SNS COLLEGE OF NURSING

SARAVANAPATTI,COIMBATORE-35

DEPARTMENT : Department of Nursing

COURSE NAME : B. Sc. (Nursing) II Year IV Semester

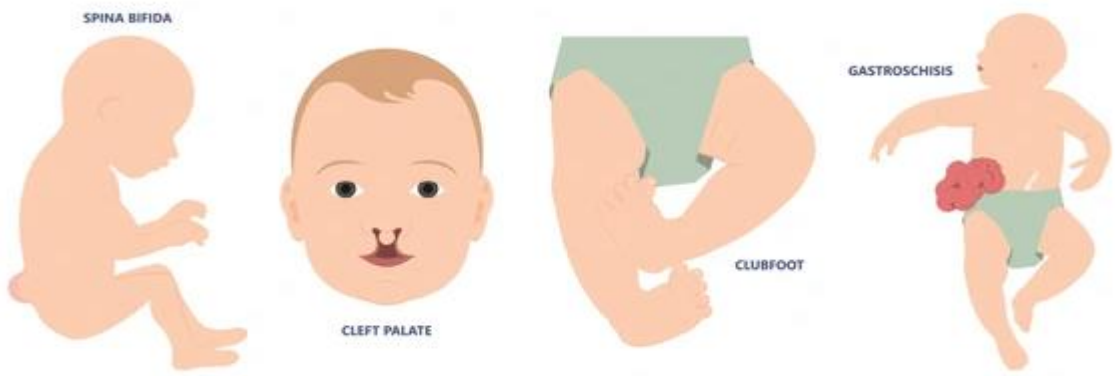
SUBJECT : Genetics

UNIT III : Genetic Testing for the Neonates and Children

TOPIC : Congenital Abnormalities

Introduction

- Congenital deformities include broad range of physical abnormalities existing from birth.
- The impacts of congenital deformities can be primary such as developmental delays (delays in motor & language skills)
- Or can be secondary (Social ostracism or low self esteem)
- However surgical procedures may help, but children may need multiple surgeries



Developmental Delay Definition

- Congenital disorders can be defined as structural or functional anomalies that occur during intrauterine life. Also called birth defects, congenital anomalies or congenital malformations, these conditions develop prenatally and may be identified before or at birth, or later in life. **– WHO**
- Congenital anomalies, previously referred to as birth defects, are structural (how the body is built) or functional (how the body works) anomalies present at birth that can cause physical disability, intellectual and developmental disorders, and other health problems. **- NICHD**

INCIDENCE

Global incidence - About 30 to 70/1000 live birth.

In India - 2.5 to 4 %

Most common type of birth defect-CNS abnormalities(22%)

CAUSES

A) GENETIC FACTORS:

- *Chromosomal abnormalities-eg.Down's syndrome*
- *Single gene disorders*
  - * Autosomal inheritance
    - .Dominant traits-0ne affected parent
    - .Recessive traits-Both parents
  - * X- linked or sex linked inheritance
    - .Dominant traits-daughter affected
    - .Recessive traits-son affected
- *Polygenic or multifactorial inheritance*
  - .combination of polygenic & environmental factors

B) ENVIRONMENTAL:

- *Intra uterine infections* – STORCH (Syphilis,Toxoplasmosis,Rubella,cytomegalaovirus and Herpes Virus)
- *Drugs intake during pregnancy* - Steroids,Anticonvulsants,Cocaine,Lithium,etc.,
- *X-Ray exposure during pregnancy*
- *Maternal diseases* - DM, CF, endocrine abnormalities , iodine deficiency, folic acid deficiency, malnutrition.,
- *Abnormal intrauterine environment* - bicornuate uterus,septed uterus,polyhydramnios,etc.,
- *Maternal addiction* - alcohol, tobacco & smoking
- *Environmental pollution* - air.

DIAGNOSIS

PRENATAL

TESTS	RATIONALE
◦ Maternal and family history ◦ Physical examination ◦ Biochemical assay ◦ Cytogenic study ◦ Blood test ◦ Hormonal assay ◦ Radiography ◦ USG	◦ Early detection ◦ Appropriate management

POST NATAL

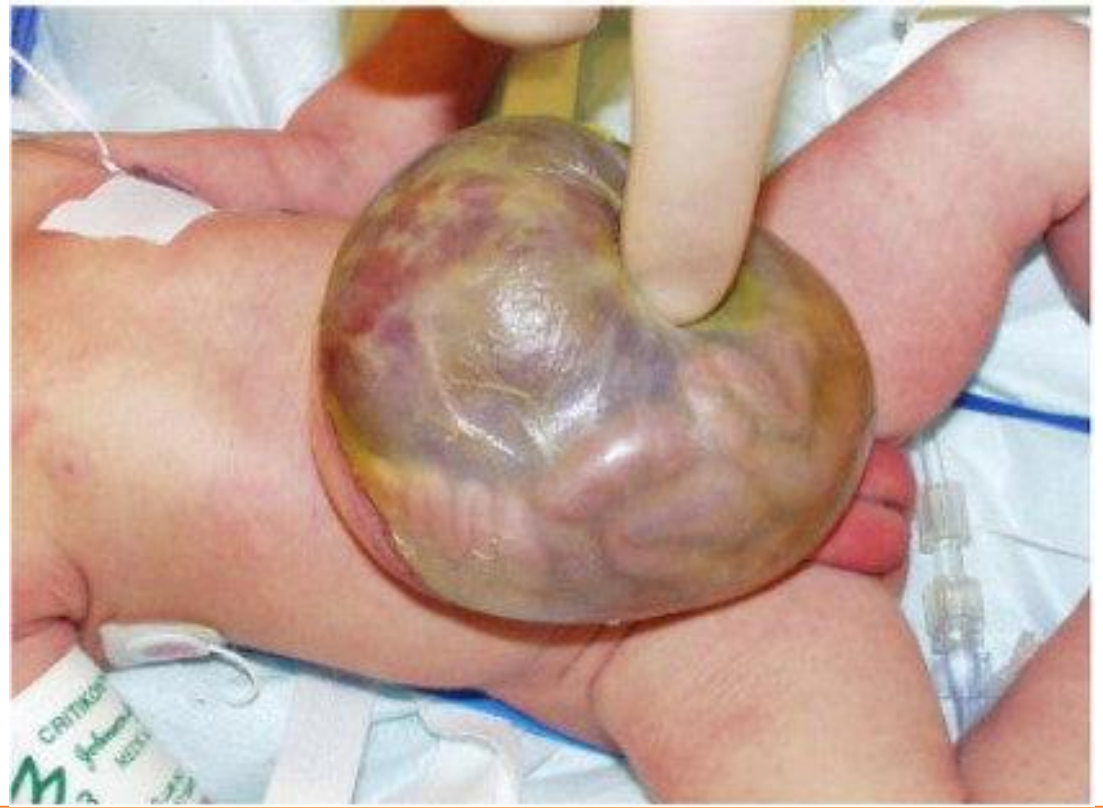
TESTS	RATIONALE
◦ Maternal and family history ◦ Physical examination ◦ Biochemical assay ◦ Cytogenic study ◦ Blood test ◦ Hormonal assay ◦ Radiography ◦ USG	◦ Early detection ◦ Appropriate management

