

Congenital and Genetic Abnormalities

The etiology of birth defects is not completely understood, malformations may occur from Genetic factors, such as change in the chromosome number, mutation, or structural abnormalities, or environmental factors such as irradiation, infection, and drugs. The most frequent malformations occur from interactions between multiple genetics and environmental factors. Birth defects are common, costly, and critical conditions that affect 1 in every 33 babies born in the United States each year. Every 4 ½ minutes, a baby is born with a birth defect in the United States. That means nearly 120,000 babies are affected by birth defects each year.

Abnormalities include but are not limited to:

Congenital cardiac anomalies

Congestive heart failure

Cleft palate/lip (failure of the bone to fuse)

Hypospadias (urethra opens on the undersurface)

Epispadias (urethra is on the dorsal surface)

Ambiguous genitalia (uncertain male/female)

Craniosynostosis (premature closures)

Spina bifida (lack of one or more vertebral arches)

Hydrocephalus (increased CSF/enlarged head)

Anencephaly (complete or partial absence of brain)

Microcephaly (smaller head and brain)

Obstructions of the alimentary tract

Diaphragmatic hernia

Pyloric stenosis

Omphalocele

Gastroschisis

Imperforated anus

Esophageal atresia

Chromosomal anomalies

Trisomy 13 or d- lifespan <1 year

Trisomy 18 or e- lifespan <6 months

Trisomy 21 or down's syndrome- 1 in 500 births

Musculoskeletal disorders

Talipes equinovarus (Clubfoot)

Congenital hip dysplasia

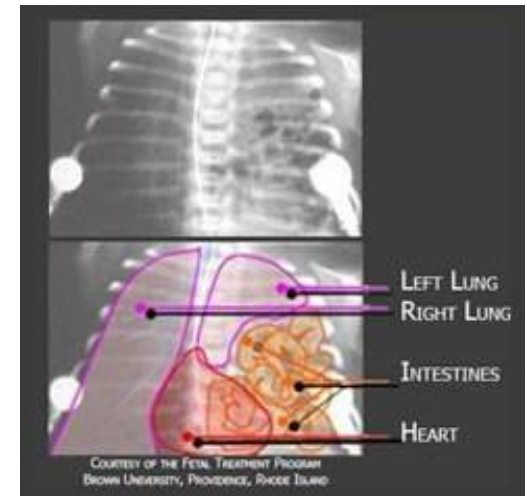
Scoliosis

Polydactyly (extra digits)

Diaphragmatic Hernia:

Caused by a defect in the development of the diaphragm that allows abdominal organs to herniate into the thoracic cavity. The majority of cases involve the left leaf of the diaphragm. The mortality rate is high. Stabilization of the infant prior to a surgical repair requires placement of an ng tube to decompress the abdomen, correction of acidosis, administration of oxygen, and ventilatory assistance.

If the defect is small, it can be easily repaired. This must be done soon after birth because the herniated abdominal organs interfere with adequate respiration. If the defect is large, the mortality is higher. The defect may be repaired by using a graft patch since not enough diaphragm is there to complete the repair.



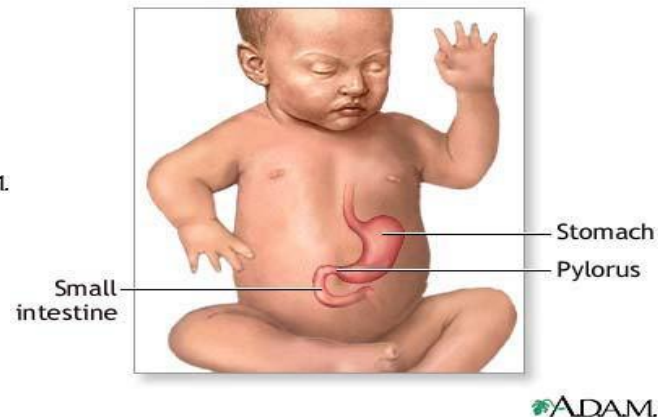
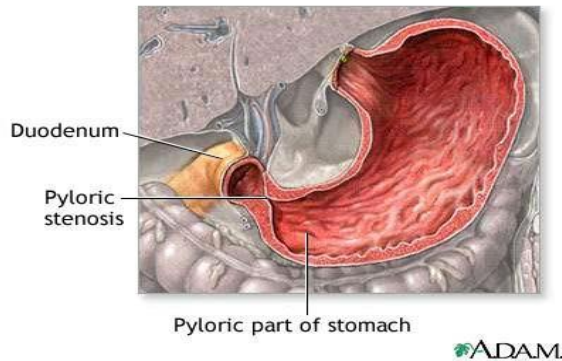
Pyloric Stenosis:

The incidence of pyloric is 2 to 5 per 1000 live birth.

It usually manifests its symptoms between the second and sixth week by the onset of vomiting that becomes projectile and occurs within 30 minutes after every feeding. The infant loses weight, bowel elimination lessens, highly colored urine becomes scanty, and the symptoms of dehydration appear. Upon examination, gastric peristalsis is found, and the pyloric "acorn like" tumor may be palpated. Surgery is the treatment of choice. The operation is not usually an emergency, leaving sufficient time for supportive

treatment to correct any dehydration or electrolyte imbalance beforehand. Fluids, electrolytes, and blood replacement may be necessary, depending on the condition of the infant. The surgery is usually performed laparoscopically.

Gastric lavage, from 1 to 2 hours before operation, may be done until returns are clear. Maintaining body heat before and after surgery is essential.



Omphaloceles

Occurs in 1 in 5000 births, in which an amount of abdominal contents protrudes at the base of the umbilicus.

