

CONGENITAL ABNORMALITIES

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LEARNING OUTCOMES

- ◉ Define congenital abnormalities
- ◉ State the significance
- ◉ Outline the common life threatening congenital abnormalities

DEFINITION

- ◉ Congenital malformations are anatomical defects present at birth
- ◉ Defects of the CNS and heart account for more than half of the total congenital malformations
- ◉ These are major causes of stillbirths and neonatal deaths
- ◉ More importantly, they are a cause of death in infancy

SIGNIFICANCE

- ◉ 1–2% of all babies are born with serious congenital malformations .
- ◉ Are major causes of Perinatal and infant death to the extend of infant mortality rate
- ◉ The incidence of serious defects and chromosomal anomalies amongst spontaneous abortions is very high.
- ◉ Some of them are predisposing factors to diseases e.g UTI in congenital obstruction of the urinary tract

- ◉ Some cause permanent disability e.g congenital dislocation of the hip and spina bifida.

COMMON LIFE THREATENING CONGENITAL ANORMALIES

- ◉ **CHOANAL ATRESIA** - congenital disorder where the back of the nasal passage (choana) is blocked,
- ◉ This is usually by abnormal bony or soft tissue (membranous) due to failed recanalization of the nasal fossae during fetal development
- ◉ Presents with;-
 - respiratory distress in the delivery room.
 - Apnoea ,
 - inability to pass a nasogastric tube through the nasal nares

COMMON LIFE THREATENING CONGENITAL ANORMALIES..

When one notices choanal atresia then suspect the following congenital anomalies (CHARGE Syndrome)

- ◉ C—Coloboma of the eyes
- ◉ H—Heart anomaly
- ◉ A—Atresia of the choana
- ◉ R—Retardation
- ◉ G—Genital anomaly
- ◉ E—Ear anomaly

COMMON LIFE THREATENING CONGENITAL ANORMALALIES..

◎ **PIERRE ROBIN SYNDROME;**

- Is a condition in which an infant has a smaller than normal lower jaw,
- a tongue that falls back in the throat, and difficulty breathing.
- It is present at birth and presents with micrognathia, cleft palate, air way obstruction

◎ **DIAPHRAGMATIC HERNIA**

- Is a birth defect in which there is an abnormal opening in the diaphragm.
- The diaphragm is the muscle between the chest and abdomen that helps you breathe.

COMMON LIFE THREATENING CONGENITAL ANORMALIES..

- The opening allows part of the organs from the belly to move into the chest cavity near the lungs
- It presents with a scaphoid abdomen, bowel sounds present in the chest and Respiratory distress

◎ **TRACHEOESOPHAGEAL FISTULA.**

- Is an abnormal connection between these two tubes.
- As a result, swallowed liquids or food can be aspirated (inhaled) into your child's lungs.

COMMON LIFE THREATENING CONGENITAL ANORMALIES..

- Feeding into the stomach directly can also lead to reflux and aspiration of stomach acid and food.
- It presents with polyhydramnious, aspiration pneumonia and excessive salivation, Inability to pass a NGT in to the stomach
- **Suspect VATER Syndrome**
 - V=Vertebral defects
 - A=anal may be imperforate
 - T-E= Tracheo -Esophageal fistula
 - R=Radial and Renal dysplasia

COMMON LIFE THREATENING CONGENITAL ANORMALIES..

⦿ Intestinal obstruction

- Is significant mechanical impairment or complete arrest of the passage of contents through the intestine
 - This is due to pathology that causes blockage of the bowel.
- ## ⦿ It presents with Polyhydramnious, bile stained emesis, and abdominal distension