Thi – Qar University
Dentistry College
Second Stage

Congenital anomalies

Dr. Sada Ghalib

Congenital Anomaly

Congenital Anomaly (CA): is an anomaly that affects a body part or physiologic function and is present at birth.

It is caused by the abnormal ontogenetic development of the fetus.

The process is affected by genetic, environmental or both factors.

The disturbance of the regulation and development cascades take

place on the level of tissue, cell or molecule

Dr. Sada Ghalib

Other „synonyms“

Congenital Malformation is a congenital anomaly of the structure of some body part.

Birth Defect or **Congenital Disorder** are nearly synonyms for the term Congenital Anomaly.

Chromosomal Aberrations are the numerical or structural abnormalities of the karyotype.

Genetic Disorders are the conditions caused by the mutation of the gene(s).

Nomenclature – type of anomaly

Malformation: is caused by an abnormal development of the organ / tissue, that is abnormal from the beginning.

Disruption: is caused by destructive process, that affects an organ / tissue, that started to develop normally.

Deformation: is caused by an abnormal physical force, that damages healthy organ / tissue.

Dysplasia: is caused by an abnormal organization of the cells in the organ / tissue.

Nomenclature – association of anomalies

Isolated anomaly: an anomaly that is not associated with any other conditions (e.g. **isolated polydactyly**).

Sequence: multiple anomalies that result from the pathologic cascade caused by a primary insult (e.g. **Potter's sequence**).

Association: selected congenital anomalies that tend to develop all together – in an association (e.g. **VATER association**).

Syndrome: complex of phenotypic traits (anomalies) that are typical for defined clinical diagnosis (e.g. **Down syndrome**).

Types of Polydactyly

Teratogenesis

Teratogene: is an agent that is able to affect normal ontogenetic development and lead to a congenital anomaly.

Mutagene: is an agent that is able to affect the genetic information on the level of DNA or on the level of chromosomes.

Mutagens cause mutations.

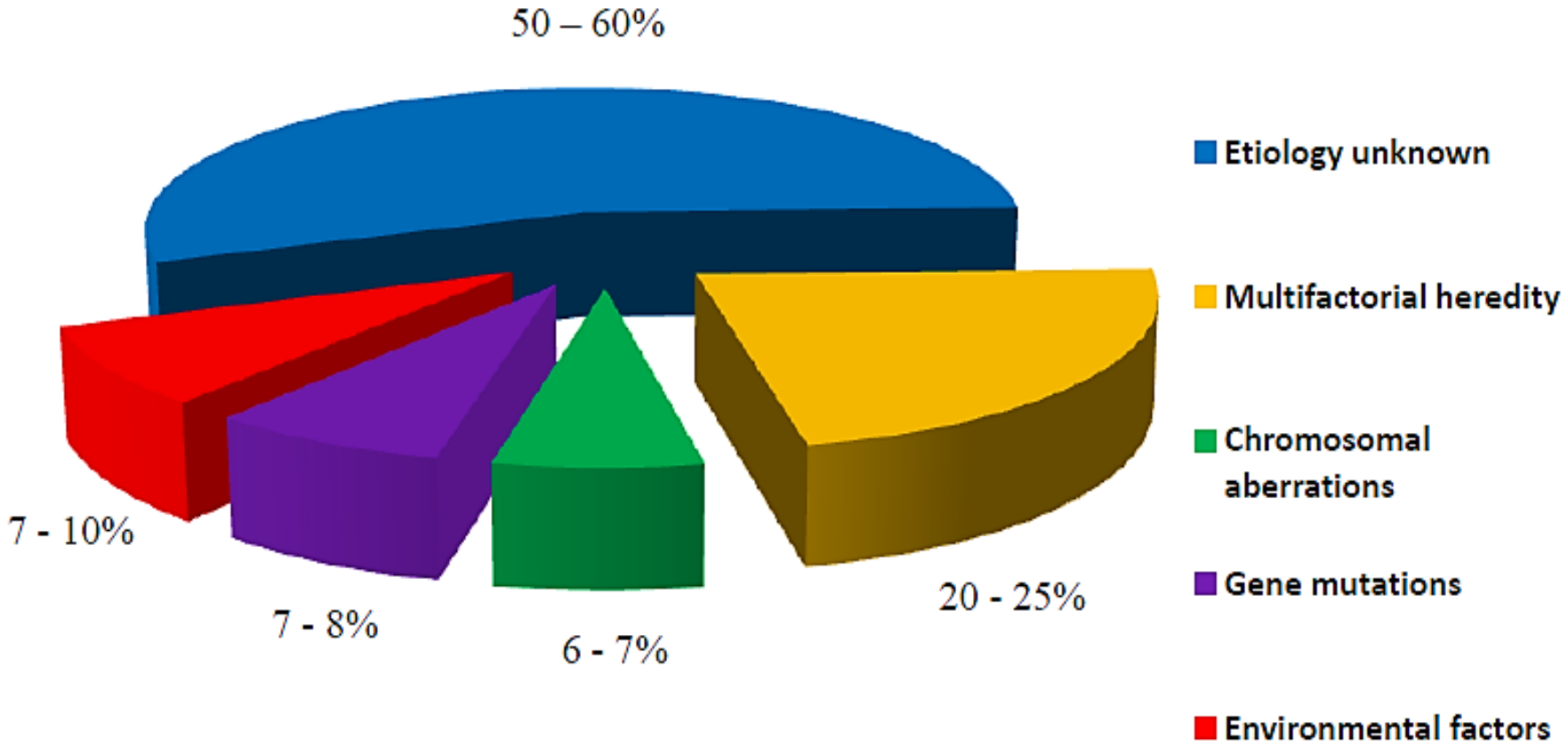
Teratogens cause congenital anomalies.

Some mutagens are also teratogens. However, not all teratogens are mutagens.

What causes congenital anomalies?

- Genetic factors
- Environmental factors
- Unknown factors

Etiology of the congenital anomalies in man



From: Moore K. L., Persaud T. V. N.; The Developing Human: Clinically Oriented Embryology, 6th Edition; 1998

Dr. Sada Ghalib