Omphaloceles develop between the 8th and 10th week of fetal life. The mass is covered with a layer of peritoneum and amnion and may rupture at delivery.

Omphaloceles are often seen in conjunction with other cardiac, genitourinary, and extra intestinal anomalies. Treatment requires covering the mass with sterile gauze soaked in saline, and placement, and immediate total or staged surgical repair. Sepsis is a serious and potential complication.



Abdominal viscera herniating into base of umbilicus (omphalocele)



Occurs 1 in 4,000 births or less. The lesion is not covered. With membrane, and the umbilical cord protrudes lateral to the defect in the abdominal wall. Defects are fixed by primary closure after emptying the bowel contents or the defect is placed in a plastic sterile bag. The bag is placed by gravity from the KDC warmer and reduced on a daily basis. Once the defect is close to being completely back in the abdominal space the bag is removed and the abdominal wall closed. If the child does not have enough skin for closure, a patch may be put into place until it can be closed on a later date.

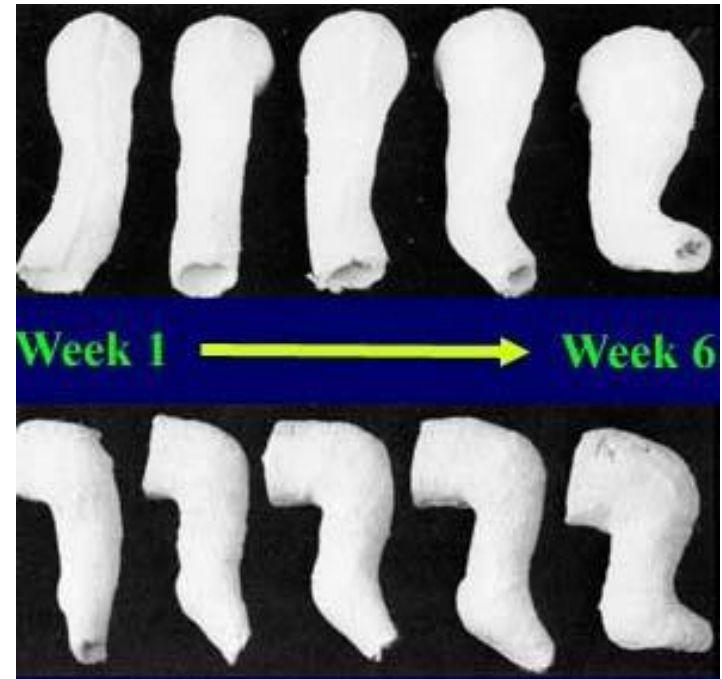


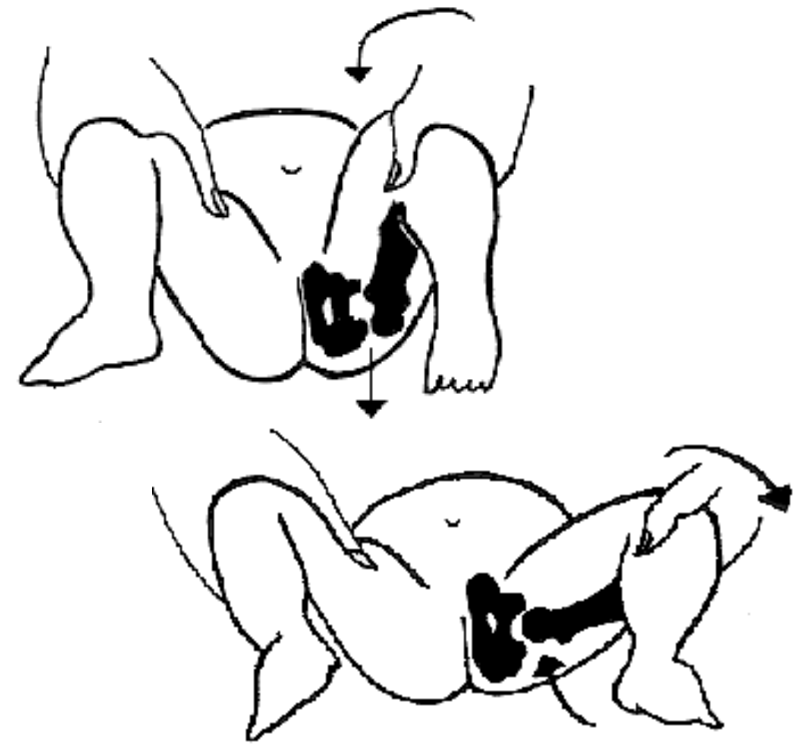
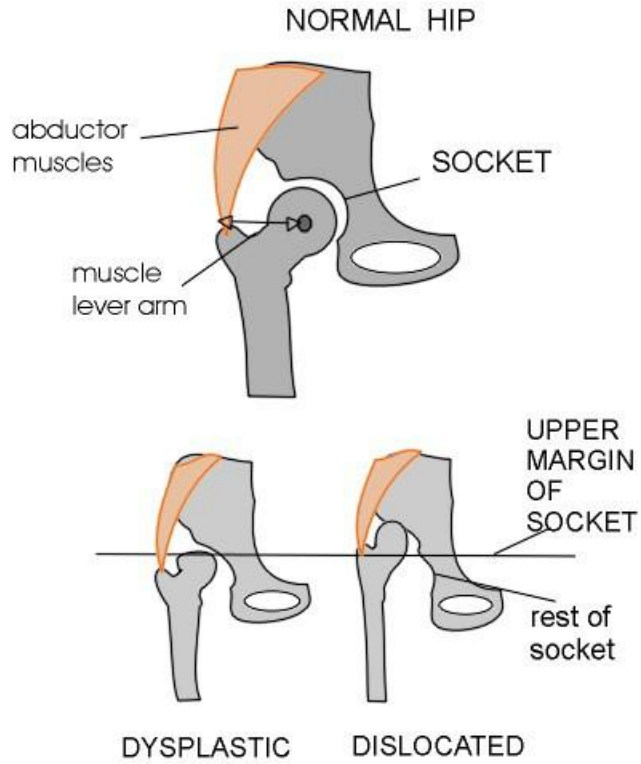
Intestine protruding through abdominal wall defect

ADAM.

