

Common Congenital Anomalies
Presenting in the Neonatal Period

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Common Congenital Anomalies
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Objectives

Describe the genesis of and general approach to congenital anomalies detected in the neonatal period

List the most common anomalies encountered in the newborn period

Recognize those anomalies that may warrant further evaluation and follow-up

Definitions

Congenital- of or pertaining to a condition present at birth, whether inherited or caused by the environment

Anomaly- a deviation from the common rule, type, arrangement, or form; an odd, peculiar, or strange condition, situation, quality, etc.

Birth defect- a physical or biochemical defect (as cleft palate, phenylketonuria, or Down syndrome) that is present at birth and may be inherited or environmentally induced

Definitions (continued)

Neonatal-of or relating to the first 28 days of an infant's life

Common- widespread; general; ordinary; of frequent occurrence; usual; (in this case 3-4%)

Definitions (continued)

Morphogenesis-formation of the structure of an organism or part; differentiation and growth of tissues and organs during development

Dysmorphism-an anatomical malformation

Normal variant- Something that differs in form only slightly from something else

Types of problems in morphogenesis

- Malformation
- Deformation
- Disruption
- Dysplasia

Types of problems in morphogenesis

Poor formation of tissue, organ or structure-
malformation

cleft lip/palate, spina bifida

sequence-a single localized poor formation of tissue that initiates a chain of subsequent defects; nearly normal to severe; recurrence risk 1-5%

Types of problems in morphogenesis

*Unusual forces on normal tissue-***deformation**

club foot, breech molding

sequence-no problem in the fetus but mechanical forces result in altered morphogenesis (oligohydramnios, breech); very good prognosis; low recurrence risk

Types of problems in morphogenesis

*Breakdown of normal tissue-***disruption**

amniotic bands, in utero stroke

normal fetus is subjected to a destructive problem (vascular, infectious or even mechanical) and its consequences; variable prognosis; very low recurrence risk

Types of problems in morphogenesis

Abnormal organization of cells into tissues-
dysplasia

skeletal dysplasia (achondroplasia)

may be specific gene defect/mutation that leads to the abnormal development of tissue; variable prognosis and recurrence

Patterns of Malformations

Syndrome-a pattern of anomalies that occur together and are pathogenetically related

Sequence-a pattern of anomalies in which a single known defect in development causes a cascade of subsequent abnormalities

Patterns of Malformations

Developmental field defect- a pattern of anomalies caused by disturbance of a region of the embryo that develops in a contiguous physical space. This region is known as a developmental field

Patterns of Malformations

Association- two or more anomalies that are not pathogenetically related and occur together more frequently than expected by chance. In general, the etiology of associations is not defined. It is possible that some represent developmental field defects

Major Malformations

Have medical and/or social implications
May often require surgical intervention

Mainz congenital birth defect monitoring system of almost 31,000 liveborn, stillborn and aborted fetuses found a 6.9% occurrence of major malformations

Minor Malformations

Mostly cosmetic significance
Rarely require surgical intervention or have medical implications
Part of normal variation in the general population
~50% of minor anomalies occur in the head and neck
Mainz system-35.8% occurrence

Malformation syndromes-Multiple congenital anomalies

Multiple structural defects that cannot be explained on the basis of a single initiating defect and its consequences, but felt to be due to a common cause, such as chromosomal abnormalities, mutant gene disorders and environmental teratogens

Malformation syndromes-Multiple congenital anomalies

Infants with three or more minor anomalies are at increased risk of having a major defect or syndrome. In two reports, a major malformation was present in 26 and 19.6 percent of infants with three or more minor anomalies

Gestational age vs. embryonic age

Gestational age is the time that has passed since the onset of the last menstruation, which generally or as standard occurs 2 weeks before the actual fertilization.

Embryonic age, in contrast measures the actual age of the embryo or fetus from the time of fertilization.

Nevertheless, menstruation has historically been the only means of estimating embryonal/fetal age, and is still the presumed measure if not else specified.

However, the actual duration between last menstruation and fertilization may in fact differ from the standard 2 weeks by several days.

Thus, the first week of embryonic age is already week three counting with gestational age.

Furthermore, the number of the week is one more than the actual age of the embryo/fetus. For example, the embryo is 0 whole weeks old during the 1st week after fertilization.

(from Wikipedia)

Relative Timing of Certain Malformations	
Central Nervous System (CNS)	
Holoprosencephaly	23 days
Anencephaly	26 days
Meningomyelocele (spina bifida)	28 days
Face	
Cleft lip	36 days
Cleft palate	10 weeks



Relative Timing of Certain Malformations	
Gut	
Esophageal atresia + TEF	30 days
Duodenal atresia	7-8 weeks
Omphalocele	10 weeks
Heart	
Transposition of great vessels	34 days
Ventricular septal defect	6 weeks

Approach to dysmorphology

Gather information-history, family history, physical examination

Which anomaly represents the earliest defect in morphology?

Approach to dysmorphology

Can all the anomalies be explained on the basis of a single problem in morphogenesis that leads to a cascade of subsequent defects?

Attempt to arrive at a specific overall diagnosis and counsel accordingly

Etiology of Congenital Malformations

Frequency

(69,277 infants of gestational age ≥ 20 weeks)

- Not known 43.1 percent
- Multifactorial 22.8 percent
- Familial 14.4 percent
- Chromosomal 10.1 percent
- Single gene 4.1 percent
- Teratogen 4.1 percent
- Uterine factors 2.5 percent
- Twinning 0.4 percent

Common Congenital Anomalies Presenting in the Neonatal Period	
	Occurrence
Cardiac (1/3 to 1/4 of all defects)	100-200
Hemangiomas/birthmarks	10-20
Hypospadias	140-325
Polydactyly	100-500
Cleft lip/palate	700-100
Clubfoot	333-1000

Common Congenital Anomalies Presenting in the Neonatal Period	
	Occurrence
Single unilateral palmar crease	10-20
Ear tags/pits	10-100
Single umbilical artery	150-500
Spina bifida	500-1500
Omphalocele	5000
Gastroschisis	10,000

Common Congenital Anomalies Presenting in the Neonatal Period	
	Occurrence
Trisomy 21	800-1000
Klinefelter's syndrome	1000
Turner syndrome	2500
Trisomy 18	3000
VACTERL association	6250
Trisomy 13	10,000




