## Solutions

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

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

**2.1**

47,XY,+21: **Down syndrome**

45,X: **Turner syndrome**

47,XXY: **Klinefelter syndrome**

**2.2**

**47,XX,+18**

**46,XY**

**2.3**

the two chromosomes of a pair do not separate

so gametes have 24 or 22 chromosomes

a 24-chromosome gamete joined by a normal 23 gives 47: a trisomy

**2.4**

chromosomes 14 and 21 are joined into one

no important genes are lost, so the genetic material is balanced

**3.1**

(a) $\dfrac{1/100}{1/1250} = \mathbf{12.5}$ times

(b) $10\,000/100 = \mathbf{100}$

(c) true positives $0.99 \times 100 = 99$; false positives $0.001 \times 9900 \approx 10$; $99/109 \approx \mathbf{91}\%$

(d) NIPT is a screening test: some positives are false, and the DNA partly comes from the placenta

**3.2**

(a) after fertilisation, in an early mitotic division, so only the cells from that line carry the extra 21

(b) many tissues have normal cells

(c) the parent looks normal but can make many trisomic gametes, so the risk for the next child is higher than the population risk

**3.3**

(a) all genetic material is present, just joined; at meiosis the joined chromosome may go with an extra 21, so an egg can carry too much



(b) an egg with a normal 14 and a normal 21 (or with the joined chromosome alone): healthy; an egg with the joined chromosome and a normal 21: Down syndrome

(c) the risk depends on which parent carries it; sperm carrying the unbalanced set are less often able to fertilise or survive

(d) a full karyotype; it shows a translocation, so the parents and their relatives can be tested and given their own risks



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