GASTROSCHISIS

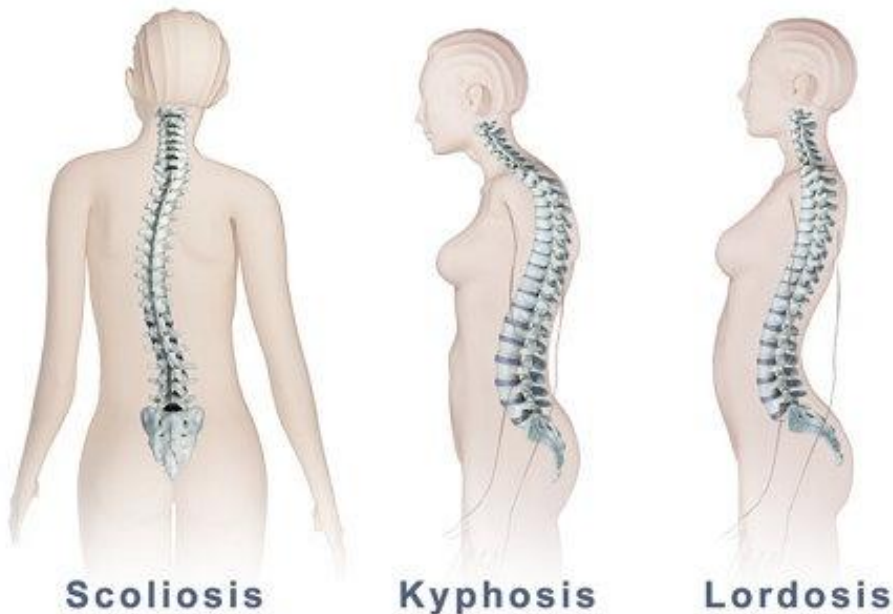
Talipes Equinosvarus (Club Foot) 1:5,000 births





Hip Dysplasia
 Checked with Barlow's and
 Ortolani's maneuvers

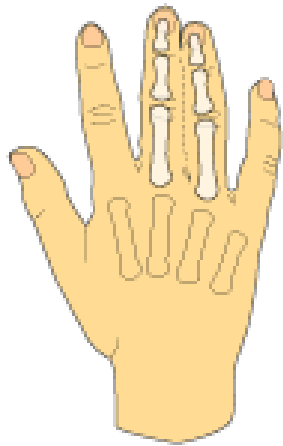
Scoliosis



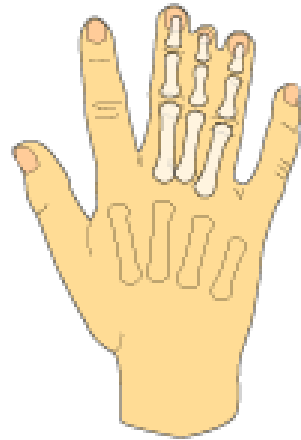
Scoliosis is a lateral (toward the side) curvature in the normally straight vertical line of the spine. When viewed from the side, the spine should show a mild roundness in the upper back and shows a degree of swayback (inward curvature) in the lower back. When a person with a normal spine is viewed from the front or back, the spine appears to be straight. When a person with scoliosis is viewed from the front or back, the spine appears to be curved.

Approximately 2% to 3% of Americans at age 16 have scoliosis. Less than 0.1% have spinal curves measuring greater than 40 degrees, which is the point at which surgery becomes a consideration.

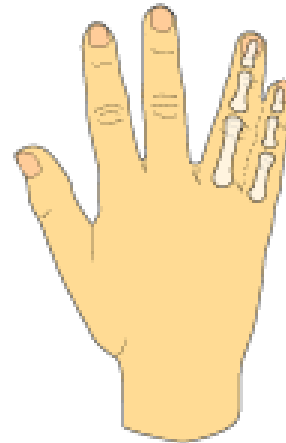
Polydactyly (extra digits)



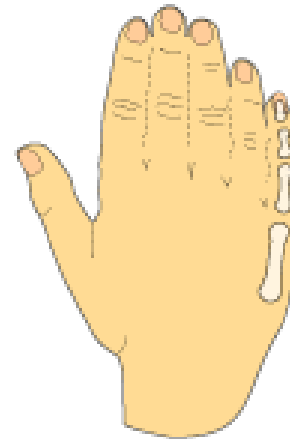
-Type I
-Common type
-2-3/10,000 newborns
-Also called zygodactyly
-2 Loci: 2q34; 3p21.31



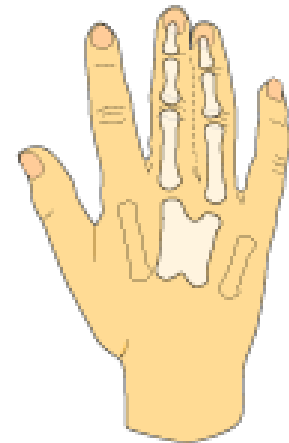
-Type II
-Also called synpolydactyly
-HOXD13 mutations
-2q31-32



-Type III
-Connexin 43 mutations (GJA1 gene)
In Oculo-dento-digital dysplasia type
-6q21



-Type IV
-Rare
-locus



-Type V
-Rare
-HOXD13 (some cases)

Types of Craniosynostosis

The types of craniosynostosis depend on what sutures join together early.

