





CONGENITAL ANOMALIES

- 
- Congenital abnormalities are those defects and diseases which are substantially determined before or during birth and recognizable in early life.
 - Some disorders are detected at birth, some are obvious in early life and some may become apparent until much later in life.



Definition


- All biochemical, structural and functional disorders present at birth.
 - Birth defects are structural or functional abnormalities present at birth that cause physical or mental disability. Some may be fatal.
- 

Incidence

1. 2-3 per 100 children are born with birth defects around the world
2. 2.5/1000 babies are born with Neural Tube Defects
3. 2.7/1000 babies are born with Club foot, Gastrointestinal tract abnormalities and defective diaphragm
4. 1.9/1000 babies are born with Cleft lip, Cleft palate and Congenital Heart Defects
5. Birth defects incidence in India has not reduced over the last 8 years




causes

- Genetic problems caused when one or more genes doesn't work properly or part of a gene is missing
 - Problems with chromosomes, such as having an extra chromosome or missing part of a chromosome
 - Environmental factors that a woman is exposed to during pregnancy, such as rubella or German measles while pregnant, or using drugs or alcohol during pregnancy.
- 



Risk factors

- Advanced maternal age
 - Consanguinity
 - Maternal malnutrition
- 

Common congenital anomalies

1. Central nervous system:

- Anencephaly
- Spina bifida
- Hydrocephalus
- Microcephaly , Macrocephaly
- Syringomyelia, Diastematomyelia
- Porencephaly , schizencephaly
- Agenesis of cranial nerves

