Human embyology

Congenital malformation

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- **abnormal structure resulting from disorder of embryonic development.**
- **Teratology: research on congenital malformation, branch of embryology.**

I Classification

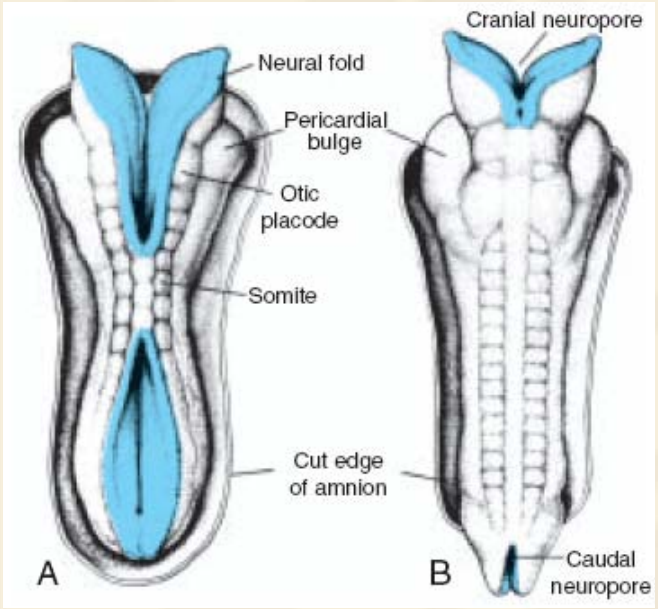
(1) Developmental malformation of whole embryonic body

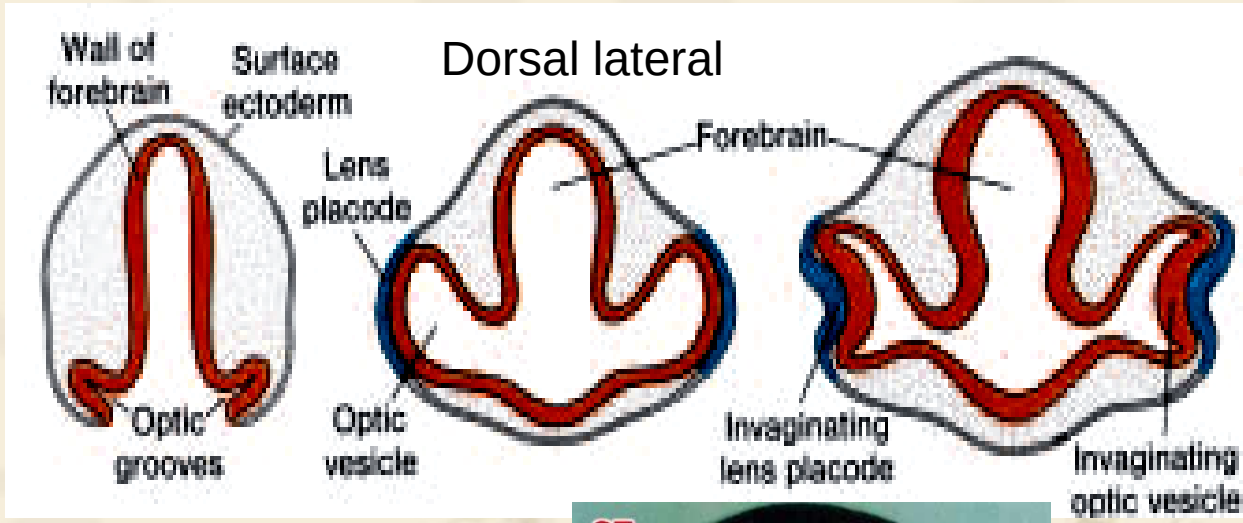
- severe genetic defect
- Death in early period (spontaneous abortion)

(2) Partly developmental malformation

- **developmental disorder in a part of embryonic body**
- **Cyclopia/Monophthalmia/ Monophthalmus**

***Mono-* +*ophthalmos* eye**





ventral lateral

A pair of optic grooves located lateral of unclosed cephalic neural tube fuse together.

- **Synpodia/ sirenomelus /sirenomelia**
- ***Seiren* siren+ *melos* limb**
- **“mermaid symptom”**

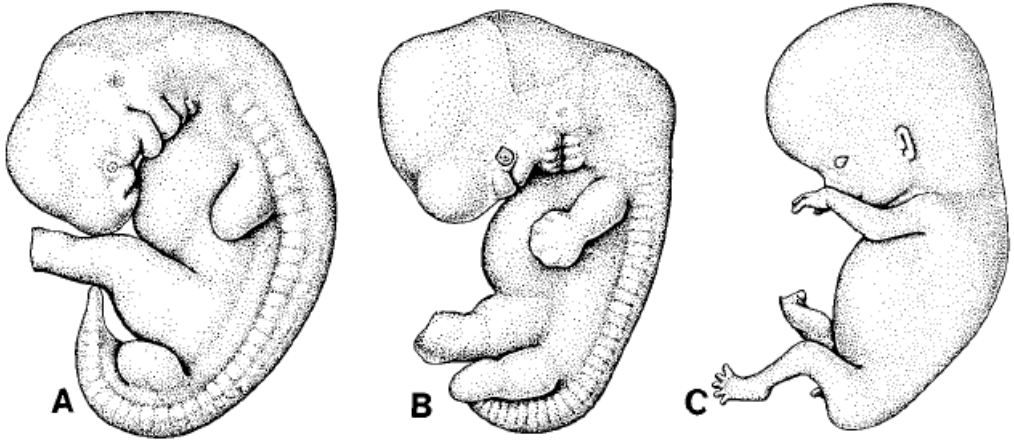
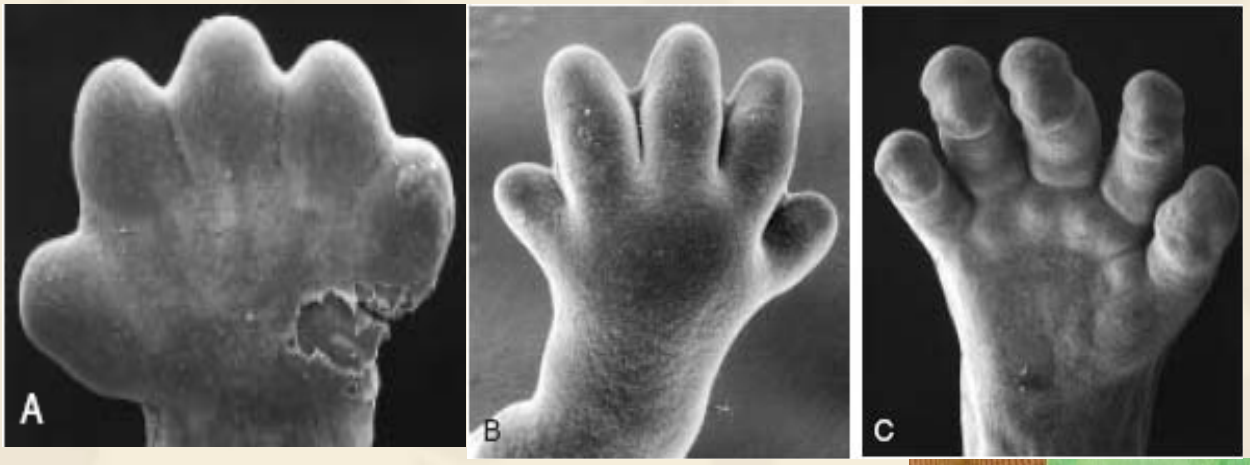


Figure 8.12 Development of the limb buds in human embryos. **A.** At 5 weeks. **B.** At 6 weeks. **C.** At 8 weeks. The hindlimb buds are less well developed than those of the forelimbs.

