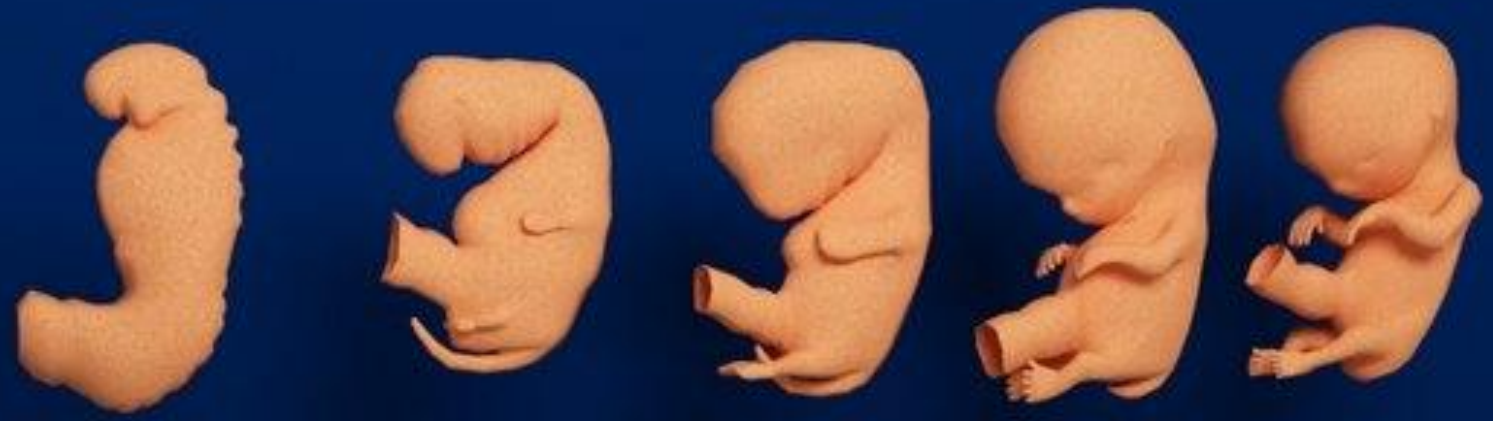
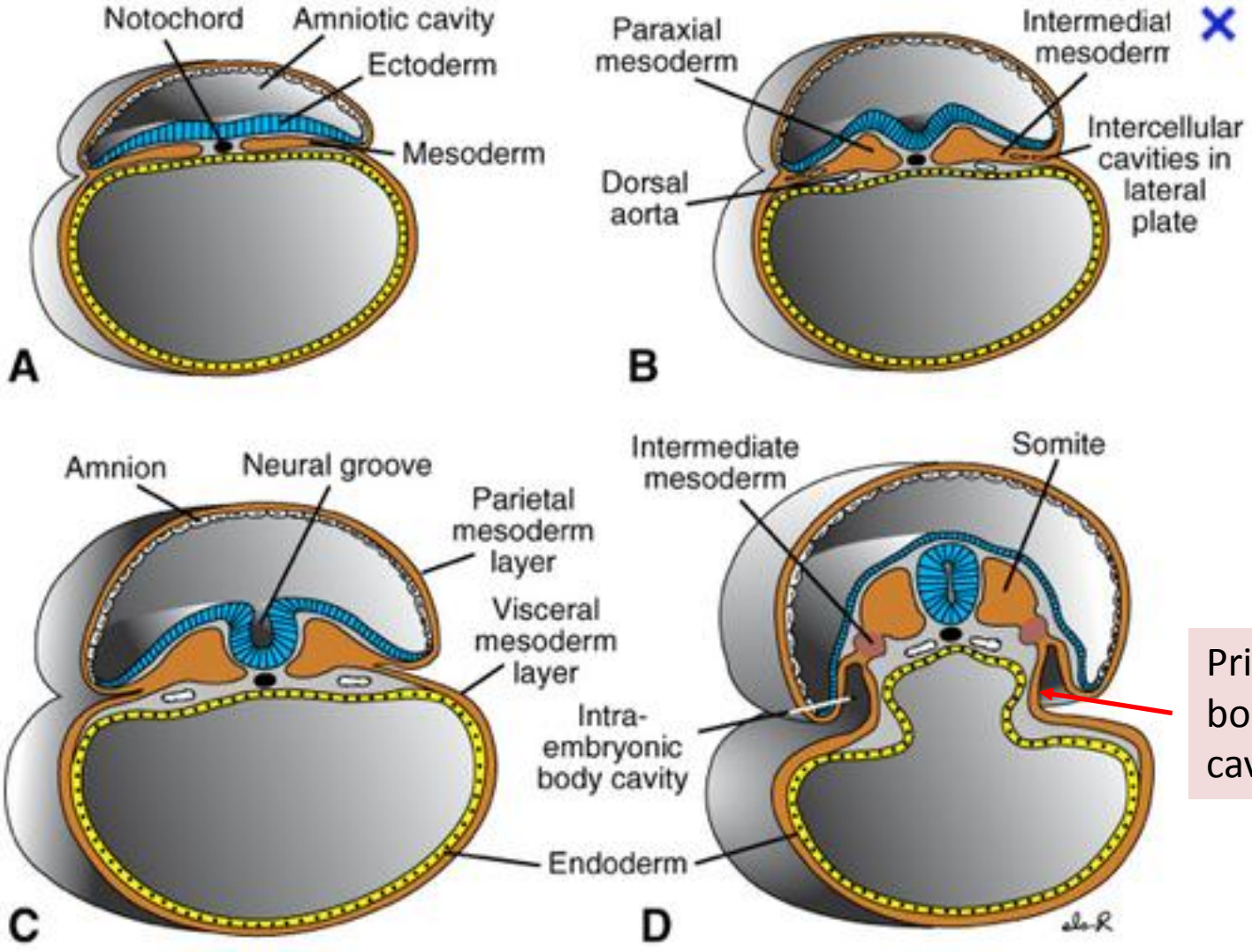
embryology



Clinical correlation

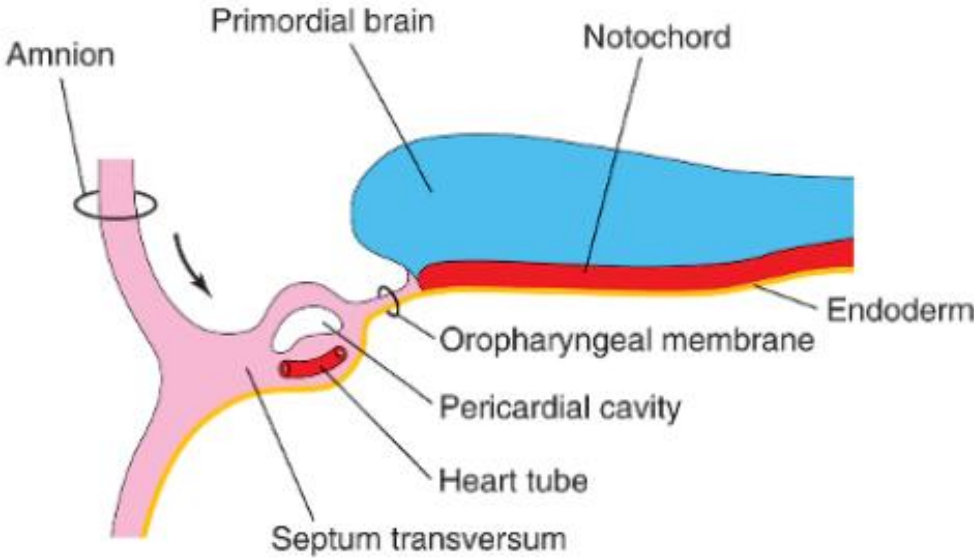
summary

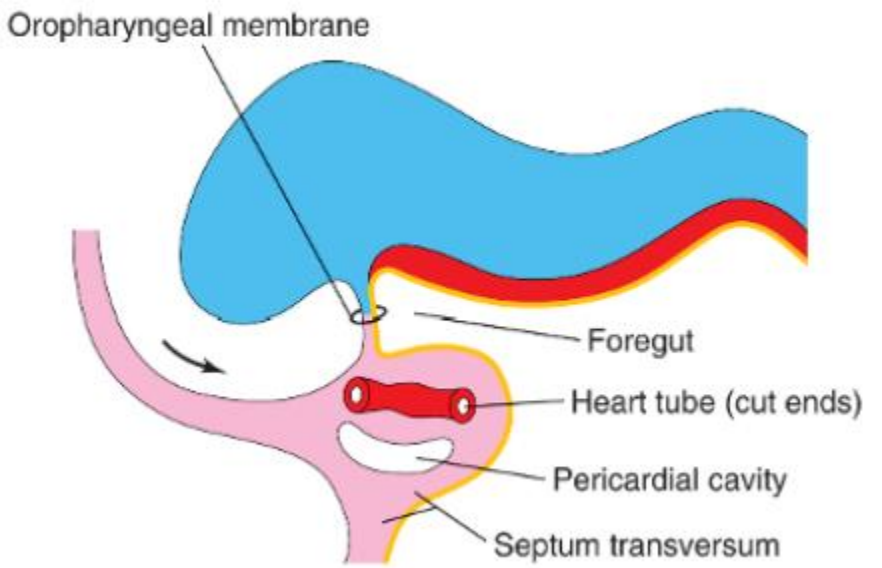


Primitive body cavity

The embryonic coelomic cavity gives rise to the serous cavities: pleural, peritoneal, and pericardial.

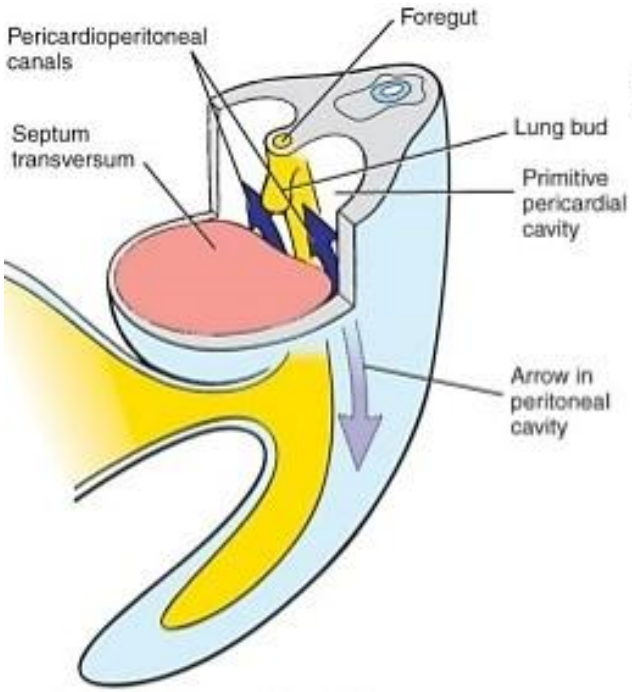
Longitudinal sections through the cranial half of human embryos during the **fourth week** of development.

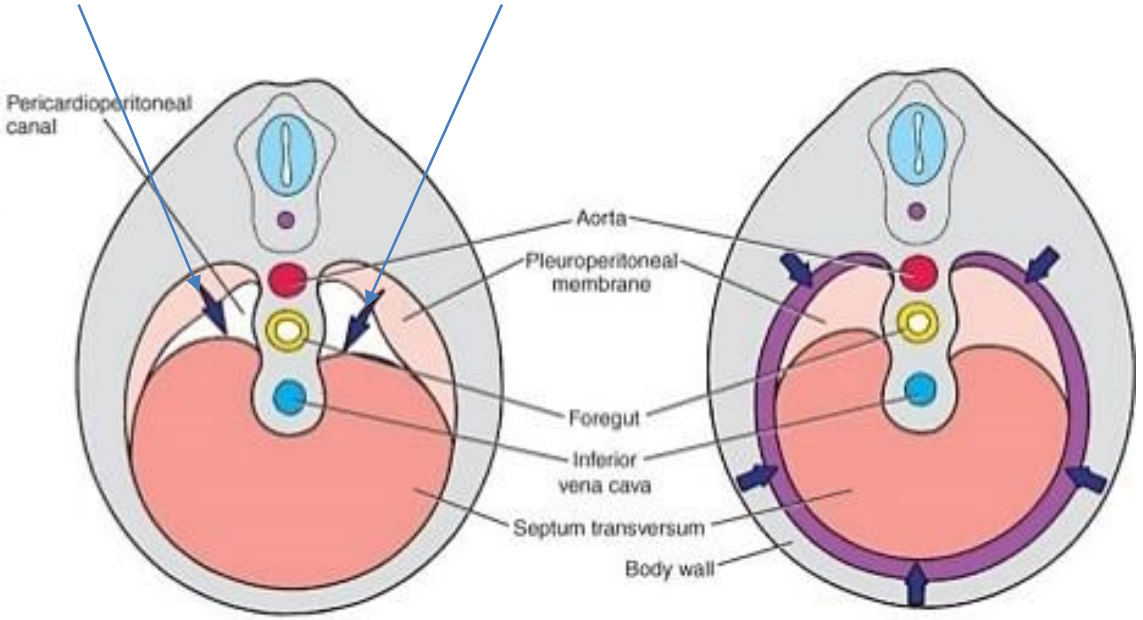


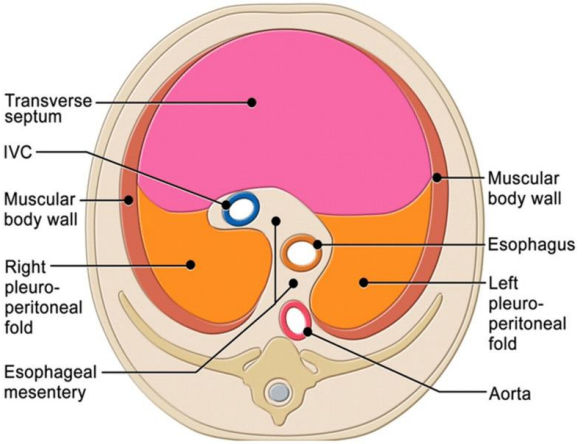


The **septum transversum** is a thick mass of cranial mesenchyme, formed in the embryo, that gives rise to parts of the thoracic diaphragm

