathbf{1/150}$

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