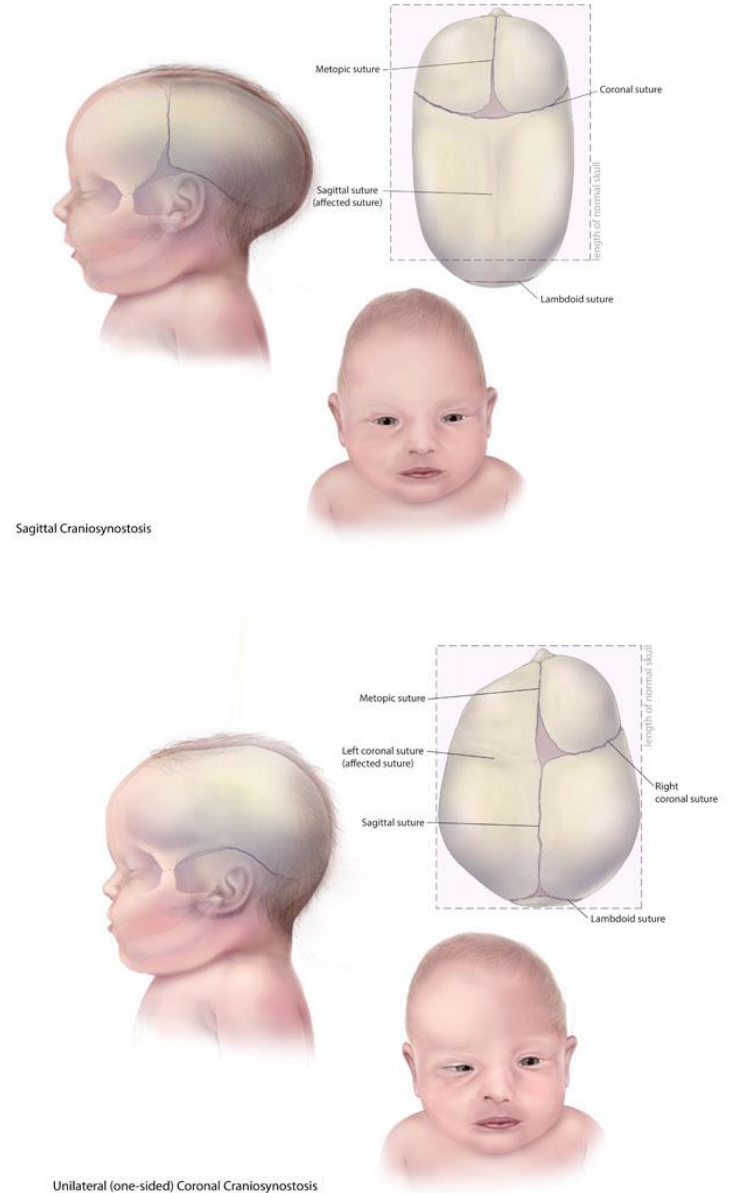
Sagittal synostosis— The sagittal suture runs along the top of the head, from the baby's soft spot near the front of the head to the back of the head. When this suture closes too early, the baby's head will grow long and narrow (scaphocephaly). It is the most common type of craniosynostosis.

Coronal synostosis – The right and left coronal sutures run from each ear to the sagittal suture at the top of the head. When one of these sutures closes too early, the baby may have a flattened forehead on the side of the skull that closed early (anterior plagiocephaly). The baby's eye socket on that side might also be raised up and his or her nose could be pulled toward that side. This is the second most common type of craniosynostosis.

Bicoronal synostosis – This type of craniosynostosis occurs when the coronal sutures on both sides of the baby's head close too early. In this case, the baby's head will grow broad and short (brachycephaly).

Lambdoid synostosis – The lambdoid suture runs along the backside of the head. If this suture closes too early, the baby's head may be flattened on the back side (posterior plagiocephaly). This is one of the rarest types of craniosynostosis.

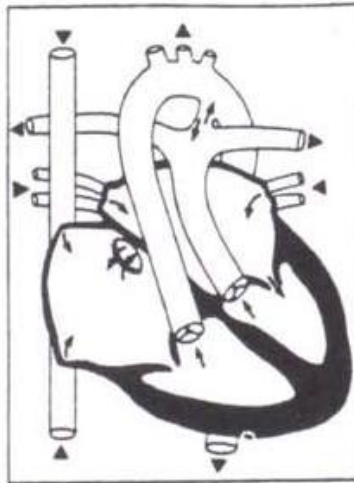
Metopic synostosis – The metopic suture runs from the baby's nose to the sagittal suture at the top of the head. If this suture closes too early, the top of the baby's head shape may look triangular, meaning narrow in the front and broad in the back (trigonocephaly). This is one of the rarest types of craniosynostosis.



Congenital Heart Anomalies

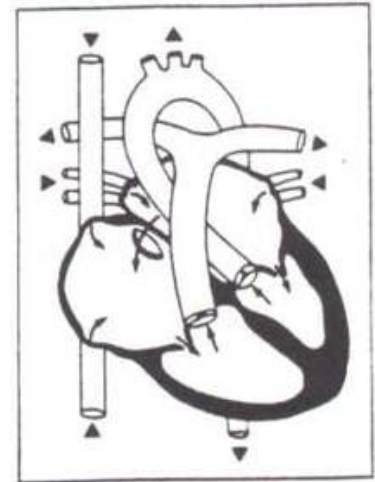
Transposition of the great arteries

This anomaly is an embryologic defect caused by a straight division of the bulbar trunk without normal spiraling. As a result, the aorta originates from the right ventricle, and the pulmonary artery from the left ventricle. An abnormal communication between the two circulations must be present to sustain life.