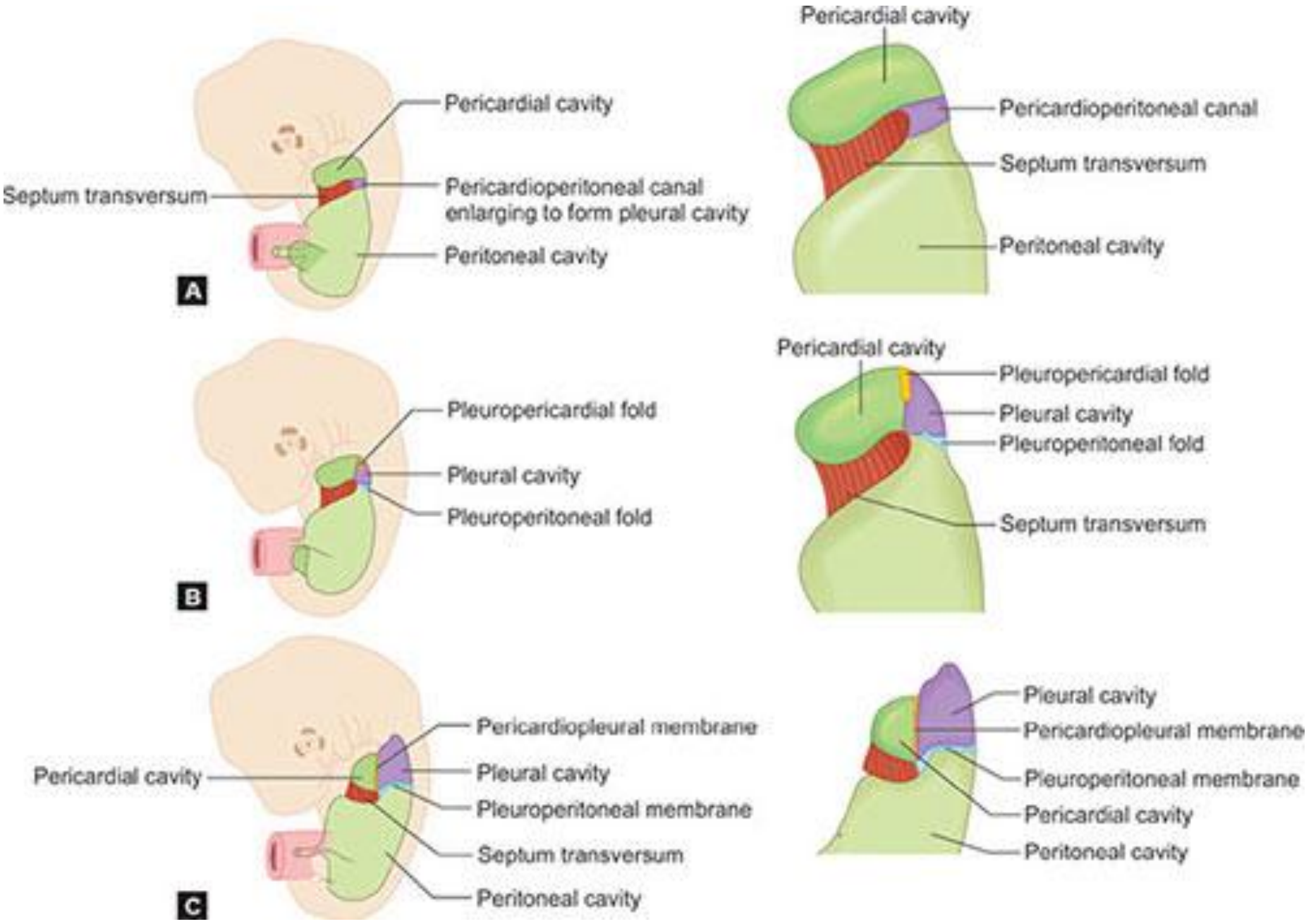
Pericardial-peritoneal channels



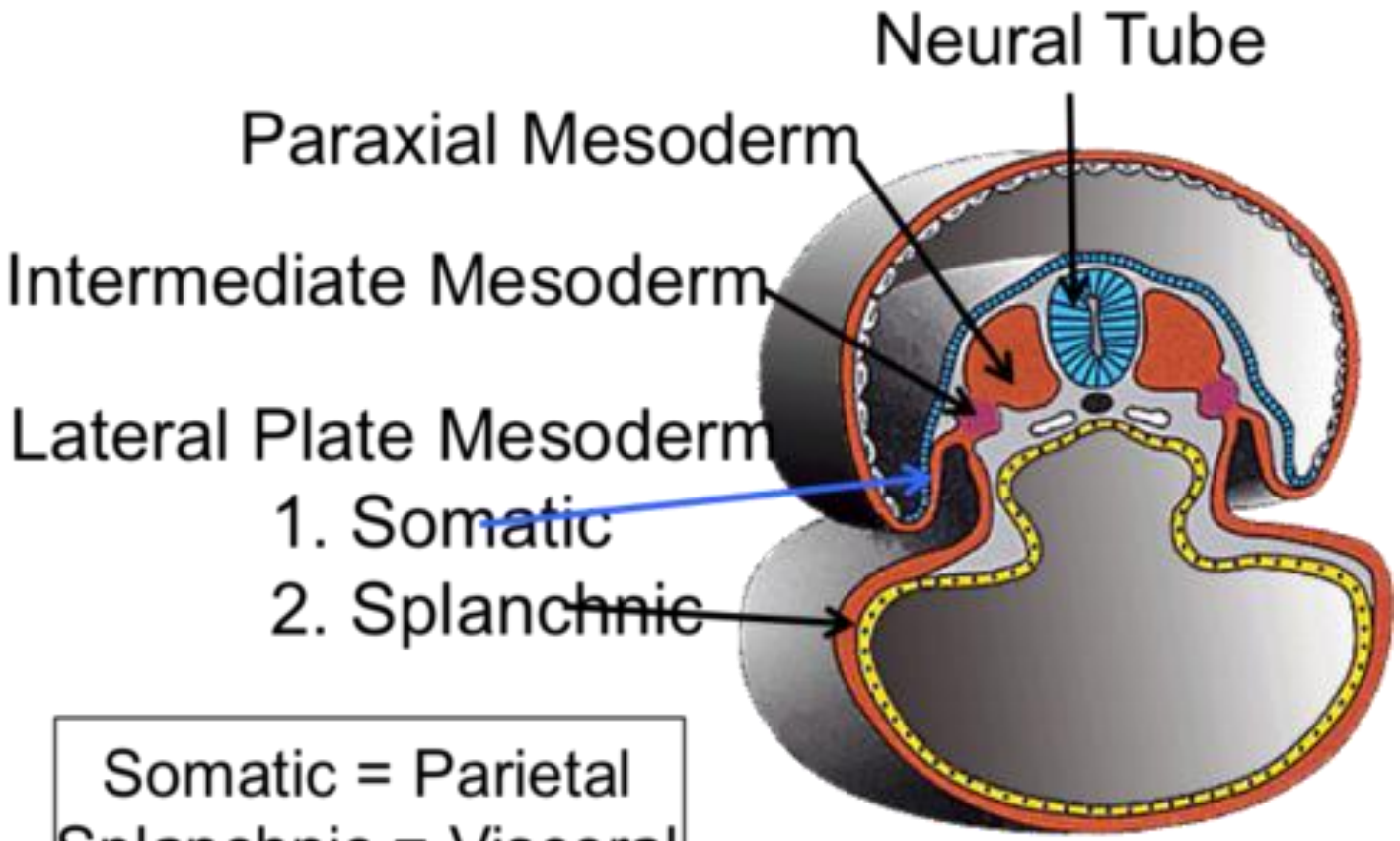




The diaphragm develops from a combination of embryonic structures: the septum transversum, pleuroperitoneal folds, dorsal mesentery of the oesophagus, and somites.

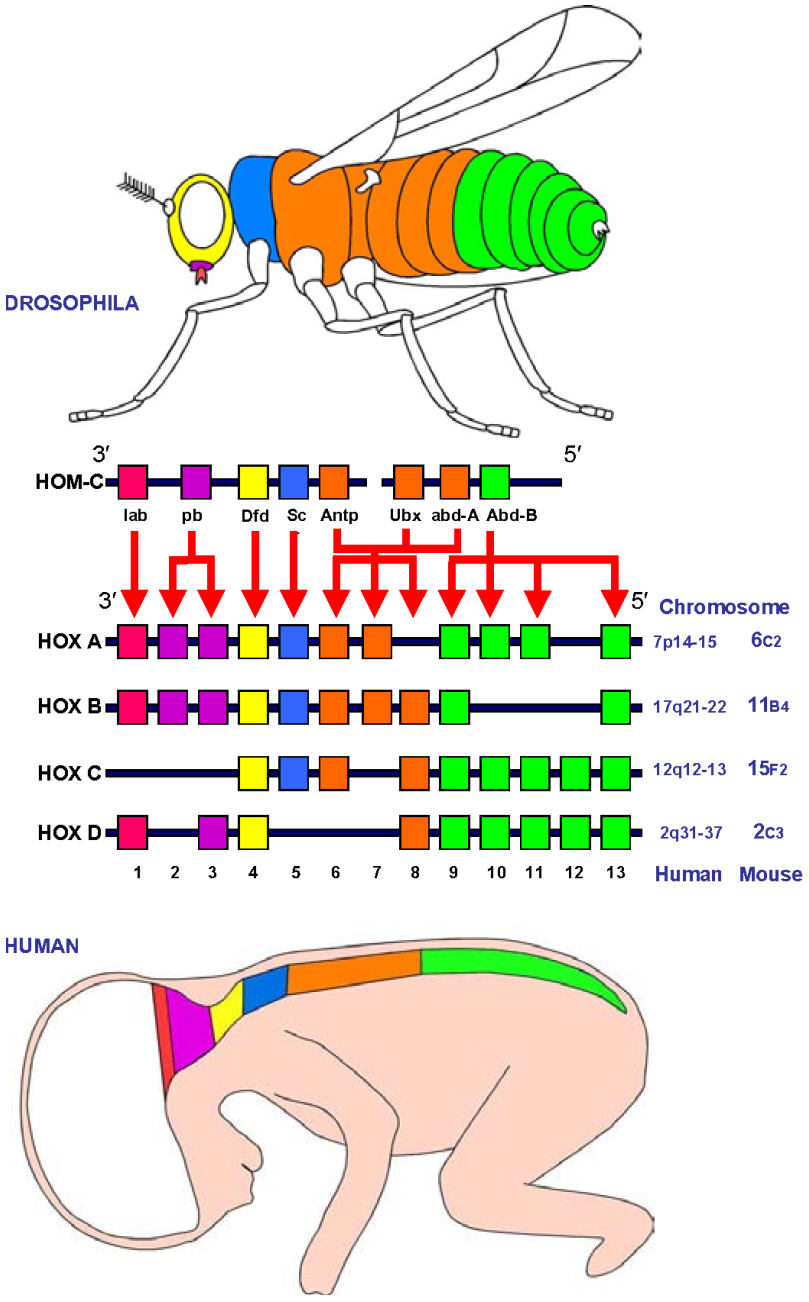

Pericardial cavity
Septum transversum
Pericardioperitoneal canal enlarging to form pleural cavity
Peritoneal cavity
A
Pericardial cavity
Pericardioperitoneal canal
Septum transversum
Peritoneal cavity
Pleuropericardial fold
Pleural cavity
Pleuroperitoneal fold
B
Pericardial cavity
Pleuropericardial fold
Pleural cavity
Pleuroperitoneal fold
Septum transversum
Pericardiopleural membrane
Pericardial cavity
Pleural cavity
Pleuroperitoneal membrane
Septum transversum
Peritoneal cavity
C
Pleural cavity
Pericardiopleural membrane
Pleuroperitoneal membrane
Pericardial cavity
Peritoneal cavity

Intraembryonic cavity is lined with lateral plate mesoderm



Serous membranes develop from parietal and visceral lateral plate mesoderm

Patterning of the anteroposterior axis: Regulation by homeobox genes



Hox genes are a group of highly conserved genes that encode transcription factors crucial for establishing the body plan along the anterior-posterior axis in bilateral animals. They play a key role in development by directing cell differentiation and tissue formation, essentially defining where structures will be located along the body.

Primordial germ cells and teratomas

Primordial germ cells (PGCs) are **derived from a population of pluripotent epiblast cells**

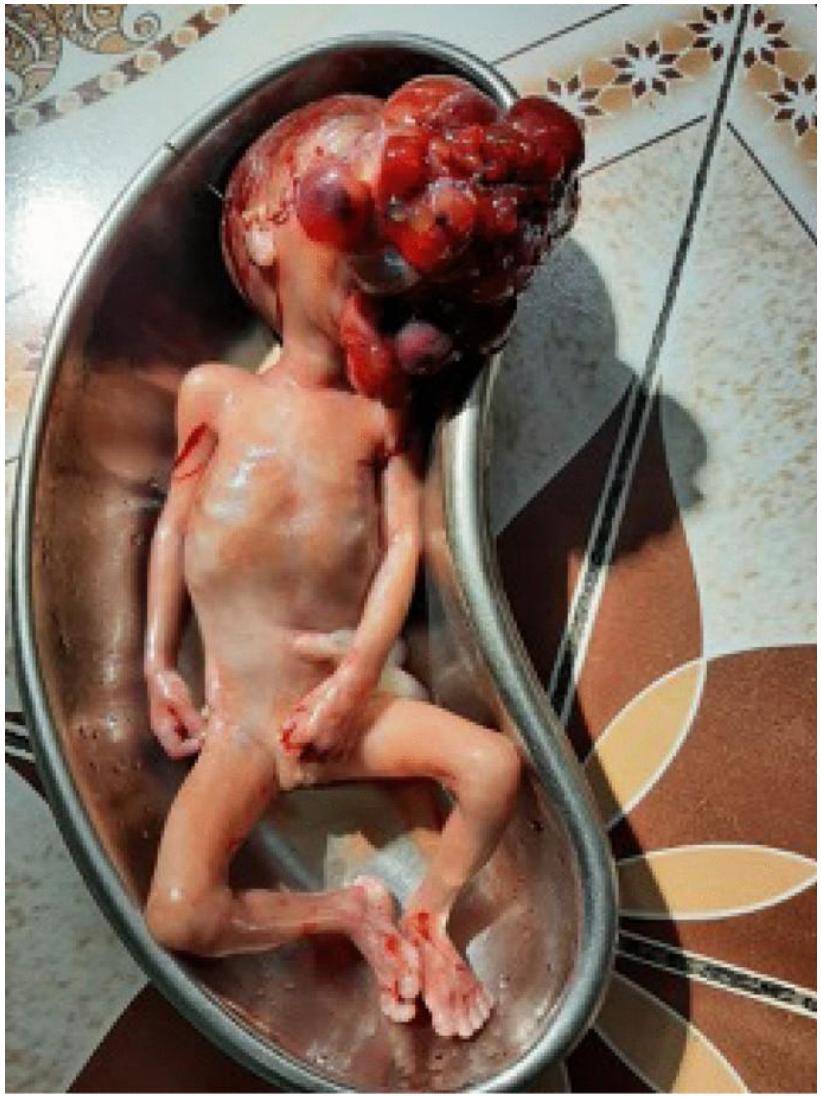
Teratomas are tumours consisting of **all three layers of germ cells** and account for around one-third of all neonatal tumours

They can be located along the midline of the body from the head to the pelvis, but occur most often in the **sacrococcygeal** region.

Remnants of the **primitive streak** can persist in the sacrococcygeal region

Teratomas can be diagnosed with the use of ultrasounds in utero.

Maternal serum AFP is a commonly used test during second trimester to assess a possible neural tube defect.



oropharyngeal teratoma

Fetal Med. 8, 237–241 (2021). https://doi.org/10.1007/s40556-021-00319-1

Chromosomal abnormalities

numerical

Trisomy 21 (Down syndrome)
Trisomy 18
Trisomy 13
Klinefelter syndrome
Turner syndrome
Triple X syndrome

Klinefelter syndrome 47XXY
Turner syndrome 45X
Triple X syndrome

abnormal number of sex chromosomes

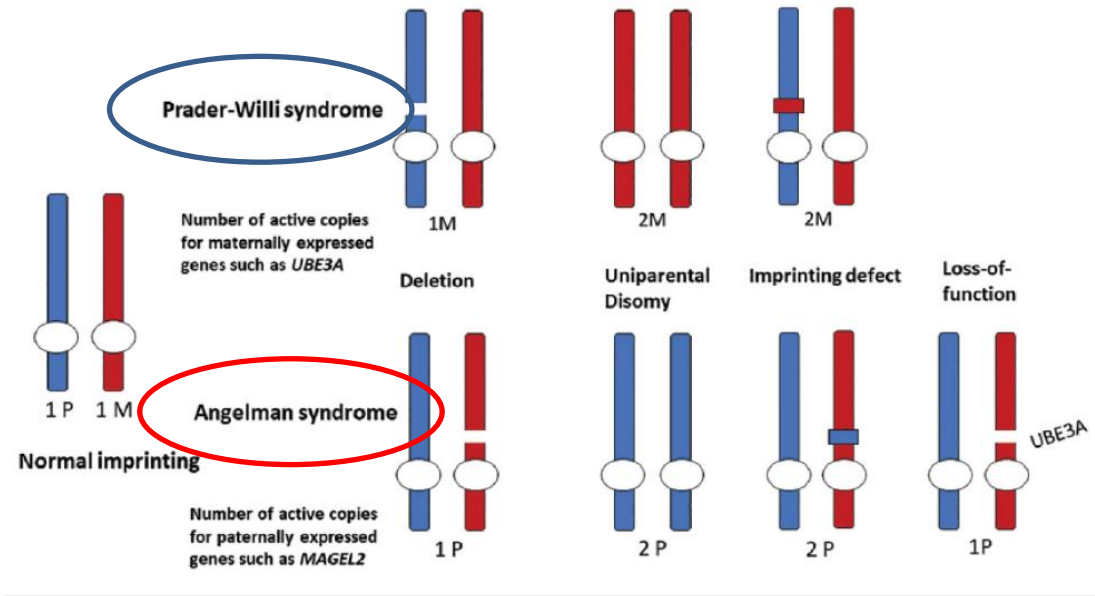
Klinefelter syndrome 47XXY
Turner syndrome 45X

Gonadal dysgenesis
dysgenesis is a genetic condition due to errors in cell division and or alterations in genetic material, leading to complete or partial loss of gonadal development.

47,XX,+21 karyotype
45,X/47,XY,+21 karyotype
Gonadal dysgenesis

Chromosomal abnormalities

structural



microdeletions

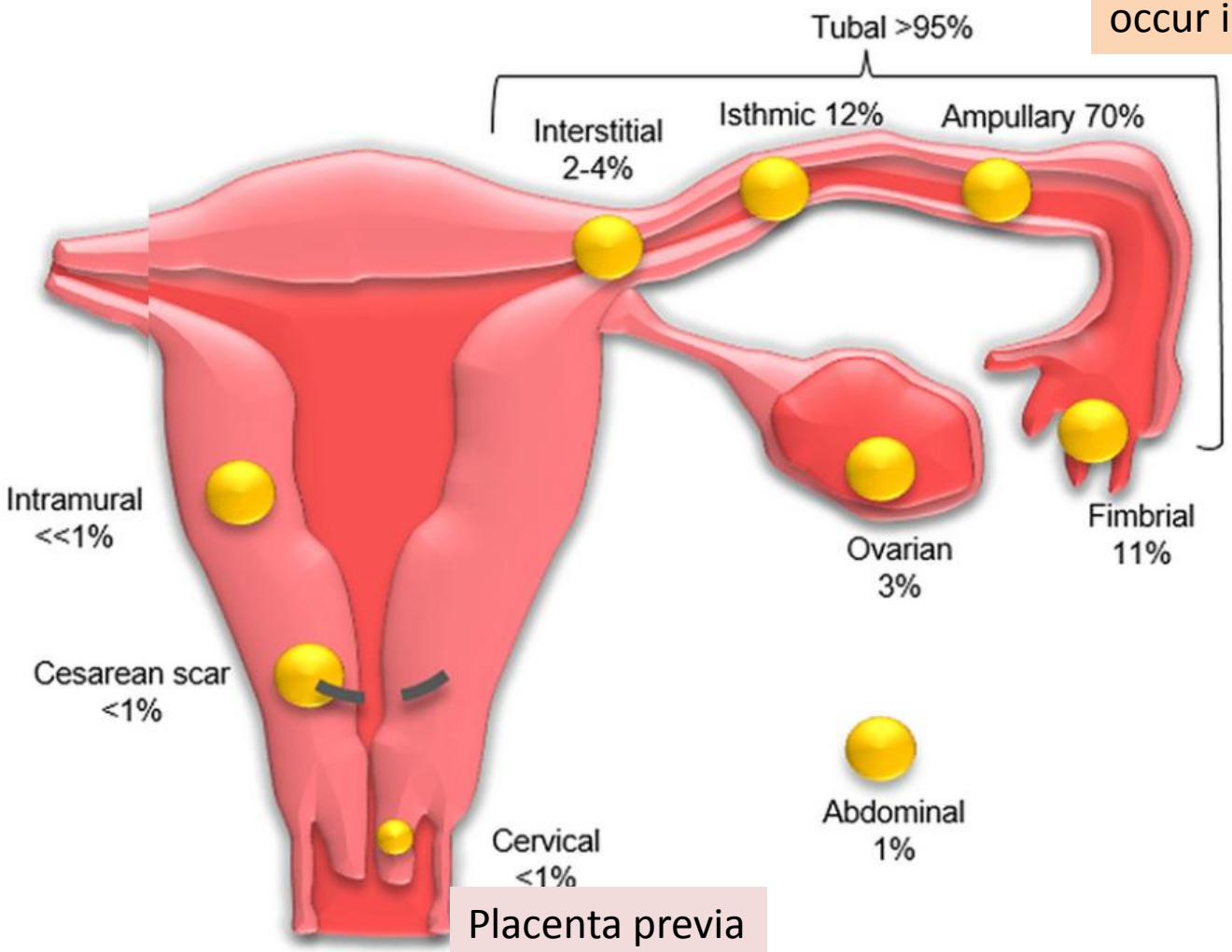
Genomic imprinting is the process by which only one copy of a gene in an individual (either from their mother or their father) is expressed

monoallelic inheritance

cri-du-chat syndrome results from missing a piece of chromosome number 5. The syndrome's name is based on the infant's cry, which is high-pitched and sounds like a cat.

