Course- B.Sc. (Botany Honours), Part -3

Paper-VI (Group-B), Molecular Biology

Topic- Genetic Counseling.

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Genetic Counseling

Genetic counselling is a communication process, which aims to help individuals, couples and families understand and adapt to the medical, psychological, familial and reproductive implications of the genetic contribution to specific health conditions.

a. Prospective Genetic Counselling:

Identification of Heterozygous individuals and explaining to them about risk of it to their children if they marry another homozygous, e.g., Sickle cell disease and Thalassaemia.

b. Retrospective Genetic Counselling:

Counselling to be conducted in connection with congenital mental retardation, psychiatric illness and inborn errors.

Methods which could be suggested under retrospective genetic counseling are:

1. Contraception

2. Pregnancy termination

3. Sterilisation

This process integrates the following:

- Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence.
- Education about the natural history of the condition, inheritance pattern, testing, management, prevention, support resources and research.
- Counselling to promote informed choices in view of risk assessment, family goals, ethical and religious values.
- Support to encourage the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder

Reasons for Genetic Counseling

Based on your personal and family health history, your doctor can refer you for genetic counseling. There are different stages in your life when you might be referred for genetic counseling:

- **Planning for Pregnancy**: Genetic counseling before you become pregnant can address concerns about factors that might affect your baby during infancy or childhood or your ability to become pregnant, including
    - Genetic conditions that run in your family or your partner's family
    - History of infertility, multiple miscarriages, or stillbirth
    - Previous pregnancy or child affected by a birth defect or genetic condition
    - Assisted Reproductive Technology (ART) options

- **During Pregnancy**: Genetic counseling while you are pregnant can address certain tests that may be done during your pregnancy, any detected problems, or conditions that might affect your baby during infancy or childhood, including
    - History of infertility, multiple miscarriages, or stillbirth
    - Previous pregnancy or child affected by a birth defect or genetic condition
    - Abnormal test results, such as a blood test, ultrasound, Chorionic Villus Sampling (CVS), or amniocentesis
    - Maternal infections, such as Cytomegalovirus (CMV), and other exposures such as medicines, drugs, chemicals, and x-rays
    - Genetic screening that is recommended for all pregnant women, which includes cystic fibrosisexternal icon, sickle cell disease, and any conditions that run in your family or your partner's family

- **Caring for Children:** Genetic counseling can address concerns if your child is showing signs and symptoms of a disorder that might be genetic, including
    - Abnormal newborn screening results
    - Birth defects
    - Intellectual disability or developmental disabilities
    - Autism spectrum disorders (ASD)

- Vision or hearing problems

- **Managing Your Health**: Genetic counseling for adults includes specialty areas such as cardiovascular, psychiatric, and cancer. Genetic counseling can be helpful if you have symptoms of a condition or have a family history of a condition that makes you more likely to be affected with that condition, including
  - Hereditary breast and ovarian cancer (HBOC) syndrome
  - Lynch syndrome (hereditary colorectal and other cancers)
  - Familial hypercholesterolemia
  - Muscular dystrophy and other muscle diseases
  - Inherited movement disorders such as Huntington's diseaseexternal icon
  - Inherited blood disorders such as sickle cell disease