Ultrasonography for pregnant women

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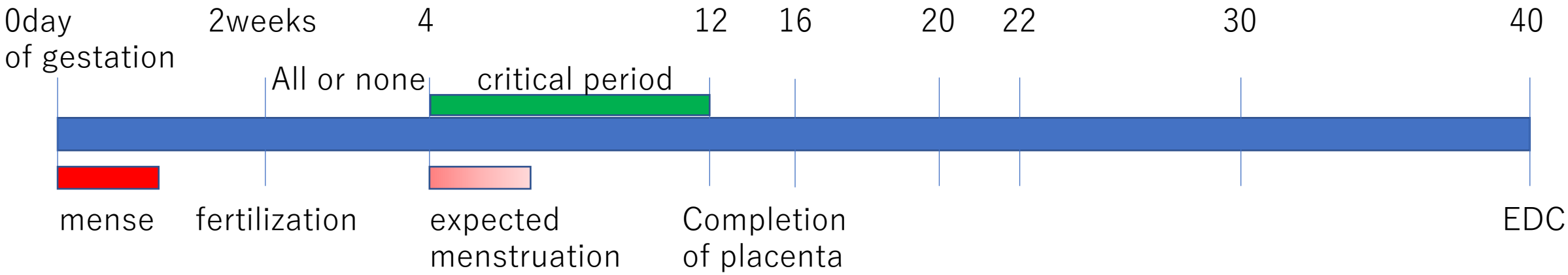
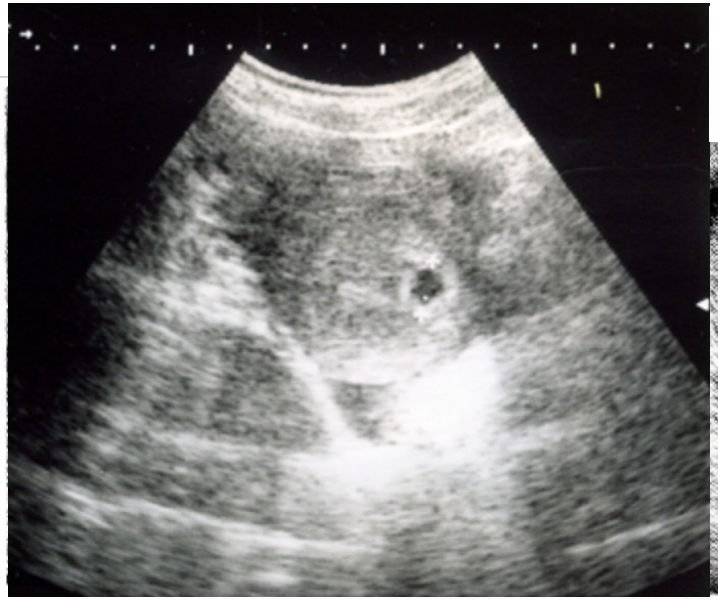
In the beginning of pregnancy by transabdominal ultrasonography

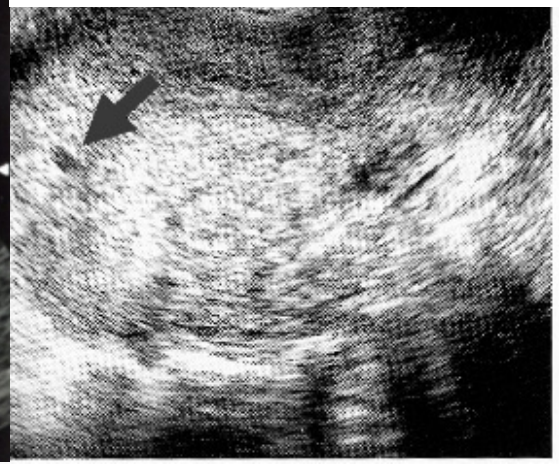
- **fertilization ・implantation～8th weeks of gestation：embryo**
  - 5th～6th ：gestational sac（GS）visible（pseudo GS in ectopy）
  - 7th～10th ：GS（gestational weeks-4cm）
  - 7th ：yolk sac（YS）visible
  - 8th ：fetal heart beat visible

- **after 8th weeks of gestation：fetus**
  - 10th ：CRL is accurate（gestational weeks-7cm）