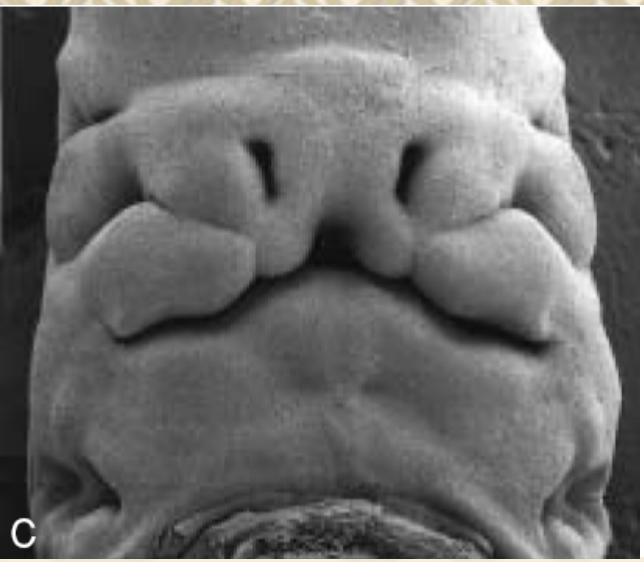
Because of the fusion of two hindlimb buds

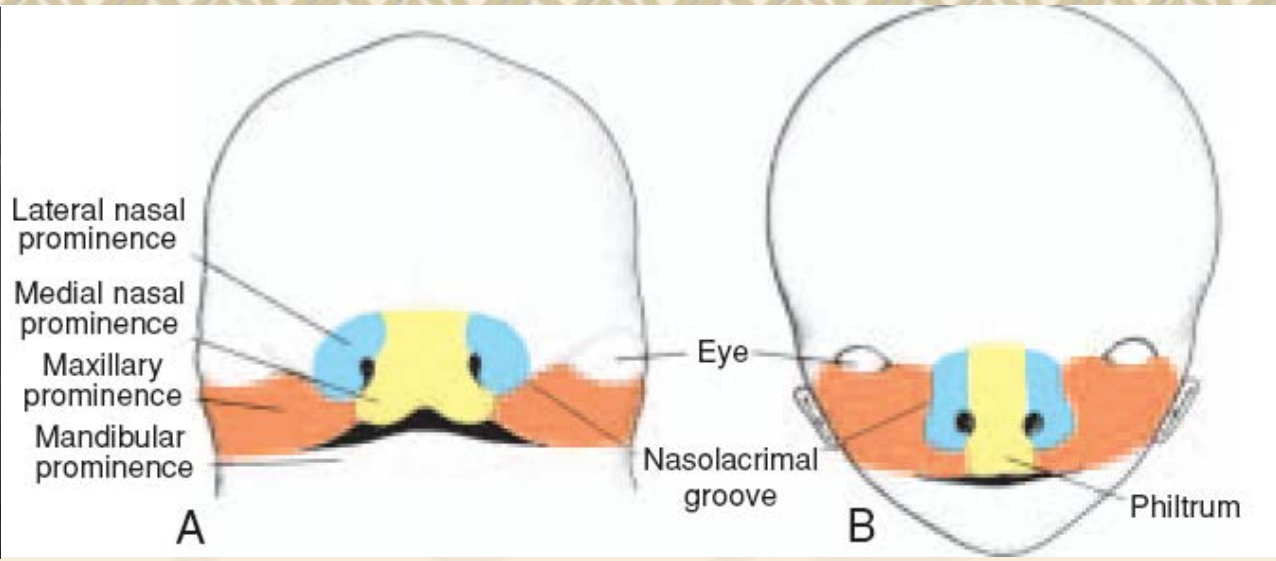
Syndactyly/syndactylism



Scanning electron micrographs of human hands. **A.** At 48 days. Cell death in the apical ectodermal ridge creates a separate ridge for each digit. **B.** At 51 days. Cell death in the interdigital spaces produces separation of the digits. **C.** At 56 days. Digit separation is complete. The finger pads will create patterns for fingerprints.

Because no apoptosis occurs between two cartilaginous digital rays





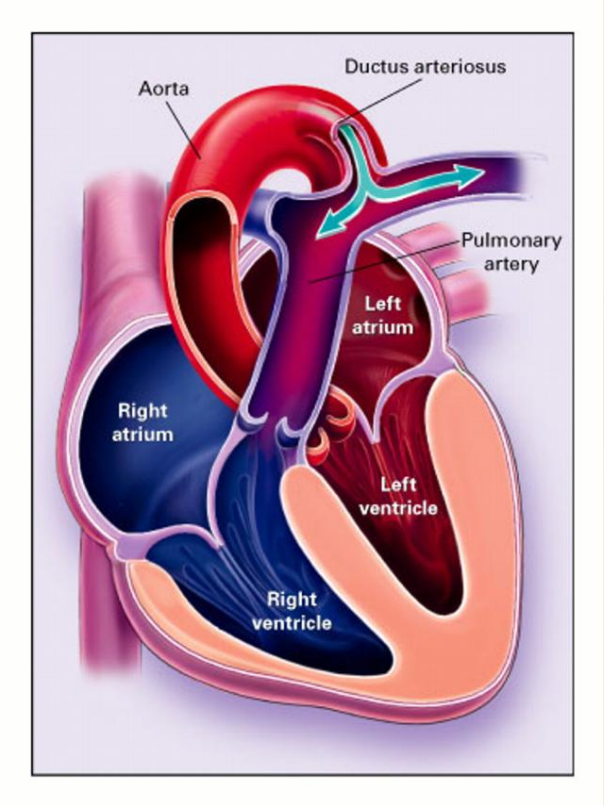
The cause of lateral cleft lip is maxillary prominence dose not fuse with homolateral medial nasal prominence.

Cheiloschisis/chiloschisis/Cleft lip

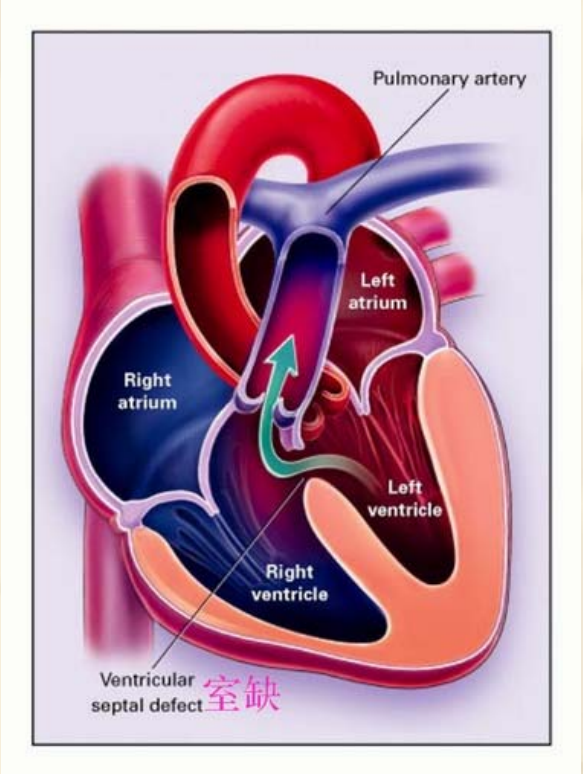
***Cheilo*- lip/edge+ *schisis* cleft**

3) Partly deformation of organs

- failure of organic development or underdeveloped organ



Because ductus arteriosus/arterial duct remain open over afterbirth, there is connection between aorta and pulmonary artery.

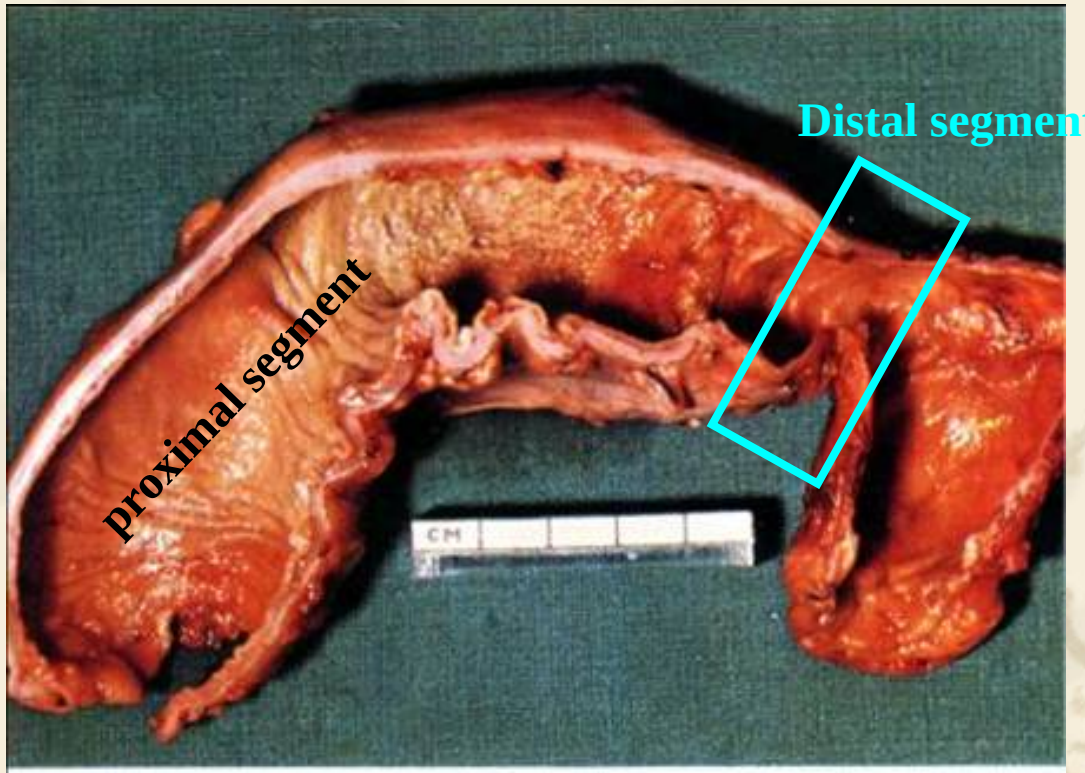


Because of interventricular septal defect in membranous part , there is connection between left ventricle and right ventricle.

