Solutions

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

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

2.1

A: rapid **FISH or QF-PCR**, then a **karyotype**

B: **microarray** (or exome)

C: a **targeted test** for the family variant

2.2

CVS: **11–14 weeks**

amniocentesis: **from 15 weeks**

2.3

no DNA is gained or lost in a balanced translocation

a microarray only finds gains and losses (copy number)

2.4

the test is usually delayed until he is an adult

no treatment in childhood would change the outcome

he keeps the right to decide for himself whether to know

3.1

(a) **100**

(b) **99**

(c) 1% of 99 900 = **999**

(d) $\dfrac{99}{99+999} \approx$ **9%**

(e) most positive results are false, because the disease is rare compared with the false positive rate

3.2

(a) $\frac{1}{2}$

(b) testing relatives for the known family variant, moving out through the family as each new carrier is found

(c) yes: BRCA1 is on an autosome, so each child has a $\frac{1}{2}$ chance

(d) keep her confidence but discuss why it matters and help her tell them herself, for example with a family letter

3.3

(a) $\frac{1}{4}$

(b) **CVS**; it samples placental tissue (chorionic villi)

(c) the baby will be healthy, with mild anaemia at most

(d) the couple get balanced information about all options; they make the decision, and the counsellor does not push one choice

(e) IVF with **preimplantation genetic testing**: only embryos without two variant copies are put in the womb

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