

PREVENTIVE MEASURES FOR CONTROL OF HEREDITARY DISEASES

1. Eugenics (GENETIC MANIPULATION)

- It is the study of Hereditary Improvement of Human Race by Controlled Selective Breeding
- Improvement of Human Hereditary Traits is done through various forms of Interventions
- Aims - to improve the Genetic Composition of Human Population
- I. Negative Eugenics > People who are suffering from Serious Hereditary Diseases are sterilized from producing

children

- Lowering Fertility of Genetically Disadvantaged through Abortions, Sterilization & other methods

- II. Positive Eugenics > Encouraging the carriers of Desirable Genotypes for parenthood

2. Euthenics (ENVIRONMENTAL MANIPULATION)

- Improvement / Manipulation in an Environment, which enables the Genes to Express Themselves readily

- Adapting Environment to his Gene^{*}. Deals with the human improvement through altering External Factors such as

* Controllable Environment • Prevention and removal of Contagious Disease & Parasites

- Education regarding Home Economics, Sanitation & Housing

- Dysgenics - Environment is changed that lead to Deterioration in expression of Genes i.e., growth of population

3. Genetic Counseling

- Genetic counseling is the process of investigating Individuals & Families affected by or at risk of Genetic Disorders, to help them understand and adapt to the Medical, Psychological & Familial implications of Genetic contributions to

Disease.

I. Prospective Genetic Counseling • Identifying Heterozygous Individuals & explaining to them the risk of having affected Children if they marry another Heterozygote

- Done in - Sickle Cell Anemia, Thalassemia II.

Retrospective Genetic Counseling • Most of the Genetic Counseling at present are Retrospective i.e., Hereditary Disorder has already occurred within the family

4. Prevention of Consanguineous Marriages

- When Blood Relatives marry each other, there is an increased risk in offspring of traits controlled by Recessive Genes May

cause

- Albinism, Alkaptonuria, Phenylketonuria, Premature Death, Congenital Malformation

- Lowering of Consanguineous Marriages will improve Community Health.

5 Prevention of Late Marriages - Down's Syndrome/Mongolism is more frequent in Children born of Elderly Mothers

6. Protection against Mutagens - Mutagens such as - Ionizing Radiations & Chemical Mutagens Rh Hemolytic Disease in the Newborn is prevented by Immunization with Anti - D Globulin

- 7. Detection of Carriers - Female Carriers of Duchenne Muscular Dystrophy are detected by elevated Serum Creatine

Kinase

8. Prenatal Diagnosis (Amniocentesis)

- Examination of sample of Amniotic Fluid for prenatal diagnosis of Chromosomal Anomalies • Culture & Karyotyping of Fetal Cells from Amniotic Fluid - for diagnosis of fetal anomalies. • Biochemical Analysis of Amniotic Fluid - for diagnosis of Metabolic Disorders.

Amniocentesis can be done as early as 14th week of Pregnancy for prenatal diagnosis of • Down's Syndrome • Inborn Error Metabolism - a Thalassemia, Neural tube defects, Tay Sach's disease, Galactosemia

- Indicators of Prenatal Diagnosis • Advanced Maternal Age > Amniocentesis (14 - 16 weeks) Chorionic Villous Sampling (11 - 13 weeks • Biochemical Disorders > Protein Assay • Congenital Anomaly >

sonography, TIFFA • Screening of Neural Tube Defects & Trisomy > Maternal Serum Alpha Feto Protein & Chorionic Gonadotropin •

9. Screening of Newborns. • Sick Cell Disease - Hemoglobin Electrophoresis using Guthrie Blood Spot • Cystic Fibrosis - Immunoreactive Trypsin in Guthrie Blood Spot

• 10. Recognizing Pre - Clinical Cases. • Phenylketonuria - Phenylalanine Tolerance Test • Diabetes - Urine Examination for Sugar • Gout - Serum Uric Acid • Punnett Square Test - to find Genotype of Offspring • Founder Effect - describes distribution of disease on the basis of Genetics