CONGENITAL DEFORMITIES

NAME	SYSTEM	MANIFESTATION
GASTROSCHISIS	GI system	Defect of the abdominal wall with extrusion of bowel that is not covered by the peritoneum

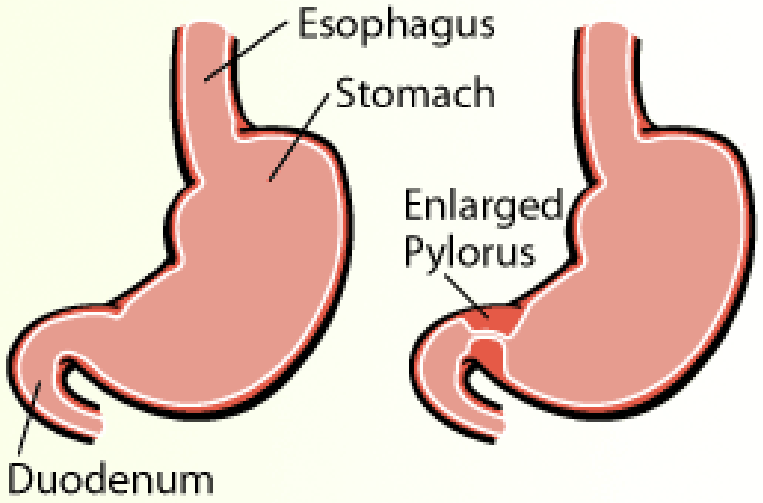
CONGENITAL DEFORMITIES

NAME	SYSTEM	MANIFESTATION
OMPHALOCELE	GI system	Defect in which the bowel / viscera protrudes through the umbilicus



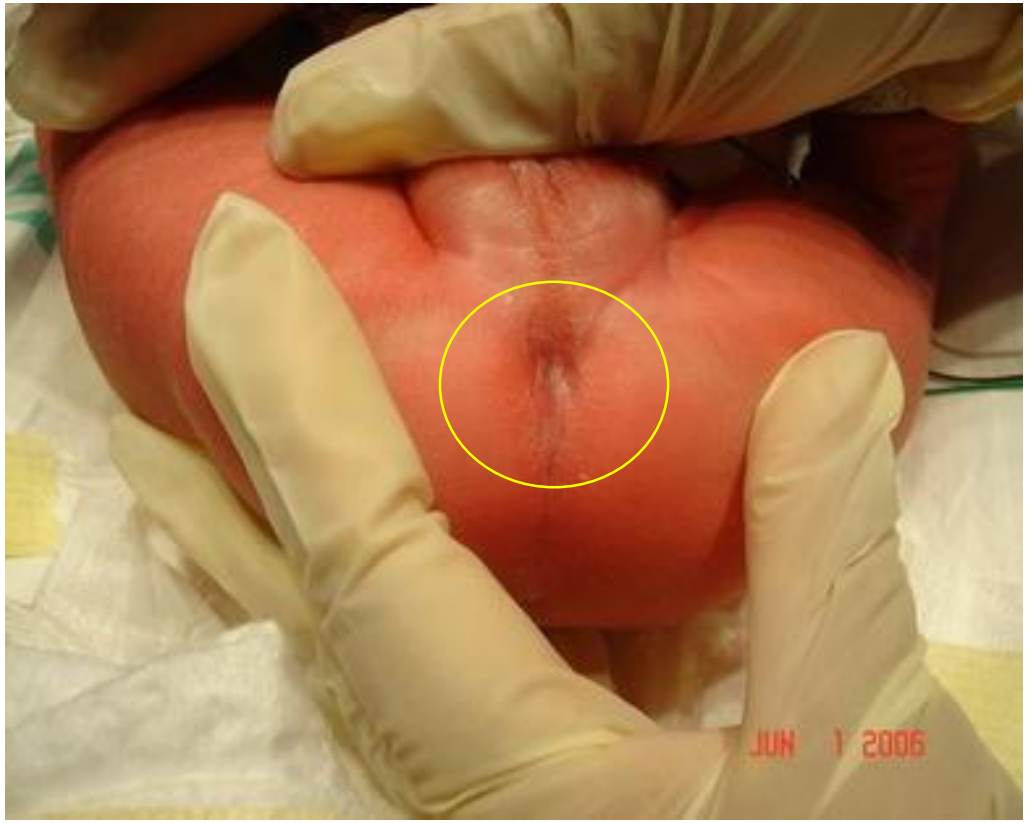
CONGENITAL DEFORMITIES

NAME	SYSTEM	MANIFESTATION
PYLORIC STENOSIS	GI system	Hypertrophy of the muscle of the pyloric sphincter