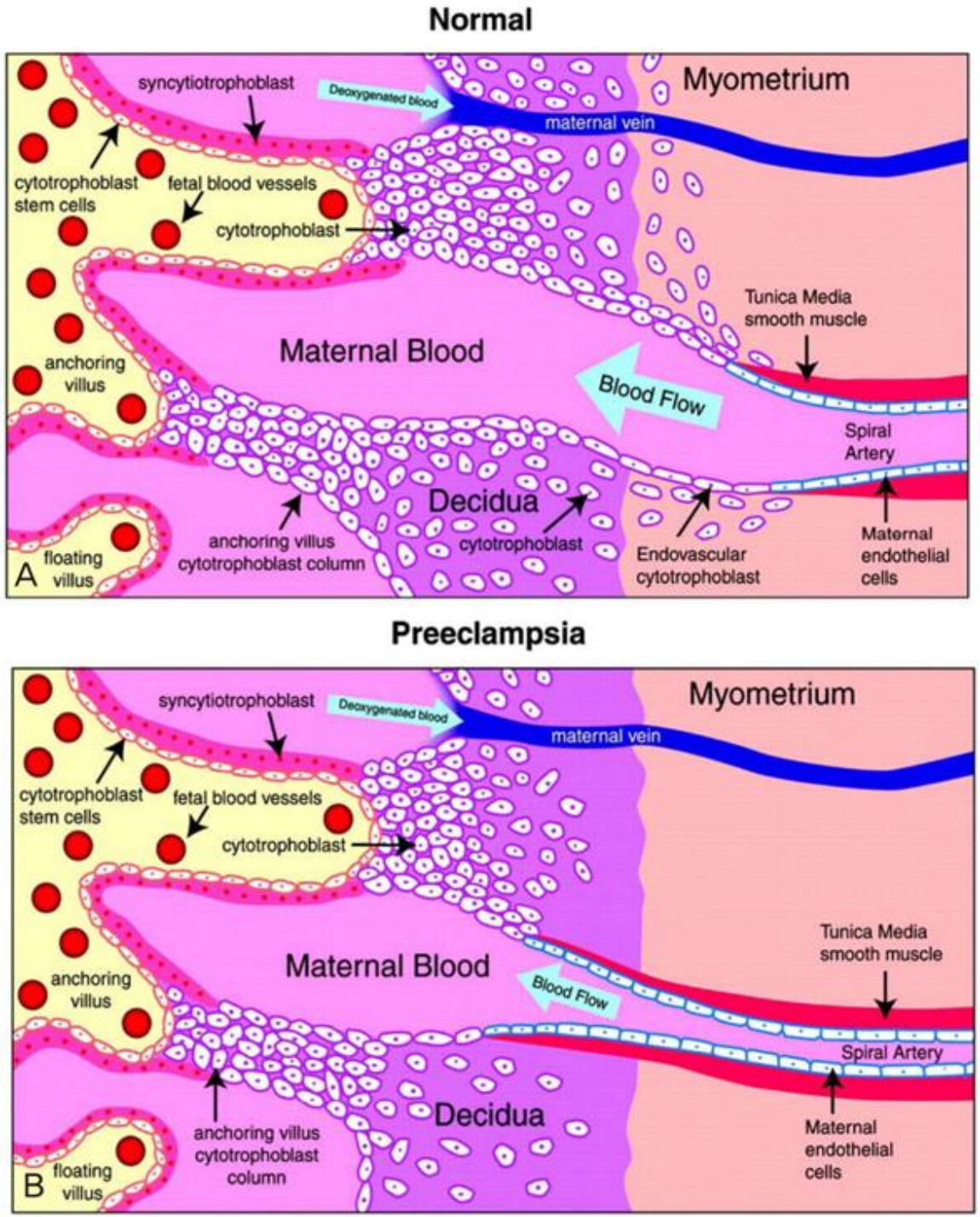
Abnormal implantation

Most frequently **AI** occur in the **ampulla**



Placenta previa

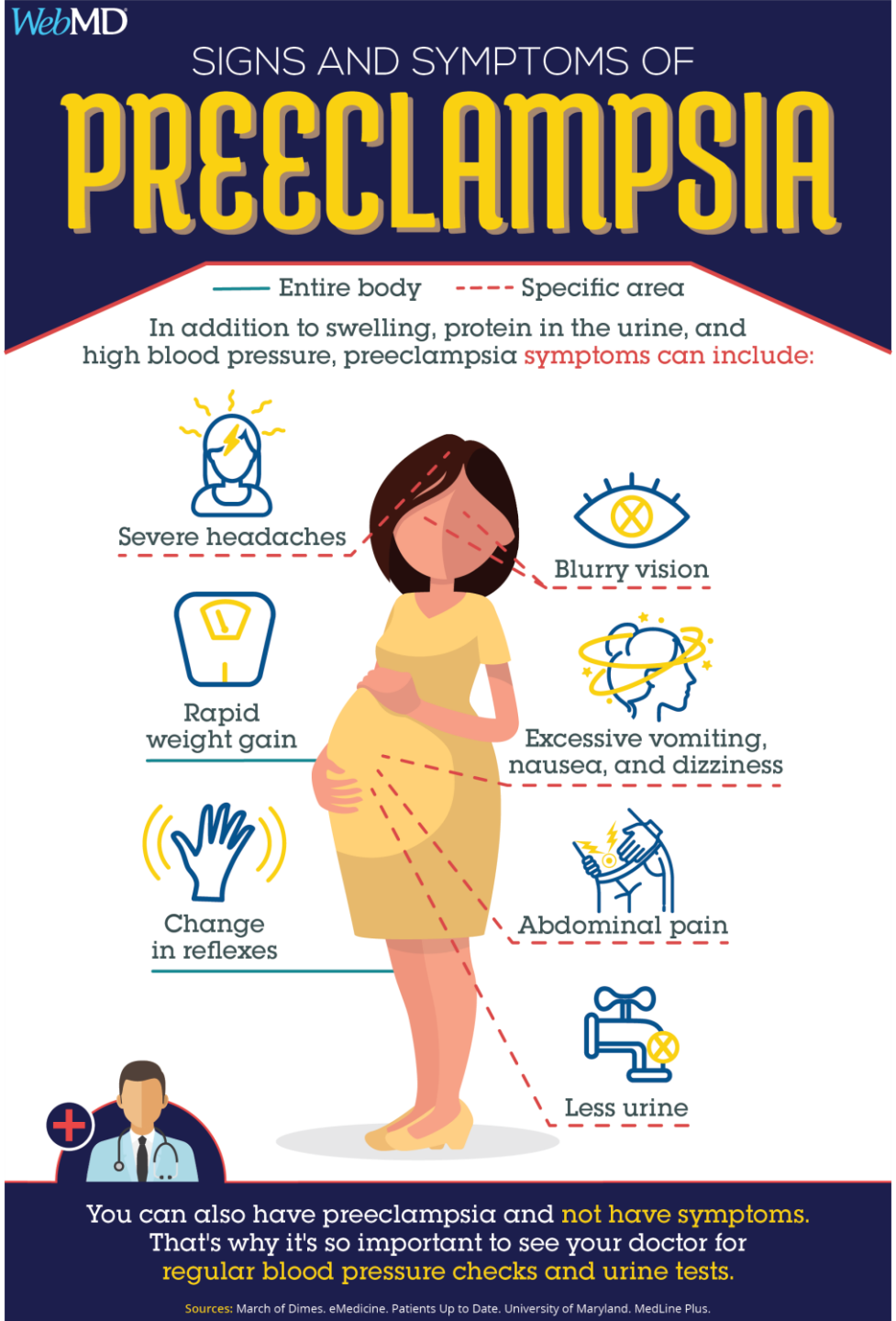
Normally - along the anterior or posterior wall of the body of the uterus



The developing placenta undergoes a process of vascular mimicry (referred to as pseudovasculogenesis) as cytotrophoblasts convert from an epithelial to an endothelial phenotype.

Pre-eclampsia is a condition that affects some **pregnant women**, usually during the second half of pregnancy (from 20 weeks) or soon after their baby is delivered

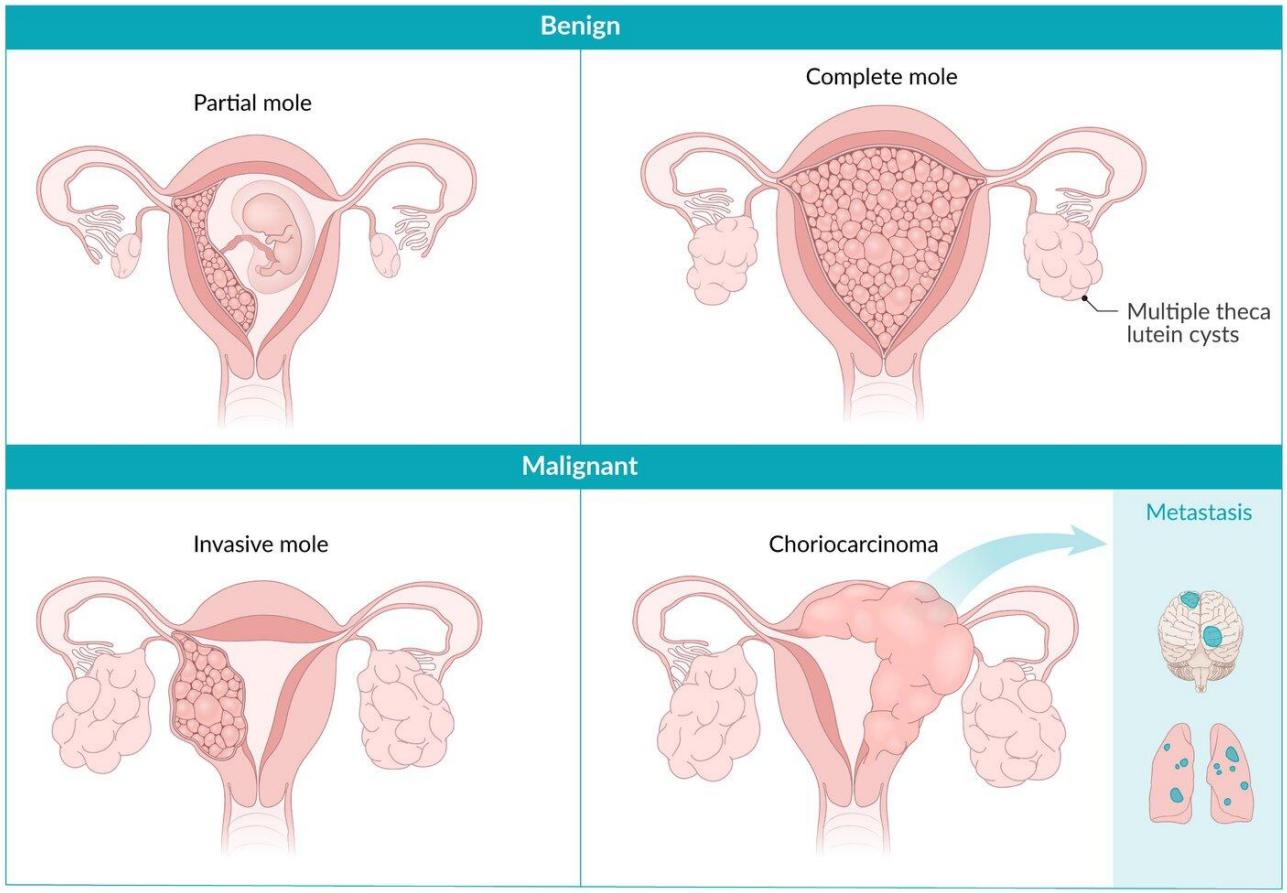
Early signs of **pre-eclampsia** include having high blood pressure (**hypertension**) and protein in urine (proteinuria)



Preeclampsia begins suddenly anytime from approximatel 20 weeks gestation to term.

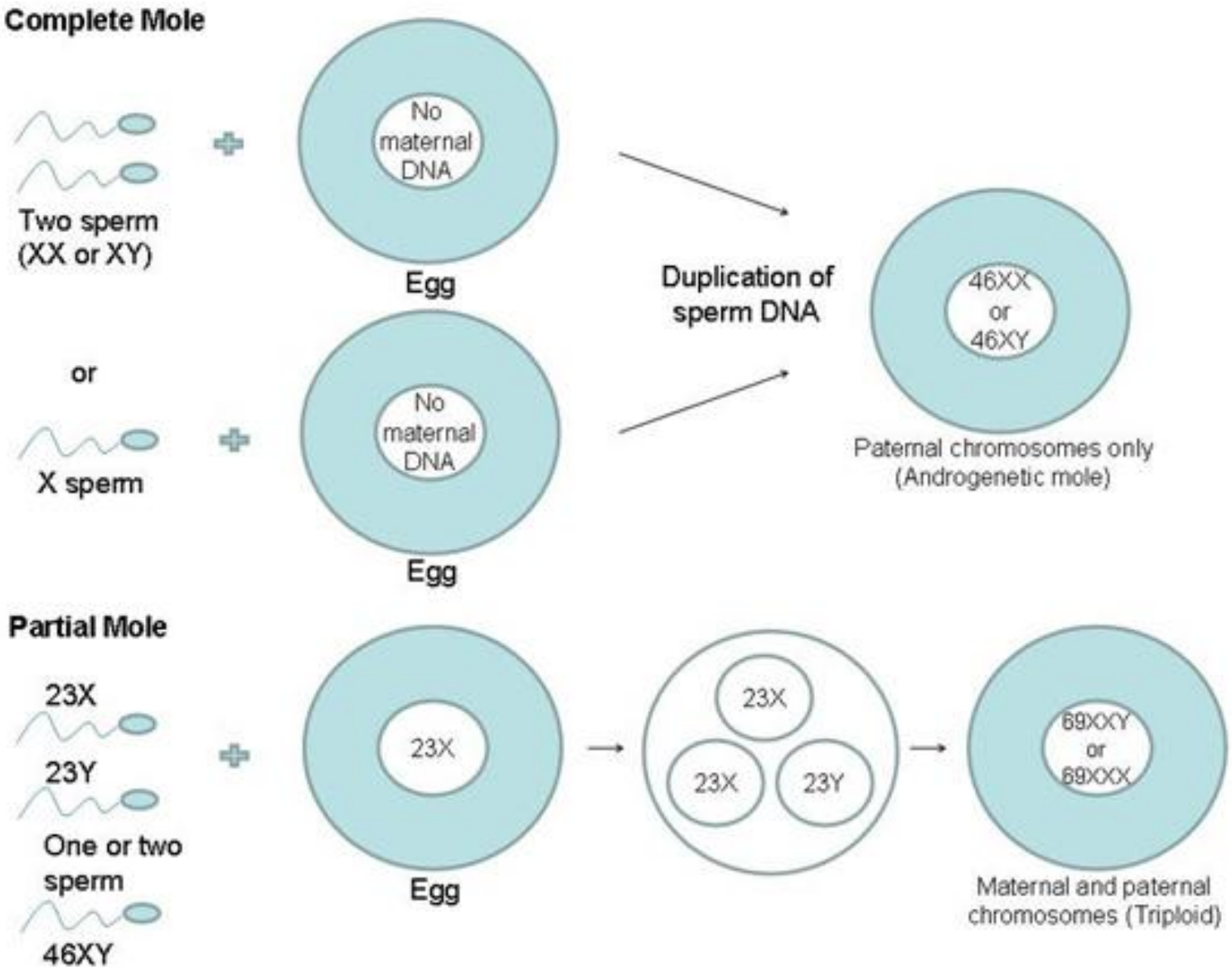
Preeclampsia commonly occurs in women with hydatidiform moles

Hydatidiform mole



Complete hydatidiform moles (CHM) are **abnormal pregnancies with no fetal development** resulting from having two paternal genomes with no maternal contribution.

