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Q4(c)

What is genetic counseling? Briefly discuss various steps involved in it.

UPSC Civil Services Mains 2024 · Anthropology GS 1 · 15 marks · Genetic imprints in human disease, genetic screening, genetic counseling, human DNA profiling, gene mapping and genome study.

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Facts & figures for this PYQ only.

Numbers move. Confirm the latest Budget, Economic Survey or census round. This sheet is for revision, not a substitute for the model answer.

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Sheldon Reed

Introduced the term genetic counselling in the mid-twentieth century and promoted an educational rather than eugenically directive relationship.

Three-generation pedigree

Documents affected status, biological relationships, reproductive history and consanguinity to frame inheritance and testing.

Screening versus diagnosis

A screening result changes probability; diagnostic testing seeks to establish the condition. Counselling must keep the distinction explicit.

Variant of uncertain significance

A VUS should not be presented as a confirmed disease cause or used alone for irreversible clinical decisions.

Cascade testing

After a familial pathogenic variant is established, targeted voluntary testing can identify at-risk relatives more efficiently.