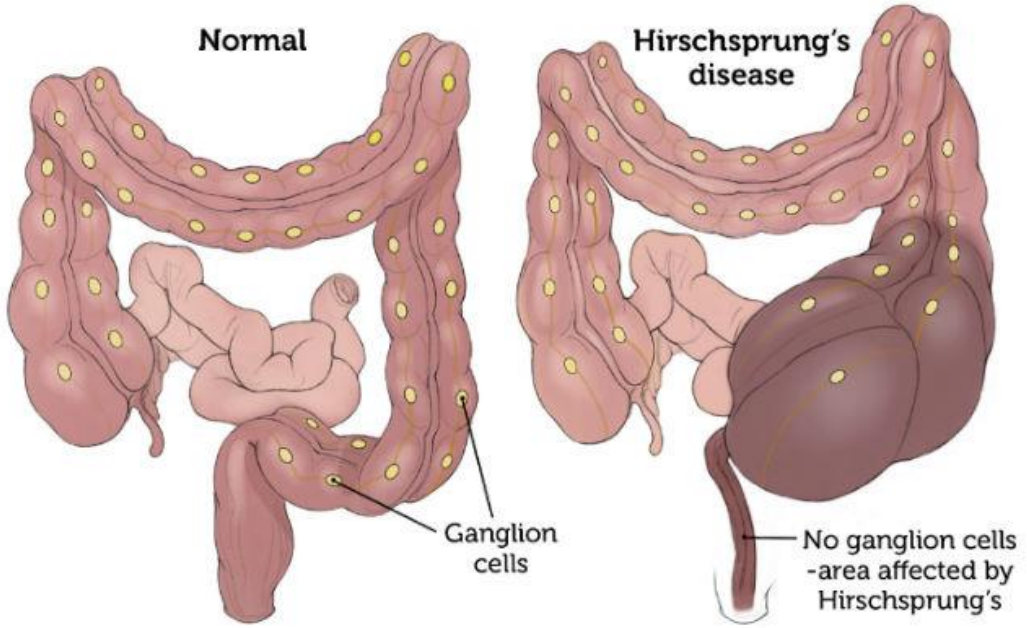
CONGENITAL DEFORMITIES

NAME	SYSTEM	MANIFESTATION
Rectal Atresia	GI system	Imperforate anus



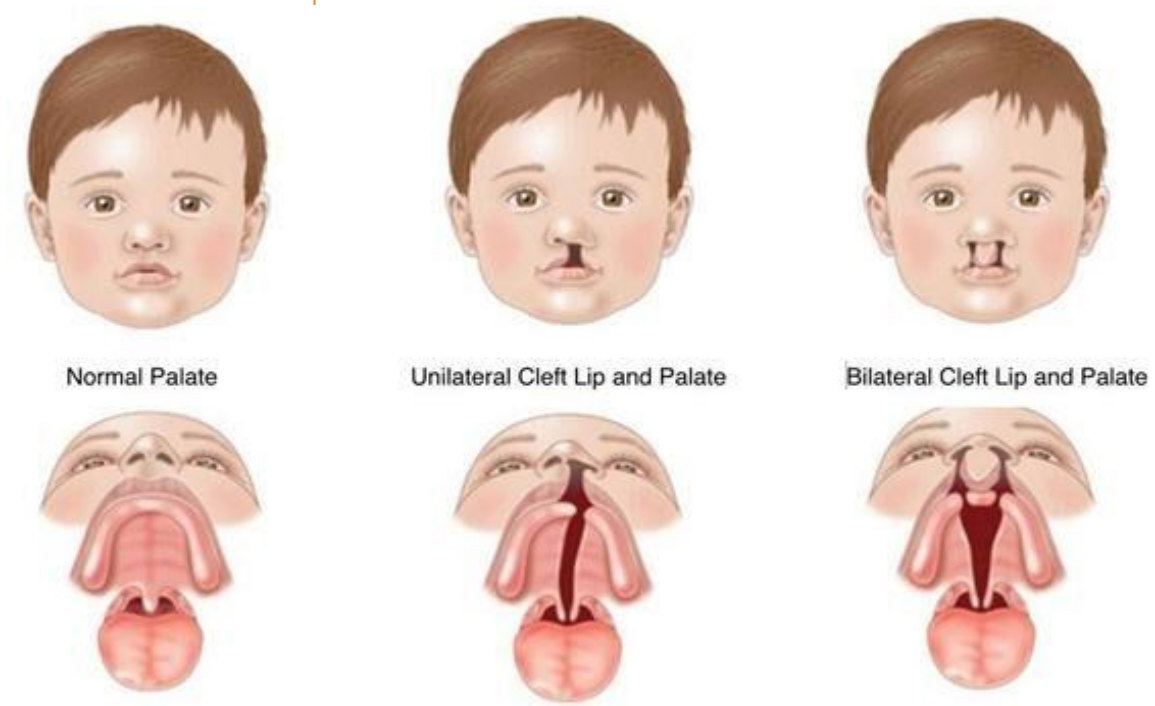
CONGENITAL DEFORMITIES

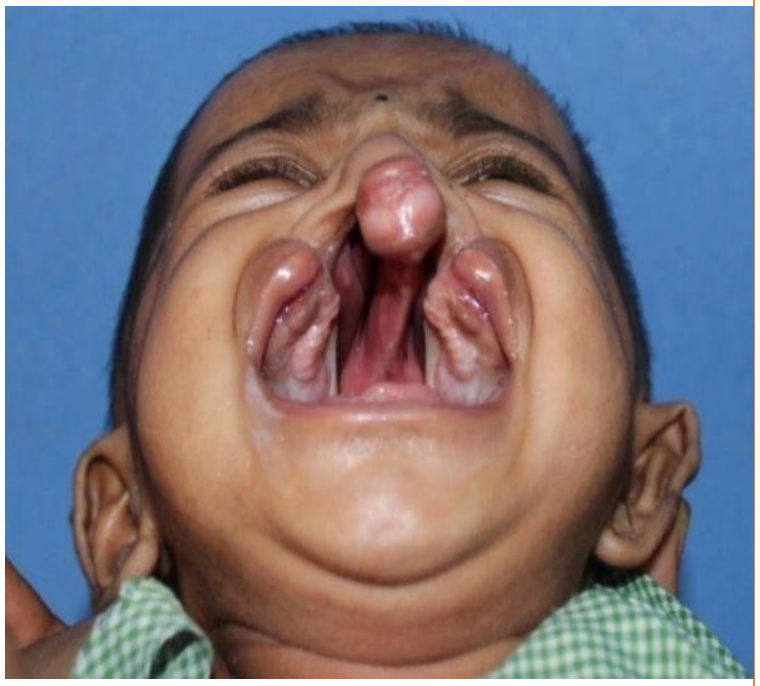
NAME	SYSTEM	MANIFESTATION