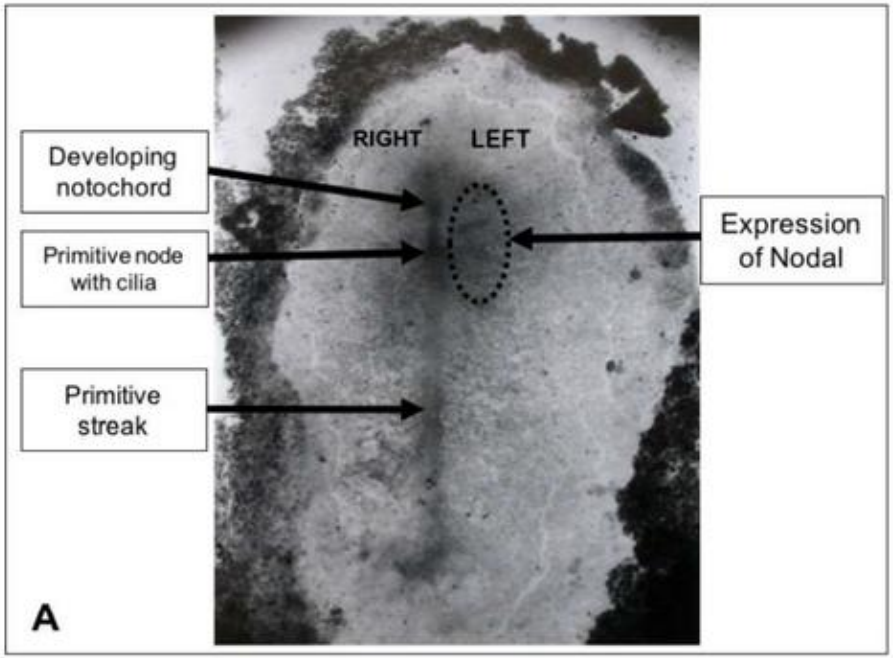
It involves unusual growth of trophoblasts
Little or no embryonic tissue is present
It secretes high leels of hCG
May produces benign or malignant (inasive mole, choriocarcinoma) tumors

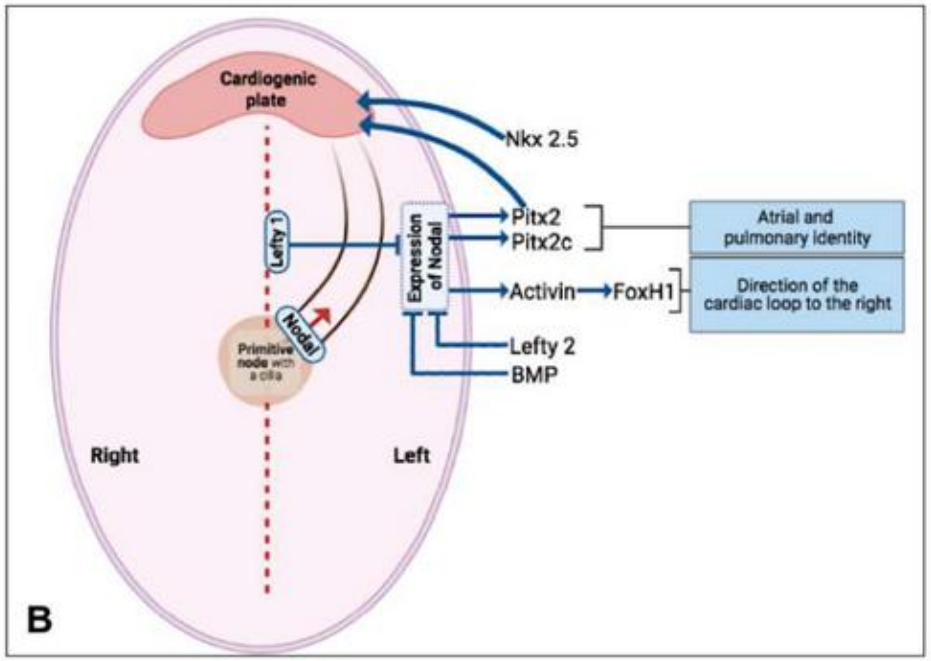

Complete Mole
Two sperm
(XX or XY)
No maternal DNA
Egg
Duplication of sperm DNA
46XX or 46XY
Paternal chromosomes only
(Androgenetic mole)
or
X sperm
No maternal DNA
Egg
Partial Mole
23X
23Y
One or two sperm
46XY
23X
Egg
23X
23X
23Y
69XXY or 69XXX
Maternal and paternal chromosomes (Triploid)

determination of the left-right axis of the embryo

Nodal is a member of the TGF-beta family and together with Lefty both are involved in the initial left-right (L-R) patterning of the axis of the embryo during gastrulation.

Serotonin signaling is a very early step in patterning of the left-right axis

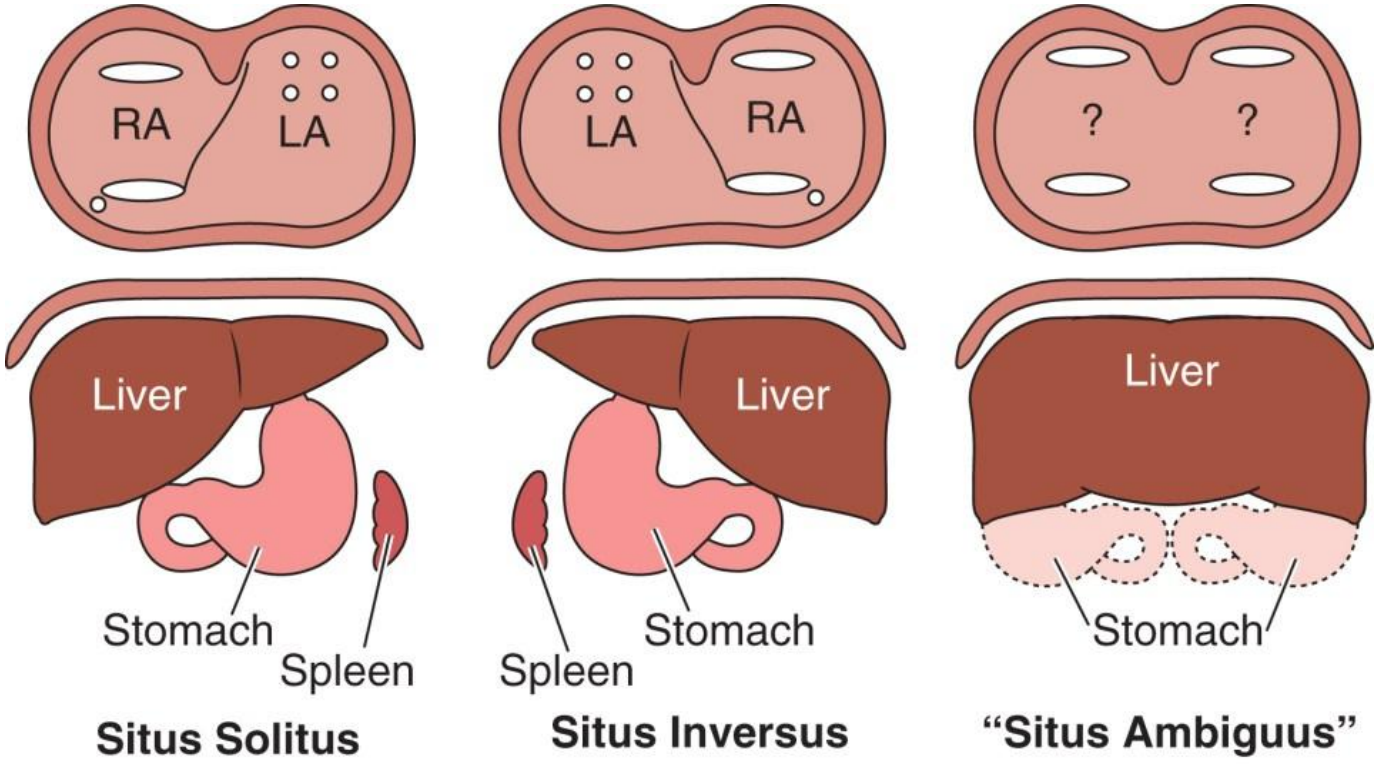




Birth defects associated with laterality

Situs inversus

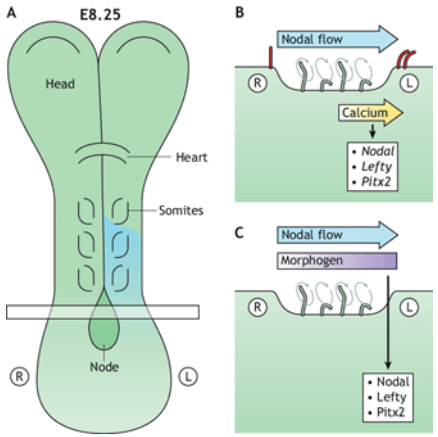
Condition where the positioning of all organs is reversed in a mirror image arrangement

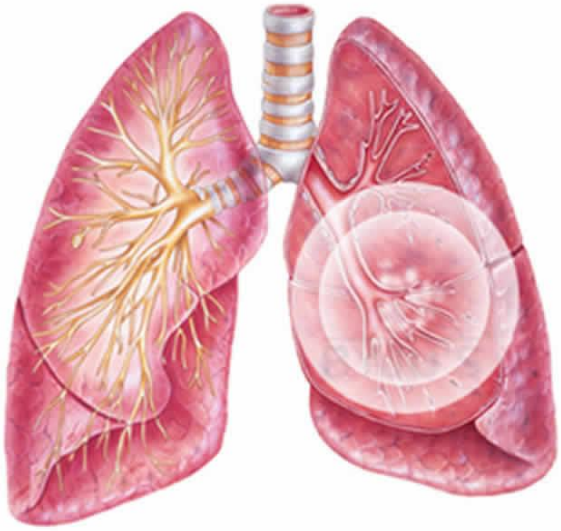


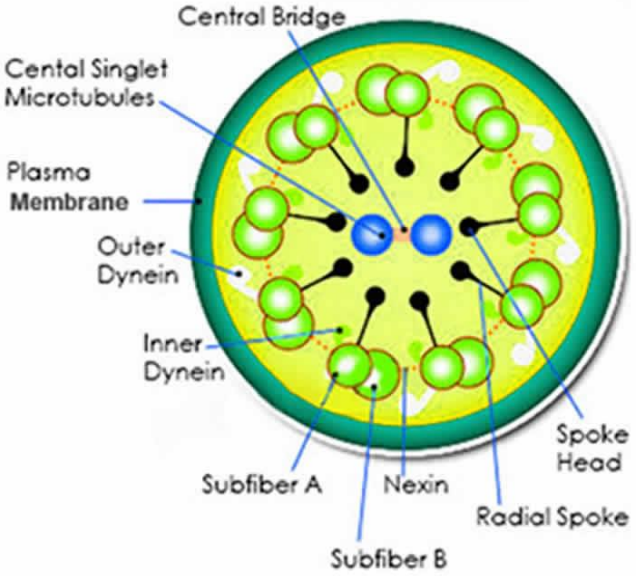
Situs ambigus or heterotaxy

is a disturbance in the usual left-right distribution of the thoracic and abdominal organs that does not entirely correspond to the complete mirror image (situs inversus)

PRIMARY CILIARY DYSKINESIA







Cilia are normally present on the entral surface of the primitive node and are involved in L-R patterning

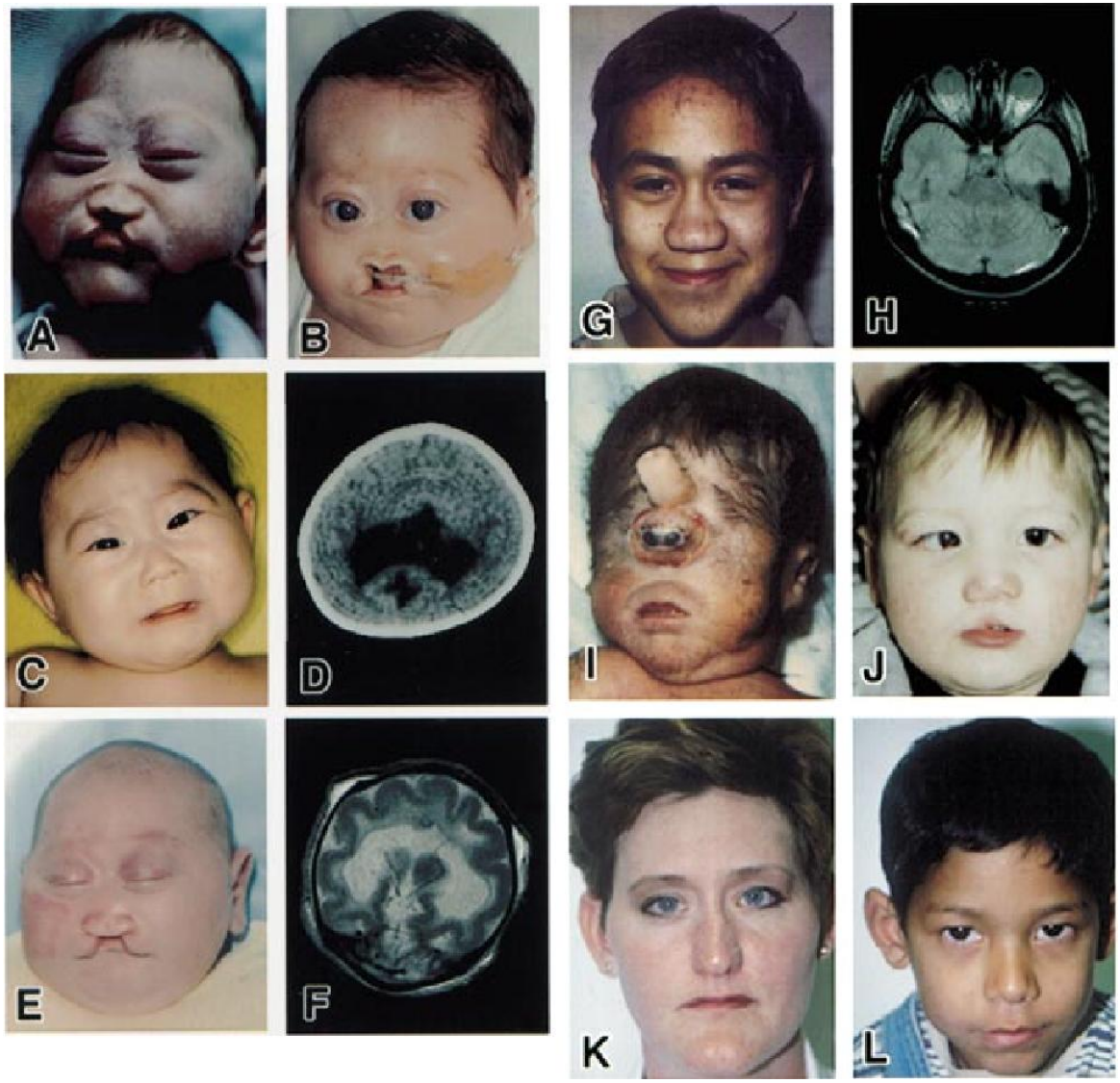
The triad of **situs inversus**, chronic sinusitis and bronchiectasis is called **Kartagener's syndrome**

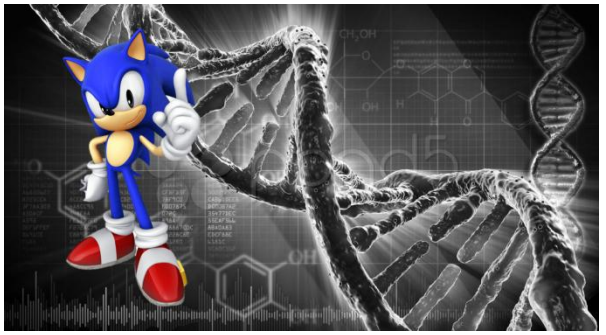
Chronic sinusitis is a long-lasting inflammation of the sinuses

Bronchiectasis is caused by the airways of the lungs becoming damaged and widened.

https://healthjade.net/kartagener-syndrome/

Holoprosencephaly





Sonic hedgehog

Holoprosencephaly (HPE) in individuals with Sonic Hedgehog (SHH) mutations

The high doses of alcohol at the beginning of the third week of development kill cells in the anterion midline of the germ disc

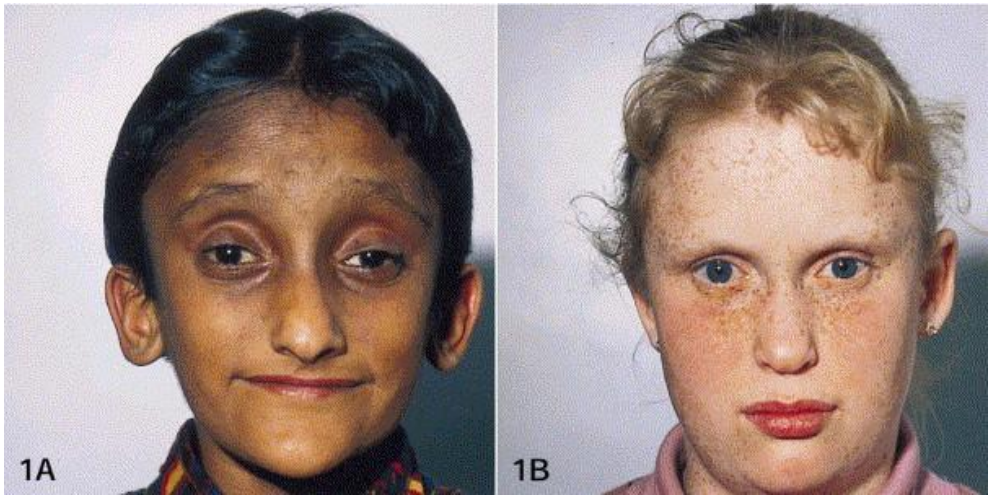
DOI:10.1093/HMG/8.13.2479Corpus ID: 15468979

Alagille syndrome

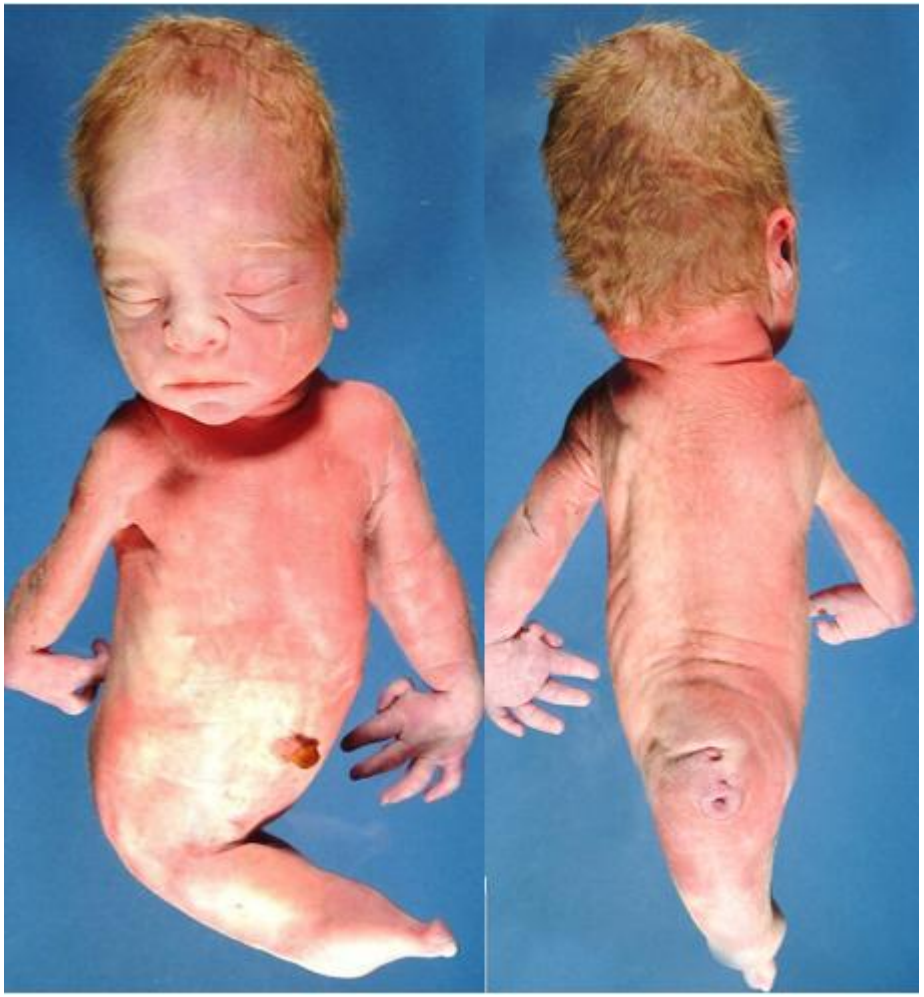
Rare genetic disorder that can affect multiple organ systems of the body including the liver, heart, skeleton, eyes and kidneys

Common symptoms, which often develop during the first three months of life, include blockage of the flow of bile from the liver (cholestasis), yellowing of the skin and mucous membranes (jaundice), poor weight gain and growth.