Atrial septal defect

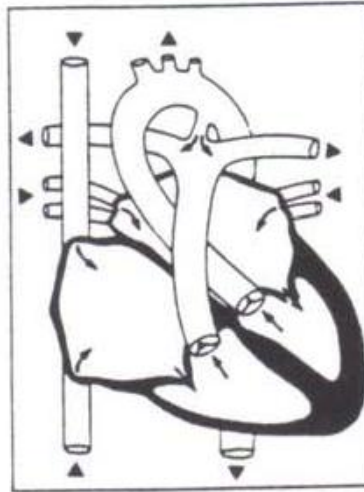
An atrial septal defect is an abnormal opening between the right and left atria. Basically, three types of abnormalities result from incorrect development of the atrial septum. An incompetent foramen ovale is the most common defect. The high ostium secundum defect results from abnormal development of the septum secundum. Improper development of the septum primum produces a basal opening known as an ostium primum defect, frequently involving the atrioventricular valves. In general, left to right shunting of blood occurs in all atrial septal defects.



Congenital Heart Anomalies

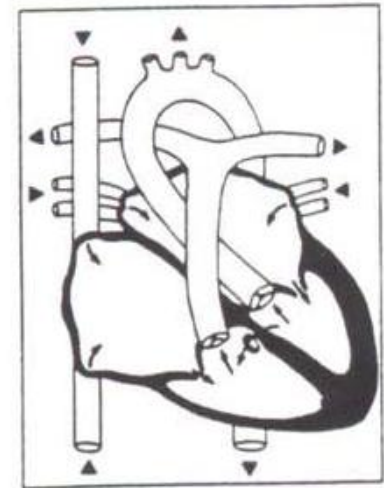
Patent ductus arteriosus

The patent ductus arteriosus is a vascular connection that, during fetal life, short circuits the pulmonary vascular bed and directs blood from the pulmonary artery to the aorta. Functional closure of the ductus normally occurs soon after birth. If the ductus remains patent after birth, the direction of blood flow in the ductus is reversed by the higher pressure in the aorta.



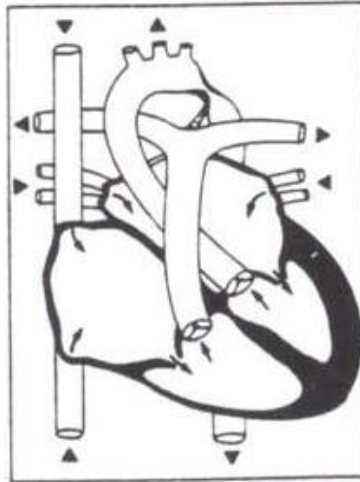
Ventricular septal defect

A ventricular septal defect is an abnormal opening between the right and left ventricle. Ventricular septal defects vary in size and may occur in either the membranous or muscular portion of the ventricular septum. Due to higher pressure in the left ventricle, a shunting of blood from the left to right ventricle occurs during systole. If pulmonary vascular resistance produces pulmonary hypertension, the shunt of blood is then reversed from the right to the left ventricle resulting in cyanosis.



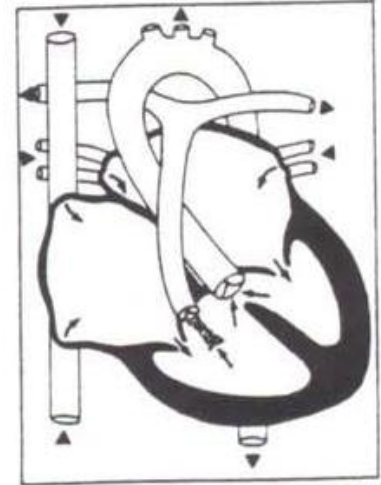
Coarctation of the aorta

Coarctation of the aorta is characterized by a narrowed aortic lumen. It exists as a preductal or postductal obstruction, depending on the position of the obstruction in relation to the ductus arteriosus. Coarctations exist with great variation in anatomical features. The lesion produces an obstruction to the flow of blood through the aorta causing an increased left ventricular pressure and work load.



Tetralogy of Fallot

Tetralogy of Fallot is characterized by the combination of four defects—(1) pulmonary stenosis, (2) ventricular septal defect, (3) overriding aorta, and (4) hypertrophy of right ventricle. It is the most common defect causing cyanosis in patients surviving beyond two years of age. The severity of symptoms depends on the degree of pulmonary stenosis, the size of the ventricular septal defect, and the degree to which the aorta overrides the septal defect.