HIRSCHSPRUNG'S DISEASE	GI system	A ganglionic section of the large intestine is absent, peristalsis does not occur.



CONGENITAL DEFORMITIES

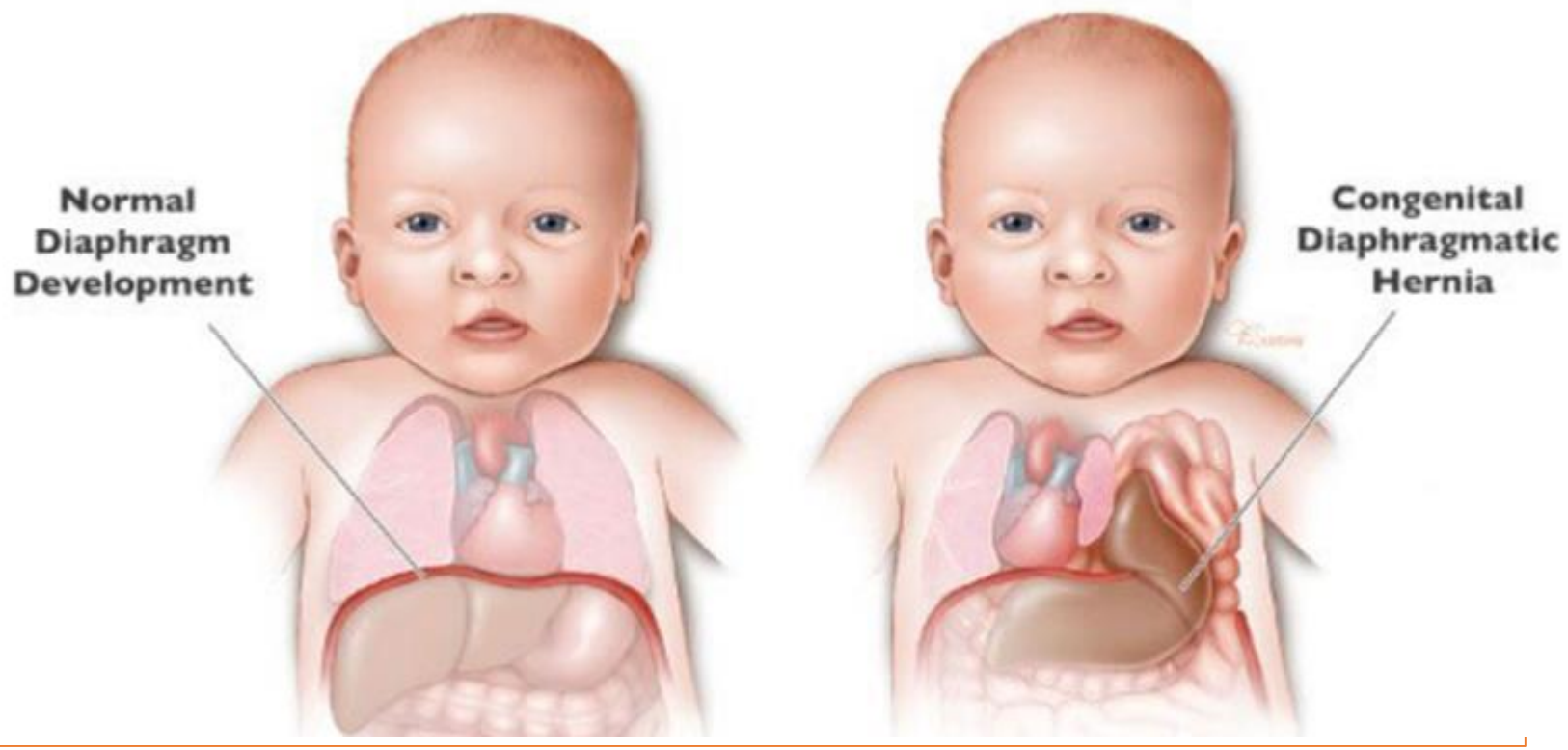
NAME	SYSTEM	MANIFESTATION
CLEFT LIP AND CLEFT PALATE	GI system	Clefts in the palate may affect both the hard palate, soft palate or both.