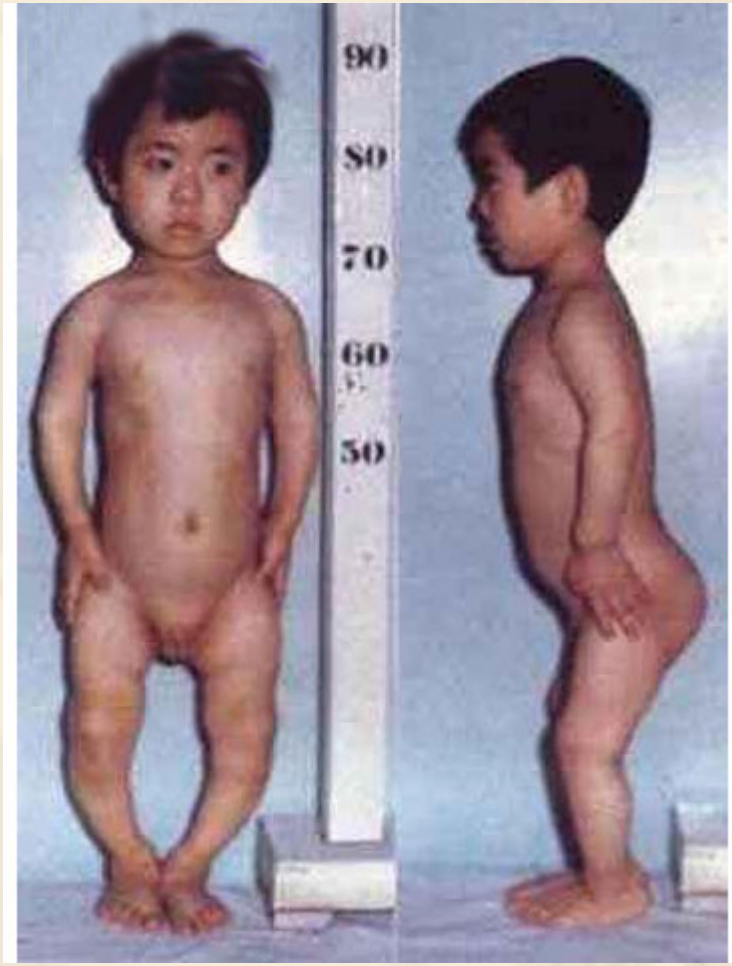
(4) Defective tissue differentiation

- ❖ **in the later stage of embryonic development**
- ❖ **congenital megacolon** ***mega-*** **big**

Because neural crest cells fail to migrate into or penetrate wall of developing colon, defective parasympathetic innervation of distal colon is formed. The last contraction of the distal colon results in feces is accumulated in proximal colon, causing megacolon.



4.55 Congenital aganglionic megacolon



Achondroplasia

Achondroplasia in a 15-year-old boy. Note dwarfism of the short limb type, the limbs being disproportionately shorter than the trunk. The limbs are bowed; there is an increase in lumbar lordosis; and the face is small relative to the head.

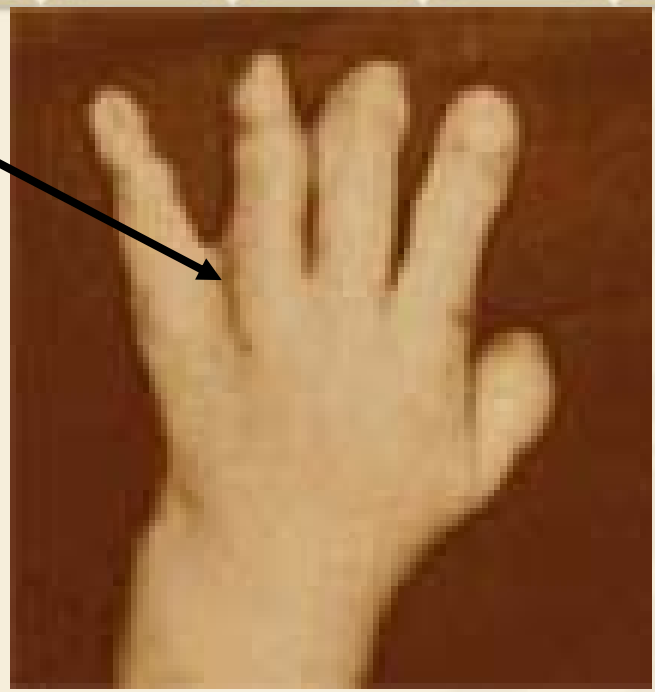
(5) Hypergenetic teratosis

- excessive development of a part of organ or the whole organ
- polydactyly

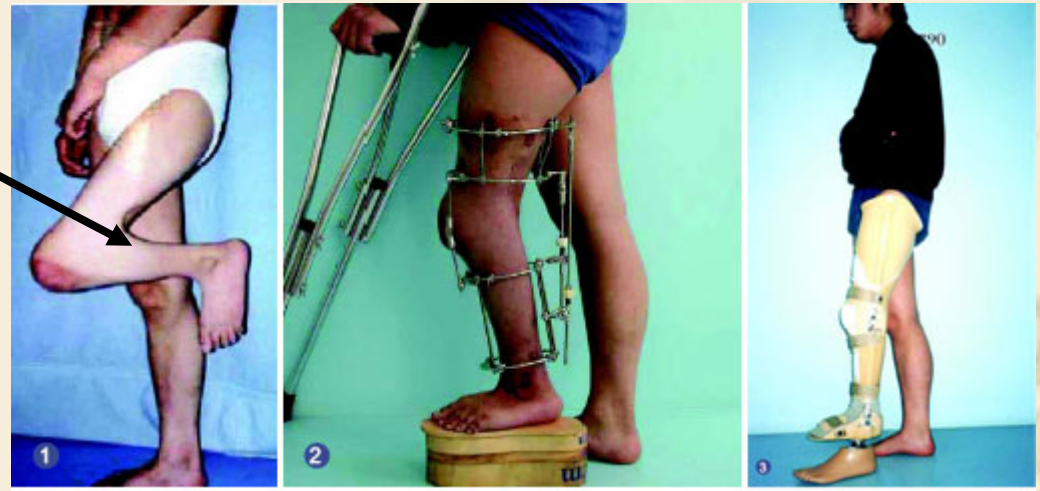
(6) Defective absorption

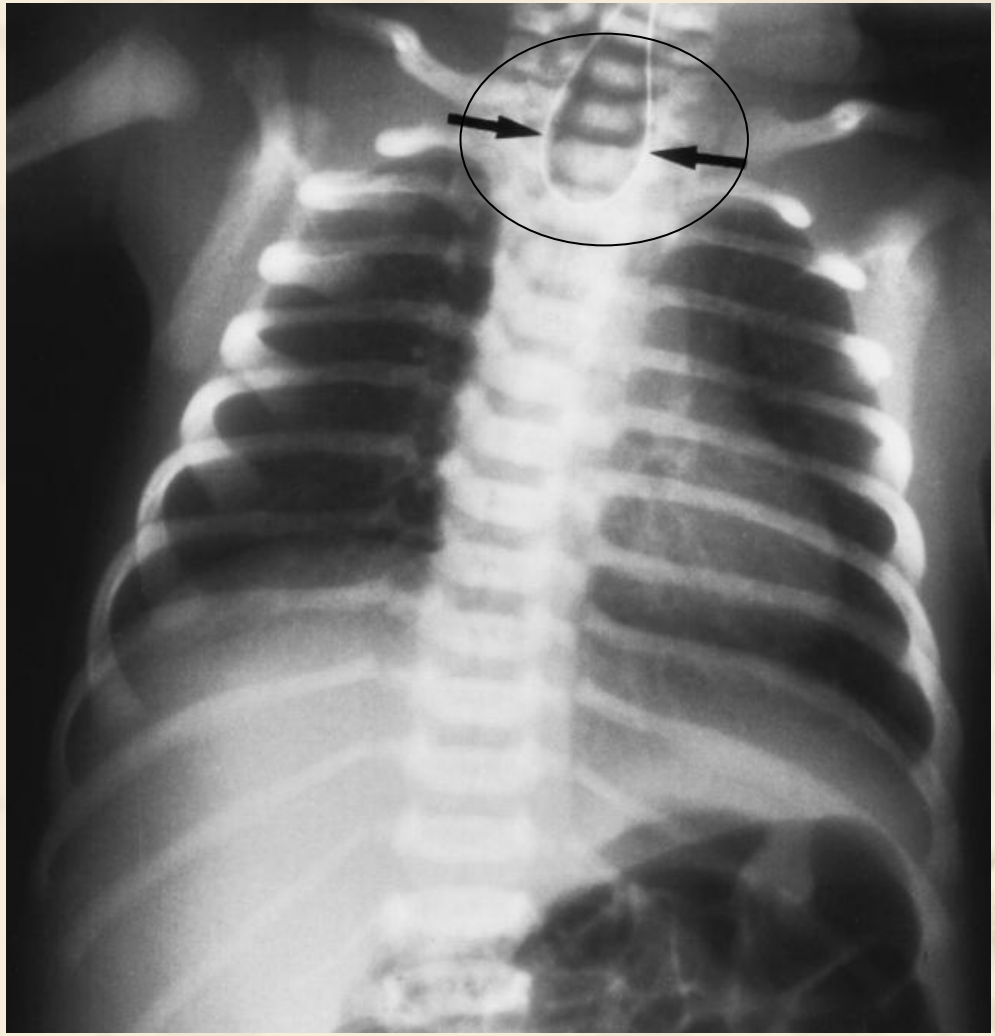
- **Some structures are not absorbed .**
- **Unabsorbed caudal vertebra-----a tail**

