CONGENITAL MALFORMATION

Congenital malformations may be regarded as one ear. form of reproductive failure. The favorable environmental and genetic factors result in normal reproduction and the unfavorable factors lead to sterility, abortion, stillbirth, preterm births or neonatal death.

Congenital anomalies represent a relative reproductive success when compared to sterility or abortion.

Prevention of congenital defects must be achieved by special attention to environmental factors rather than by attempting to improve heredity towards successful reproductive outcome.

Definitions

The terms congenital anomaly and congenital malformation are used interchangeably, but they are different in meaning.

A WHO document described the terms, the congenital anomaly being used to include all biochemical, structural and functional disorders present at birth and the congenital malformation should be confined to structural defects only, present at birth.

The birth defects or congenital defects may be inherited genetically or acquired during gestation or inflicted during parturition.

Incidence

The global incidence of congenital disorders is estimated about 30 to 70 per 1000 live birth (1989). Approximately half of these infants have fatal outcome or lifelong chronic diseases. Actual numbers vary widely between countries.

The most common congenital defects are congenital heart diseases and central nervous system malformations.

In India, the incidence of congenital defects is about 2,5 to 4% among children.

Congenital anomalies are also considered as one of the important cause of physical and mental handicapped conditions among survivors.

Most common type of birth defect is CNS abnormalities, approximately 22% of all defects.

Risk Factors

Some factors are considered to be significantly associated with incidence of congenital anomalies.

- Advanced maternal age, e.g. elderly mother has risk of birth of baby with Down's syndrome or other congenital anomalies.
- Consanguinity, i.e. baby born out of consanguineous marriages (among blood relations), are at risk of congenital defects like mental retardation.
- Maternal malnutrition especially folic acid deficiency can lead to CNS defects and iodine deficiency can lead to mental retardation or other congenital anomalies.

ETIOLOGY OF CONGENITAL ANOMALIES

The causes of most of the congenital anomalies are not fully understood. Majority of the causes are unknown or due to complex interaction between genetic and environmental

Common Congenital Malformations

1. Skeletal defects

A limb anomaly is called dysmelia.

It includes

- Amelia (absence of arm or leg)
- Ectrodactyly (absence of one or more central digits of hand or foot)\
- Phocomelia (hand or feet attached to abbreviated arms and legs)
- Polymelia (more than normal number of limbs)
- Polydactyly (having more digits or fingers in hand or feet)
- Syndactyly (two or more digits are fused together
- Achondroplasia (disorder of bone growth leading to dwarfism)
- Aplasia (absence of an organ or tissue).

2. Central nervous system defects

Congenital anomalies of nervous system include

- Neural tube defects like spina bifida (failure of formation of bony arch around spinal cord) who includes meningocele (failure of formation of bony arch around spinal cord without

defective development of spinal cord) and myelomeningocele (failure of formation of bony arch around spinal cord with protrusion of cord through the defect)

- Encephalocele (protrusion of brain and meninges through opening in the skull)
- Anencephaly (absence of large part of brain and skull) Hydrocephalus (enlargement of skull due to accumulation of CSF) Microcephaly (abnormally small head and brain)
- Megalencephaly (abnormally large brain)
- Amold-chiari malformation
- Dandy Walker malformation

3**. Gastrointestinal defects**

It includes

- Cleft lip (failure of fusion of maxillary processes with nose elevation on frontal prominence defect
- cleft palate (failure of fusion of secondary palate with each other and with primary palate)
- esophageal atresia (failure of esophagus to form a continuous passage from pharynx to stomach), pylonic stenosis (narrowing of pylorus),
- anorectal malformation (malformed structure of anus and the rectum),
- exomphalos (herniation of abdominal organs into umbilical cord with evisceration into the sac) and megacolon (enlarged colon due to absence of ganglionic cells).

4. Respiratory system defects

It includes

- Choanal atresia (back of nasal passage is blocked)
- TEF or tracheoesophageal fistula (abnormal connection between trachea and esophagus)
- Diaphragmatic hernia (protrusion of abdominal organs through a defect in diaphragm into the thoracic cavity).

5. Cardiovascular defects

Neonates may be born with cyanotic and acyanotic cardiac defects like

- Atrial septal defect (abnormal communication between right and left atrium)

- Ventricular sepal defect (abnormal communication between right and left ventricle)
- Aortic stenosis (obstructive lesion in aorta that interferes with outflow of blood from the left ventricle)
- Pulmonary stenosis (obstructive lesion in pulmonary artery that interferes with outflow of blood from right ventricle)
- Tetralogy of fallot (It includes 4 defects ventricular sepal defect, aortic over riding, pulmonary
- stenosis and right ventricular hypertrophy)
- Dextroposition (heart on right side).

