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Q3(a)

What is meant by karyotype? How does its analysis help in diagnosis of the chromosomal aberrations in man?

UPSC Civil Services Mains 2024 · Anthropology GS 1 · 20 marks · Numerical and structural aberrations (disorders).

(a) Numerical and structural aberrations (disorders).

Facts & figures for this PYQ only.

Numbers move. Confirm the latest Budget, Economic Survey or census round. This sheet is for revision, not a substitute for the model answer.

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Standard complements

46,XX and 46,XY are conventional chromosome notations; notation describes chromosomes and should not be treated as a complete account of sex or development.

G-banding

Giemsa produces reproducible band patterns that identify homologues and localise large structural breakpoints.

Robertsonian translocation

Fusion involving acrocentric chromosomes may be balanced in a parent but create aneuploid gametes, including translocation Down syndrome.

Mosaicism

Two or more cell lines require analysis of multiple cells and sometimes multiple tissues; low-level mosaicism may be missed.

Resolution limit

Conventional karyotyping sees megabase-scale change, not most sequence variants. Microarray detects smaller imbalances but not all balanced rearrangements.

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