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FACULTY OF NURSING

Chapter-07

Congenital Malformatio

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Definition

Congenital anomalies are also known as birth defects, congenital disorders or congenital malformations (WHO).

Congenital anomalies can be defined as structural or functional anomalies, including metabolic disorders, which are present at the time of birth

Epidemiology

Congenital anomalies affect an estimated 1 in 33 infants and result in approx 3.2 million birth defect-related disabilities every yr.

About 270 000 newborns die during the first 28 days of life every year from congenital anomalies.

may result in long-term disability, which may have significant impacts on individuals, families, health-care systems and societies.

most common severe congenital anomalies are heart defects, neural tube defects and Down syndrome.

Epidemiol…

Although congenital anomalies may be genetic, infectious or environmental in origin, most often it is difficult to identify the exact causes.

Many congenital anomalies can be prevented. eg vaccination, adequate intake of folic acid and iodine, and adequate antenatal care are keys for prevention.

Causes and risk factors

- Genetic/chromosomal
- Environmental
- Socioeconomic status
- Infections
- Maternal nutrition status

Genetics

Genetic/ mutations

Consanguinity (relationship by blood) increases the prevalence of rare genetic congenital anomalies and nearly doubles the risk for neonatal and childhood death, intellectual disability and serious birth anomalies in first cousin unions.

Race e.g. Ashkenazi Jews or Finns, have comparatively high prevalence of rare genetic mutations, leading to a higher risk of congenital anomalies.

Syndromes

Environmental factors

Maternal exposure to
- pesticides
- medications
- alcohol
- tobacco
- psychoactive substances
- certain chemicals
- high doses of vitamin A during the early pregnancy,
- high doses of radiation.
- Working or living near or in waste sites, smelters, or mines.

Maternal infections

TORCHES (Toxoplasmosis, Rubella, Cytomegalovirus, Herpes, Syphillis)

Varicella

HIV

Socioeconomic factors

congenital anomalies are more frequent among resource constrained families and countries.

It is estimated that about 94% of severe birth defects occur in middle- and low-resource countries,

where mothers are more susceptible to macronutrient and micronutrient malnutrition and may have

increased exposure to agents or factors that induce or increase the incidence of abnormal prenatal development, particularly infection and alcohol.

Advanced maternal age also increases the risk of some chromosomal abnormalities including Down syndrome.

Maternal nutritional status

Iodine deficiency

folate insufficiency

Obesity

diabetes mellitus

Infectious Agents

- Rubella (German Measles)
    - Malformations of the eye
        - Cataract, Microphthalmia
    - Malformations of the ear
- Congenital deafness
    - Due to destruction of cochlea
- Malformations of the heart
    - Patent ductus arteriosus
    - Atrial septal defects
    - Ventricular septal defects

Infectious Agents (cont.)

- Rubella (German measles)
  - May be responsible for some brain abnormalities
    - Mental retardation
  - Intrauterine growth retardation
  - Myocardial damage
  - Vascular abnormalities

Infectious Agents (cont.)

- Cytomegalovirus
  - Disease is often fatal early on
  - Malformations
    - Microcephaly
  - Cerebral calcifications
  - Blindness
    - Chorioretinitis
  - Kernicterus (a form of jaundice)
  - multiple petechiae of skin
  - Hepatosplenomegaly
  - Mother asymptomatic

Infectious Agents (cont.)

- Herpes Simplex Virus
  - Intrauterine infection of fetus occasionally occurs
  - Usually infection is transmitted close to time of delivery
  - Abnormalities (rare)
    - Microcephaly
    - Microphthalmos
    - Retinal dysplasia
    - Hepatosplenomegaly
    - Mental retardation
  - Usually child infected by mother at birth
    - Inflammatory reactions during first few weeks

Infectious Agents (cont.)

- Varicella (chickenpox)
  - Congenital anomalies
    - 20% incidence following infection in 1st trimester
    - Limb hypoplasia
    - Mental retardation
    - Muscle atrophy
- HIV/AIDS
  - Microcephaly
  - Growth retardation
  - Abnormal facies (expression or appearance of the face)

Infectious Agents (cont.)