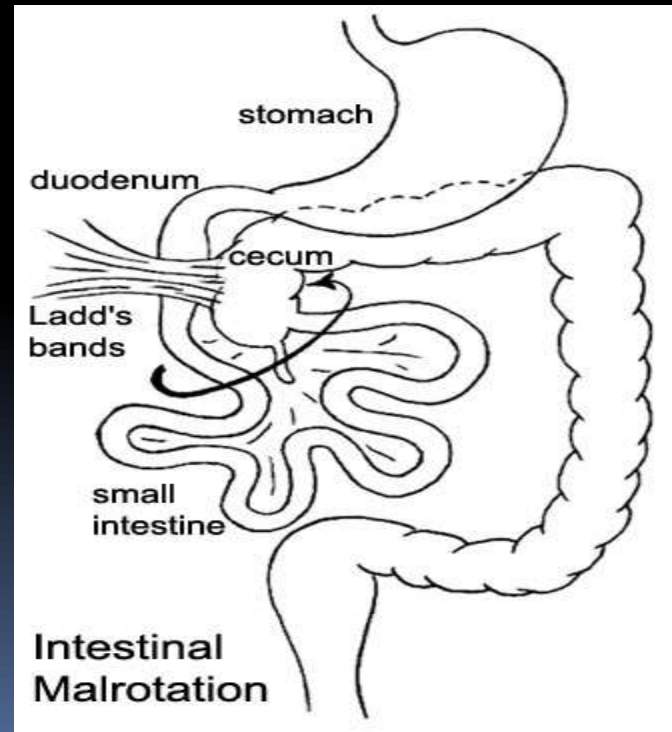
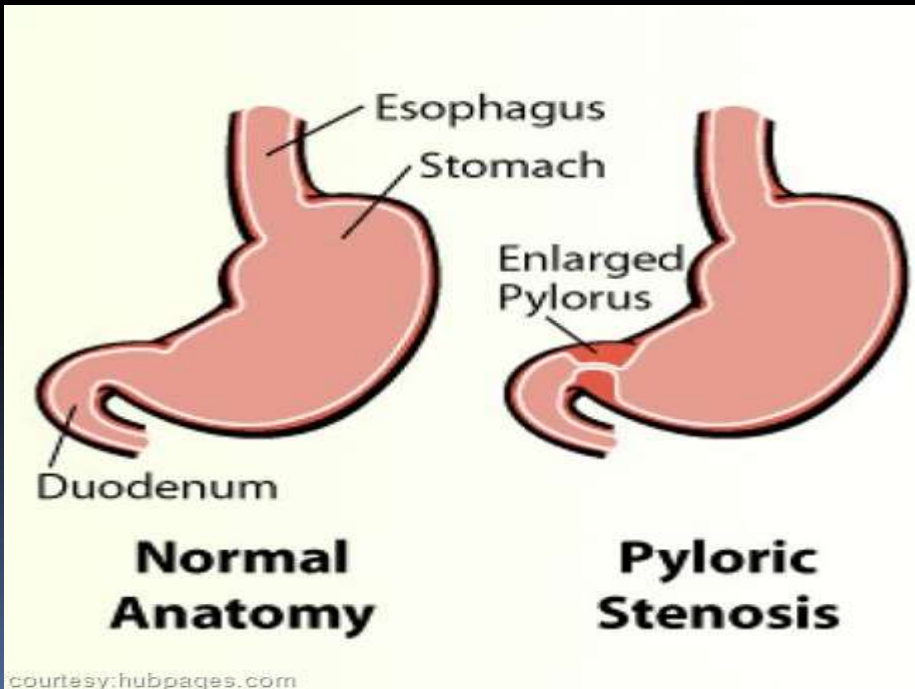
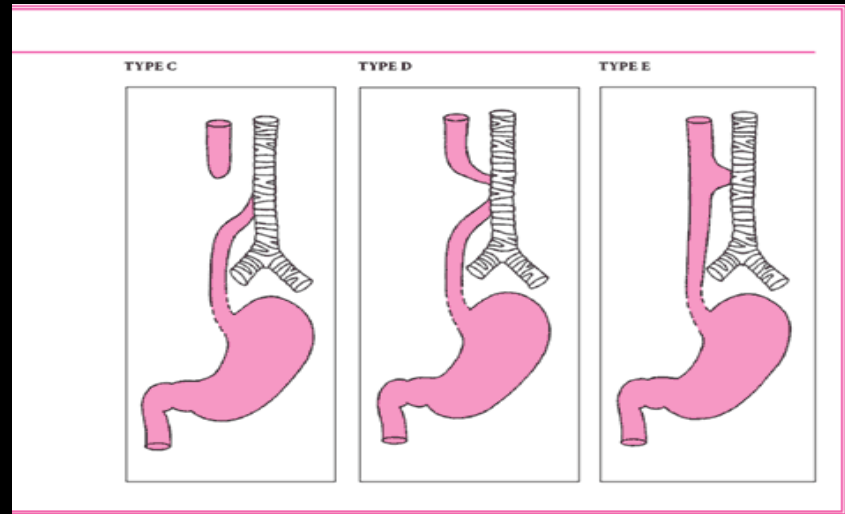
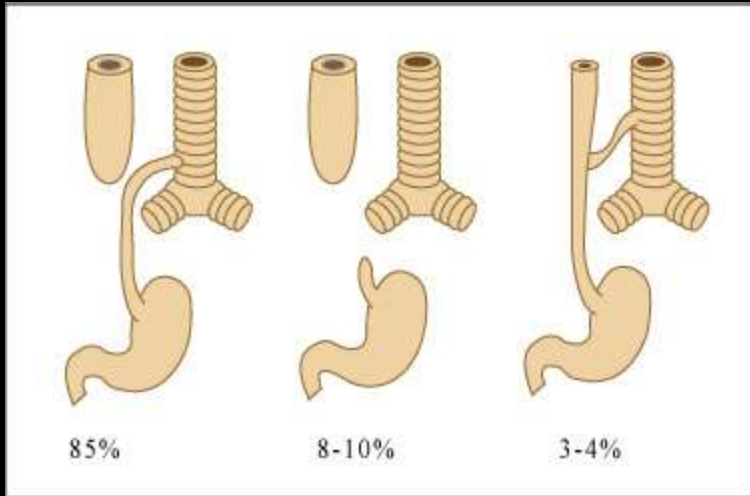