Caused by the mutation within NOTCH pathway



Caudal dysgenesis = sirenomelia



Insufficient **mesoderm** is formed in the caudalmost region of the embryo

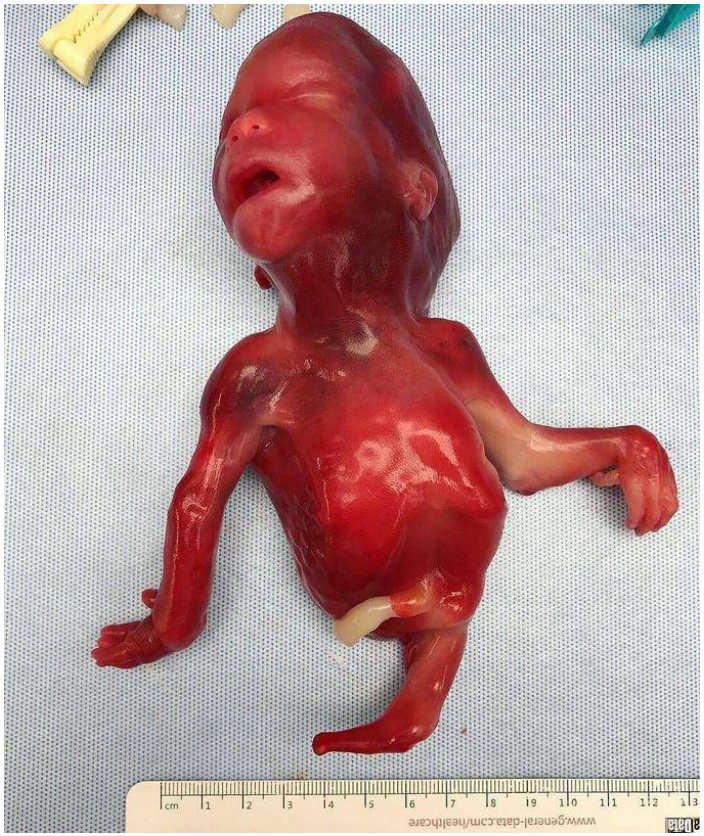
The mesoderm contributes to formation of:
Lower limbs
Urogenital system
Lumbosacral vertebrate

extremely rare congenital developmental disorder characterized by anomalies of the lower spine and lower limbs. Affected babies are born with partial or total leg fusion. Sirenomelia is thought to affect one in every 60,000 to 100,000 infants.

Sirenomelia

Caudal dysgenesis

- a rare congenital anomaly characterized by a single lower extremity which is associated with abnormalities in other organ systems, commonly affecting the gastrointestinal and the urogenital systems



- defect in the formation of the primitive streak during late gastrulation could lead to caudal body malformations

- loss of **mesoderm** in the lumbosacral region has resulted in fusion of the buds and other defects

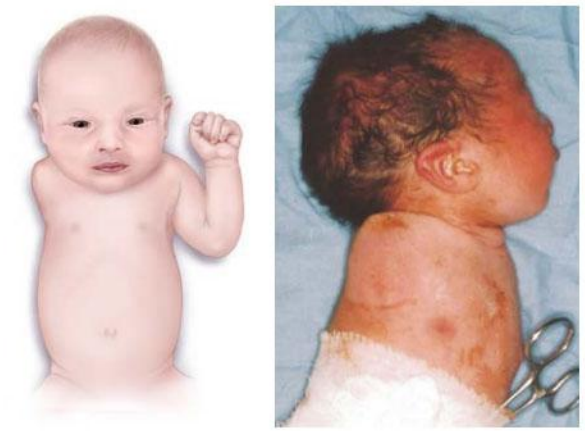
Meromelia

- defined broadly as the partial absence of at least one limb

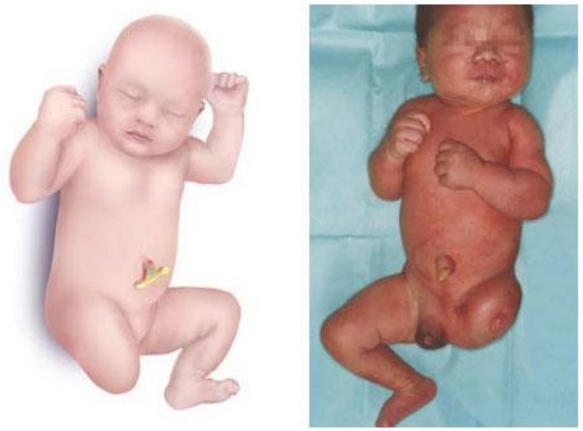
Amelia

- congenital anomaly characterized by the complete absence of one or more limbs

Amelia of the upper limb (Q71.0)



Amelia of the lower limb (Q72.0)



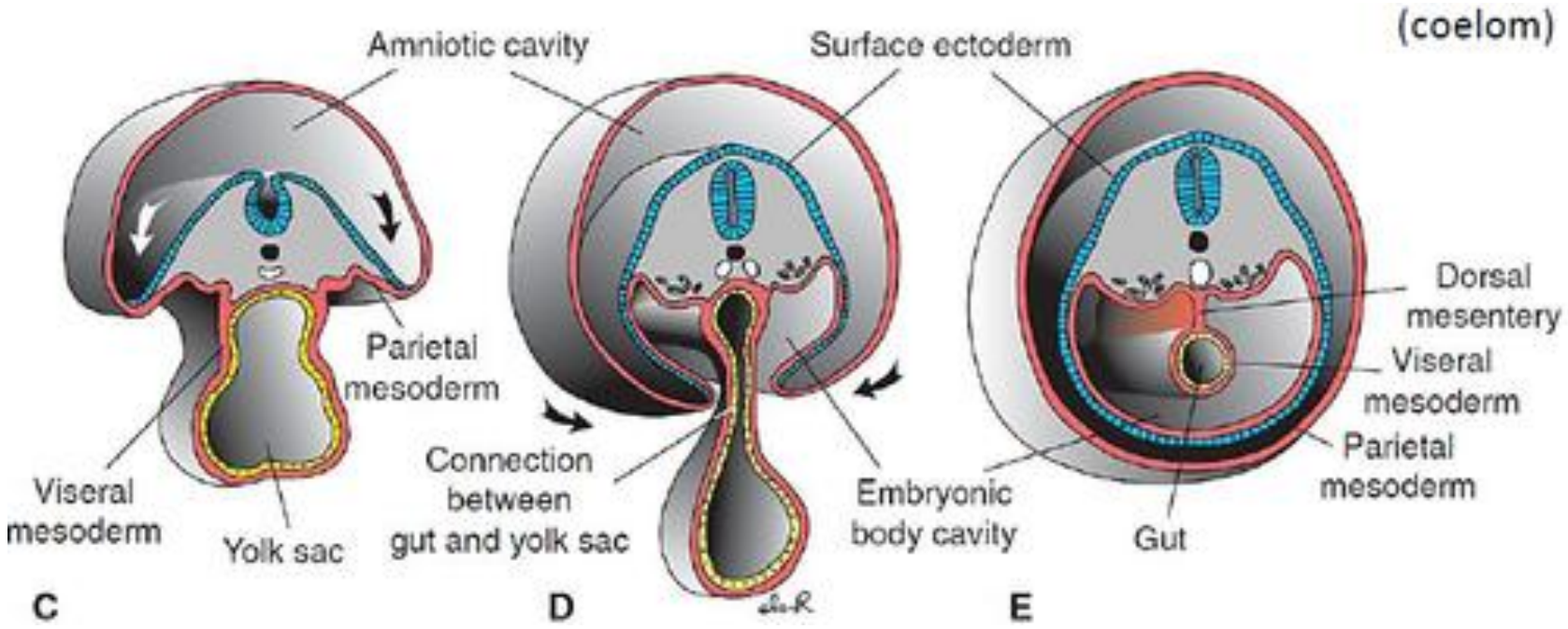
Etiological factors include genetic, teratogens (infamous-**thalidomide** use), vascular disruptions and ischemia, chemicals and radiation exposure

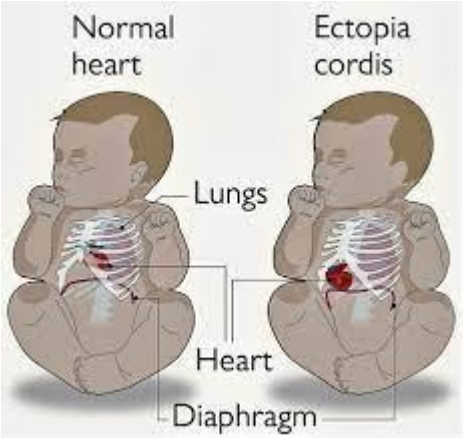
Thalidomide was a widely used drug in the late 1950s and early 1960s for the treatment of nausea in pregnant women. It became apparent in the 1960s that thalidomide treatment resulted in severe birth defects in thousands of children.

More recently, thalidomide has proven useful for treating cancer and leprosy and is approved for these uses.

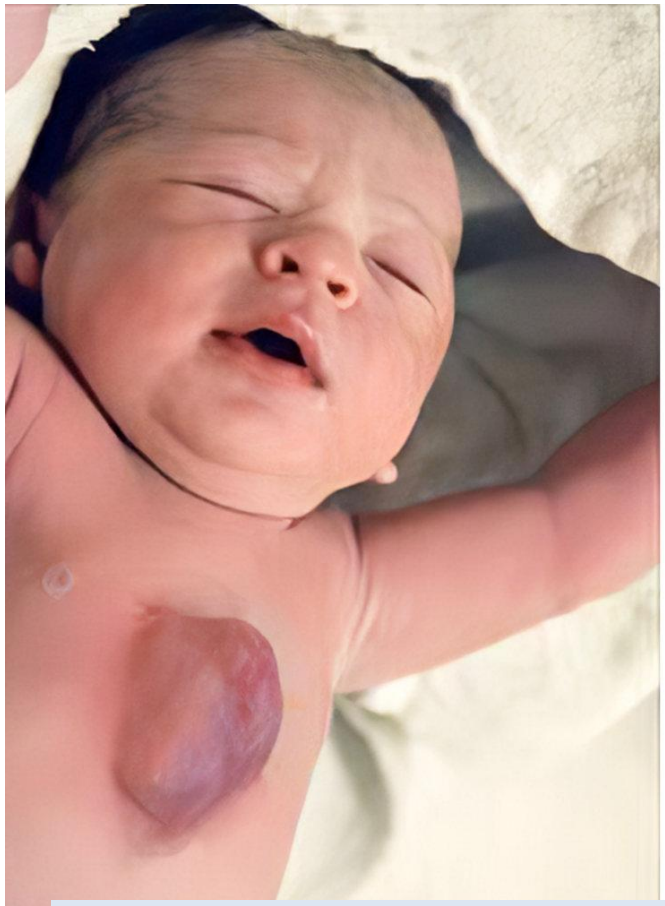
Leprosy is an infectious disease that causes severe, disfiguring skin sores and nerve damage in the arms, legs, and areas around your body.
Leprosy (Hansen disease)

Ventral body wall defects





Ventral body wall defects



Ectopia Cordis

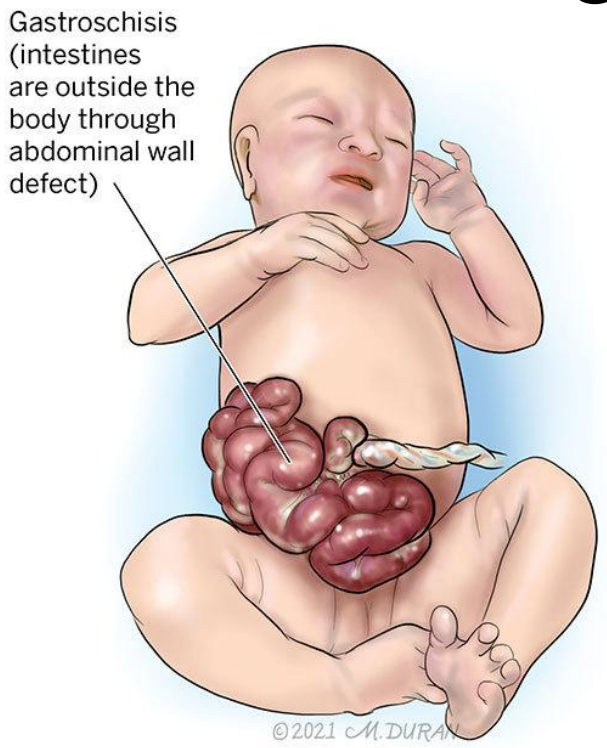
is a rare type of malformation where the heart is not located normally. It may be partially or completely located outside the thoracic cavity and can be associated with other congenital abnormalities. It results from failure of maturation of midline mesoderm and ventral body formation during embryogenic formation. The exact etiology remains unknown.

Although surgical correction may be attempted, the prognosis is generally poor and depends on the severity of intracardiac malformations and the presence of associated abnormalities. Most infants are stillborn or die within the first hours or days of life.

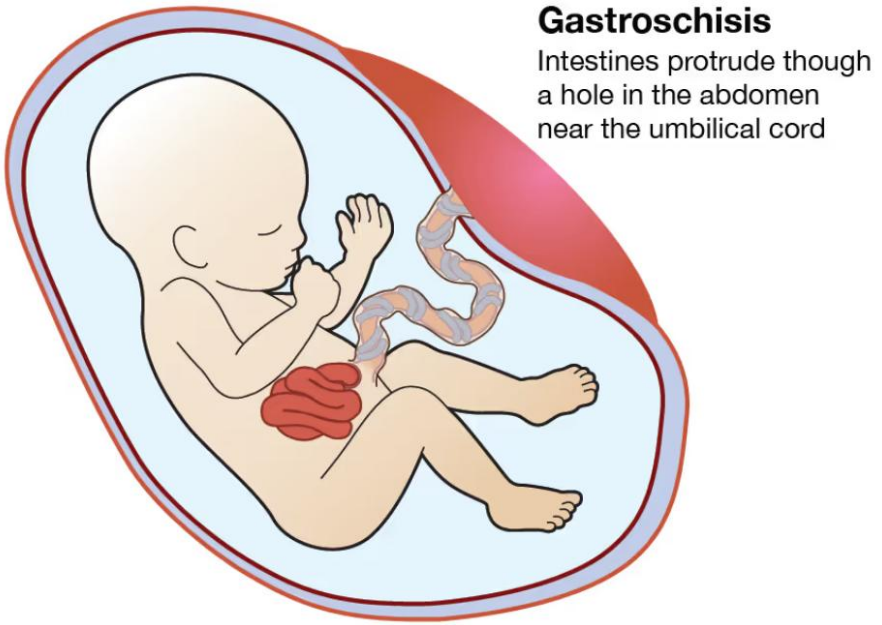
Ectopia cordis may occur as an isolated malformation or associated with a larger category of ventral body wall defects that affect the thorax, abdomen, or both.

Ventral body wall defects

Gastroschisis



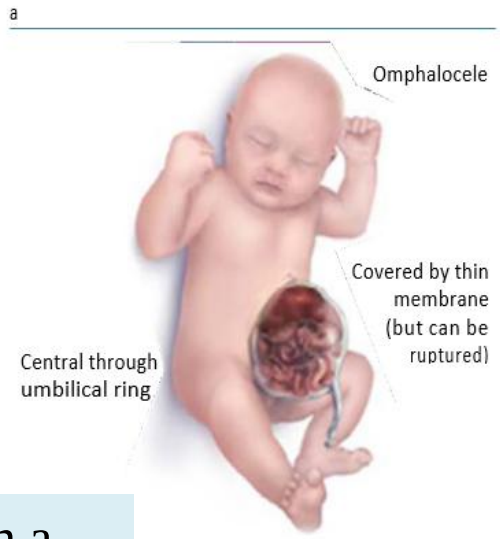
Gastroschisis is an abdominal wall defect most commonly located to the right of an intact umbilical cord; rarely, the defect is to the left of the umbilical cord.