- Toxoplamosis
  - Protozoa parasite (Toxoplama gondii)
    - Sources
      - Poorly cooked meat
      - Domestic animals (cats)
      - Contaminated soil with feces
- Syphilis
  - Congenital deafness
  - Mental retardation
  - Diffuse fibrosis of organs (eg. liver & lungs)
- In general most infections are pyrogenic
  - Hyperthemia can be teratogenic
    - Fever
    - Hot tubs & Saunas

Radiation

- Teratogenic effect of ionizing radiation well established
  - Microcephaly
  - Skull defects
  - Spina bifida
  - Blindness cleft palate
  - Extremity defects
- Direct effects on fetus or indirect effects on germ cells
- May effect succeeding generations
- Avoid X-raying pregnant women

Radiation

- Studies of offspring of Japanese women who were pregnant at the time of the atomic bomb explosions over Hiroshima & Nagasaki who survived the blast
  - 28% aborted
  - 25% gave birth to children who did not survive their first year
  - 25% of the surviving children had abnormalities of CNS
    - e.g. Microcephaly & mental retardation

Chemical agents/Drugs

- Role of chemical agents & drugs in production of anomalies is difficult to assess
  - Most studies are retrospective
    - Relying on mother's memory
  - Large # of pharmaceutical drugs used by pregnant women
    - NIH study – 900 drugs taken by pregnant women
      - Average of 4/woman during pregnancy
      - Only 20% of women use no drugs during pregnancy
  - Very few drugs have been positively identified as being teratogenic

Drugs

Thalidomide

- Antinauseant & sleeping pill
- Found to cause amelia & meromelia
  - Total or partial absence of the extremities
- Intestinal atresia
- Cardiac abnormalities
- Many women had taken thalidomide early in pregnancy (in Germany in 1961)

Drugs (cont.)

Aminopterin

- Antagonist of Folic Acid
- Antineoplastic agent which inhibits mitosis
- Defects
  - Anencephaly
  - Meningocele
  - Hydrocephalus
  - Cleft lip & palate

Drugs (cont.)

- Anticonvulsants (to treat epilepsy)
  - Diphenylhydantoin (phenytoin)
    - Craniofacial defects
    - Nail & digital hypoplasia
    - Growth abnormalities
    - Mental deficiency
    - The above pattern is know as “fetal hydantoin syndrome”
  - Valproic acid
    - Neural tube defects
    - Heart defects
    - Craniofacial & limb anomalies

Drugs (cont.)

Trimethadione (syndrome)

- Malformed ears
- Cleft palate
- Cardiac defects
- Urogenital anomalies
- Skeletal anomalies

Drugs (cont.)

- Antipsychotic drugs (major tranquilizers)
  - Phenothiazine & lithium
    - Suspected teratogenic agents
- Antianxiety drugs (minor tranquilizers)
  - Meprobamate, chlordiazepoxide,
    - Severe anomalies in 11-12% of offspring where mothers were treated with the above compared to 2.6% of controls
  - diazepam (valium)
    - Fourfold ⇑ in cleft lip with or without cleft palate

Drugs (cont.)

- Anticoagulants
  - Warfarin (A.K.A cumadin or cumarol)
    - Teratogenic
    - Hypoplasia of nasal cartilage
    - Chondrodysplasia
    - Central nervous system defects
      - Mental retardation
      - Atrophy of the optic nerves
- Antihypertensive agents
  - angiotensin converting enzyme (ACE) inhibitor
    - Growth dysfunction, renal dysfunction, oliogohydramnios, fetal death

Drugs (cont)

- Propylthiouracil
  - Goiter
  - Mental retardation
- Potassium iodide
  - Goiter
  - Mental retardation
- Streptomycin
  - deafness
- Sulfonamides
  - kernicterus
- Imipramine (antidepr.)
  - Limb deformaties
- Tetracyclines
  - Bone & tooth anomalies
- Amphetamines
  - Oral clefts
  - CV abnormalities
- Quinine
  - Deafness
- Aspirin
  - Potentially harmful in large doses

Drugs (cont.)

- Isotretinoin (13-*cis*-retinoic acid)
  - Analogue of vitamin A
  - Drug is prescribed for treatment of cystic acne & other chronic dermatoses
  - Highly tertogenic
    - Reduced & abnormal ear development
    - Flat nasal bridge
    - Cleft palate
    - Hydrocephaly
    - Neural tube defects
    - Heart anomalies

Recreational drugs

- PCP angel dust
  - Possible malformations & behavioral disturbances
- Cocaine-vasoconstrictor $\Rightarrow$ hypoxia
  - Spontaneous abortion
  - Growth retardation
  - Microcephaly
  - Behavioral problems
  - Urogenital anomalies
  - gastroschisis

Alcohol

- Relationship between alcohol consumption & congenital abnormalities
- Fetal alcohol syndrome
  - Craniofacial abnormalities
    - Short palpebral fissures
    - Hypoplasia of the maxilla
  - Limb deformities
    - Altered joint mobility & position
  - Cardiovascular defects
    - Ventricular septal abnormalites
  - Mental retardation
  - Growth deficiency

Cigarette Smoking

Has **not** been linked to major birth defects

- Smoking does contribute to intrauterine growth retardation & premature delivery
- Some evidence that is causes behavioral disturbances