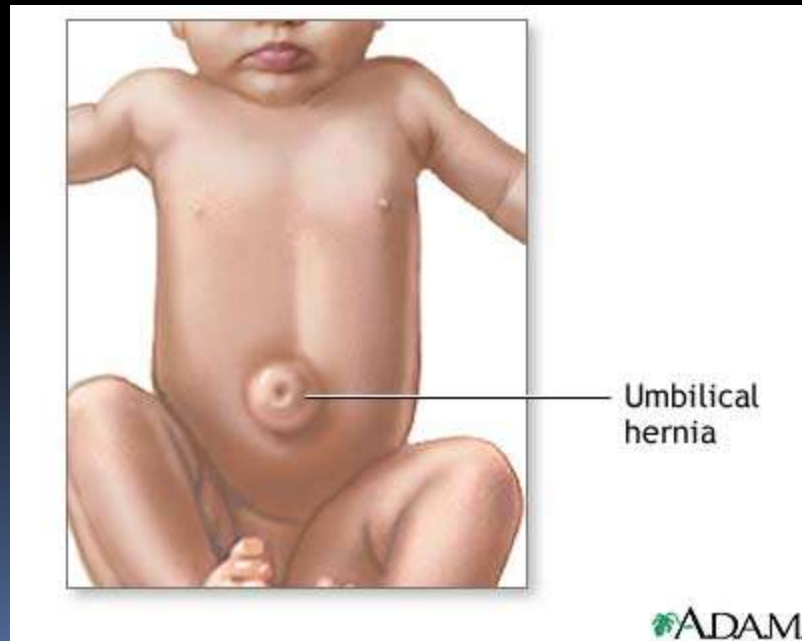
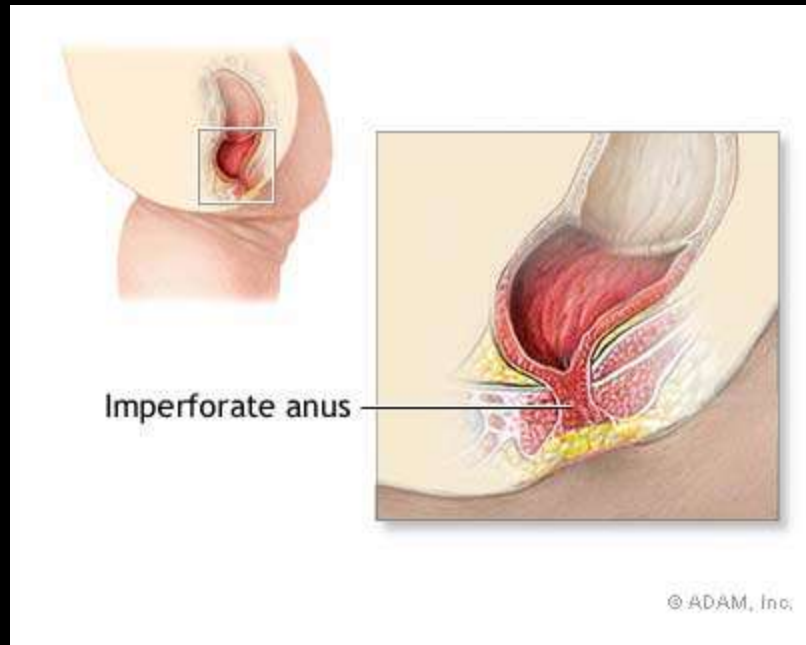
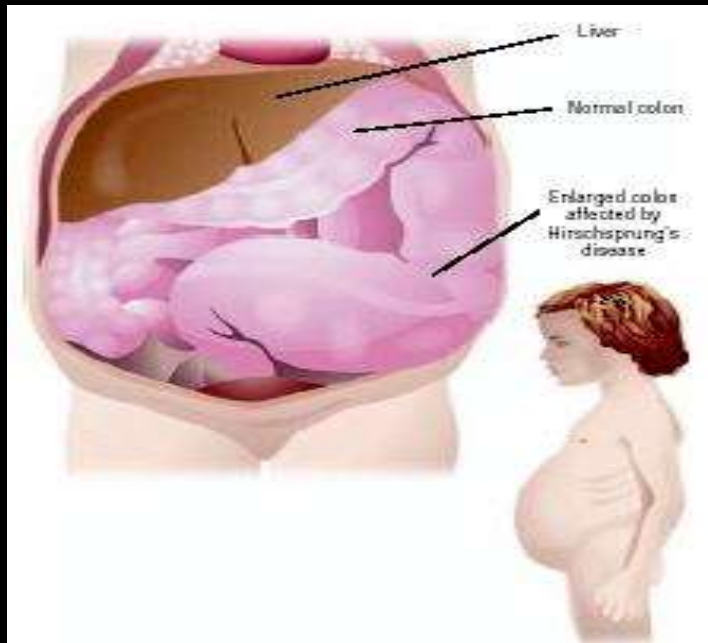
2. Congenital heart diseases:

