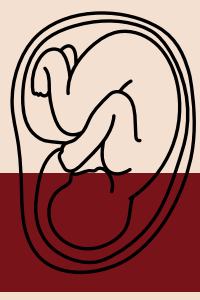
Sex and Gender Differences

BIRTH DEFECTS

Congenital anomalies are any physical and/or mental impairment/dysfunction/alteration during embryonic/fetal development



Risk Factors

- Risk factors **before conception**: parental genetics, infertility problems, use of **a**ssisted **r**eproductive **t**echnologies (**ART**s), advanced age, and genetic inter-relatedness
- Risk factors **during pregnanc**y: maternal substance use, folic acid deficiency, obesity, uncontrolled pre-gestational diabetes, phenylketonuria, teratogenic prescription drug use, infections, exposures (ionizing radiation, organic mercurials, and lead)

Female

- **Female** fetuses at higher risk of congenital heart disease (i.e. Atrioventricular septal defects in Down Syndrome)
- **Protective factor:**
  - Higher catecholamine levels provide support during hypoxia at preterm delivery, therefore resulting in less oxidative stress

Male

- **Male** fetuses at higher risk secondary to maternal smoking
- Pulmonary and cardiac disorders (i.e. Tetralogy of Fallot)
  - Preterm males at higher risk due to **delayed** lung maturation

Prevalence & Presentation

- In the US, the most common birth defects are clubfoot, cleft palate, limb deformities, pulmonic stenosis/atresia, and Down Syndrome
- 50% of first-trimester miscarriages have chromosomal anomalies

1 in 33 babies born in the US have a birth defect

Females have higher rates of:

- neural tube defects (e.g. anencephaly)
- endocrine system defects (e.g. congenital hypothyroidism)
- choledochal cyst
- hip dysplasia
- Trisomy 18
- atrial septal defect
- patent ductus arteriosus

Males have higher rates of:

- omphaloceles
- pyloric stenosis
- clubfoot
- cleft palate
- sex organ defects
- urinary tract defects
- ventricular septal defect
- pulmonic stenosis and atresia

Pathophyisology

- May be the result of one or more genetic, infectious, nutritional, or environmental factors
  - Often difficult to identify the exact causes
- **Males: more** commonly affected by **X-linked variants**
- **Females: less** commonly affected and phenotypically **less** severe
  - Unless homozygous for the deleterious allele

Screening

- Available screening tests include:
  - ultrasound, blood and urine tests, amniocentesis, genetic testing, imaging, etc
- Noninvasive prenatal testing (NIPT):
  - analyzing fetal DNA in maternal blood (AFP, hCG, estriol, inhibin)

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SEX and GENDER HEALTH COLLABORATIVE of the American Medical Women's Association

To learn more or get involved, visit **amwa-doc.org/sghc** or email us at **sghc@amwa-doc.org**

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Definitions & Disclaimers **References**