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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Core demand of the question

- Define genetic counselling and its ethical orientation
- Describe the sequence from referral and diagnosis to follow-up
- Distinguish screening, risk estimation and diagnostic testing
- Use Indian population examples and discuss consent, stigma and limits

Revision summary

Genetic counselling helps families understand hereditary and psychosocial implications and choose voluntarily.

The sequence is referral, pedigree, diagnosis, risk assessment, consented testing, communication, options and follow-up.

Screening estimates risk; a positive screen is not automatically a diagnosis.

Risk explanations must include penetrance, variable expression and uncertainty.

Thalassaemia and sickle-cell programmes require confirmatory tests and non-stigmatising counselling.

Privacy, coercion, sex selection and unequal access are central ethical concerns.

Introduction

Genetic counselling is a communication process that helps individuals and families understand the medical, hereditary and psychosocial implications of genetic conditions and make informed decisions. **Sheldon Reed**, who introduced the term, stressed counselling rather than eugenic command; modern practice is non-directive and autonomy-centred.

Body

Steps in genetic counselling

1. **Referral and contract:** Clarify the client's concern-affected child, family history, recurrent pregnancy loss, carrier screening or prenatal finding-and the limits of confidentiality.

2. **History and pedigree:** Record a three-generation pedigree, diagnoses, ancestry where medically relevant, consanguinity, reproductive history and exposures. Social genealogy must not be assumed to equal biological parentage.

3. **Clinical diagnosis:** Review records and phenotype; involve relevant specialists. A precise diagnosis precedes meaningful recurrence risk.

4. **Risk assessment:** Identify Mendelian, chromosomal, mitochondrial or multifactorial inheritance. Calculate risk from the pedigree and population data while explaining penetrance, variable expression and uncertainty.

5. **Testing discussion and consent:** Explain no-test, screening and diagnostic options; possible results include positive, negative, uncertain and incidental findings. Choose karyotype, biochemical assay, targeted variant test, panel, microarray or sequencing according to the question.

6. **Communication:** Use plain language and absolute risk, check understanding and distinguish a carrier from an affected person. Provide a written summary.

7. **Options and decision support:** Discuss treatment, surveillance, cascade testing and reproductive choices such as natural conception, prenatal diagnosis, IVF with PGT, donor gametes or adoption where available. The counsellor informs but does not prescribe.

8. **Psychosocial care and follow-up:** Address guilt, family disclosure, grief, insurance or marriage stigma; revisit uncertain variants as knowledge changes and refer to support services.

Indian and anthropological relevance

Thalassaemia and sickle-cell programmes illustrate carrier screening followed by confirmatory testing and counselling. Consanguineous or endogamous populations may have elevated frequencies of particular recessives, but community labels are poor substitutes for testing. Prenatal screening such as NIPT estimates probability; CVS or amniocentesis can provide diagnostic material.

Counselling must guard against coerced testing, sex selection, caste/tribal stigmatisation, breach of genomic privacy and pressure to terminate pregnancy. Limited laboratories and uncertain variants can make "choice" unequal. Family benefit must be balanced with the client's confidentiality.

Flow diagram

Referral -> History and three-generation pedigree

History and three-generation pedigree -> Clinical diagnosis

Clinical diagnosis -> Risk assessment

Risk assessment -> Testing options and consent

Testing options and consent -> Communicate results

Communicate results -> Support voluntary choices

Support voluntary choices -> Follow-up and family care

Conclusion

Good genetic counselling is not a laboratory result delivered with advice. It is a cycle of verified diagnosis, transparent risk, voluntary testing, supported choice and follow-up. Its anthropological quality lies in joining inheritance to kinship, stigma, language and unequal access without reviving eugenics.

Facts & figures

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.