

# 6.2 Mendelian inheritance and pedigrees

Name: \_\_\_\_\_ Class: \_\_\_\_\_ Date: \_\_\_\_\_ **Total: 23 marks**

**KEY WORDS** pedigree

## Objective

Identify inheritance patterns from pedigrees and compute segregation risks.

### 1 Key knowledge and worked examples

■ **The signatures**

AD: vertical, father-to-son. AR: horizontal, consanguinity, 1/4. X-linked recessive: no male-to-male. X-linked dominant: affected father → ALL daughters, no sons ✓.

■ **Worked example: risk down the pedigree**

AD parent (Aa) × aa → each child 1/2; an affected child’s children again 1/2 ✓ – vertical 1/2s.

### 2 Practice

**2.1** Which pattern never shows male-to-male transmission? [3]

**2.2** Two carriers’ affected-child risk? [3]

**2.3** An affected father (X-linked dominant) has daughters – risk? [3]

### 3 Exam-style questions

**3.1** A woman (28, unaffected) has an affected parent with an adult-onset AD condition (fictional).

Compute her prior risk (1/2). Explain how being unaffected at 28 modifies it (onset distribution – Bayes). State what a predictive test would change. [7]

**3.2** Achondroplasia in a child of two average-height parents (fictional).

Explain the de-novo mutation logic (~80 %, paternal age). Give the sib recurrence (near population) and the child’s own offspring risk (1/2). State the germline-mosaicism caveat. [7]