

1. Nondisjunction at division I of meiosis gives which gametes?
 - ☐ Four gametes with $n+1$
 - ☐ Four normal gametes
 - ☐ Two with $n+1$ and two with $n-1$; none normal
 - ☐ One $n+1$, one $n-1$ and two normal
2. Which pedigree most suggests autosomal recessive inheritance?
 - ☐ Only males affected, linked through healthy mothers
 - ☐ All daughters of an affected father affected
 - ☐ Two affected siblings with healthy parents who are first cousins
 - ☐ An affected person in every generation, including father to son
3. A pedigree shows affected mothers, affected children of both sexes and no transmission from any father. Which pattern fits?
 - ☐ Mitochondrial inheritance
 - ☐ X-linked recessive
 - ☐ Autosomal recessive
 - ☐ Autosomal dominant
4. A single-letter change turns a codon into a stop codon. What type of variant is it?
 - ☐ Missense
 - ☐ Frameshift
 - ☐ Nonsense
 - ☐ Synonymous
5. In a population at Hardy–Weinberg equilibrium, $q = 0.1$. What percentage of people are carriers (Aa)?
_____ %
6. Which test can show a balanced translocation?
 - ☐ A CFTR carrier panel
 - ☐ Sanger sequencing of one gene
 - ☐ Chromosomal microarray

7. The recurrence risk is the same after one affected child as after two.

☐ True ☐ False

8. An egg with 24 chromosomes is fertilised by a normal sperm. How many chromosomes does the embryo have?

_____ chromosomes

9. Two carriers have a child with a recessive condition. Their healthy daughter wants to know her chance of being a carrier. Give it as a percentage.

_____ %

10. All children of a mother with a mitochondrial DNA variant are affected to the same degree.

☐ True ☐ False

1. **Two with n+1 and two with n−1; none normal**

The whole pair goes to one side at division I, so both daughter cells are abnormal and so are all four gametes.

2. **Two affected siblings with healthy parents who are first cousins**

Recessive conditions cluster in one generation, with carrier parents, and consanguinity makes two carriers more likely.

3. **Mitochondrial inheritance**

Mitochondrial DNA comes only with the egg, so fathers pass nothing on.

4. **Nonsense**

A new stop codon cuts the protein short. A frameshift needs an insertion or deletion.

5. **18 %**

$p = 0.9$, so $2pq = 2 \times 0.9 \times 0.1 = 0.18 = 18\%$.

6. **Karyotype**

Nothing is gained or lost in a balanced translocation, so copy-number tests miss it; the karyotype sees the swapped pieces.

7. **False**

A second affected child shows the family sits further along the liability curve; neural-tube risk rises from about 3% to about 10%.

8. **47 chromosomes**

$24 + 23 = 47$, a trisomy.

9. **66.7 %**

Of AA, Aa, Aa and aa, she cannot be aa. Two of the three remaining boxes are carriers, so $2/3 \approx 67\%$.

10. **False**

Each egg samples a different fraction of mutant copies, so severity differs between children.