

Exercise walk-through

6.1

Chromosomes and karyotypes

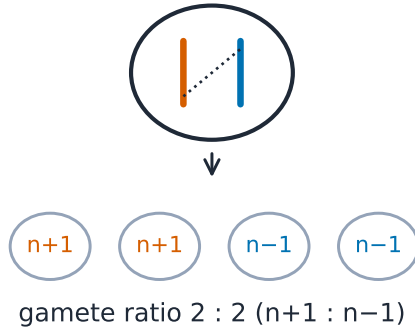
University Medicine

Chromosomes: the machine and its misfires

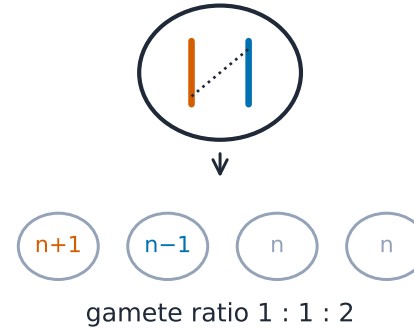
- 1 **Count** $46 = 23$ pairs; **meiosis** (1 duplication, 2 divisions) $\rightarrow$ 4 haploid gametes.
- 2 **Ratios Nondisjunction** I $\rightarrow$ 2:2 ($n+1 : n-1$); II $\rightarrow$ 1:1:2 ($n+1 : n-1 : n, n$).
- 3 **Survivors** Trisomy 21 $\sim 1/700$ ($\sim 95\%$ meiosis-I); 45,X the only viable monosomy; sex-chromosome aneuploidies buffered.

Chromosomes - the figure

nondisjunction I



nondisjunction II



Mis-segregation windows I vs II: gamete ratios 2:2 and 1:1:2.

Exercises 2.1-2.3

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]

Exercises 2.1-2.3

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

2.1

- $2(n+1) : 2(n)$ ✓.

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

2.2

- $47,XX,+21$ ✓.

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

2.3

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

Exercise 3.1

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

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

[7]

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3.1

- Meiosis I ($\sim 95\%$ 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.

Exercise 3.2

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.

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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.