

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 $\rightarrow$ 2:2; division-II $\rightarrow$ 1:1:2 ✓. Trisomy 21 ($\sim 1/700$, $\sim 95\%$ 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]

Solutions

2.1

- $2(n+1) : 2(n) \checkmark$.

2.2

- $47,XX,+21 \checkmark$.

2.3

- The X carries essential genes; no X = non-viable.

3.1

- Meiosis I (~95 % of cases).
- Recurrence $\approx 1\%$ (above population for a young mother) - a small predisposition to nondisjunction persists.
- A Robertsonian translocation 21;14 would raise recurrence to 10–15 % (carrier parent) - karyotype the parents if family history or young maternal age.

3.2

- The nondisjunction occurred in an early mitotic division of the embryo.
- Milder: some tissue is euploid (46).
- If a parent has germline mosaicism, apparently de-novo cases can recur - counselling acknowledges it.