Solutions

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

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

2.1

30: **normal**

100: **premutation**

350: **full mutation**

2.2

it passes only from mothers

both sons and daughters of an affected mother can be affected; affected men do not pass it on

2.3

some genes here work only from one parent's copy (imprinting)

deletion on the **paternal** copy: Prader–Willi syndrome

deletion on the **maternal** copy: Angelman syndrome

2.4

the disease starts earlier or is more severe in each generation

the repeat grows longer each time it is passed on

3.1

(a) the premutation was unstable and expanded to a full mutation when passed from mother to son

(b) the **mother**

(c) **1/2**

(d) $\frac{1}{2} \times 0.8 = \mathbf{0.4}$

(e) methylation switches off the FMR1 gene, so its protein is missing

3.2

(a) all children of affected mothers can be affected, sons and daughters; the affected son's children are not affected

(b) cells hold a mix of normal and variant mtDNA (heteroplasmy); the share differs between people and tissues, and disease appears above a threshold

(c) almost none: sperm do not pass on mitochondria

3.3

(a) **full penetrance** (40 or more)

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(b) sperm come from many cell divisions, and the repeat is unstable at each division, so it tends to grow

(c) **juvenile** onset, in childhood or the teens

(d) uniparental disomy: with two maternal copies, there is no working paternal copy of the imprinted genes

(e) the effect depends on which parent the copy came from, not only on the genotype