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Q5(b)

Down's Syndrome.

UPSC Civil Services Mains 2015 · Anthropology GS 1 · 10 marks · Autosomal aberrations

(c) Autosomal aberrations-Down syndrome, Patau, Edward and Cri-du-chat syndromes.

Core demand of the question

- Write a note on Down's Syndrome
- Keep the note to a 10-mark length of about 150 words

Revision summary

Down's syndrome is usually trisomy 21 from meiotic **non-disjunction**.

Lejeune linked the phenotype to the extra chromosome.

Translocation and mosaic forms exist.

Maternal age raises risk of the common form.

Counselling uses karyotype; racial labels are obsolete.

Introduction

Down's syndrome is a chromosomal condition, usually trisomy 21. Anthropology meets it in human genetics, growth, and counselling.

Body**Cause**

- **John Langdon Down** described the phenotype. **Lejeune** showed an extra chromosome 21.
- Most cases are **non-disjunction** in meiosis, risk rising with maternal age.
- **Translocation** Down's can be inherited from a balanced carrier. **Mosaics** are milder in some tissues.

Features and care

- Hypotonia, a flat facial profile, single palmar crease, and a risk of heart defects are common.
- It is not a 'race type'. Older racial labels were a scientific error.
- Genetic counselling uses karyotype, and now **NIPT**, without treating the child as a social rank.

Flow diagram

Down syndrome -> Trisomy 21

Down syndrome -> Translocation

Down syndrome -> Mosaic

Trisomy 21 -> Meiotic nondisjunction

Conclusion

Down's syndrome is trisomy 21 in most cases. The anthropological task is accurate cytogenetics and humane counselling, not a racial cartoon.

Facts & figures

Lejeune

1959 demonstration of trisomy 21 as the usual cause.

Non-disjunction

Failure of chromosomes to separate in meiosis, the common origin.

Translocation

An inherited rearrangement that can recur in a family even in young mothers.

NIPT

Non-invasive prenatal testing of cell-free DNA, a screening step before diagnostic karyotype.

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