Finger web



Knee web



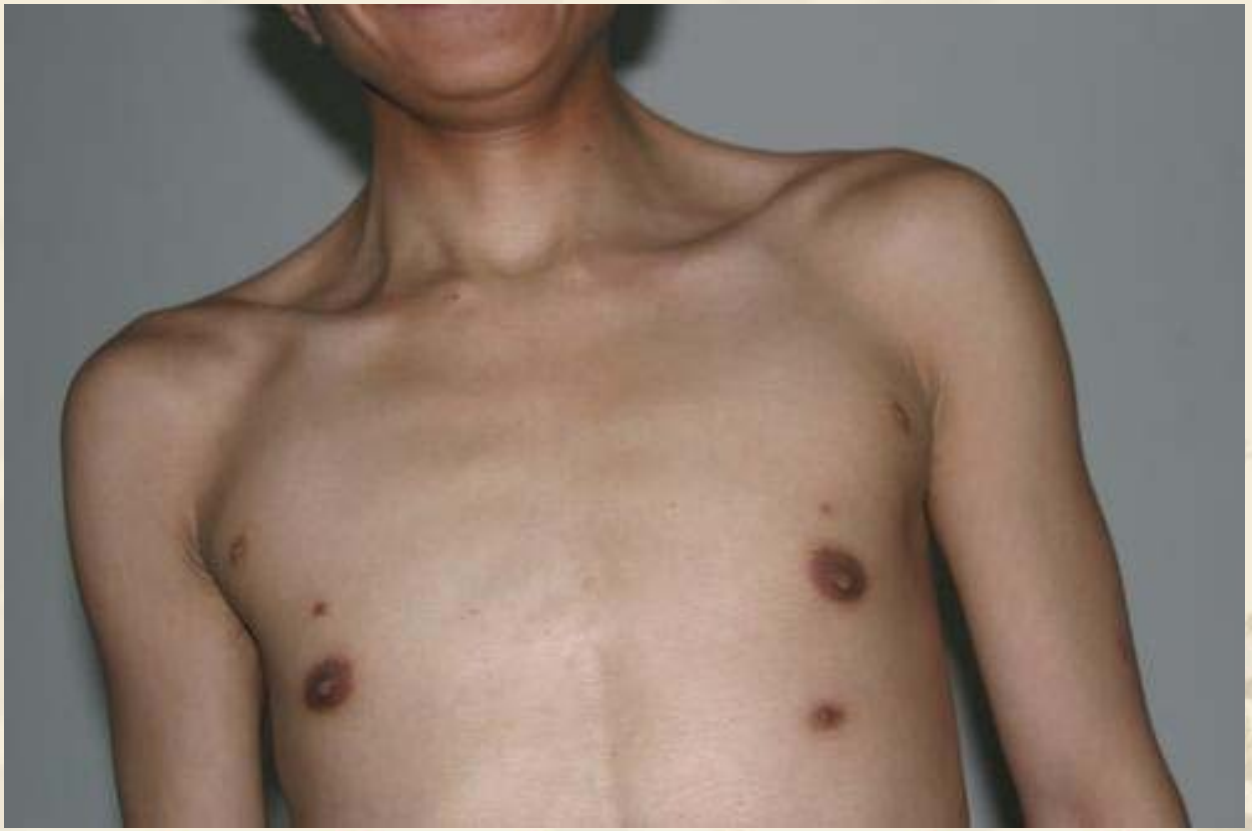


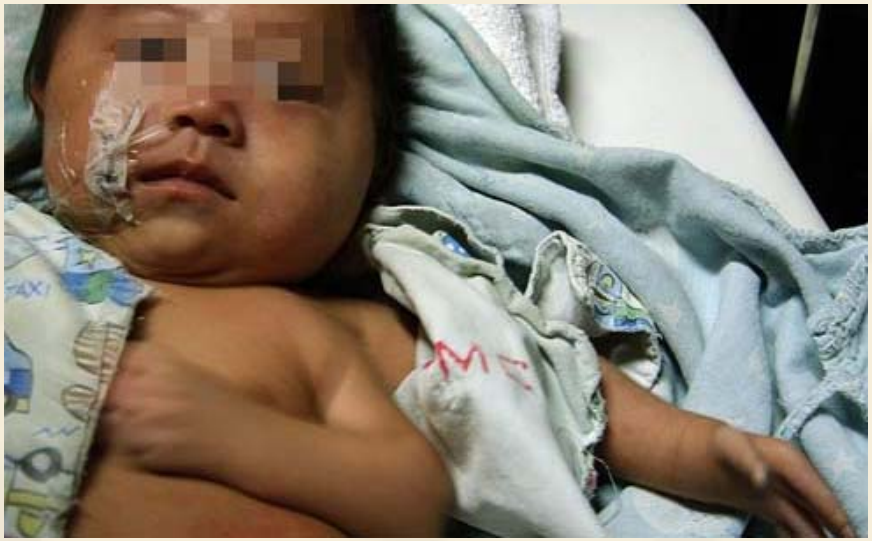
esophageal atresia, irradiated iodine visualization

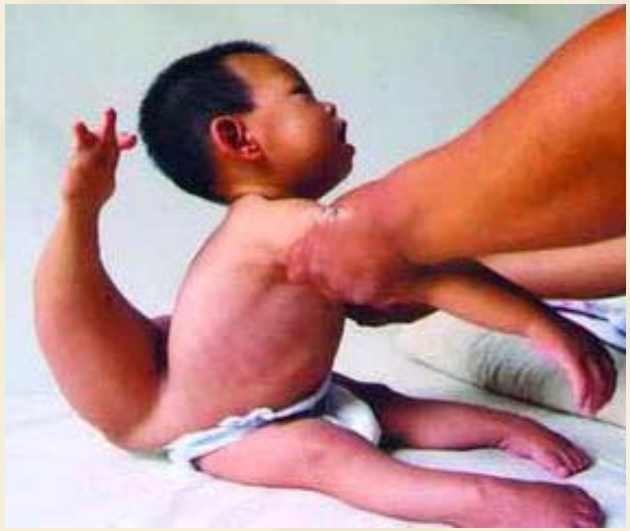
（7）Ectopic teratism & superfluous development

- The number of organ primordium is more than the normal.
- Organ primordium is formed at abnormal position

