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Q1(d)

Pedigree analysis in genetic counselling

UPSC Civil Services Mains 2022 · Anthropology GS 1 · 10 marks · Genetic imprints in human disease, genetic screening, genetic counseling, human DNA profiling, gene mapping and genome study.

(d) Genetic imprints in human disease, genetic screening, genetic counseling, human DNA profiling, gene mapping and genome study.

Core demand of the question

- Write a note on pedigree analysis in genetic counselling
- Keep the note to a 10-mark length of about 150 words

Revision summary

A pedigree uses **standard symbols** to show how a trait runs in a family.

Dominant, recessive, and X-linked patterns can be read from the chart.

Garrod linked such patterns to inborn errors of metabolism.

Counselling uses the chart for recurrence risk, especially with consanguinity.

Molecular tests refine, they do not replace, the family drawing.

Introduction

A pedigree is a family tree drawn with **standard symbols** so that a Mendelian pattern can be seen. Genetic counselling uses it before any laboratory test.

Body

How the chart works

- Squares are males, circles are females, shading marks the phenotype, and a double bar marks consanguinity.
- **Archibald Garrod** and later clinical geneticists used such charts for inborn errors. Anthropology still uses them for **PTC**, **albinism**, and **haemophilia**.
- Autosomal dominant traits appear every generation. Recessive traits skip. X-linked traits, as in European royal haemophilia, spare fathers-to-sons.

Counselling use

- The chart estimates recurrence risk for a couple, especially where cousin marriage is common, as in many Indian communities studied by **L. D. Sanghvi**.
- It cannot replace molecular tests for incomplete **penetrance**, but it still frames the interview and the ethics of disclosure.

Flow diagram

Pedigree -> Mendelian pattern
Mendelian pattern -> Recurrence risk
Recurrence risk -> Counselling

Conclusion

Pedigree analysis is the first Mendelian instrument of counselling. It turns a family story into a risk figure without pretending that genes are fate.

Facts & figures

Standard symbols

Square male, circle female, filled affected, arrow the proband, double line consanguinity.

Haemophilia A

X-linked recessive; a classic counselling pedigree in European royal houses.

Sanghvi

Mapped consanguinity and genetic load in Indian populations, which makes pedigree work locally urgent.

Penetrance

A dominant pedigree can hide non-penetrant carriers, so the chart is a hypothesis, not a diagnosis.

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