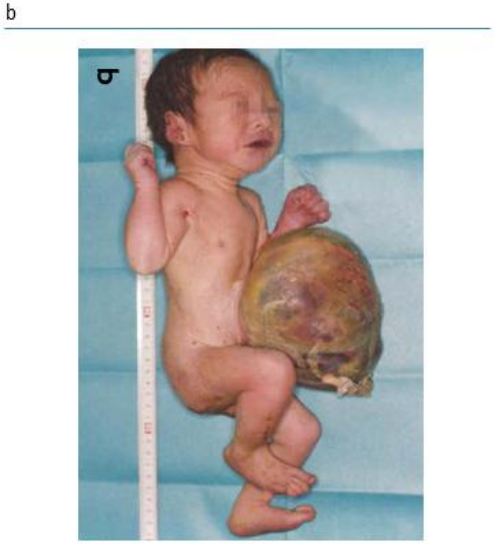


Ventral body wall defects

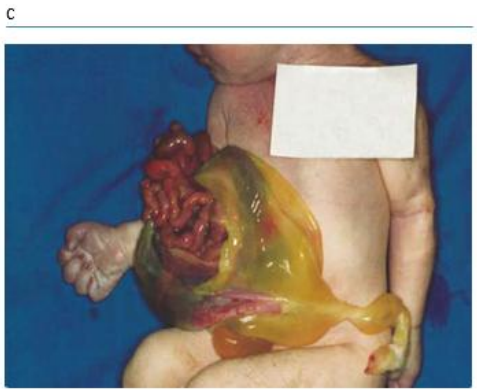
Omphalocele

It is a protrusion of the abdominal contents covered with peritoneum through the base of the umbilical cord.





Omphalocele **does not arise** from a failure in body wall closure. It originates when physiological umbilical herniation fails to return to the abdominal cavity.



Ventral body wall defects

Protrudes into umbilical cord
Covered by amnion

After delivery
exposed to air
No peritoneal layer

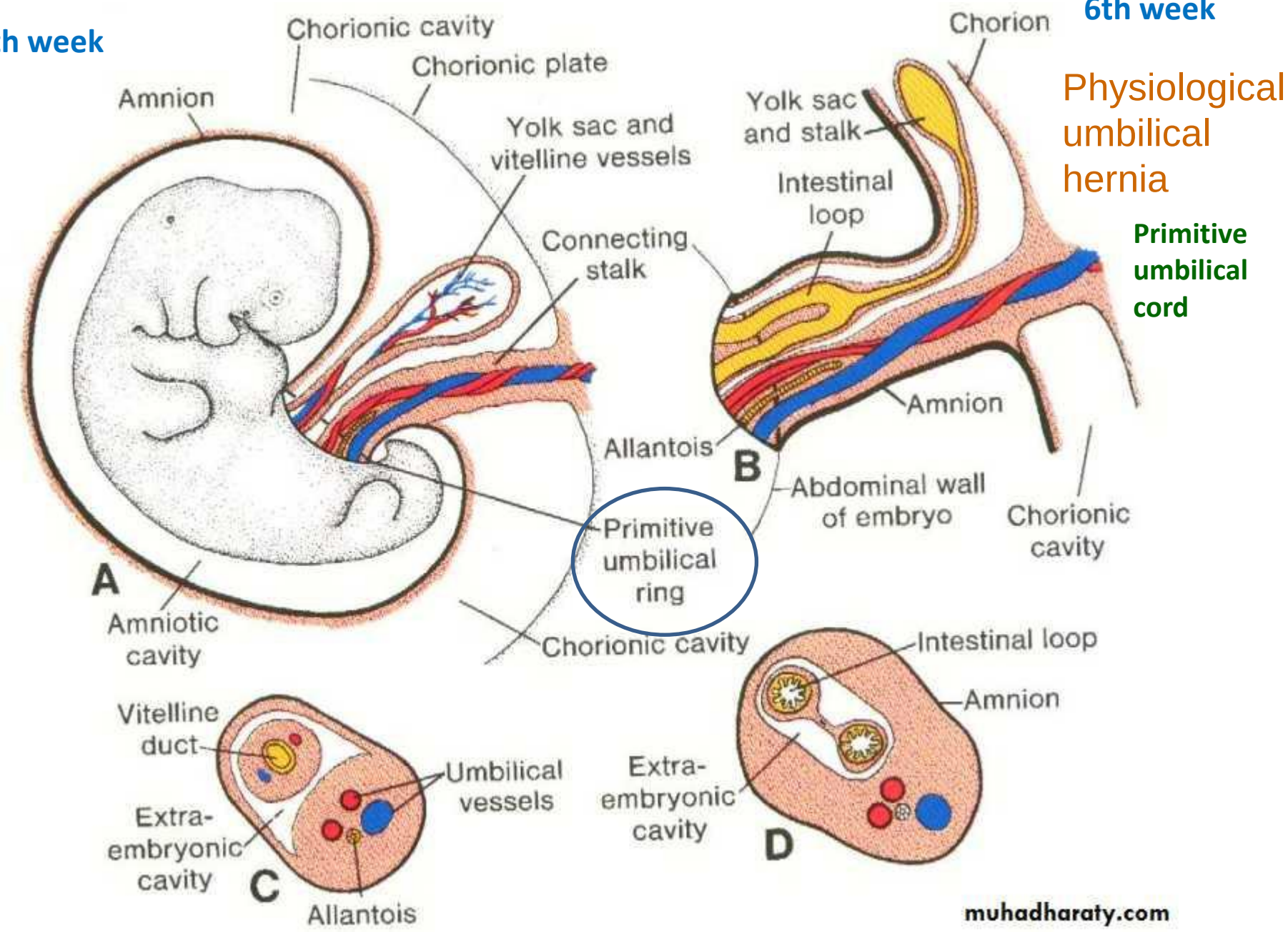
Body wall closure fails in the abdominal region

It originate when portion of the gut tube that normally herniates into the umbilical cord during the 6th to the 10th weeks fails to return to the abdominal cavity

The ventral body wall closes completely exept for the umbilical region where the connecting stalk and yolk sac duct remain attached

Fifth week

6th week

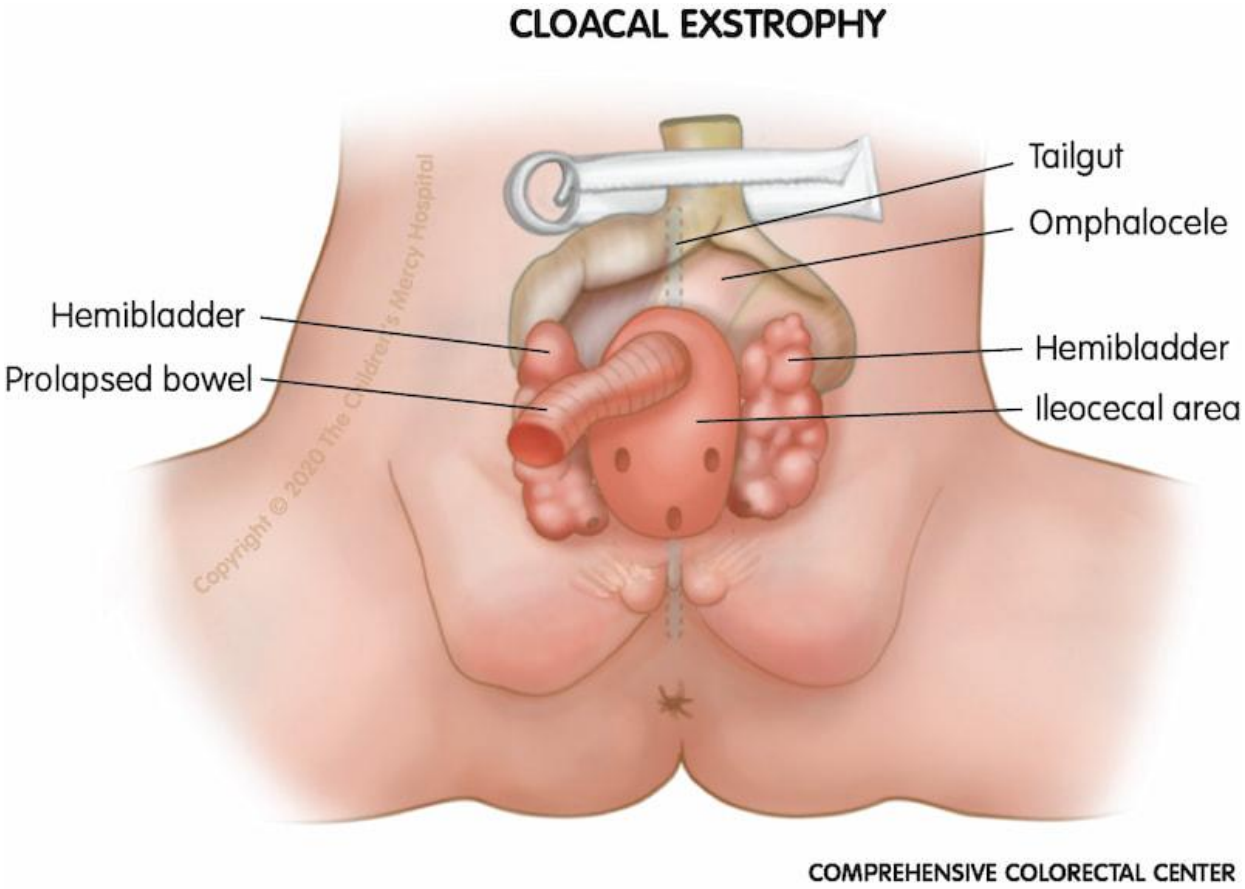


Physiological umbilical hernia

Primitive umbilical cord

muhadharaty.com

Ventral body wall defects



affects the urinary, digestive and reproductive systems. It causes an inside-out bladder (exstrophy) and organs that push through a hole in the abdomen (omphalocele). A child with cloacal exstrophy also has divided genitals and an incomplete anus. Infants need staged corrective surgeries to treat each of these problems.

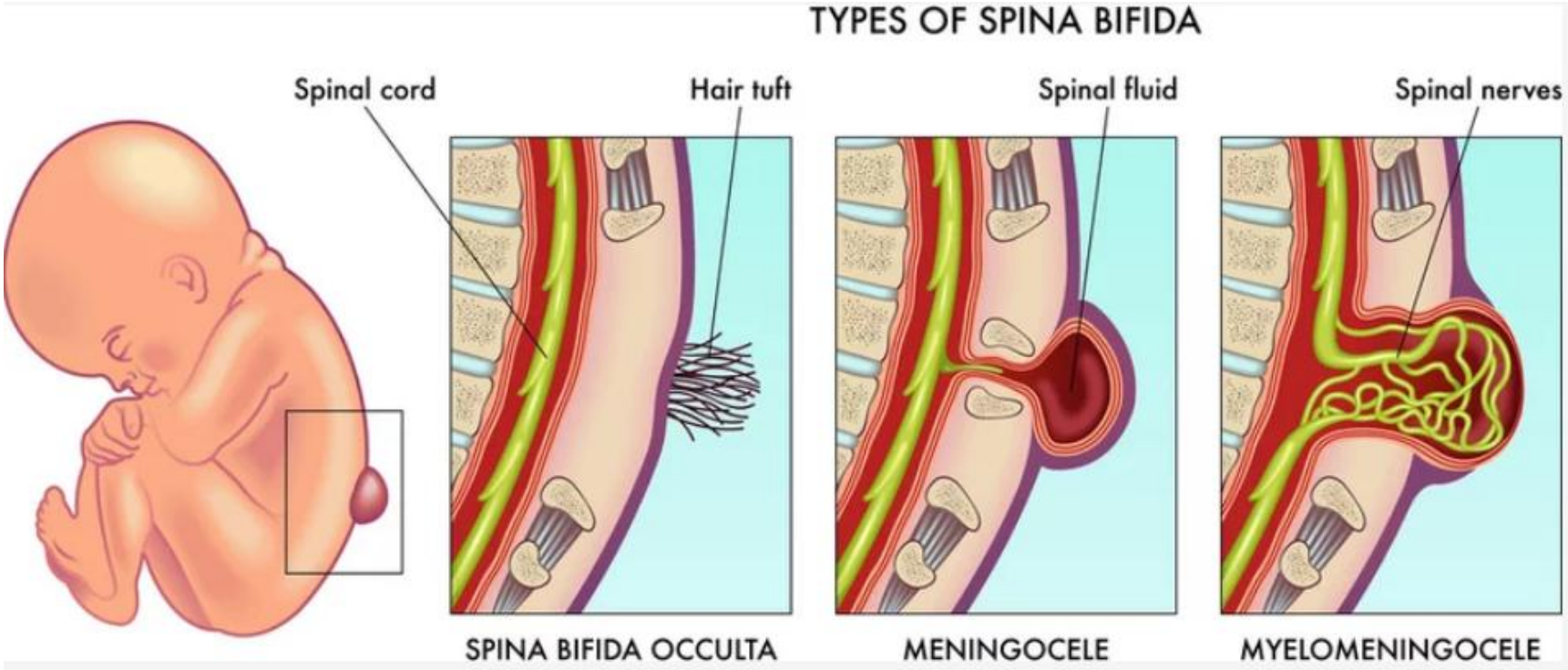
Neural tube defects

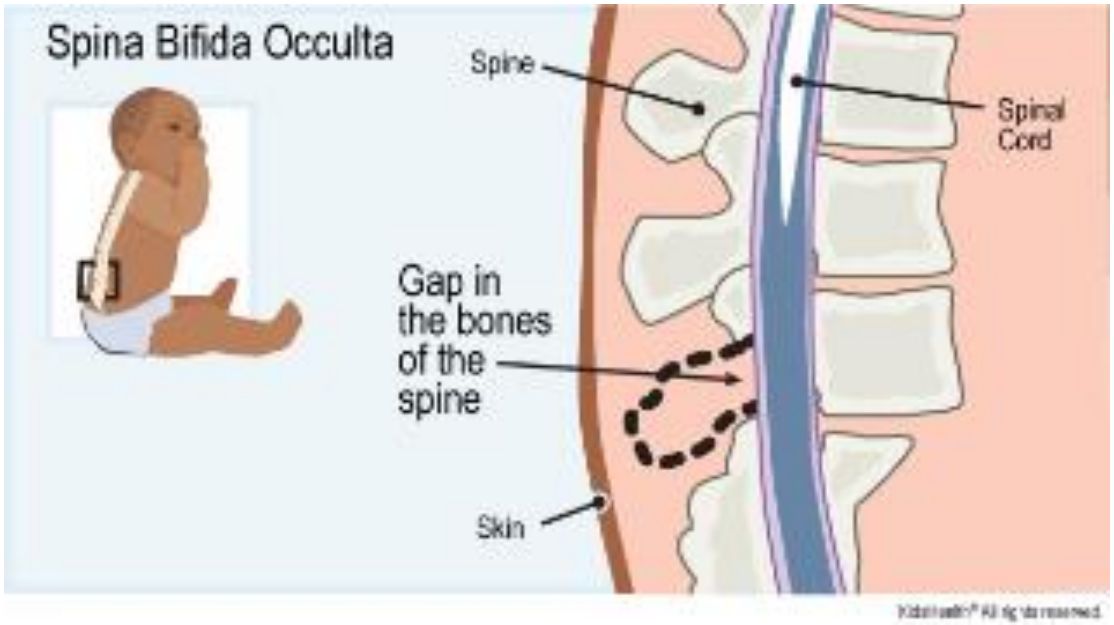
a group of birth defects that occur when the neural tube, the structure that develops into the brain and spinal cord, fails to close completely during early pregnancy

Spina bifida

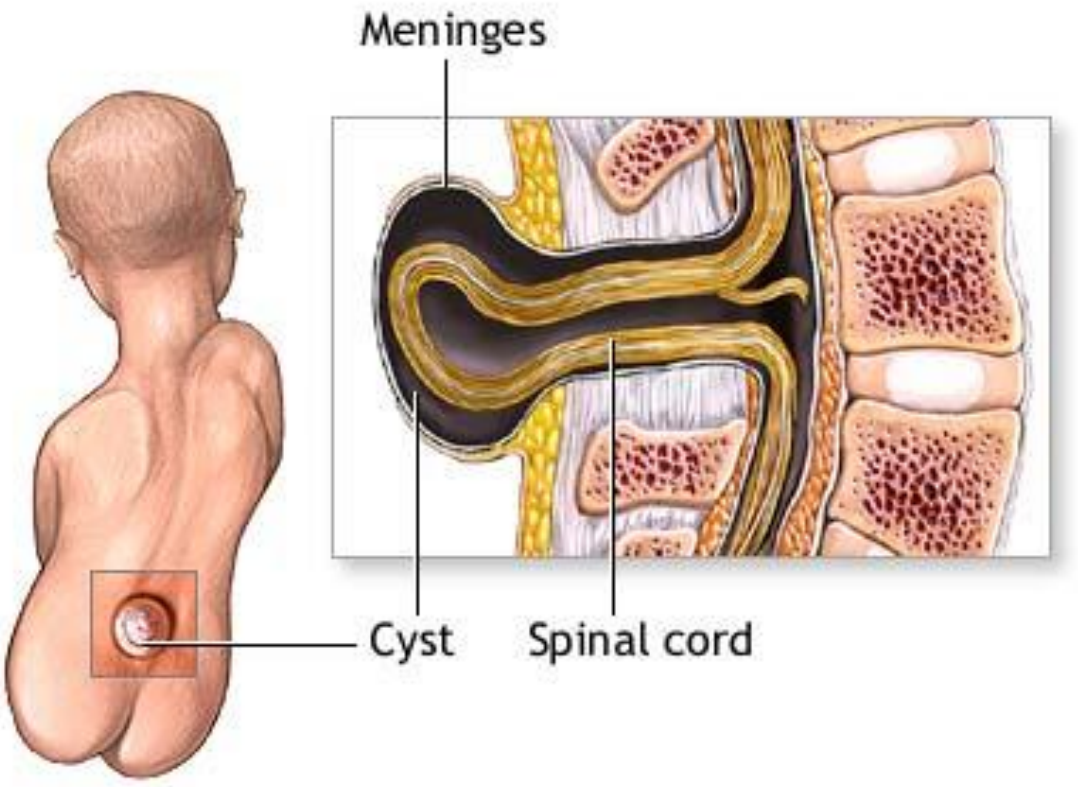
Spina bifida is the most common type of neural tube defect (NTD). It happens when the neural tube doesn't close completely somewhere along the spine during fetal development.

