Q4(c)

"Chromosomal aberrations can play havoc with the human body and mind." Explain with suitable examples

UPSC Civil Services Mains 2021 · Anthropology GS 1 · 15 marks · Numerical and structural aberrations (disorders).

(a) Numerical and structural aberrations (disorders).

Core demand of the question

- "Chromosomal aberrations can play havoc with the human body and mind." Discuss with examples
- Write about 200 words for 15 marks

Revision summary

Aberrations are numerical or structural changes of chromosomes.

Down syndrome is trisomy 21 with body and cognitive effects.

Turner is 45,X; Klinefelter is 47,XXY; both alter gonads and sometimes learning.

Deletions and unbalanced translocations can be as severe as extra chromosomes.

Outcomes vary; counselling and care matter as much as the karyotype name.

Introduction

Chromosomal aberrations are changes in number or structure of chromosomes. They can alter development of body and brain because many genes move together.

Body

Numerical change

- **Down syndrome** (trisomy 21) produces distinctive facies, hypotonia, heart defects, and a range of intellectual disability. Maternal age raises risk of nondisjunction.
- **Turner syndrome** (45,X) affects females: short stature, ovarian dysgenesis, and sometimes spatial and social-cognitive difficulties. Intelligence is often near typical range, but the body is marked.
- **Klinefelter syndrome** (47,XXY) affects males: taller stature, hypogonadism, infertility, and a raised rate of language and learning problems.
- Other aneuploidies, such as trisomy 13 and 18, are often lethal early. Sex-chromosome aneuploidy is more survivable because of X inactivation.

Structural change

- Deletions, duplications, inversions, and translocations can be balanced in a parent and havoc in the child. Cri-du-chat (5p deletion) affects larynx and cognition.
- Reciprocal translocations explain some recurrent miscarriage and, if unbalanced, malformation syndromes.

Body and mind

- **The phrase in the question is clinical:** heart, gonad, stature, and language networks are all gene-rich. Havoc is variable, not a single fate.
- Counselling uses karyotype, now often microarray and NGS, plus pedigree. Anthropology adds stigma, schooling, and care, not a racial reading of the karyotype.

Flow diagram

Chromosomal aberration -> Aneuploidy

Chromosomal aberration -> Deletion translocation

Aneuploidy -> Down Turner Klinefelter

Down Turner Klinefelter -> Body and mind phenotype

Conclusion

Aneuploidy and rearrangements can disrupt growth, gonads, heart, and cognition. Down, Turner, and Klinefelter are the teaching examples. Severity is a spectrum, not a sentence.

Facts & figures

Down syndrome

Trisomy 21; most frequent viable autosomal aneuploidy; associated with congenital heart disease.

Turner syndrome

45,X; short stature and streak gonads; not a uniform intellectual profile.

Klinefelter syndrome

47,XXY; male hypogonadism and a raised rate of language delay.

Nondisjunction

Failure of chromosomes to separate in meiosis; the usual origin of trisomy.