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Q6(c)

Discuss chromosomal aberrations in man illustrating with examples

UPSC Civil Services Mains 2015 · Anthropology GS 1 · 15 marks · Numerical and structural aberrations (disorders).

(a) Numerical and structural aberrations (disorders).

Core demand of the question

- Discuss chromosomal aberrations in man illustrating with examples
- Write about 200 words for 15 marks

Revision summary

Numerical aberrations are aneuploidies from **non-disjunction**.

Down, Edwards, Patau, Turner, and Klinefelter are type cases.

Structural changes include deletion, inversion, and translocation.

Familial Down's may follow a **Robertsonian** 14;21.

Microarray and FISH extend the old karyotype.

Introduction

Chromosomal aberrations are changes in chromosome number or structure. They are a major class of human genetic disease and of counselling cases.

Body

Numerical

- **Aneuploidy from non-disjunction: trisomy 21** (Down), **trisomy 18** (Edwards), **trisomy 13** (Patau).
- **Sex chromosomes: XO Turner, XXY Klinefelter, XYY, and XXX.**
- **Polyploidy** is rare in liveborn humans and usually lethal.

Structural

- **Deletion: cri-du-chat** (5p).
- **Duplication** and **inversion** may be balanced in a parent and unbalanced in a child.
- **Translocation: Robertsonian** 14;21 can cause familial Down's. Reciprocal translocations appear in recurrent miscarriage.
- **Isochromosome** and **ring** chromosomes are rarer teaching types.

Detection and meaning

- Karyotype, **FISH**, and now chromosomal microarray catch what light microscopy misses.
- Mosaics, as in some Turner and Down cases, blunt the textbook phenotype.

- Anthropology uses these examples to teach that 'type' is cytogenetic, not a racial grade.

Flow diagram

Chromosomal aberration -> Numerical aneuploidy
Chromosomal aberration -> Structural deletion translocation
Numerical aneuploidy -> Down Turner Klinefelter
Structural deletion translocation -> Cri-du-chat Robertsonian

Conclusion

Human chromosomal aberrations are numerical or structural. Down, Turner, Klinefelter, **cri-du-chat**, and **Robertsonian** translocation are the standard illustrations.

Facts & figures

Non-disjunction

The usual origin of free trisomy 21 and many sex-chromosome aneuploidies.

Turner 45,X

Short stature, ovarian dysgenesis; often mosaic.

Cri-du-chat

Deletion of 5p; cry and developmental delay.

Robertsonian

Fusion of two acrocentrics; a classic cause of translocation Down syndrome.