Q5(b)

Genetic imprinting in human diseases

UPSC Civil Services Mains 2022 · Anthropology GS 1 · 10 marks · Applications of Anthropology

12. Applications of Anthropology: Anthropology of sports, Nutritional anthropology, Anthropology in designing of defence and other equipments, Forensic Anthropology, Methods and principles of personal identification and reconstruction, Applied human genetics-Paternity diagnosis, genetic counselling and eugenics, DNA technology in diseases and medicine, serogenetics and cytogenetics in reproductive biology.

Core demand of the question

- Write a note on genetic imprinting in human diseases
- Keep the note to a 10-mark length of about 150 words

Revision summary

Imprinting silences one parental copy of a gene.

Haig tied the logic to parental conflict over growth.

Paternal versus maternal loss at 15q yields Prader-Willi or Angelman.

Beckwith-Wiedemann and Silver-Russell are growth-side examples.

Counselling must record which parent transmitted the region.

Introduction

Imprinting is parent-of-origin silencing of a gene. The same deletion can therefore cause two different diseases according to whether the chromosome came from the mother or the father.

Body

The mechanism

- Epigenetic marks, not a change of DNA letters, switch one parental copy off. **David Haig** linked this to parental conflict over foetal growth.
- Chromosome 15q is the teaching human region.

Disease pairs

- **Prader-Willi syndrome** follows loss of the paternal 15q contribution. **Angelman syndrome** follows loss of the maternal UBE3A copy.
- **Beckwith-Wiedemann** and **Silver-Russell** growth disorders involve imprinted loci on 11p.
- Hydatidiform moles and some cancers show imprinting errors. Pedigree counselling must ask which parent transmitted the chromosome.

Anthropological note

- This is non-Mendelian at the phenotype even though the deletion can be mapped. A simple dominant chart fails.

Flow diagram

Imprint -> Father copy on

Imprint -> Mother copy on

Father copy on -> Prader Willi

Mother copy on -> Angelman

Conclusion

Imprinting makes parent of origin a clinical fact. Prader-Willi and Angelman are the pair every counsellor names.

Facts & figures

Prader-Willi

Hypotonia, hyperphagia, and intellectual disability after missing paternal 15q material.

Angelman

Severe developmental disorder after missing maternal UBE3A function.

Haig

Kinship theory of imprinting: paternal genes favour foetal extraction, maternal genes restrain it.

11p

Imprinted growth cluster involved in Beckwith-Wiedemann and Silver-Russell syndromes.