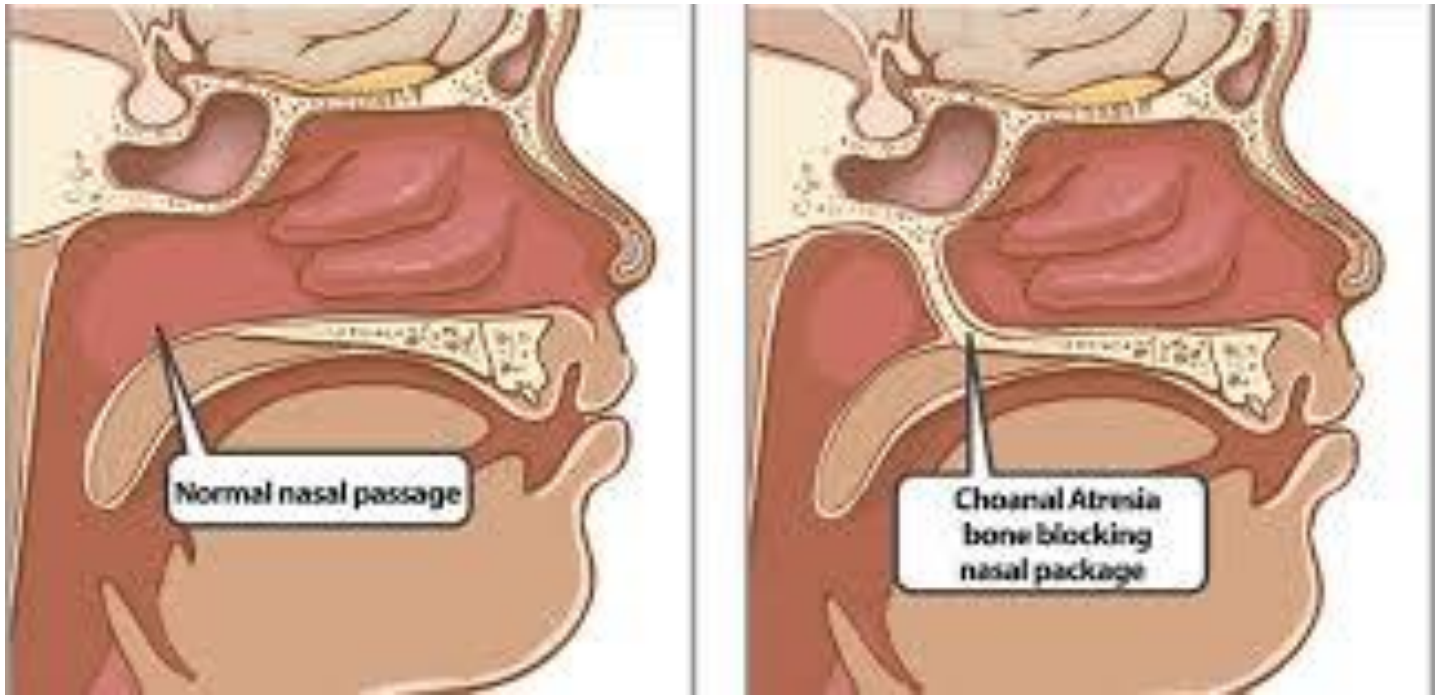
CONGENITAL DEFORMITIES

NAME	SYSTEM	MANIFESTATION
DIAPHRAGMATIC HERNIA	GI system	Defect In the diaphragm that allows the herniation of the abdominal contents into the thoracic cavity



CONGENITAL DEFORMITIES

NAME	SYSTEM	MANIFESTATION
CHOANAL ATRESIA	Respiratory system	Unilateral / bilateral narrowing of the nasal passage with a web of tissue occluding nasopharynx

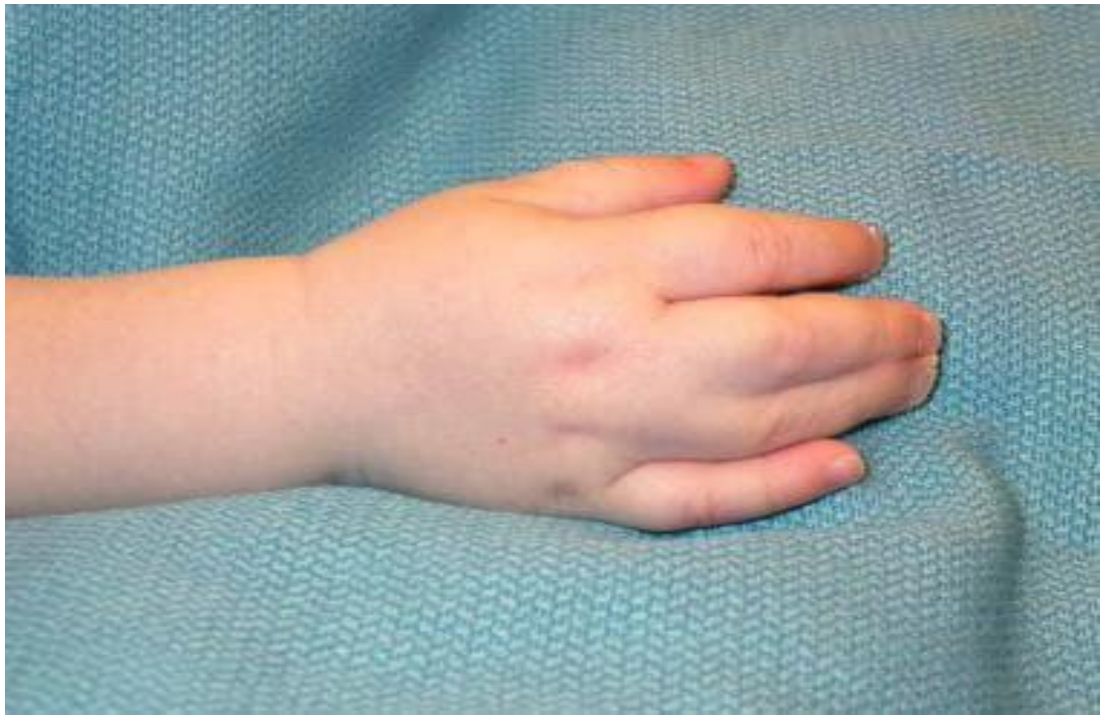


CONGENITAL DEFORMITIES

NAME	SYSTEM	MANIFESTATION
1. POLYDACTYLY 2.SYNDACTYLY	Musculo skeletal system	Extra digit of hand / foot Webbing of digit of hand / foot

1.