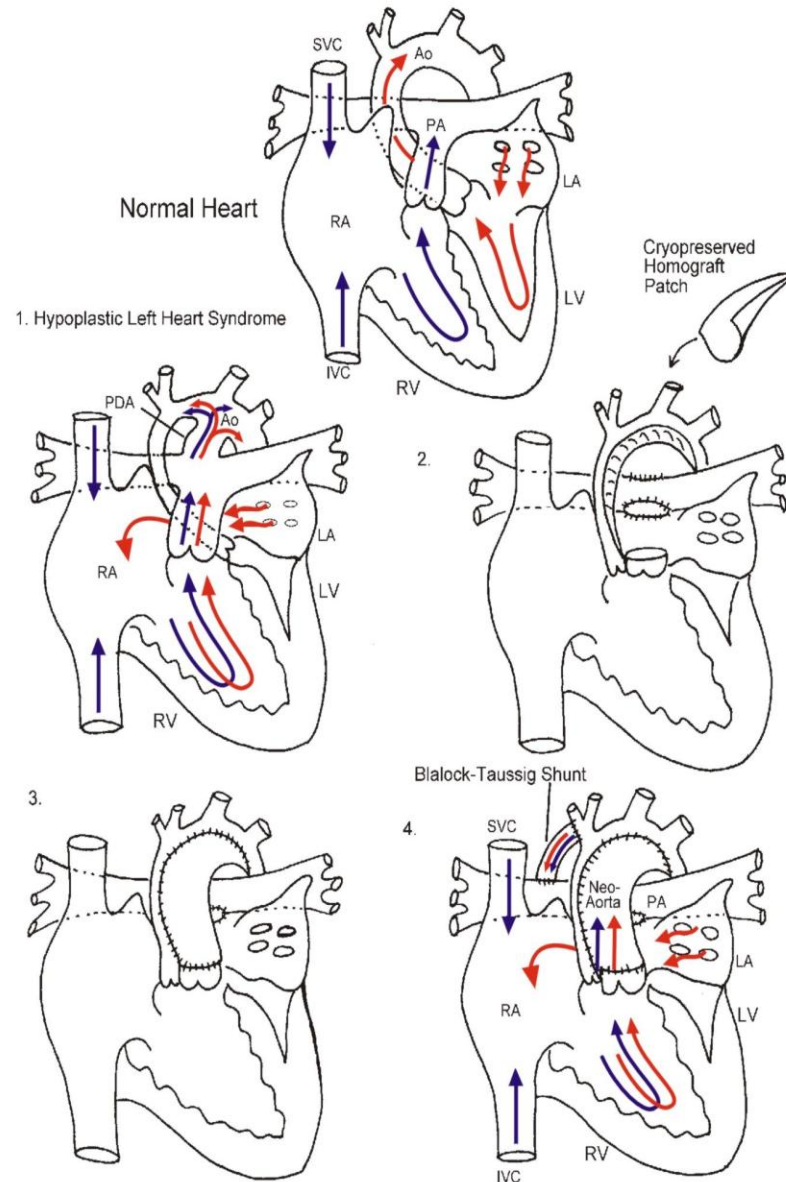


Congenital Heart Anomalies

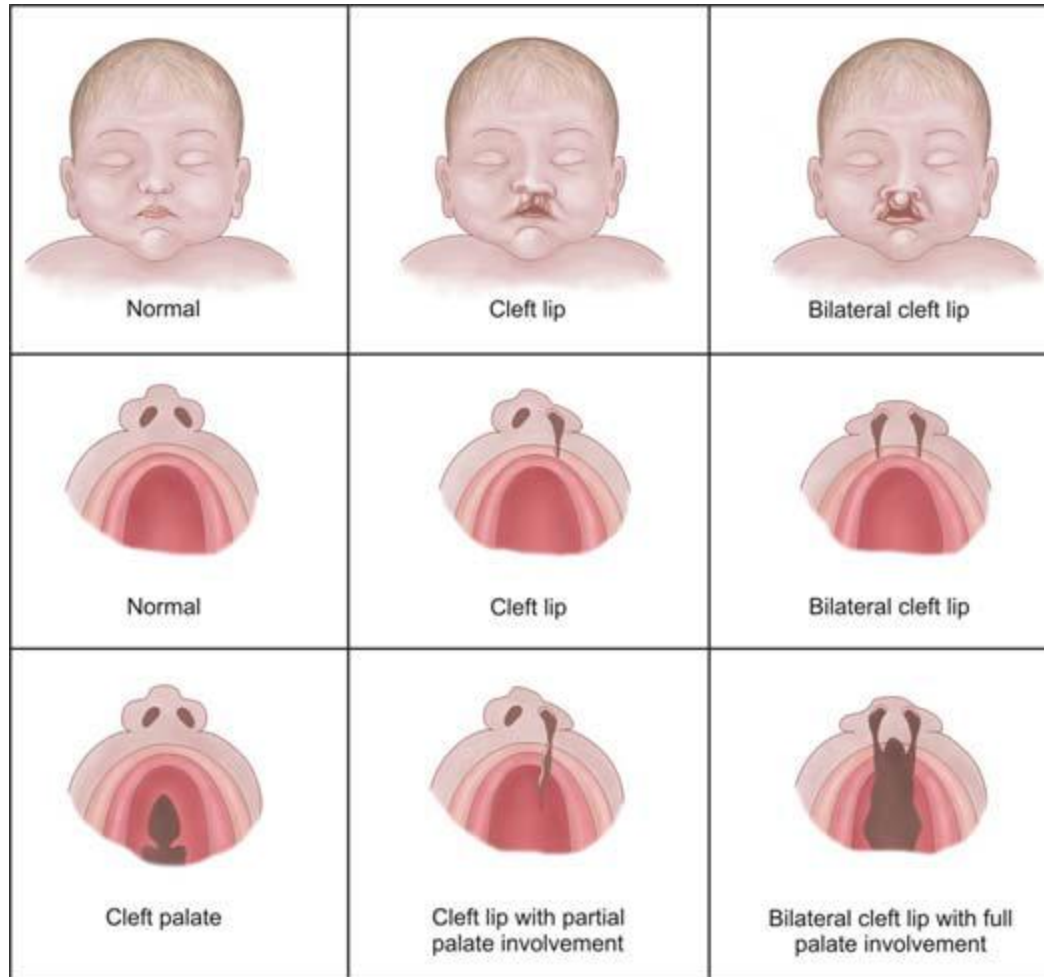
Congenital Heart Anomalies

HLHS
Norwood
BDG
Fontan

Norwood Procedure for Hypoplastic Left Heart Syndrome



Cleft Lip and Palate



Hypospadias, Epispadias, Ambiguous Genitalia

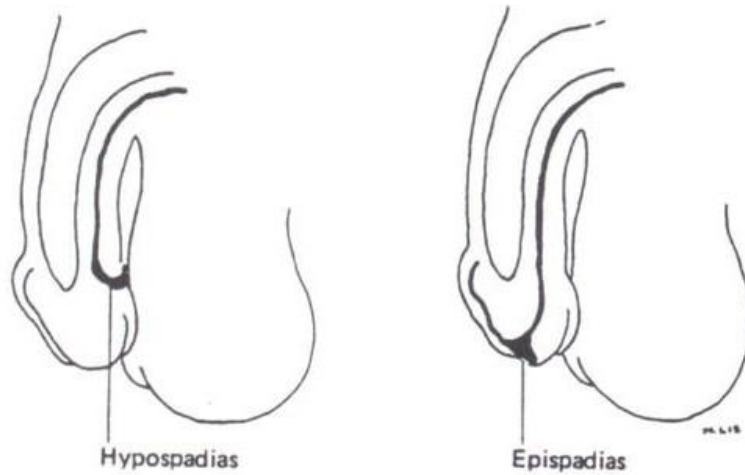


FIGURE 45-5

Illustration of hypospadias and epispadias.



