

6.5 Population genetics

Name: _____ Class: _____ Date: _____

Total: 23 marks

KEY WORDS Hardy-Weinberg equilibrium -

Objective

Compute carrier frequencies and couple risks from Hardy-Weinberg.

1 Key knowledge and worked examples

■ The equation

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

■ Worked example: the couple

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]

Solutions

2.1

- $q = \sqrt{1/10\ 000} = 1/100$; carriers $2pq \approx 1/50$ ✓.

2.2

- $q \approx 1/60$ ($2pq \approx 1/30 \rightarrow q \approx 1/60$).

2.3

- Heterozygote advantage against malaria (selection).

3.1

- Residual = $(1/25 \times 0.05) / (1/25 \times 0.05 + 24/25 \times 1) \approx 0.002 \approx 1/500$ ✓ (Bayes).
- Child risk = $1 \times 1/500 \times 1/4 = 1/2000$ ✓.
- Not zero: no panel detects every variant (CNVs, regulatory, rare missense).

3.2

- $q = \sqrt{1/40\ 000} = 1/200$; carriers $\approx 1/100$ ✓.
- Two carriers meeting: $(1/100)^2 = 1/10\ 000$; with the $1/4$ affected-child factor that IS the $1/40\ 000$ disease rate ✓.
- A founder effect concentrates the allele in a subgroup: carriers $1/30 \rightarrow q^2$ -based expectations rise orders of magnitude.