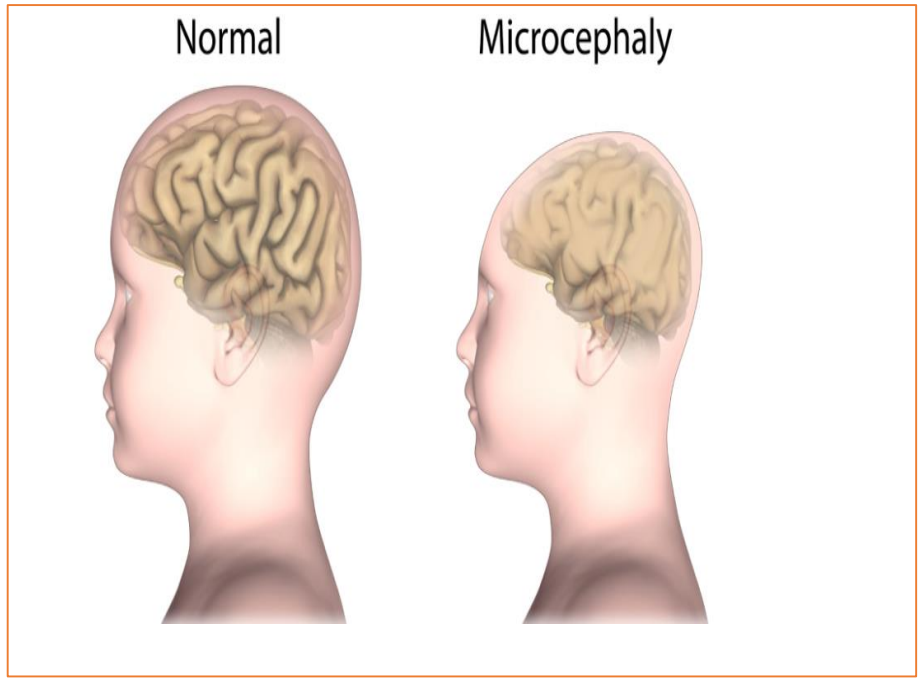
2.



CONGENITAL DEFORMITIES

NAME	SYSTEM	MANIFESTATION
1. Microcephaly 2. Anencephaly	CNS system	Head circumference is < 2 percentile Occurs when the fetal brain and skull don't fully develop in the uterus.

1.



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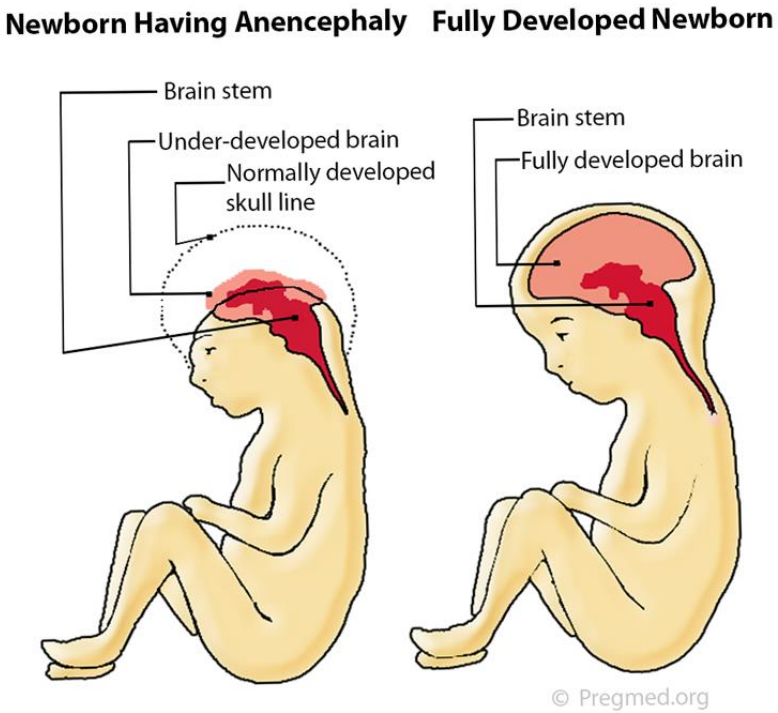
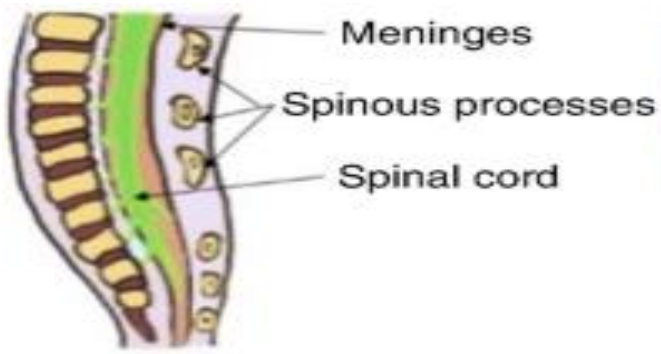
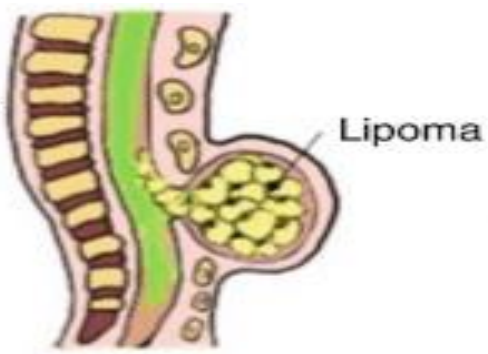