- VSD
- ASD
- TGA
- Tricuspid atresia
- Tricus arteriosus
- Tetralogy of fallot
- Aortic stenosis
- Pulmonic stenosis
- Aortic or pulmonary artery dilatation
- Mitral or aortic regurgitation
- Ebstein's anomaly
- Dextrocardia

3. GI system abnormalities

- Tracheo esophageal fistula
- Oesophageal atresia
- Congenital pyloric stenosis
- Meconium ileus
- Malrotation of gut
- Congenital megacolon
- Ano rectal malformations
- Exomphalos
- Umbilical hernia
- Diaphragmatic hernia
- Femoral hernia
- Congenital intestinal obstruction
- Gastroschisis







Gastroschisis




Omphalocele

Copyright the Lucina Foundation, all rights reserved.



4. Respiratory system abnormalities

- Choanal atresia
 - Tracheoesophageal fistula
 - Congenital atelectasis
 - Pulmonary agenesis
 - Congenital stridor
 - Congenital cyanosis
- 

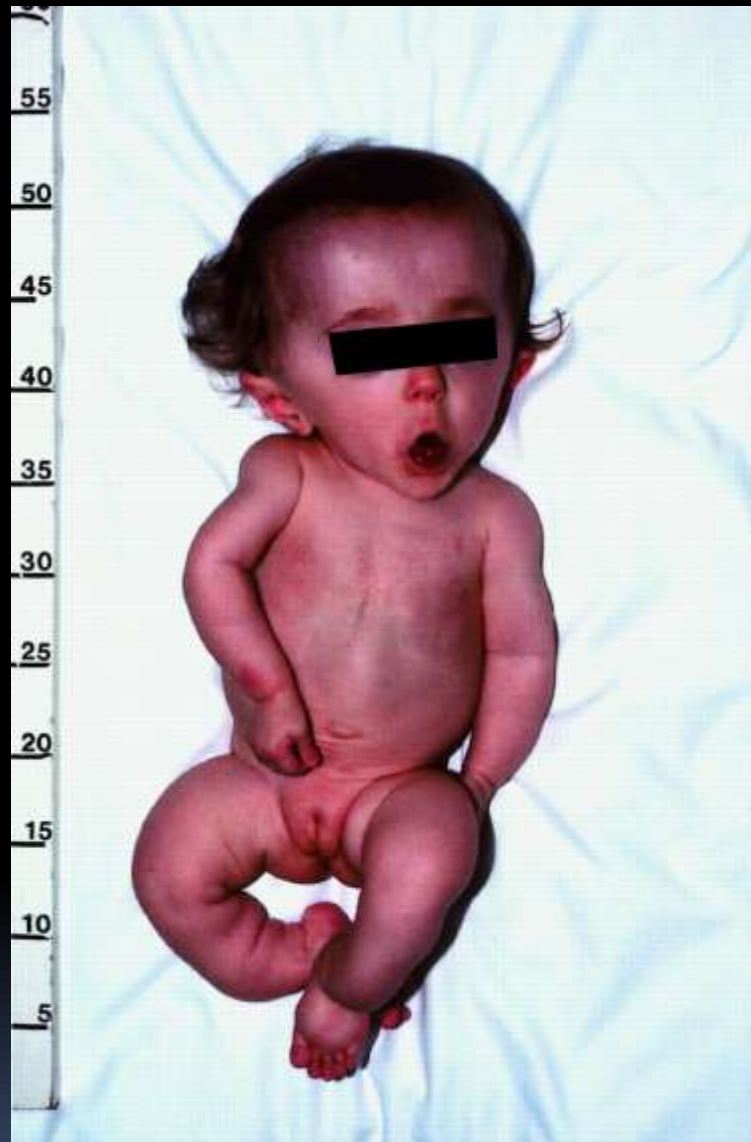
5. Genito urinary system

- Renal agenesis
- Congenital hydronephrosis
- Congenital polycystic kidney
- Horse shoe kidney
- Posterior urethral valves
- Hypospadias
- Epispadias
- Congenital phimosis
- Cong. Hydrocele
- Undescended testes
- Cong. Inguinal hernia
- Ambiguous genitalia
- Malformations of reproductive organs.

