



# Congenital Malformation



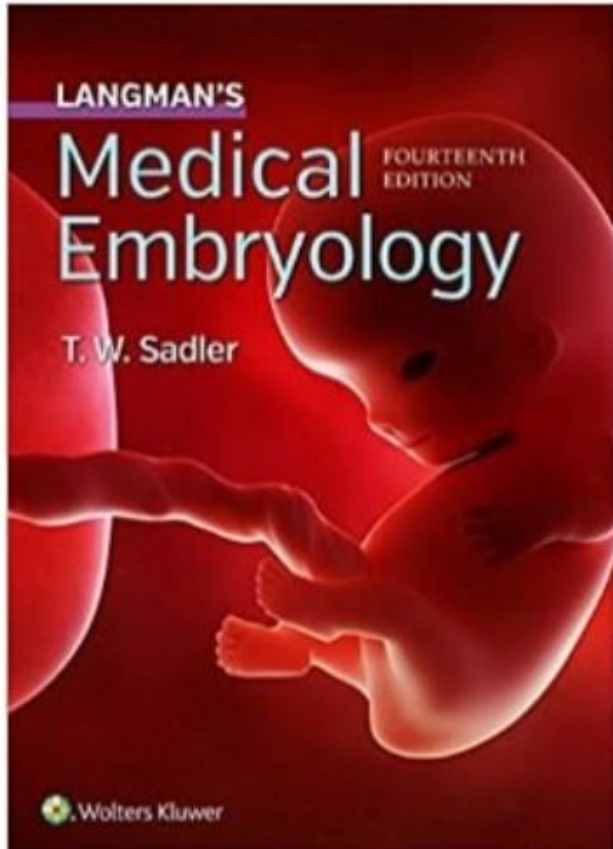
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# Learning Objectives

- Congenital Malformation
- Definition
- Incidence
- Principle
- Etiology
- Prenatal diagnosis
- Some Congenital Malformation

## **Congenital malformation**

-includes the structural defects present at birth. also known as birth defects, congenital disorder

-It consist of primary morphologic defects in an organ or body part resulting from abnormal developmental events that are directly involved in the development of that organ or body part. For example, failure of the neural groove to close results in a malformation called a neural tube defect.

## Deformation

•It consist of **secondary** morphologic defects that are imposed upon an organ or body part owing to mechanical forces; that is, deformations affect the development of an organ or body part **indirectly**. For example, if insufficient amniotic fluid forms (i.e., oligohydramnios(decrease of amniotic fluid)).

# Principle of teratology

1. Susceptibility to teratogenesis depend on the genotype of the conceptus.
2. Susceptibility to teratogens varies with the developmental stage at the time of exposure(third to eight weeks ).
3. Manifestation of teratogenesis depend on dose and duration of exposure.
4. Teratogens act in specific ways on developing cells (biochemical, molecular, cell death)
5. Manifestation of abnormal development are death, malformation, growth retardation, functional disorders.

## **Prenatal diagnosis of teratology**

1-Ultrasound

2-Maternal serum screening(measurement of several protein responsible for teratogen)

3-Amniocentesis

4-Chorionic villus sampling

## **Incidence of Congenital Malformation**

- ❖ Congenital anomalies affect 1 in 33 infants and result in 3.2 million birth defect.
- ❖ About 270000 newborn die during the first 28 days of live every year.
- ❖ Many congenital anomalies can be prevented (vaccination ,adequate intake of folic acid and iodine).

## Etiology

- 1-Infectious agents (rubella virus which cause glaucoma, heart defect and tooth abnormality)
- 2-Radiation(killing of cells)
- 3-Chemical agents(thalidamine (anticoagulant))
- 4-Hormones
- 5- Nutritional deficiency
- 6-Maternal diseases(diabetes, polyketon urea)
- 6-Obesity
- 7-Hypoxia
- 8- Heavy metals(mercury)



# **Fetal therapy**

1. Fetal transfusion
2. Fetal medical treatment
3. Fetal surgery
4. Stem cell transplantation and gene therapy

## Some Congenital Malformation



ANENCEPHALY



MENINGOENCEPHALOCELE-

- A. Normal spine
- B. Spina bifida occulta
- C. Meningocele
- D. Meningomyelocele





Meningocele



Meningomyelocele



Hydrocephalus



Microcephaly



Macrocephaly



Syringomyelia

Thank You



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