

# 6.1 Chromosomes and karyotypes

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

**KEY WORDS** nondisjunction

## Objective

Compute nondisjunction gamete ratios and write karyotypes.

### 1 Key knowledge and worked examples

■ **The ratios**

Division-I nondisjunction → 2:2; division-II → 1:1:2 ✓. Trisomy 21 (~1/700, ~95 % meiosis I); 45,X the only viable monosomy.

■ **Worked example: the sentence**

47,XY,+21 = male trisomy 21; 45,X = Turner; 47,XXY = Klinefelter ✓ – count, sex, change.

## 2 Practice

**2.1** Give the gamete ratio for division-I nondisjunction. [3]

**2.2** Write the karyotype for a female with trisomy 21. [3]

**2.3** Why is 45,Y never seen? [3]

## 3 Exam-style questions

**3.1** A couple has one child with trisomy 21 (fictional); the mother is 30.

State the likely meiotic window. Give the recurrence risk (~1 %) and explain why it is above the population figure. State what maternal-age independent risk factors would change the counselling (translocation). [7]

**3.2** A child has mild trisomy-21 features; two cell lines 46/47 are found.

Explain the timing of the error (post-zygotic mitosis). Predict the phenotype severity relative to full trisomy. Explain why parental germline mosaicism still matters for recurrence. [7]