6. **Genitourinary system defects**

Congenital defects of genitourinary system are

- Atrophy of bladder (failure of abdominal wall to close resulting in exposed urinary bladder)
- Hypospadias (urethral meatus on ventral penile surface)
- Epispadias (urethral meatus on dorsal penile surface)
- Phimosis (foreskin is narrowed and cannot be retracted over the glans)
- Wilm's tumor (malignant tumor of kidneys)
- Polycystic kidneys (presence of multiple cysts in the kidneys)

7. Blood disorders

Congenital blood disorders are:

- Thalassemia (defective hemoglobin production)
- Hemophilia (deficiency of clotting factors)
- Sickle cell anemia (RBCs are sickle shaped so breakdown is rapid, resulting in anemia)
- Hereditary spherocytosis (disorder of RBC membrane that leads to spherical RBCs which br prematurely)

8. **Metabolic disorders**

Congenital metabolic disorders include

- Cystic fibrosis (thick sticky mucous in lungs, digestive tract and pancreas, due to a defective gere
- Phenylketonuria finability to metabolise the amino acid phenylalaninei
- Galactosemia (inability of the body to metabolise galactose)
- G-6PD deficiency (deficiency of enzyme G6PD results in hemolysis of RBCS)
- Glycogen storage disease (defect in processing or glycogen synthesis or breakdown of glycogen muscles and liver
- Mucopolysaccharidosis (absence or malfunctioning of lysosome enzyme needed for carbohydrat Tay-Sach's disease (disease of nervous system characterized by deafness, decreased eye contac metabolism blindness and decreased muscle tone) etc.

9**. Endocrinal abnormalities**

Congenital endocrinal abnormalities include

- Congenital dwarfism ideficiency of growth hormone)
- Cushing's syndrome (elevated cortisol level)Cretinism (congenital hypothyroidism Goiter (diffuse or nodular enlargement of thyroid)
- Juvenile diabetes mellitus (autoimmune destruction of insulin producing beta cells resulting hyperglycemia)

10 Chromosomal anomalies

Congenital chromosomal anomalies include:

- Down's syndrome (trisomy 21)
- Patau's syndrome (trisomy 13)
- Edward's syndrome (trisomy 18) Turner's syndrome (x,0)
- Klinefelter's syndrome (XXY, XXXY)
- Cri-du-chat syndrome (partial deletion of short arm of chromosome 5)
- Prader-Willt syndrome (deletion on long arm of Paternal chromosome 15)

- Noonan syndrome (abnormality on long arm of chromosome 12)
- Rett's syndrome (X-linked dominant abnormality),

Diagnostic evaluation

It is now possible to diagnose congenital abnormalities in fetus, prior to the baby's birth. Various prenat and post natal investigations are required to diagnose congenital malformations. Prenatal diagnosis of genetic disorder has been possible because of great advances in techniques of obtaining fetal tissue well as development in cytogenetics, blochemical techniques and recombinant DNA technology.

Prenatal Diagnosis

Prenatal diagnosis can be made a using a variety of procedure like

1 **Chorionic Villus sampling:** It is a technique for obtaining fetal cells from placenta for prenatal diagnosis. The procedure is usually done transabdominally after 10 weeks of pregnancy 2 **Amniocentesis.** It is withdrawal of amniotic fluid from amniotic sac surrounding the fetes any Amniocentesis done between 11-15 weeks is useful for cytogenetic study Early in pregnancy is used for diagnosts of Down's syndrome, Trisomy 13. Trisomy 18, Fragile X ndrome and rare metabolic disorders. Syndrome Cordocentesis Withdrawing blood from umbilical vein under ultrasound guidance is called docentesis. It cordocentesis Mers in in prenatal diagnosis of genetic disorder and understanding fetal hology, development and metabolism physiology. Evelopment blood sampling can be done.

4)**Fetoscopy:** A fine endoscope is inserted into the uterus for direct visualization of fetus and fetal

5. **Embryoscopy** This is an experimental technique used in first trimester of pregnancy. A rigid endoscope is inserted through the cervix into the space between amnion and chorion; to visualize the embryo and diagnose any structural malformations

6. Ultrasonography. It is done to visualize the fetus in mother's womb. Blood tests: An elevated level of alpha fetoprotein in maternal blood indicates neural tube defect in the baby.

