**Teratology**

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**Teratology**

- The study of abnormal development in embryos and the causes of congenital malformations or birth defects

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**Birth Defects**

- Observed in 3% of newborns
- Observed in another 3% of children later
- May or may not be outwardly visible
- Etiology: genetic and environmental

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**Major and Minor Anomalies**

- Major anomalies: life/health threatening
- Minor anomalies: cosmetic

- The greater the number of minor anomalies, the greater the likelihood of a major anomaly
- Certain minor anomalies suggest specific major anomalies

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**Down Syndrome**

![Image of a child with Down Syndrome]
Turner Syndrome

Trisomy 13
- Midline defects (cleft lip and cleft palate)
- Central nervous system malformations
- Micro-ophthalmia
- Congenital heart disease
- Poor growth

Trisomy 18

Achondroplasia

Inborn Errors of Metabolism Causing Birth Defects
- Smith Lemli Opitz
- Congenital disorders of glycosylation
- Fatty acid oxidation disorders

Teratogens
- A chemical, infectious agent, physical condition, or deficiency that, on fetal exposure, can alter fetal morphology or subsequent function
- Teratogenicity depends upon the ability of the agent to cross the placenta
- The embryo is most susceptible to teratogenic agents during periods of rapid differentiation
How are agents determined to be teratogenic?
- Anecdotal data in humans
- Data from animal studies

Effect of Exposure Depends on Timing
- All or none effect early
- Effect of organogenesis during embryonic development
- Effect on size and function during fetal development

Nicotine
- IUGR
- Premature delivery
- Neurocognitive development
Fetal Alcohol Syndrome

- Characteristic facial features
- Congenital heart disease
- Growth deficiency
- Behavioral/neurocognitive deficits

Tetracycline

- Yellow/brown teeth
- Decreased bone growth

Fetal Hydantoin Syndrome

- Intrauterine growth retardation
- Microcephaly, mental retardation
- Distal phalangeal hypoplasia
- Specific facial features

Retinoic acid

- Craniofacial dysmorphisms
- Cleft palate
- Thymic aplasia
- Neural tube defects

Thalidomide Syndrome
**Congenital Rubella**

- Intrauterine growth retardation
- Micromelia
- Chorioretinitis, blindness
- Microcephaly
- Cerebral calcifications, mental retardation
- Hepatosplenomegaly

**Congenital CMV**

- Intrauterine growth retardation
- Micromelia
- Chorioretinitis, blindness
- Microcephaly
- Cerebral calcifications, mental retardation
- Hepatosplenomegaly

**Ionizing Radiation**

- Affects brain development at 10-18 weeks of gestation a HIGH dose
- No evidence of effect of exposure associated with typical diagnostic studies

**Maternal Hyperglycemia**

- Congenital heart disease
- Renal, gastrointestinal, and central nervous system malformations such as neural tube defects

**Babies of Mother’s with PKU**

- Mental retardation
- Low birth weight
- Congenital heart disease
Threshold Effect-Multifactorial