



Congenital Anomalies

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Dysmorphology

- Congenital Anomalies → alter the shape or form of one or more parts of the body of a newborn child.
- More than 20% of infant deaths were attributed to birth defects.
- They are present at birth in the development of organs or other structures.
- They are a major cause of mortality, long-term morbidity, intellectual disability, several dysfunctions.

Research

- the contribution of abnormal genes
- nongenetic, environmental influences
- developmental pathways

Medical geneticist

- to diagnose a child with a birth defect
- to suggest further diagnostic evaluations
- to give prognostic information
- to manage the expected complications
- to provide the family with an understanding of the causation of the birth defect
- to give recurrence risks

Congenital Anomalies

- Major congenital anomalies

A major anomaly can be defined as one that has an adverse outcome on either the functions or the social acceptability of the individual.

- Minor congenital anomalies

These are of neither medical nor cosmetic importance

Examples of major congenital abnormalities

Minor Congenital Anomalies

- Preauricular pit or tag
- Epicanthic folds
- Lacrimal duct stenosis
- Brushfield spots in the iris
- Lip pits
- Single palmar crease
- Fifth finger clinodactyly
- Syndactyly between second and third toes
- Supernumerary nipple
- Umbilical hernia
- Hydrocele
- Sacral pit or dimple

Incidence of structural abnormalities

Single abnormalities

- Malformation
- Disruption
- Deformation
- Dysplasia

Malformation

- A primary structural defect of an organ or part of an organ
- Only a single malformation shows generally multifactorial inheritance.
- Multiple malformations are more likely to be due to chromosomes abnormalities or single gene mutations.

Disruption

This term refers to an abnormal structure of an organ or tissue as a result of external factors disturbing the normal developmental process.

Deformation

An abnormal mechanical force that distorts an otherwise normal structure.

Dysplasia

An abnormal organization of assembly of cells into tissue.

The effects are usually seen wherever that particular tissue is present.

Multiple abnormalities

- Sequence
- Syndrome
- Association

Sequence

asosiyasyon

VACTERL

Vertebral

Anal

Cardiac

Tracheo-**E**sophageal

Renal

Limb

Environmental Factors; teratogens

- Drugs and Chemicals; alcohol, thalidomide, etc.
- Infection agents; rubella, cytomegalovirus, toxoplasmosis, etc.
- Physical agents; ionizing radiation, prolonged hyperthermia
- Maternal illness; DM, PKU, maternal epilepsy

Further reading

- Thompson&Thompson, Genetics in Medicine, eighth ed. 2016.
- Emery's Elements of Medical Genetics, 15th ed. 2017.