- hypermastia



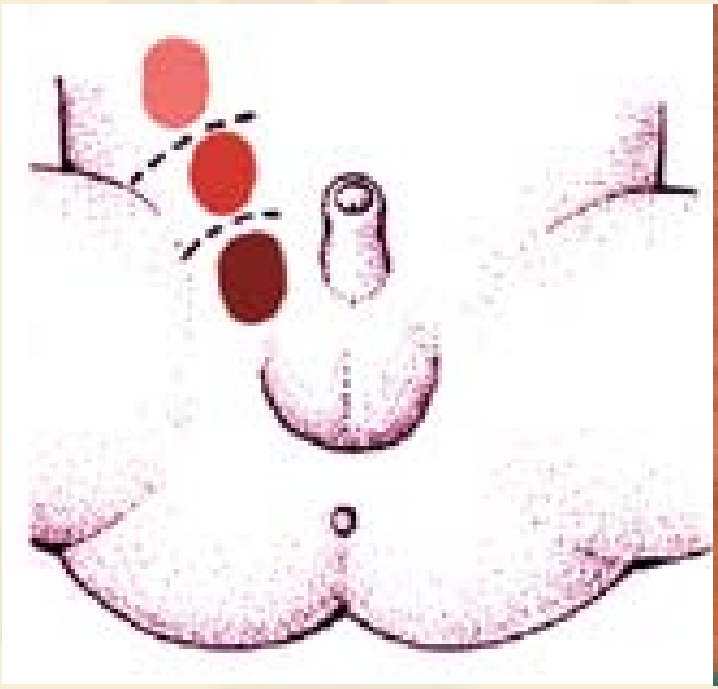


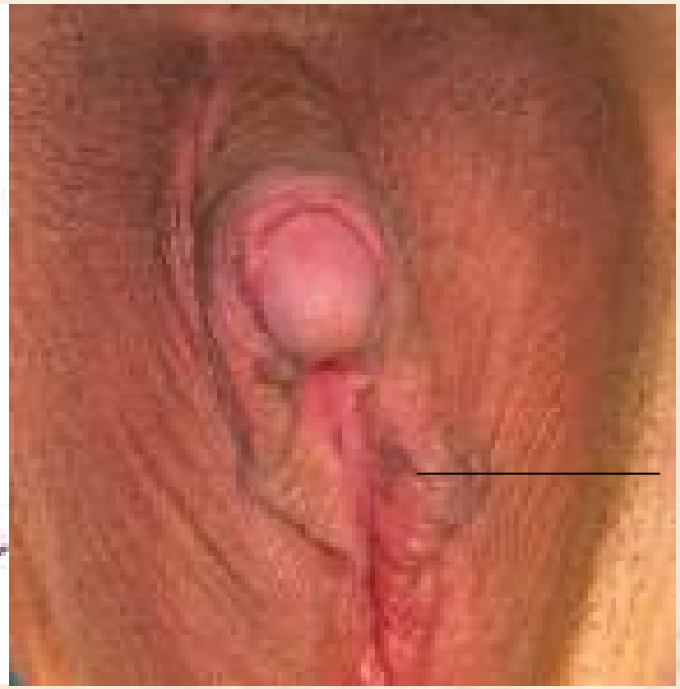


Polymelia

（8）Detained development

- Organic development stop at some point of the midway
- Cryptorchidism: undescended testis

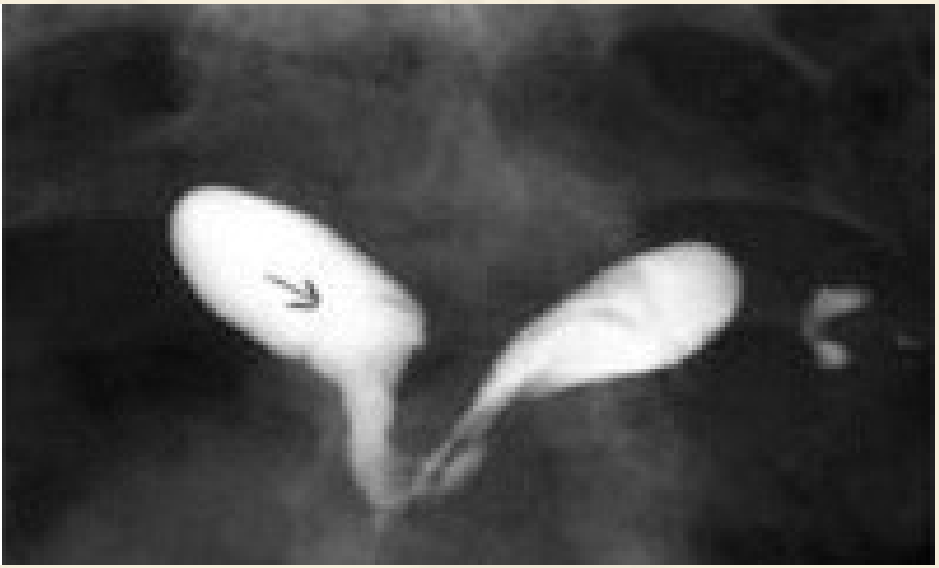




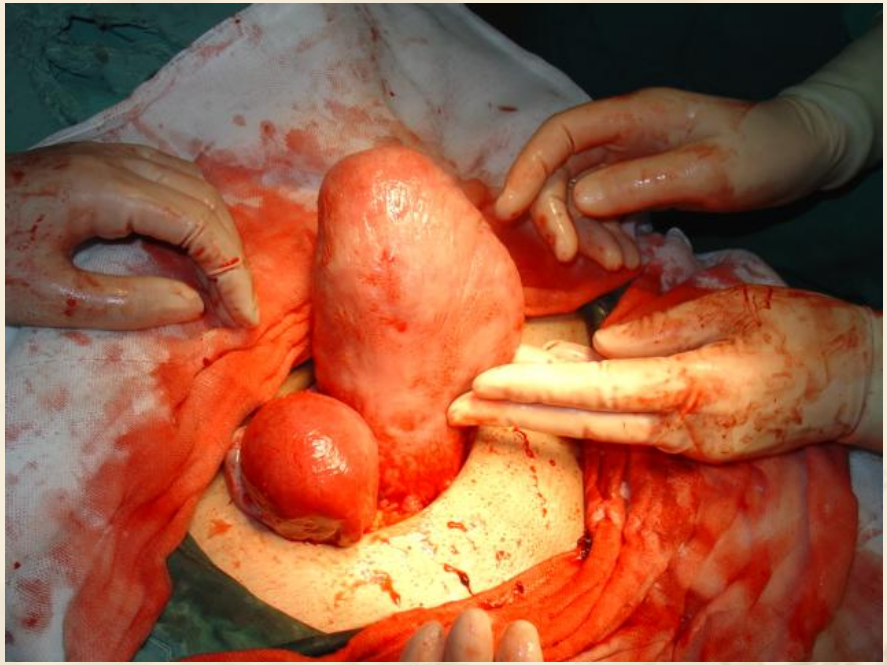
Empty scrotal sac

The testis may be hidden in inguinal groove or abdominal cavity.

One of the relatively common anomalies is the uterus bicornis, in which the uterus has two horns entering a common vagina. This condition is normal in many mammals below the primates.