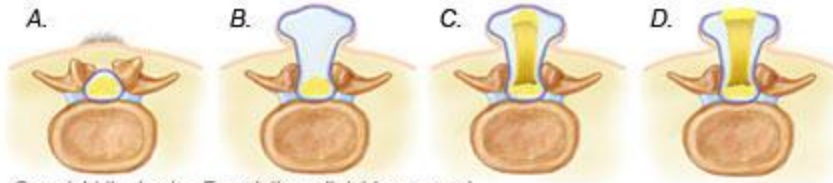
Spina Bifida: Myelomeningocele

Cyst on baby's back from spina bifida

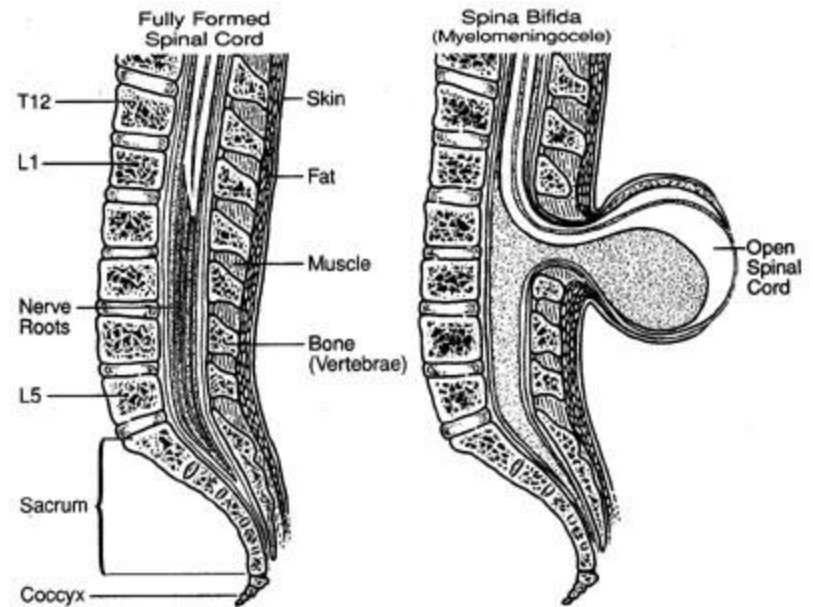


Normal newborn vertebra

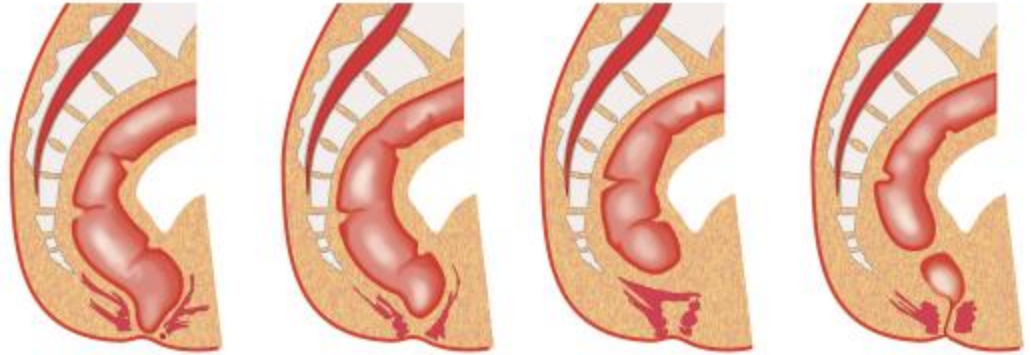
- A. Spina bifida occulta
- B. Spina bifida with meningocele
- C. Spina bifida with meningocele
- D. Spina bifida with myeloschisis