6. Musculoskeletal

- Club foot
- Cong. Dislocation of hip
- Osteogenesis imperfecta
- Polydactyly
- Webbed fingers
- syndactyly
- Phocomelia
- Cong. Scoliosis
- Marfan syndrome
- Mucopolysaccharidoses
- Muscular dystrophies










7. Blood disorders

- Thalassemia
 - Hemophilia
 - Sickle cell anemia
 - Congenital spherocytosis
- 

8. Metabolic disorders

- Cystic fibrosis
- Phenylketonuria
- G- 6PD deficiency
- Porphyria
- Cong. Lactose intolerance
- Glycogen storage diseases
- Mucopolysaccharidoses
- Tay- sachs disease
- Gaucher disease
- Wilson's disease
- Galactosemia
- Inborn errors of metabolism

9. Endocrinal abnormalities

- Cong. Hypopituitarism (dwarfism)
- Cong. Hypothyroidism (cretinism)
- Cong. Adreno genital hyperplasia
- Congenital goiter
- Diabetes mellitus



10. Chromosomal abnormalities

- Down's syndrome
- Patau's syndrome
- Edward's syndrome
- Turner syndrome
- Klienfelter's syndrome
- Cri du chat syndrome



Miscellaneous

- Cleft lip/ palate
- Cong. Cataract
- Cong. Glaucoma
- Retinoblastoma
- Color blindness
- Cong. Deafness
- Deaf and dumb
- Mental retardation
- Microagnathia
- Albinism
- Hemangioma
- Pseudohermaphroditism
- Situs inversus
- Prader- willi syndrome
- Apert syndrme
- Cong. Biliary atresia








PREVENTION OF CONGENITAL ANOMALIES

Preventive measures

- Genetic counseling is the true preventive measure of congenital anomalies.
- Reducing & discouraging consanguineous marriages.
- Avoiding late marriage of females and avoidance of pregnancy beyond the age of 35yrs.
- Promotion of health of girl child & pre pregnant health status of the females by prevention of malnutrition, anemia, folic acid deficiency, iodine deficiency etc.


- 
- Encouraging the immunization of all girl child by MMR
 - Increasing attention to the protection of individuals & whole communities against mutagens such as X ray & other ionizing radiations & also for chemical mutagens (drugs, alcohol)
 - Immunization by anti D immunoglobulin to the Rh negative mothers after abortion of first child.
 - Elimination of active & passive smoking of tobacco by mothers.
 - Avoidance of drug intake without consulting physician in the 1st trimester of pregnancy.

- 
- Prevention of intrauterine infections & promotion of sexual hygiene along with general hygienic measures.
 - efficient antenatal care
 - Promotion of therapeutic abortion of abnormal fetus & fetus with gross cong. Anomalies, after prenatal diagnosis.
 - Discouraging reproduction after birth of a baby with cong. Anomalies , without genetic counseling.
 - Increasing public awareness about the risk factors & etiological factors of cong. Anomalies.

- 
- Promotion of detection of genetic carriers . Eg:- 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.

Nursing responsibilities

- Collection of detailed history, especially history of prenatal, natal & postnatal period along with history of family illness.
- Preparation of pedigree chart by interview and home visit
- Identification of present problems, its nature & severity, for necessary interventions.
- Participation in diagnostic investigations, treatment, follow up and research project.
- Provide necessary information to the parents & family members

- 
- Motivate the family members for genetic counseling & referring to the genetic clinic.
 - Participating in genetic counseling process with special training, personal experience, knowledge & competency.
 - Provide emotional support & answer questions asked by the counselee.
 - Guide the family for rehabilitation of the child & for available social & economical support through social welfare agencies.
 - Promote public awareness about the prevention of cong. Anomalies by individual or group health education or by mass media information.