Hormones

- Androgenic Agents
  - Synthetic progestins were used frequently to prevent abortion
    - Ethisterone & norethisterone
      - Have considerable androgenic activity
        - Masculinization of female genitalia
- Diethylstilbesterol
  - Commonly used in the 1940's & 1950's to prevent abortion; in 1971 determined that DES caused increased incidence of vaginal & cervical cancer in women who had been exposed to DES in utero
  - In addition high % suffered from reproductive dysfunction
- Oral Contraceptives
  - Low teratogenic potential, discontinue if pregnancy suspected
- Cortisone-cleft palate in mice (not humans)

Maternal Disease

- Disturbances in CHO metabolism (diabetic mothers)
  - High incidence of stillbirth, neonatal deaths
  - Abnormally large infants
  - Congenital malformations
    - ⇑ risk 3-4X
    - Cardiac, Skeletal, CNS Anomalies
    - Caudal dysgensis
      - Partial or complete agenesis of sacral vertebrae in conjuction with hindlimb hypoplasia
  - Hypoglycemic episodes ⇒teratogenic (why?)
  - Oral hypoglycemic agents ⇒ maybe teratogenic

Maternal Disease (cont.)

Phenylketonuria (PKU)

- Enzyme phenylalanine hydroxylase is deficient ⇑ phenylalanine (PA) concentrations
  - Mental retardation
  - Microcephaly
- Risk can be ⇓ with low PA diet

Hypoxia

Associated with congenital malformations in a great variety of experimental animals

- In humans ???
  - Maybe smaller babies e.g. offspring at high altitude

Environmental Chemicals

Mercury

- Fish, seed corn sprayed with mercury containing fungicide
  - Multiple neurological symptoms

Lead

- ⇑ abortions
- Growth retardation
- Neurological disorders

Approach to a dysmorphic child

Pregnancy History

birth weight? SGA infants may have a chromosome anomaly or may have been exposed to a teratogen. LGA - infants of diabetic mothers or overgrowth syndrome, eg Beckwith-Wiedemann syndrome.

Was the infant full term, premature, or postmature?

maternal age is associated with an increased risk of nondisjunction leading to trisomies.

complications during the pregnancy?

Maternal medical problems? Does she take any medications? Did she smoke cigarettes, drink alcohol, or take any drugs? Neonatal hypotonia may have manifested prenatally; answers to these questions may provide information.

Family History - pregnancy or neonatal losses

Head

Enlarged fontanelle

Hair- swirls/white forelock

Eyes- congenital cataracts, extra folds, hyper/hypotelorism, upslant/downslant

Choanal atresia

Ear pits/tags, position/rotation

Cleft palate/lip

Prominent frenula/tongue tie

Neck and thorax

Cystic hygroma

Thyroglossal duct cyst

Branchial cleft cyst

Pectus excavatum

Extra mamillary tissue (3rd nipple)

Heart murmur- congenital heart disease

TEF

CHD

Abdomen

Omphalocele/gastroschesis/umbilical hernia

Scaphoid abdomen- ?congenital diaphragmatic hernia

Femoral pulses- aortic coarctation

Undescended testes

Virilized female

Fistulas, hypospadia/epispadias, imperforate anus, Hirschprung's

Posterior urethral valves (no urinating)

Extremities and skin

Congenital Hip dysplasia

Extra digits

Single palmar crease

Clubbed foot

Congenital nevi

Hemangiomas

Cleft lip and palate

Incidence: about 1 in 600 live births

Cleft lip with or without cleft palate

Syndromic: associated with another syndrome. Syndromic cleft lip/palate is more common in males

Nonsyndromic: isolated finding, not associated with any particular syndrome. Non syndromic tends to be equal between males and females.

Consider submucous cleft palate with bifid uvula

causes

Multifactorial: combination of hereditary and environmental factors involved in growth and development

Interference with normal development- within the 1st few months of development

Medications such as phenytoin, steroids, retinoids (Vitamin A derivatives)

Alcohol, hypoxia and dietary deficiencies have been implicated

Both single and multiple genes

Associated syndromes

DeGeorge/velocardiofacial/22q deletion

Pierre Robin malformation sequence

Apert syndrome

Crouzon syndrome

Treacher-Collins

complications

Feeding problems

Eustacian tube dysfunction secondary to abnormal muscle placement -> serous otitis/middle ear disease/chronic ear infections -> hearing problems

Speech problems

Dental problems

Team approach needed: medical/surgical, dental, speech and hearing

treatment

Surgical repair- usually by plastic surgery- can affect maxillofacial growth pattern

Timing of surgery controversial- often in 2 stages. One commonly used plan involves early soft palate repair at age 6 months, followed by hard palate repair at age 6 years. Others involve complete repair at a later age.

Involvement with ENT, speech therapy, following hearing tests, dental/orthodontic specialists, social supports.

- **THANK YOU**