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A



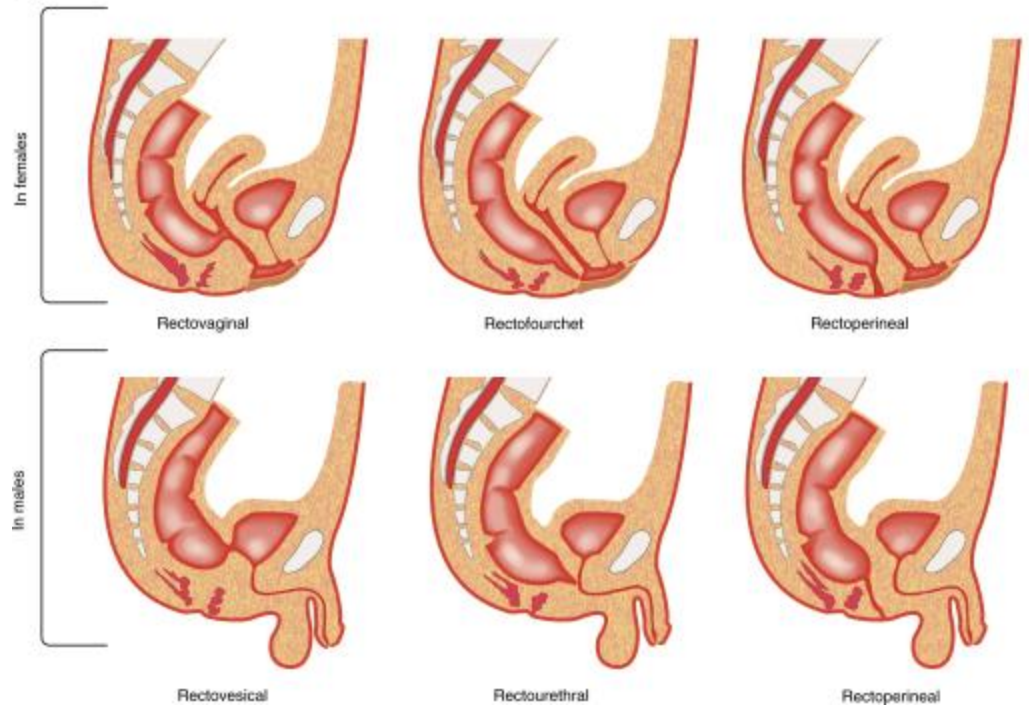
Type 1. Just a thin membrane over the anus

Type 2. Pouch is ≤ 1.5 cm from the anal dimple

Type 3. A blind pouch > 1.5 cm from the anal dimple

Type 4. Atresia of the rectum with a normal anus

B



In females

Rectovaginal

Rectofourchet

Rectoperineal

In males

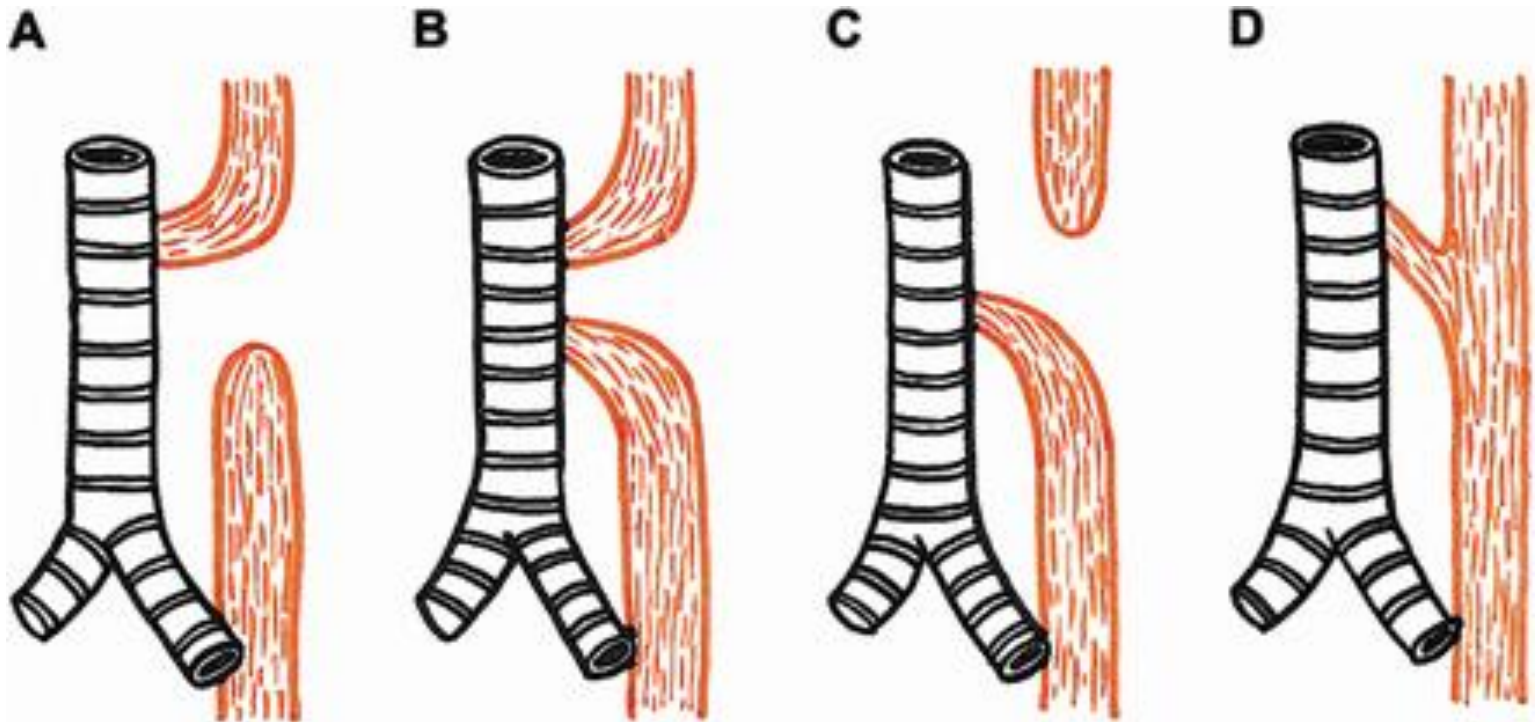
Rectovesical

Rectourethral

Rectoperineal

Imperforate Anus

Esophageal Atresia



Questions?

