

University Medicine

Name: _____ Score: _____

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

☐ Karyotype

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

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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.