irradiated iodine visualization



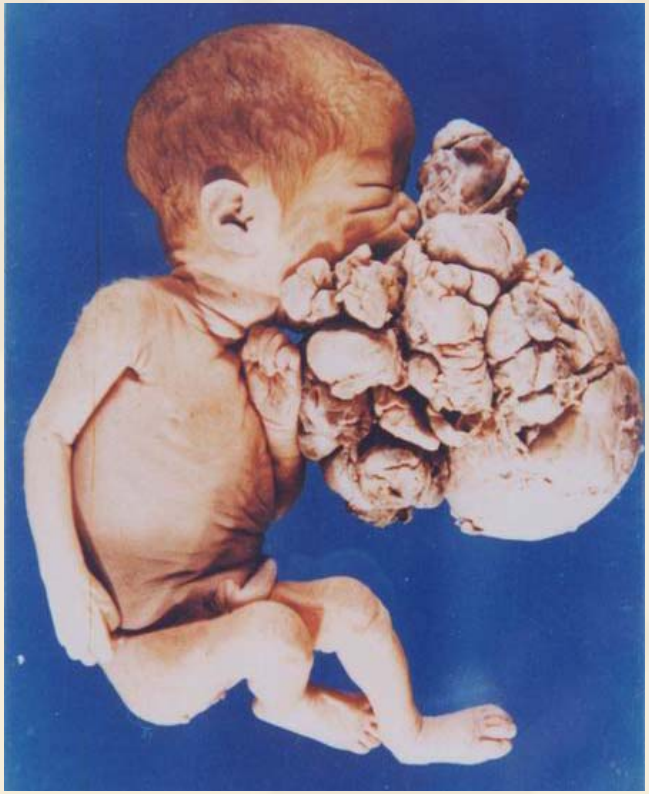
double uterus

(9) Double malformation or duplication

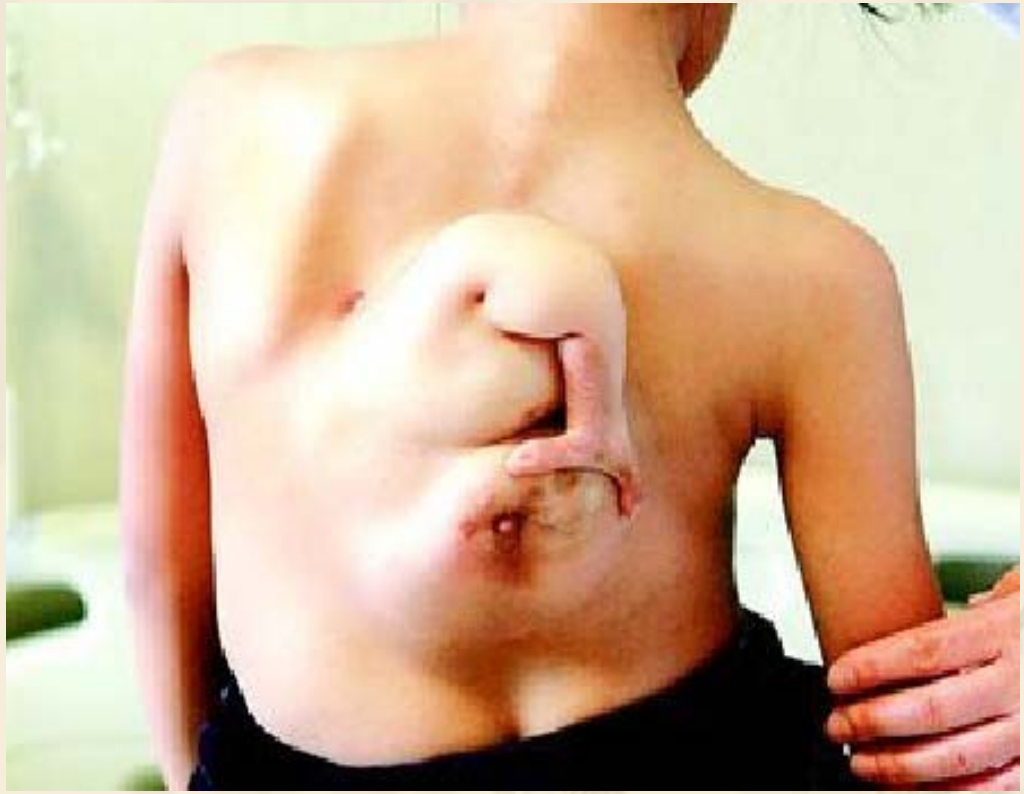
- **Symmetric conjoined twins**

（10）Parasitic malformation

Unsymmetric conjoined twins and teratoma



teratoma



Parasitic fecutus

2. Causes

genetic factor
- chromosomal aberration: number & structure
- gene mutation

environmental factor
- Biologic teratogen (terato = monster, gen = producing)
- Physical teratogen: electromagnetic wave, noise
- Chemical teratogen: water, gas
- Teratogenic medicines
- Others: smoking, excessive drinking, hypoxia, severe hypoxia

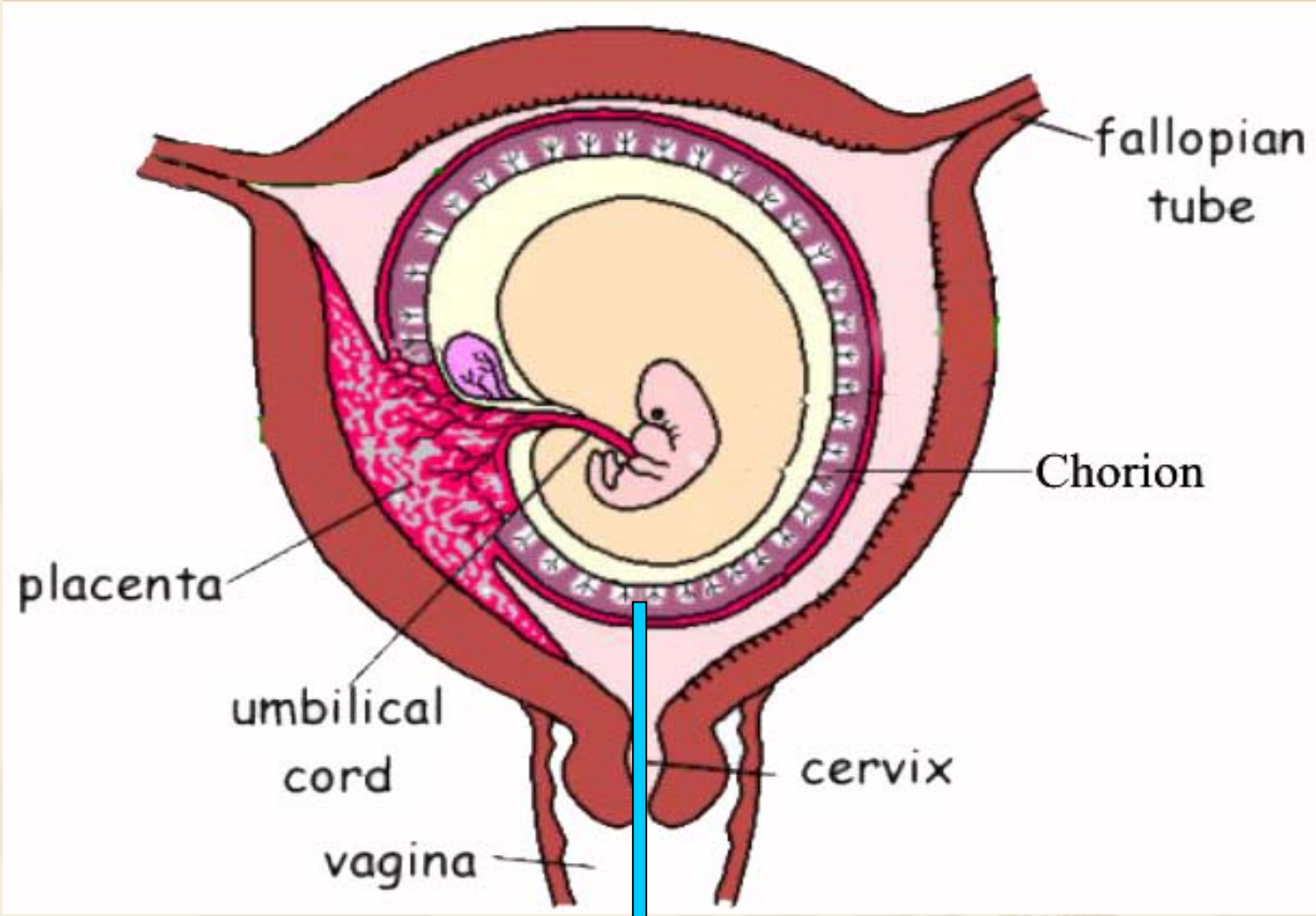
3. **Prevention & prenatal diagnosis**

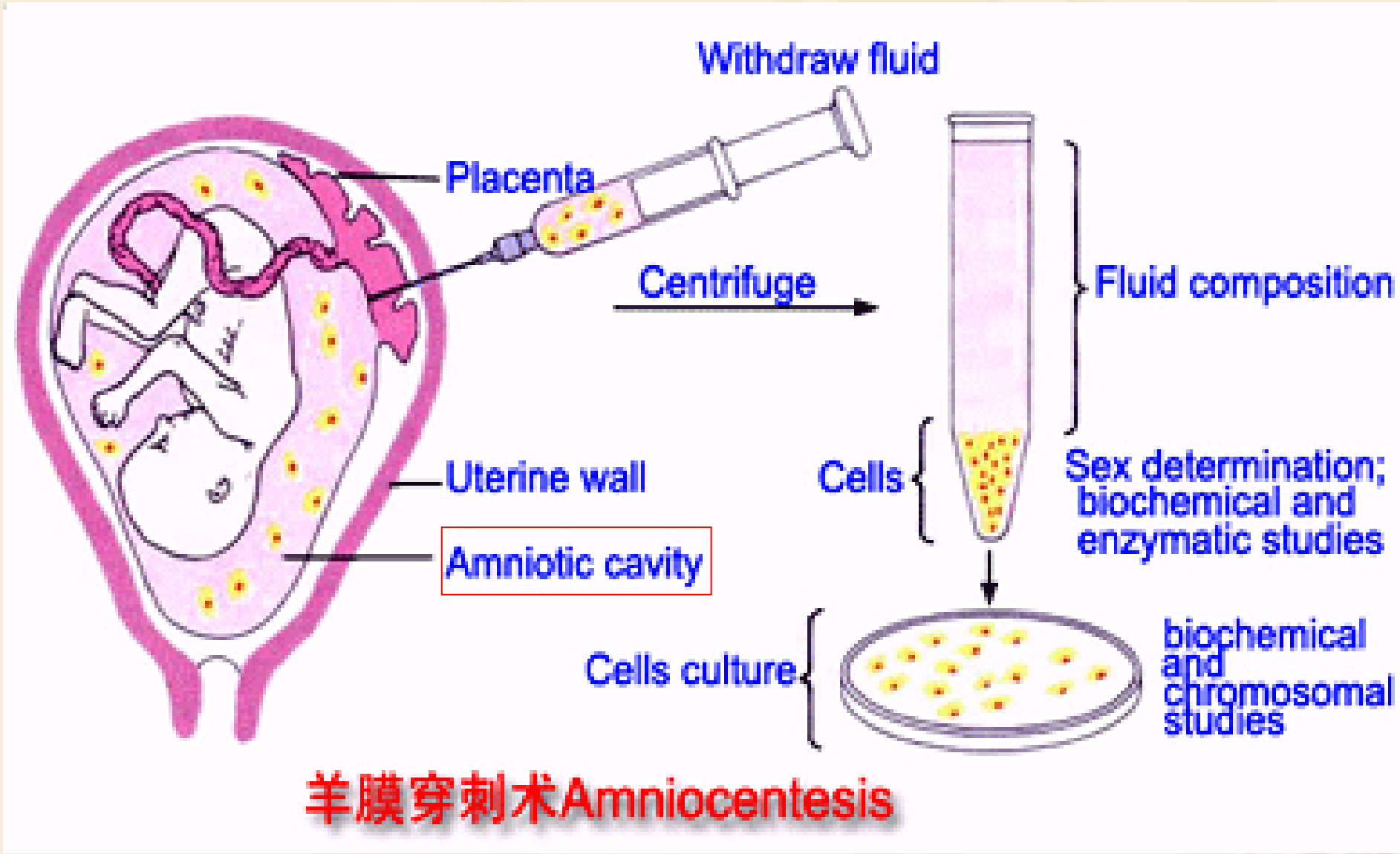
Parents

- **Genetic counseling before marriage**
- **Avoid teratogens during pregnancy**
- **Guardianship of pregnancy**