There are a few different types of spina bifida, including:



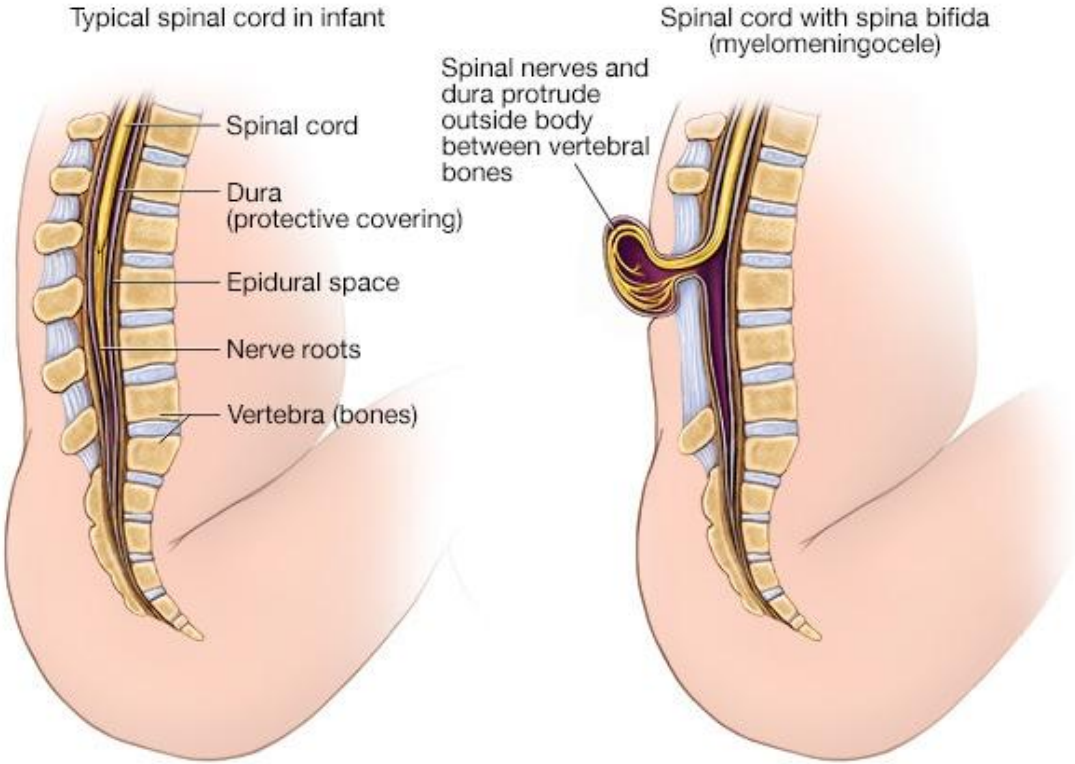


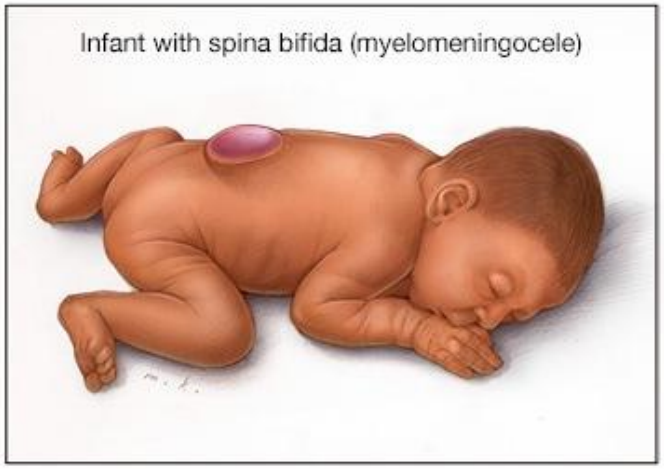
Spina bifida occulta is when a baby's backbone (spine) does not fully form during pregnancy. The baby is born with a small gap in the bones of the spine.



Meningocele: This NTD is characterized by a sac of fluid that protrudes through an opening in baby's back, but their spinal cord is not involved or damaged.

A.D.A.M.





Myelomeningocele (open spina bifida): This NTD is characterized by incomplete neural tube closure and a fluid-filled sac that protrudes (sticks out) from baby's back. The sac contains part of their spinal cord, meninges, nerves and cerebrospinal fluid (CSF). Myelomeningocele is the most severe and the most common form of spina bifida.

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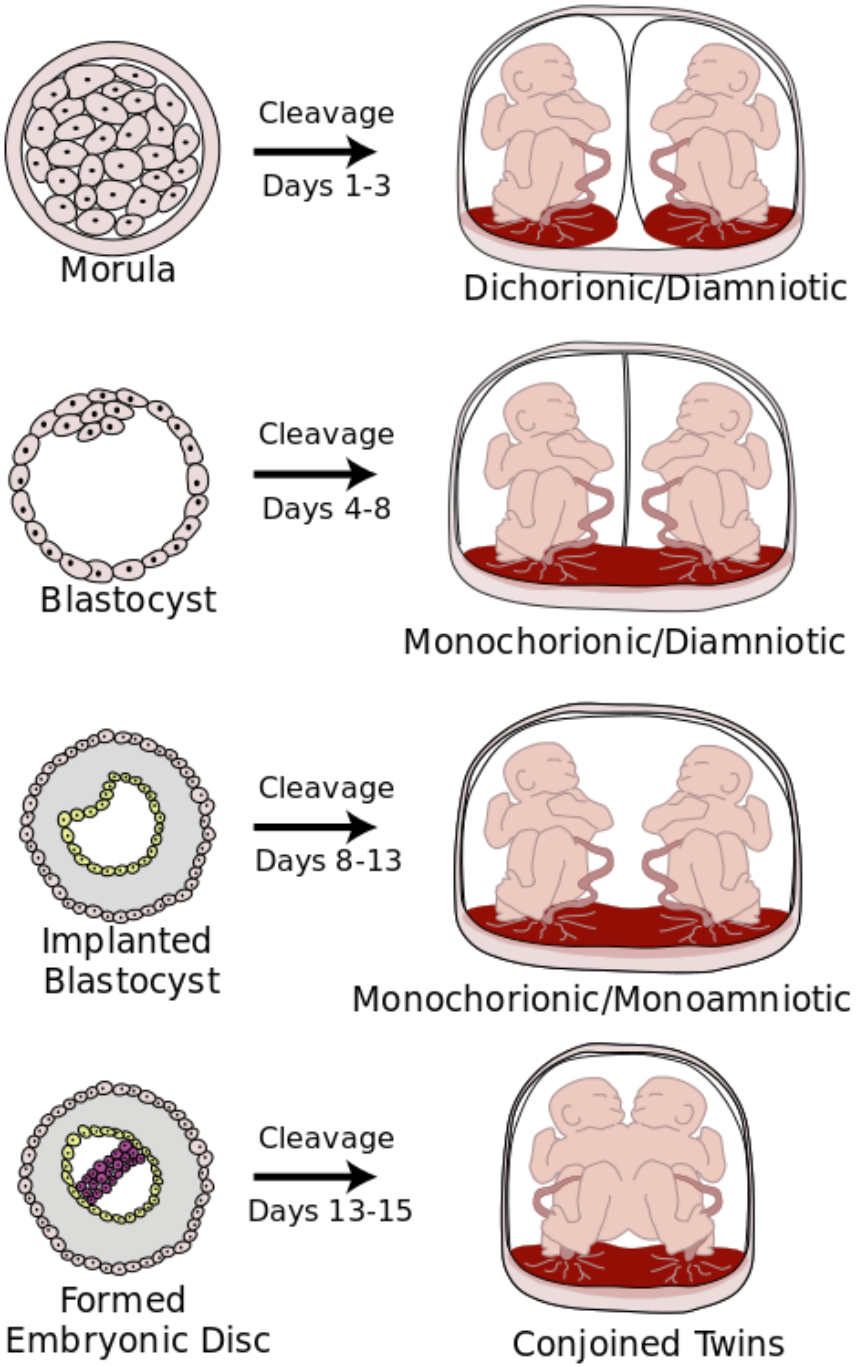
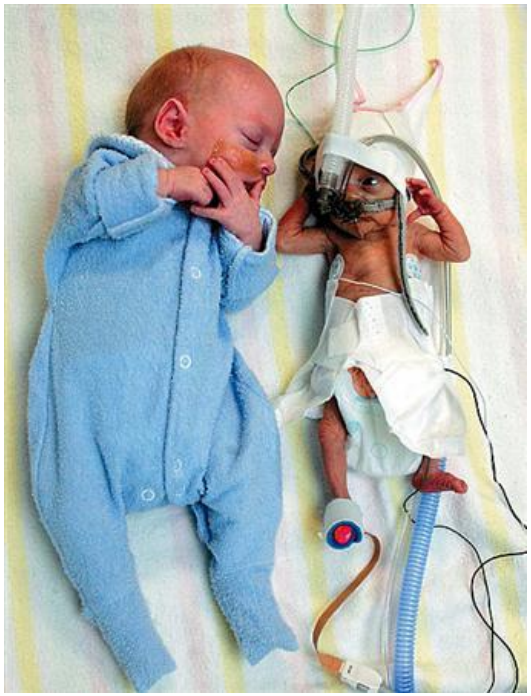
Monozygotic twins

One fertilized ovum

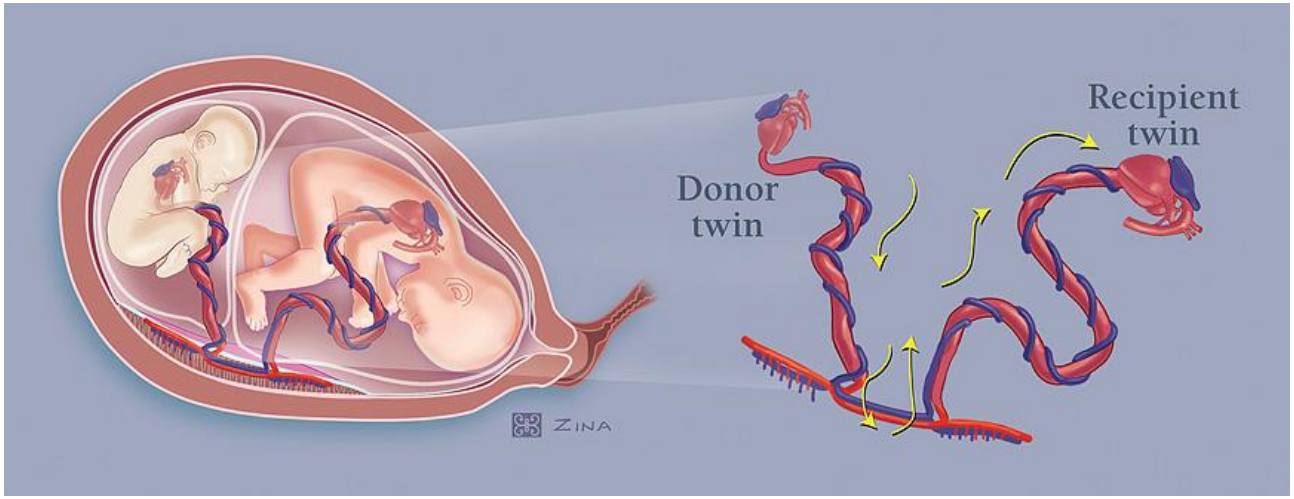
Zygote splitts at various stages of development



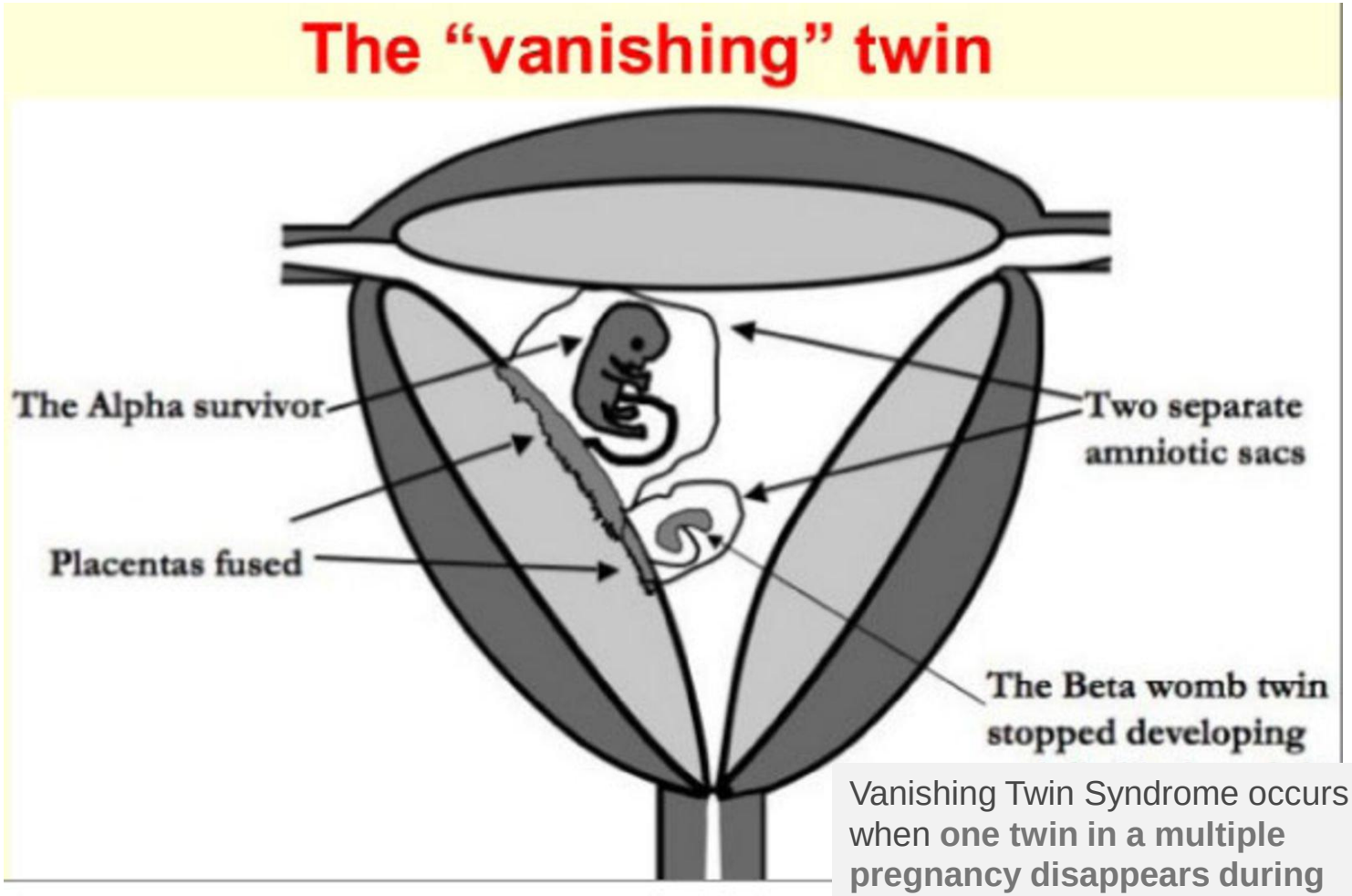
Twin-to-twin transfusion syndrome



Twin-to-twin transfusion syndrome (TTTS) is a serious condition that can occur in pregnancies involving identical twins who share a placenta. In TTTS, blood flow between the twins is unequal, with one twin (the donor) giving more blood than it receives and the other twin (the recipient) receiving too much blood. This imbalance can lead to complications for both twins, including growth differences, excess amniotic fluid in the recipient twin, and potentially death or organ damage.

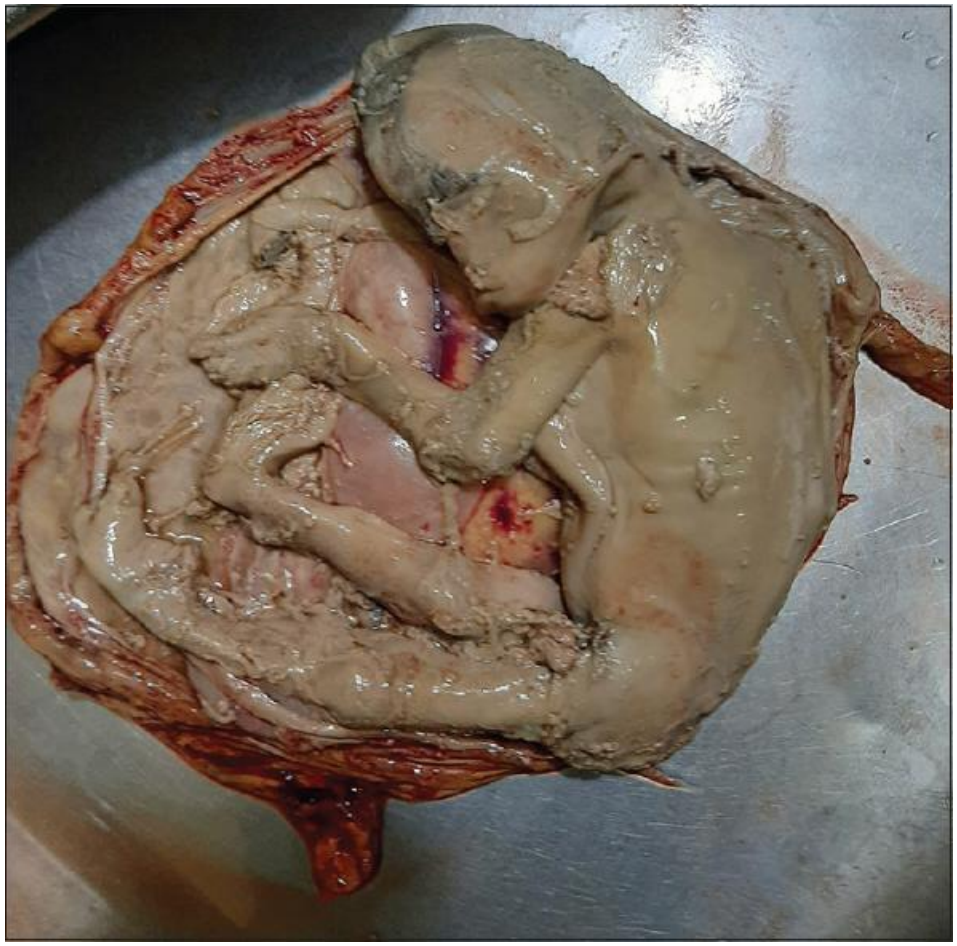


The "vanishing" twin



Vanishing Twin Syndrome occurs when **one twin in a multiple pregnancy disappears during gestation**, often in the first trimester, and is then partially or completely reabsorbed.