7. Radiography It is used to diagnose anencephaly, hydrocephaly and skeletal defects Fetal X-ray can be done in last trimester of pregnancy.

Postnatal Diagnosis

Postnatal diagnosis of congenital malformations in baby can be made by

1. Detail history of mother and family

2. Physical examination of the newborn

3. Blood tests

4 Cytogenetic study

5. Blochemical assay

6. Radiography

7 Ultrasonography

PREVENTION OF CONGENITAL ANOMALIES

Prevention of congenital anomalies can be possible by health promotional measures, specific protection, early diagnosis and specific management.

Prevention of these abnormalities is more approachable than the curative management. According to Penrose (1961), major advances in the prevention of malformations must be achieved by attention to environmental factors rather than by attempting to improve heredity.

Environmental modification is more within reach than genetic control.

Preventive Measures

The preventive measures should include the following aspects:

- Genetic counseling is the true preventive measure of congenital anomalies.
- Reducing and discouraging consanguineous marriages.
- When blood relatives marry each other there is an increased risk in the offspring of traits controlled by recessive genes and those determined by polygenes.
- Avoiding late marriage of females and avoidance of pregnancy beyond the age of 35 years.
- Promotion of health of girl child and prepregnant health status of the females by prevention of malnutrition, anemia, folic acid deficiency, iodine deficiency, etc.
- Encouraging the immunization of all girl children by MMR (Mumps, Measles, and Rubella).
- Increasing attention to the protection of individuals and whole communities against mutagens such as X-ray and other ionizing radiations and also for chemical mutagens (drugs, alcohol).

- Immunization by anti-D immunoglobulin to the 'Rh negative' mothers after abortion or first childbirth to prevent Rh-hemolytic disease of the newborn which is a genetically determined immunological disorder.
- Elimination of active and passive smoking of tobacco by mothers.
- Avoidance of drug intake without consulting the first trimester of pregnancy. physician in
- Prevention of intrauterine infections and promotion of sexual hygiene along with general hygienic measures.
- Efficient antenatal care especially removal of teratogens, periconceptional supplementation of folic acid, prevention of maternal malnutrition by adequate diet, prenatal diagnosis of suspected genetic disorders and maternal diseases, appropriate treatment of maternal diseases and infections, prevention of fetal hypoxia, etc.
- Promotion of therapeutic abortion of abnormal fetus and fetus with gross congenital anomalies, after prenatal diagnosis.
- Discouraging reproduction after birth of a baby with congenital anomalies, without genetic counseling. The risk of malformations in subsequent pregnancies is increased by 10 times.
- Increasing public awareness about the risk factors and etiological factors of congenital anomalies and their preventive measures.
- Promotion of detection of genetic carriers, e.g. both partners should arrange to test for thalassemia carrier before marriage.
- Reducing the frequency of hereditary disease and disability in the community to as low as possible by negative eugenics.
- The persons who are suffering from serious hereditary disease are debarred from producing children or sterilized; there should be no serious objection to marriage.
- Positive eugenics should be promoted to improve the genetic composition of the population by encouraging the carriers of desirable genotypes to assume the burden of parenthood.

NURSING RESPONSIBILITIES TOWARDS CONGENITAL ANOMALIES

- As a member of the team, the nurse has the responsibility of being liaison among the family, referring physician or agency and the medical-genetics team.

- The nurse is usually the first person with whom the family has contact.
- The nurse can help the family by assuring them and explaining about importance of accurate diagnosis for appropriate management.
- Nursing personnel can provide following interventions for the management of the affected child.
- Collection of details history, especially history of prenatal, natal and postnatal period along with history of family illness.
- Preparation of pedigree chart by interview and home visit. Identification of present problems, its nature and severity, for necessary interventions.
- Participation in diagnostic investigations, treatment, follow-up and research project.
- Provide necessary information to the parents and family members.
- Motivate the family members for genetic counseling and referring to the genetic clinic.
- Participating in genetic counseling process with special training, personal experience, knowledge and competency.
    - Provide emotional support and answer questions asked by the counselee.
- Guide the family for rehabilitation of the child and for available social and economical support through social welfare agencies.
- Promote public awareness about the prevention of congenital anomalies by individual or group health education or by mass media information.