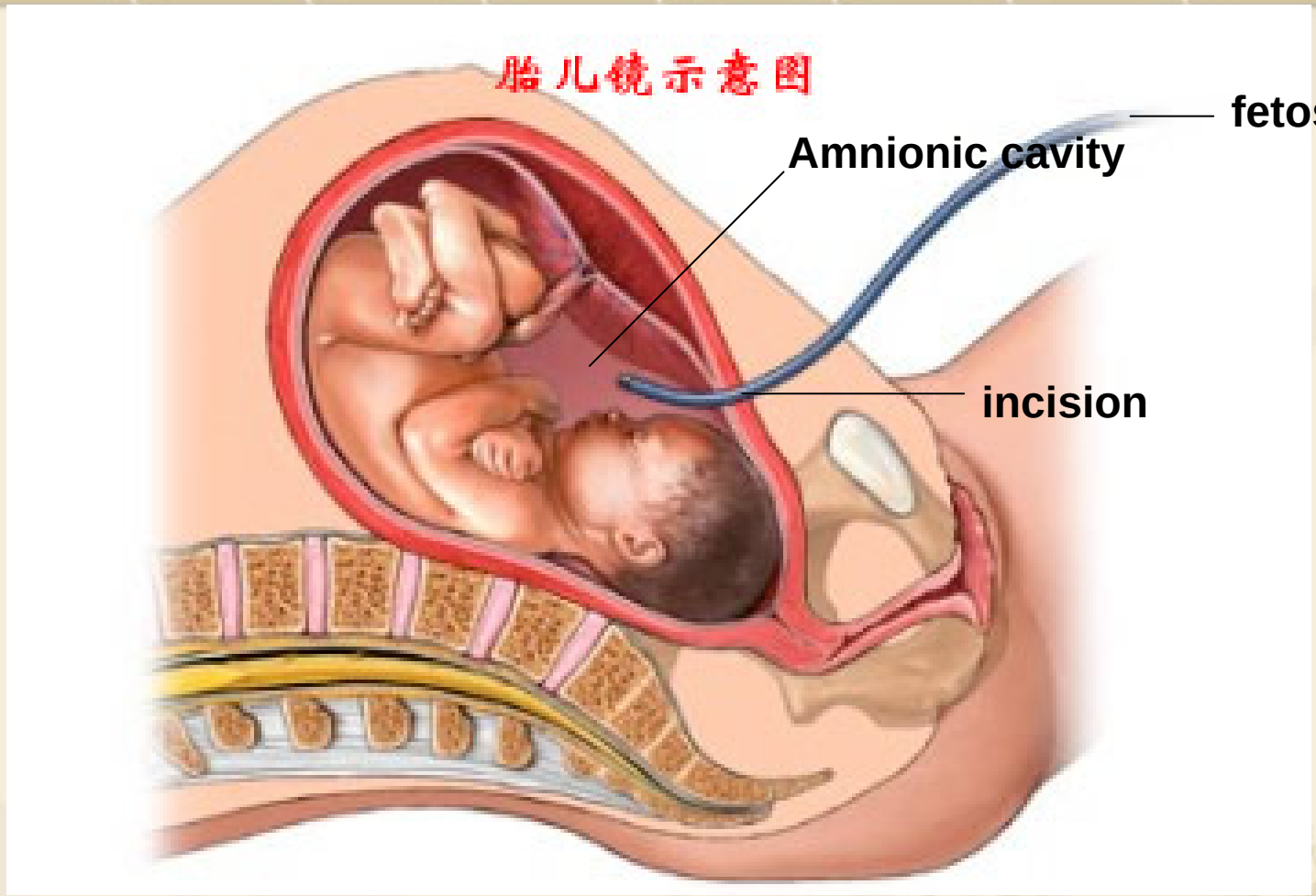
Fetus

- **1. Chorion biopsy: 40-70d**
- **2. Amniocentesis: 16th -20th week**
- **3. Fetoscopy： 15th -20th week**
- **4. Sonography: commonest, no complaints, fast, repeatability**
- **5. X－ray: bone**