CONGENITAL DEFORMITIES

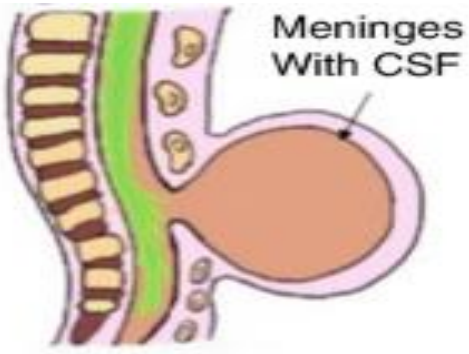
NAME	SYSTEM	MANIFESTATION
1. Spina bifida occulta 2. Closed spinal dysraphism	CNS system	A small separation or gap in one or more of the bones of the spine, called vertebrae. Failure of fusion of the vertebral bodies due to abnormal fusion of the posterior vertebral arches, with unexposed neural tissue; the skin overlying the defect is intact.
3. Meningocele		A sac of spinal fluid bulges through an opening in the spine. No nerves are affected and the spinal cord isn't in the fluid sac. Babies with meningocele may have some minor trouble with functioning, including with the bladder and bowels.
4. Myelomeningocele		The spinal canal is open along several vertebrae in the lower or middle back. Part of the spinal cord, including the spinal cord's protective covering and spinal nerves, push through this opening at birth, forming a sac on the baby's back. Tissues and nerves usually are exposed.