Fetus papyraceus



(or **mummified fetus**) is a rare complication with an incidence of 1 in 12. 000 live births seen mainly **in twin pregnancies** according to the demise of a co-twin in early gestation (usually in the 2nd trimester of pregnancy, a twin that dies in the early stages of pregnancy usually disappears completely, while the death of one at the end of a pregnancy results in fetal maceration).

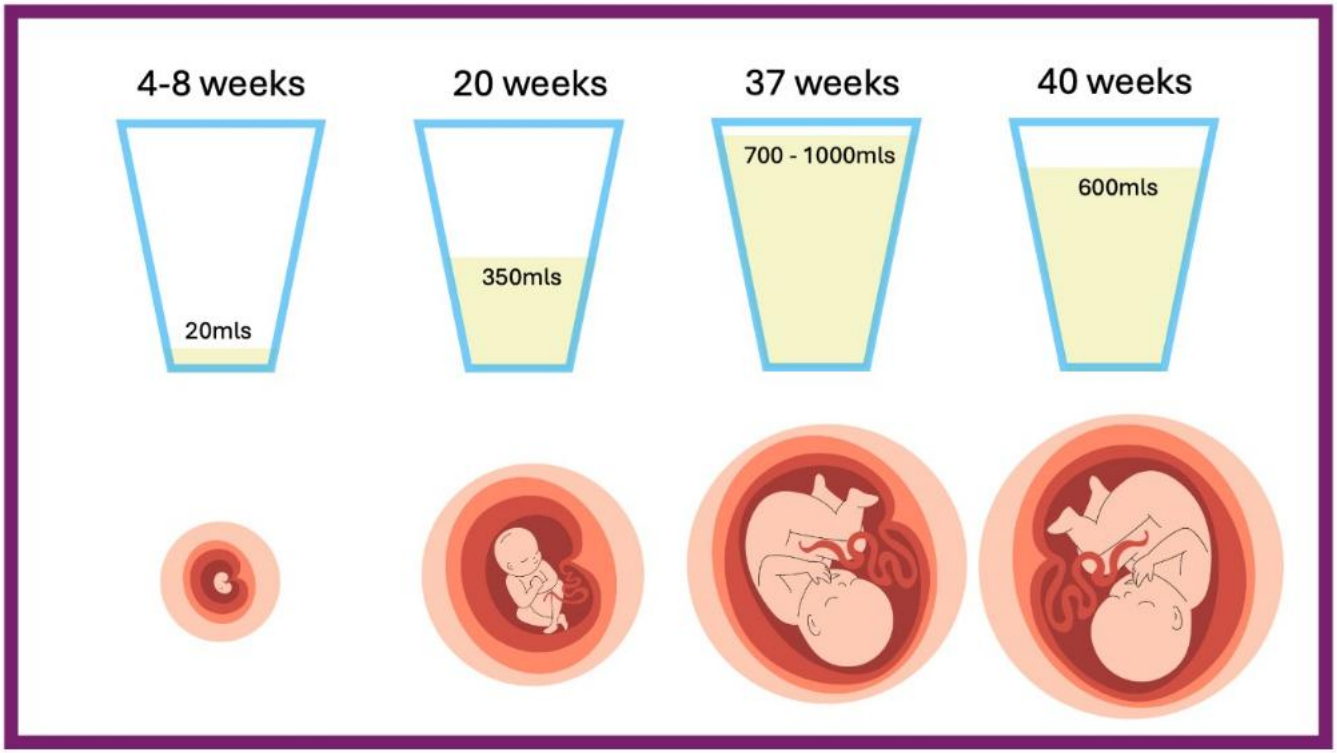
Ventral view of dead (mummified) twin with products of conception

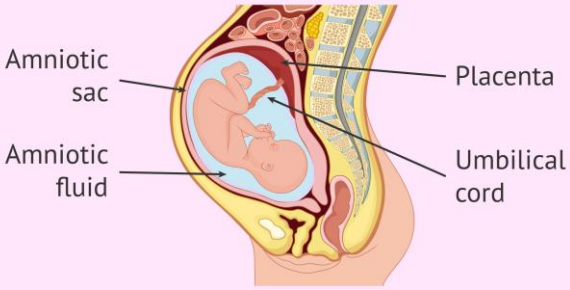
Fetal mummification complicating a dichorionic twin gestation: Clinical pearls for medical education Olusoji Edward Jagun

capillary hemangioma

Capillary infantile hemangiomas constitute about 4% to 10% of benign tumors in the pediatric age group, 80% of which occur in the head and neck region

Amniotic fluid





- amniotic fluid is an ultrafiltrate of **maternal blood plasma** and is composed of proteins, carbohydrates, carbohydrates, carbohydrates, and electrolytes that will help fetal development
- Is produced in part by amniotic cells

Oligohydramnions

disorder of amniotic fluid resulting in decreased amniotic fluid volume for gestational age.

Amniotic fluid cushions the fetus from injury, helps prevent compression of the umbilical cord

Factors that are associated with hydramnios include the following:

Maternal factors:

- Diabetes

Fetal factors:

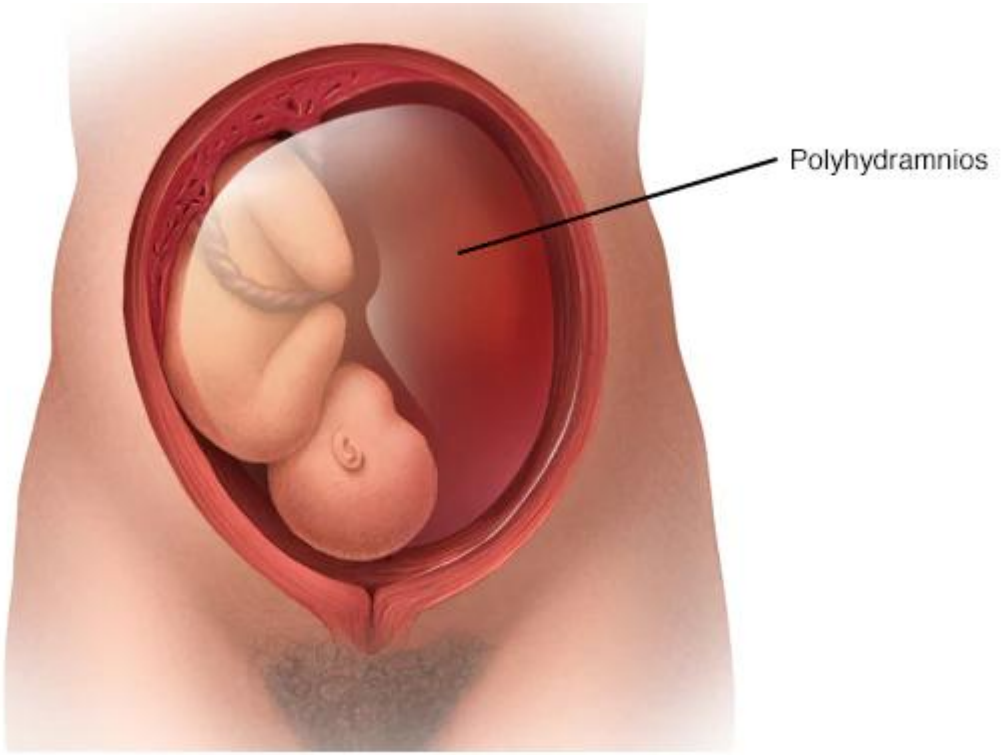
- Gastrointestinal abnormalities that block the passage of fluid
- Abnormal swallowing due to problems with the central nervous system or chromosomal abnormalities
- Twin-to-twin transfusion syndrome
- Heart failure
- Congenital infection (acquired in pregnancy)

When the embryonic kidney cells fail to develop, the result is called **renal agenesis**. It is often detected on fetal ultrasound because there will be a **lack of amniotic fluid** (called oligohydramnios).

Hydramnions

Hydramnios occurs when there's **too much** amniotic fluid around your baby during pregnancy. It can be caused by problems in both the mother and baby. It causes the uterus to grow fast.

Most cases of polyhydramnios are mild and result from a gradual buildup of amniotic fluid during the second half of pregnancy. Severe polyhydramnios may cause shortness of breath, preterm labor, or other signs and symptoms.



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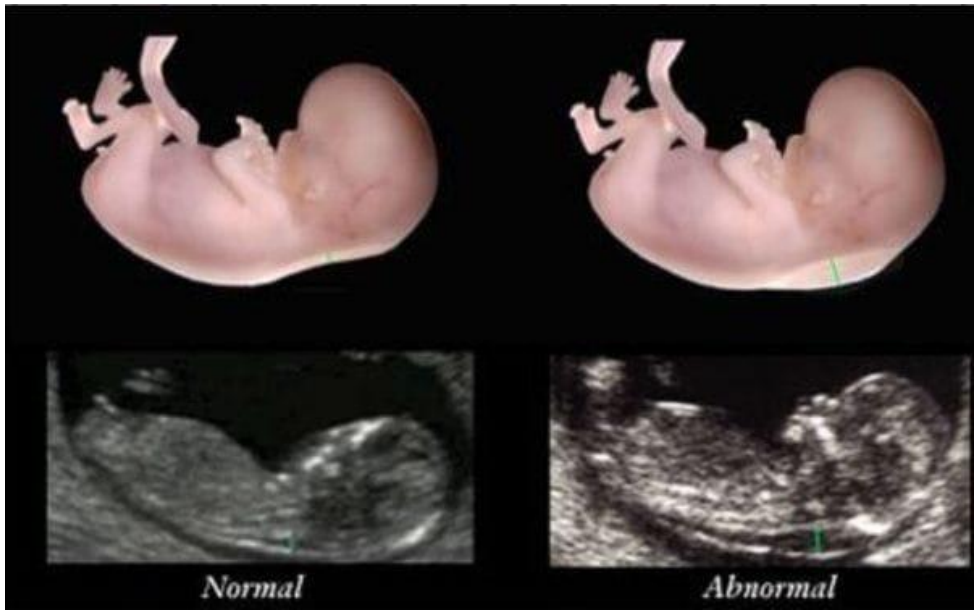
Congenital Rubella Syndrome

Is an illness in infants that results from maternal infection with rubella virus during pregnancy. When rubella infection occurs during early pregnancy, serious consequences–such as miscarriages, stillbirths, and a constellation of severe birth defects in infants–can result. The risk of congenital infection and defects is highest during the first 12 weeks of gestation and decreases thereafter

Common congenital defects of CRS include **cataracts, glaucoma, congenital heart disease, hearing loss, tooth abnormalities** and developmental delay.

prenatal diagnostic



Ultrasound – test called nuchal translucency (Down syndrome, heart defects); 11-14 weeks of pregnancy

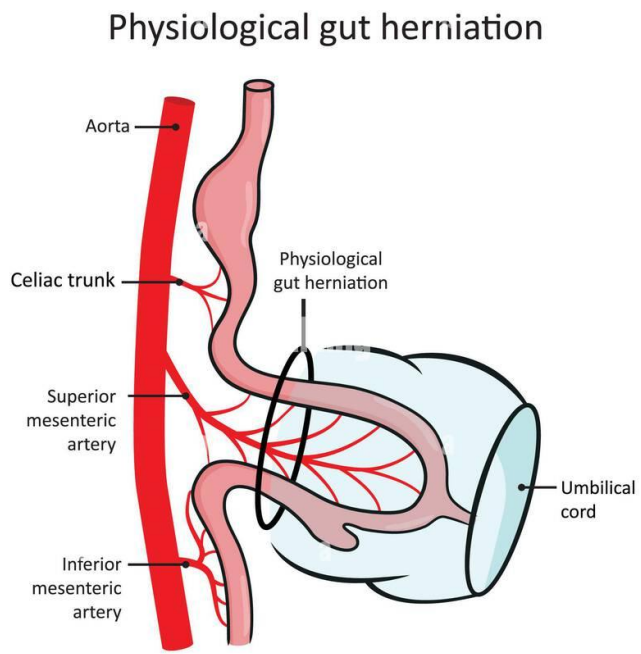
Maternal serum screening - alfa-fetoprotein concentration: **decrease** in Down syndrome, trisomy 18, sex chromosome abnormality; **increase** in neural tube defects, omphalocele, gastroschisis, bladder extrophy, others…

Amniocentesis - Withdrawing amniotic fluid with the needle (because of the amount of fluid required, the procedure is not performed before 14 weeks gestation) for analysis of biochemical factors, and metaphase karyotyping
Chorionic villus sampling

Development of the fetus

Length of pregnancy

Monthly changes



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Physiological gut herniation is **a natural phenomenon that occurs in early pregnancy**. It usually occurs from around 6-8 weeks up until 12-13 weeks in-utero, after which the bowel returns to the abdominal cavity.

12TH WEEK OF DEVELOPMENT

Primary ossification centers are present in the long bones and skull

The intestinal loops have withdrawn into the abdominal cavity

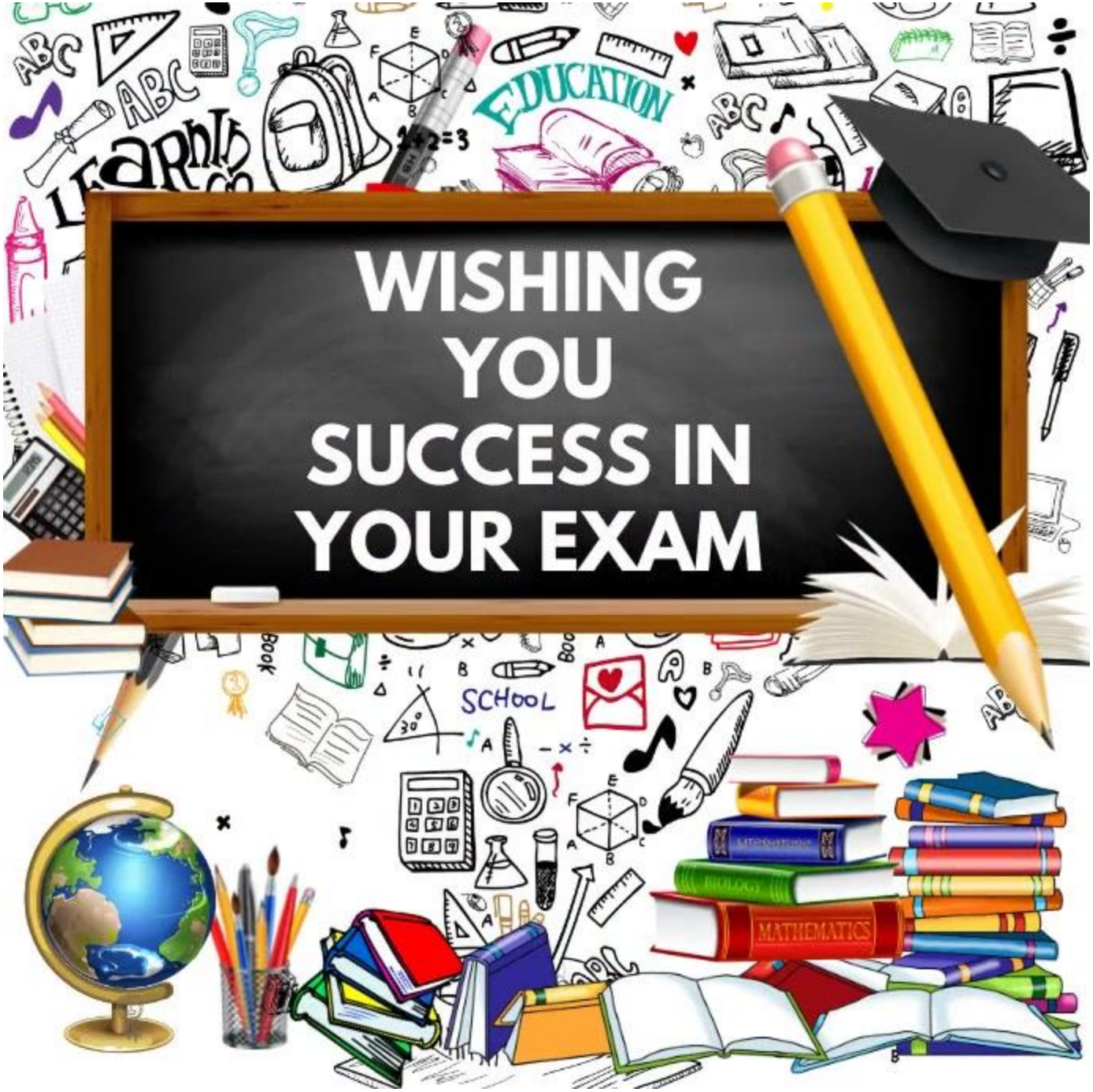
External genitalia develop to such a degree that the sex of the fetus can be determined by external examination (ultrasound)

Table 8.2 !!!!!

During the **fifth month**, movement of the fetus can be felt by the mother

During the **sixth month**, skin of the fetus is reddish with wrinkled appearance

At the **end of intrauterine life**, the skin is covered by a fatty substance – vernix caseosa)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