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

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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 × 0.9 × 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 ≈ 67%.

10. **False**

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

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