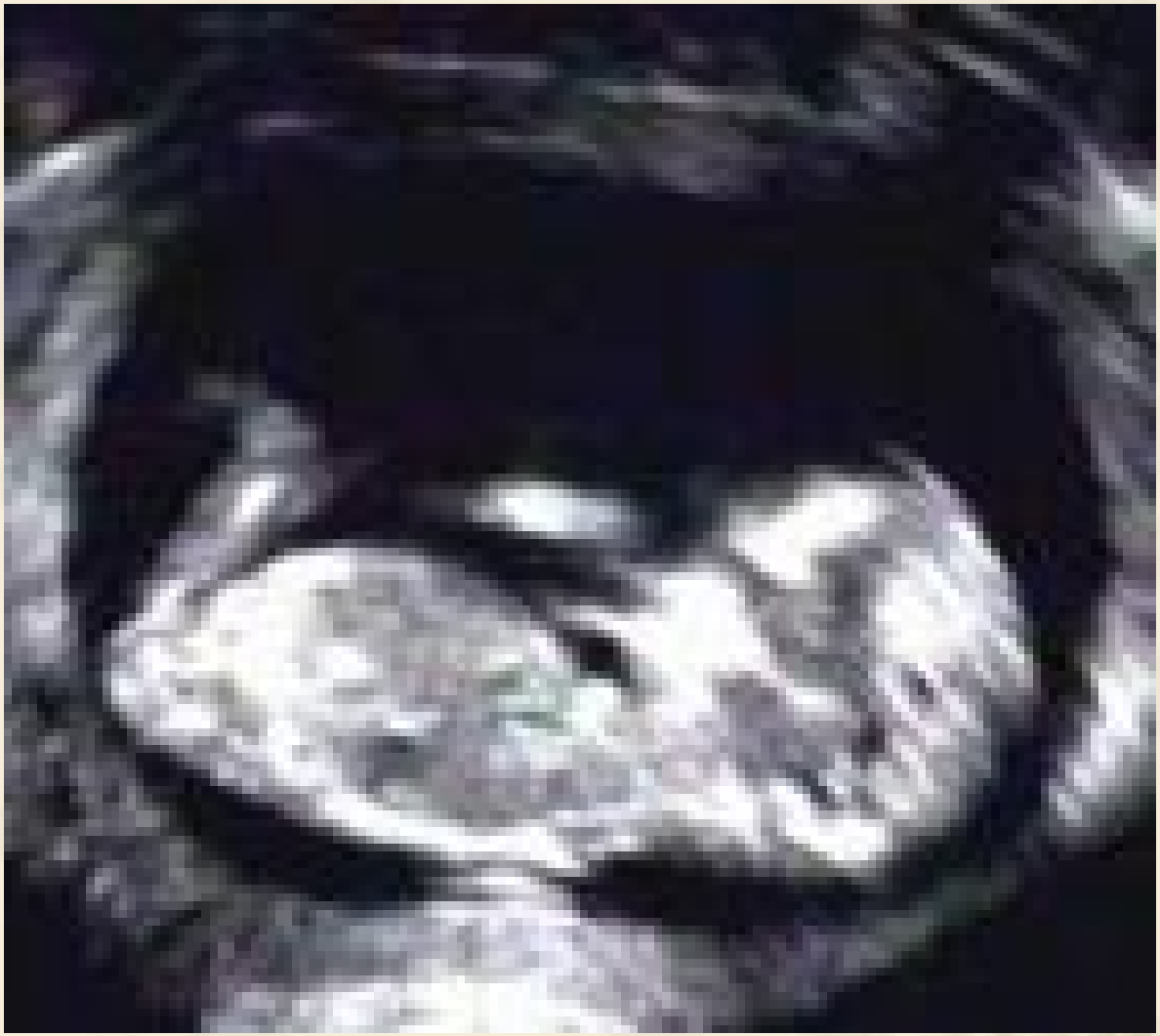

fallopian tube
Chorion
placenta
umbilical cord
cervix
vagina



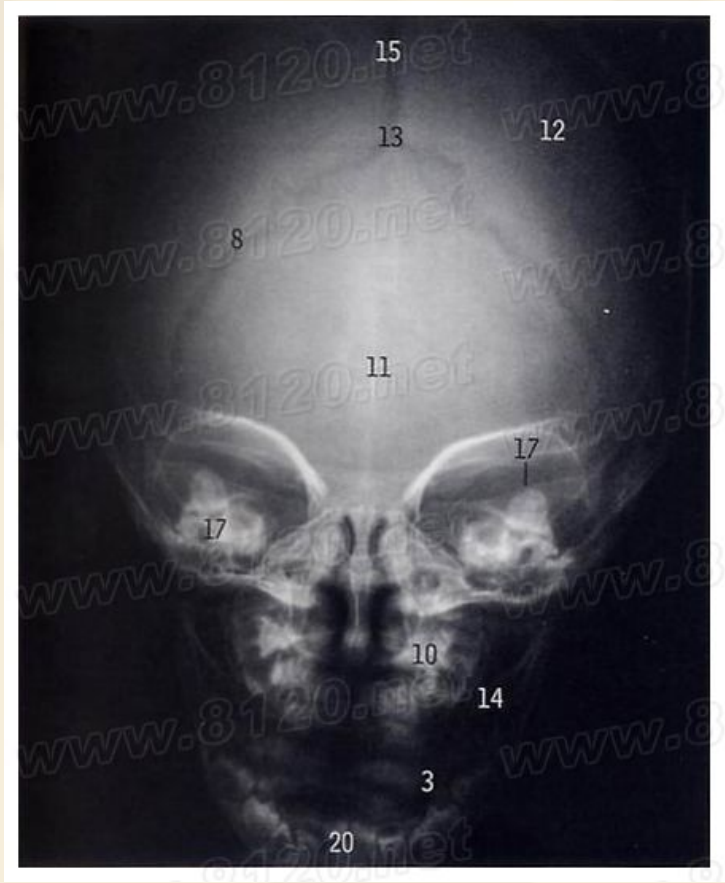
羊膜穿刺术Amniocentesis

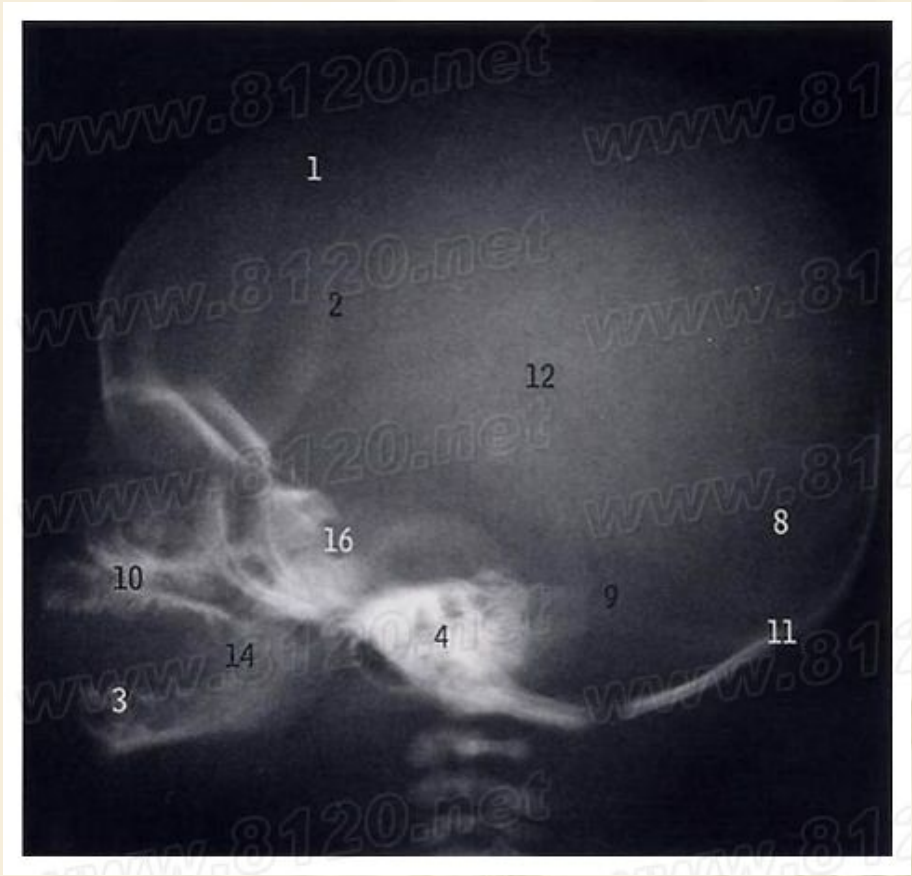


Record



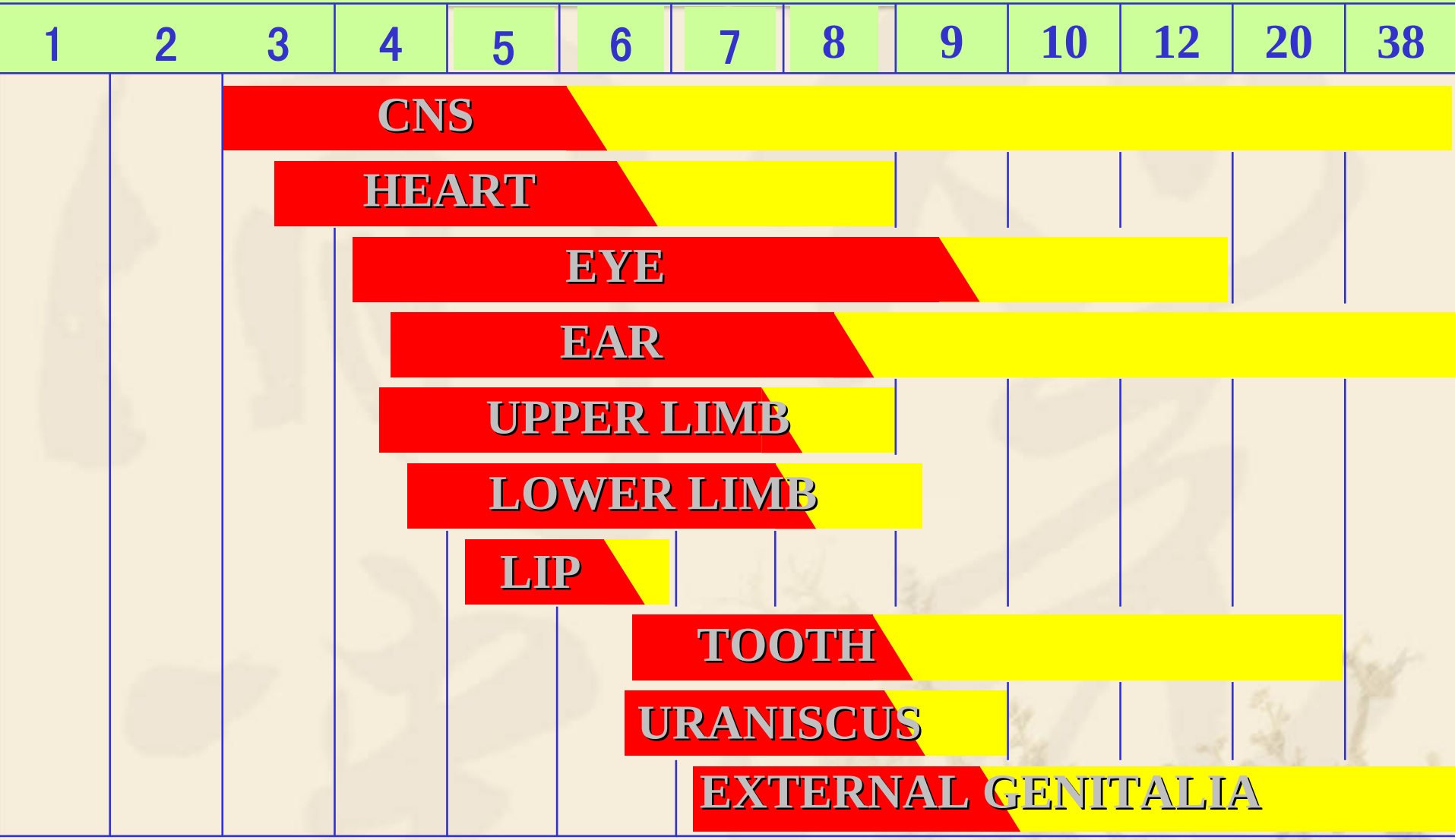
sonogram





Fetal skull detected by x-ray

The period of embryonic development（week，fertilization age）



■high sensitivity to teratogenic agent ■low sensitivity to teratogenic agent

sensitive period to teratogenic agent in human fetus: from 3rd to 9th week.

Important points

- The sensitive period of embryonic development to teratogen.