

# Inborn errors of development in human, examples, classification

Inborn errors of development or congenital malformations or birth defects are specific errors of prenatal development which we can observe in the born children.

## Ethiology

- **Monogenic disorders:** Marfan syndrome, osteogenesis imperfecta
- **Chromosomal aberrations:** down syndrome
- **Multifactorial:** hip joint dysplasia, orofacial clefts
- **Proven teratogens:** Fetal alcohol syndrome, Gregg syndrome (congenital rubella)
- **Deformation:** Amniotic bands syndrome

## The classical classifications - considering the ethiology of the defect

- **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.

## Classification, considering the multiplicity of the defect

- **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).

## Examples

- **Neural tube defects (NTD):** anencephaly, spina bifida, encaphalocoele
- **Orofacial clefts**
- **Congenital heart defects**
- **Abdominal wall defects** (omphalocele, gastroschisis)
- **Limb defects** (syndactyly, polydactyly, reduction limb defects)