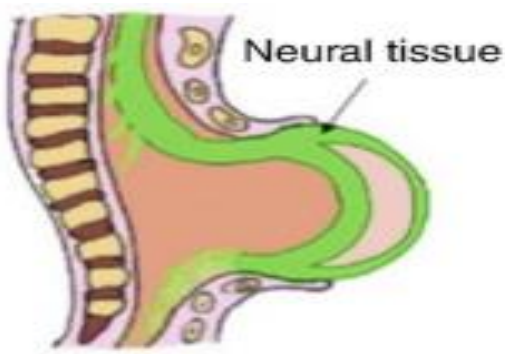
Spinal Bifida Occulta



Closed Spinal Dysraphism

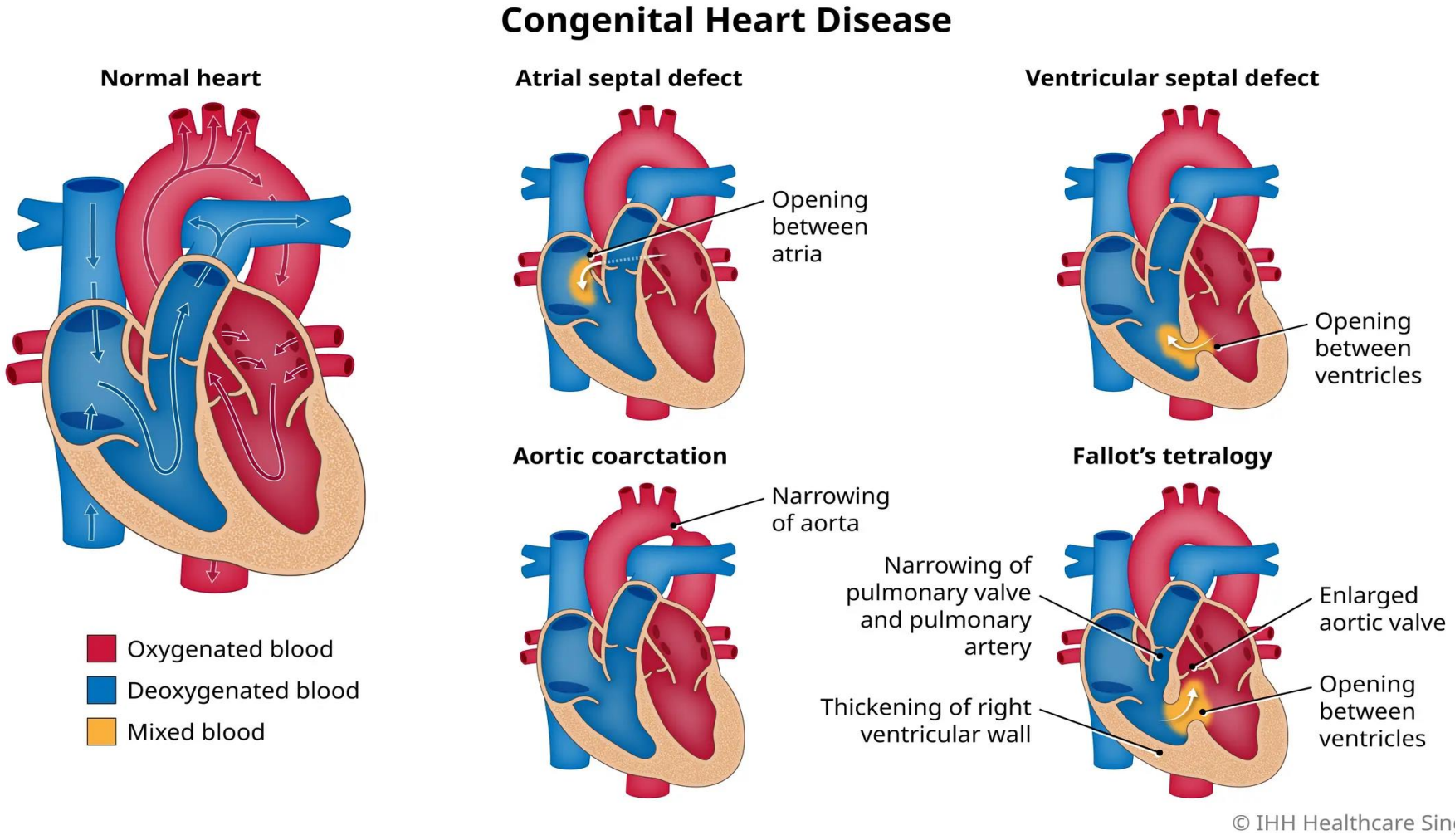


Meningocele



Myeloeningocele

Summary


Congenital Heart Disease
Normal heart
Oxygenated blood
Deoxygenated blood
Mixed blood
Atrial septal defect
Opening between atria
Ventricular septal defect
Opening between ventricles
Aortic coarctation
Narrowing of aorta
Fallot's tetralogy
Narrowing of pulmonary valve and pulmonary artery
Thickening of right ventricular wall
Enlarged aortic valve
Opening between ventricles
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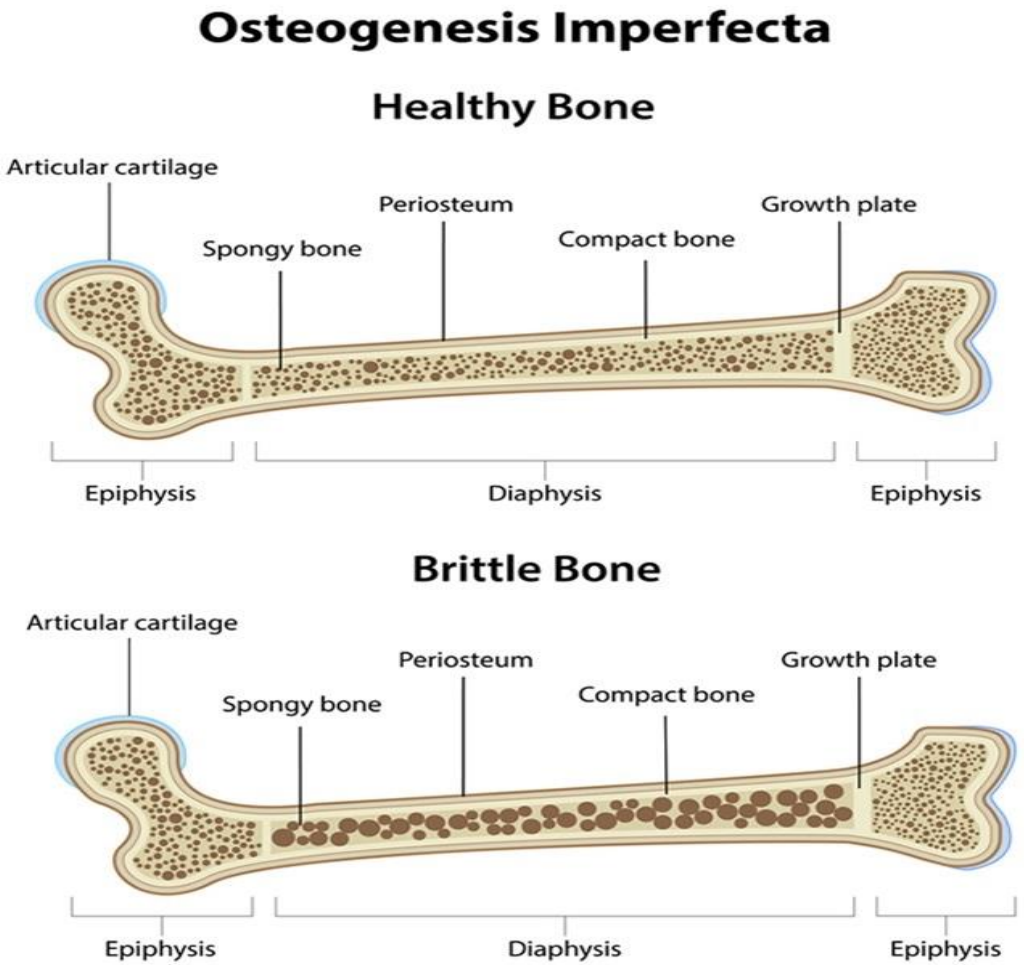
Conclusion


CONCLUSION


Osteogenesis Imperfecta
Healthy Bone
Articular cartilage
Spongy bone
Periosteum
Compact bone
Growth plate
Epiphysis
Diaphysis
Epiphysis
Brittle Bone
Articular cartilage
Spongy bone
Periosteum
Compact bone
Growth plate
Epiphysis
Diaphysis
Epiphysis


B : Blue Sclera
D : Dental imperfections (classically known as dentinogenesis imperfecta with discolored and weak teeth)
S : Sensorineural hearing loss
M : Multiple fractures

Assessment

- Define Congenital deformities.
- List out any 4 congenital deformities.
- Enumerate the CNS deformities
- Explain the etiology for Congenital deformities
- Differentiate cleft lip and cleft palate.

References

- Suresh K Sharma Textbook of Pharmacology Pathology and Genetics for Nurses 2016 edition revised 2022 Jaypee publishers
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- Chaitra K Textbook of Pathology and Genetics for BSc Nursing Students 1st edition Jaypee publishers
- Sonal Sharma Textbook of Pathology and Genetics for Nurses elsevier publishers


Thank
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