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Q7(b)

Explain the significance of screening and counseling for genetic disorders.

UPSC Civil Services Mains 2016 · Anthropology GS 1 · 15 marks · Numerical and structural aberrations (disorders).

(a) Numerical and structural aberrations (disorders).

Core demand of the question

- Explain the significance of screening and counseling for genetic disorders
- Write about 200 words for 15 marks

Revision summary

Screening searches a defined group for genetic risk.

Newborn, cascade, and prenatal programmes are the main types.

Counselling explains risk without directing the choice.

Significance is prevention, early treatment, and informed reproduction.

Stigma and eugenics are the failures to avoid.

Introduction

Screening finds people at raised risk. Counselling turns that finding into an informed, voluntary decision. Together they are the public face of medical genetics.

Body**Screening**

- **Population screening targets common, treatable, or preventable conditions: newborn TSH, sickle, thalassaemia** in high-prevalence Indian belts.
- **Cascade screening** starts from a proband and tests relatives, as in **familial cancers** or translocation Down's.
- **Prenatal tools: NIPT**, ultrasound, and diagnostic CVS or amnio after consent.
- **Significance:** early diet, transfusion programmes, and fewer surprise stillbirths. Harm: false positives and stigma if community work is crude.

Counselling

- **Non-directive** explanation of recurrence risk, using pedigrees first, as **Garrod** to modern clinics.
- **Sanghvi**-type consanguinity contexts need extra care, not blame.
- **Significance:** autonomy, timing of marriage information, and support after a diagnosis.
- **Ethics:** confidentiality, the right not to know, and no state eugenics.

Anthropology's warning

- Screening without counselling is a lab. Counselling without accessible care is empty talk.

Flow diagram

Screening -> Find high risk
Find high risk -> Counselling
Counselling -> Voluntary decision care

Conclusion

Screening detects risk early. Counselling protects choice and meaning. Their significance in India is thalassaemia, newborn, and prenatal programmes done with consent, not coercion.

Facts & figures

Thalassaemia screening

Carrier testing in high-prevalence Indian states, with prenatal options.

Newborn TSH

Congenital hypothyroidism is treatable if caught early.

Non-directive

The ethical standard of genetic counselling after the eugenics era.

NIPT

Screening, not diagnosis; positives need confirmation.

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