

Total: 23 marks

Objective

Compute carrier frequencies and couple risks from Hardy–Weinberg.

1 Key knowledge and worked examples

$$p^2 + 2pq + q^2 = 1; \text{ carriers} = 2pq. \text{ CF: } q^2 = 1/2500 \rightarrow q = 1/50 \rightarrow 2pq \approx 1/25 \checkmark.$$

Carrier $(1/25) \times$ screened-negative partner (residual $1/300) \times 1/4 \approx 1/30\,000 \checkmark$ – the carrier-screening multiplication.

2 Practice

2.1 A recessive disease at 1/10 000: compute q and the carrier rate. [3]

2.2 Tay-Sachs carrier frequency in Ashkenazi Jews (about 1/30): what is q approximately? [3]

2.3 Why does sickle heterozygosity stay common in malarial regions? [3]

3 Exam-style questions

3.1 CF carrier screening (fictional): the woman is a known carrier (1/1); her partner tests negative (panel detection ~95 %; population carrier rate 1/25).

Compute the partner's residual carrier risk after the negative test. Compute the affected-child risk. Explain why the answer is not zero. [7]

3.2 A population screens newborns for a recessive disease at $q^2 = 1/40\,000$.

Compute q and the carrier frequency. Compute the chance two unrelated carriers marry (from carrier frequencies). State how a founder effect changes the arithmetic. [7]