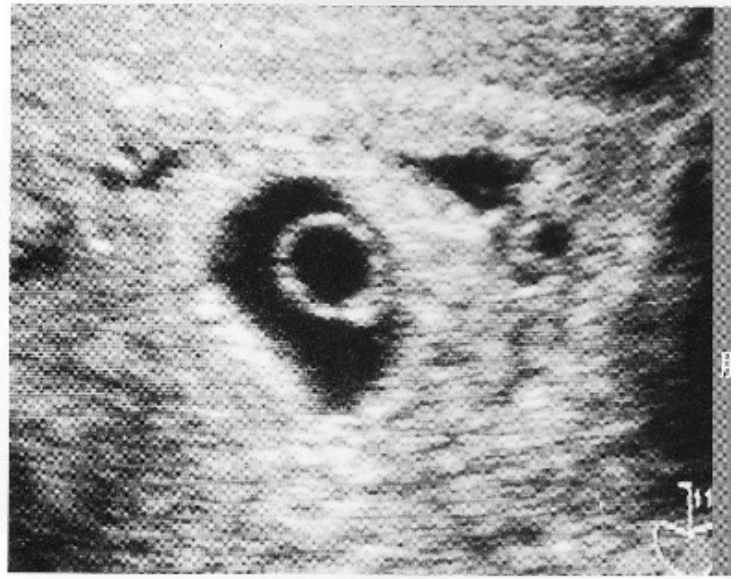


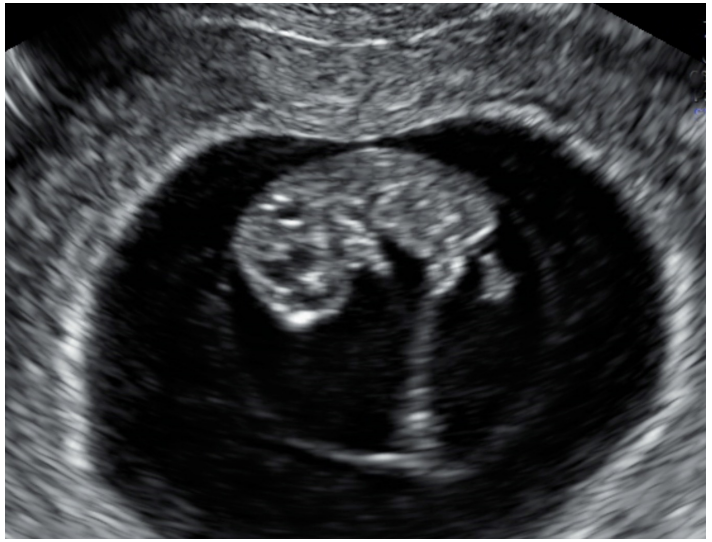


GS:
5th weeks of gestation



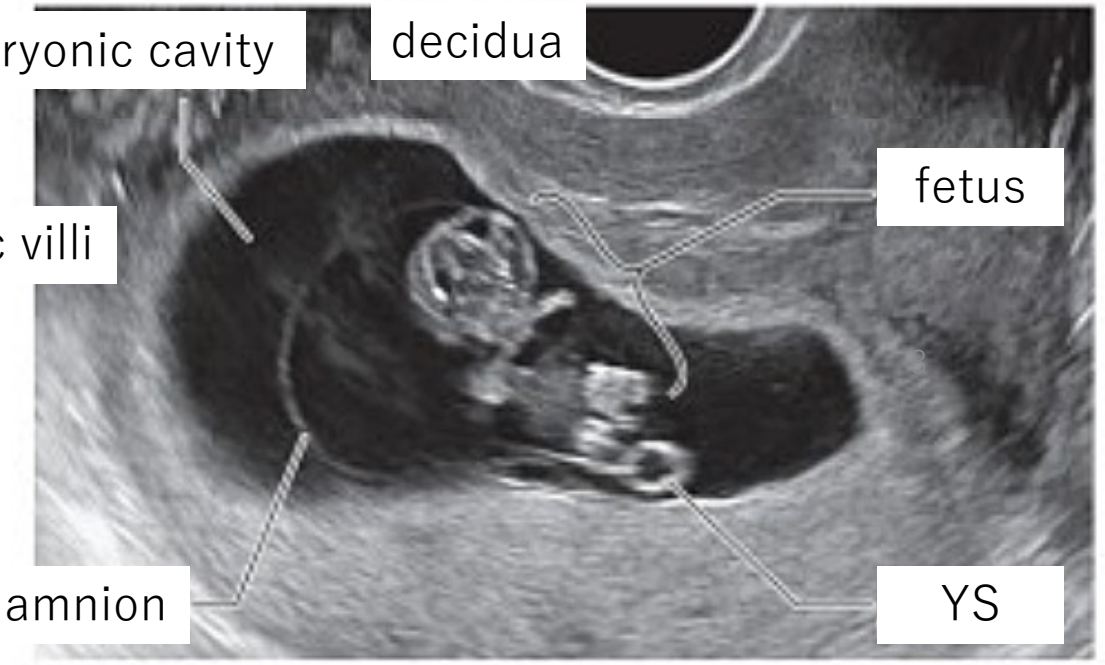
YS
6th weeks of gestation





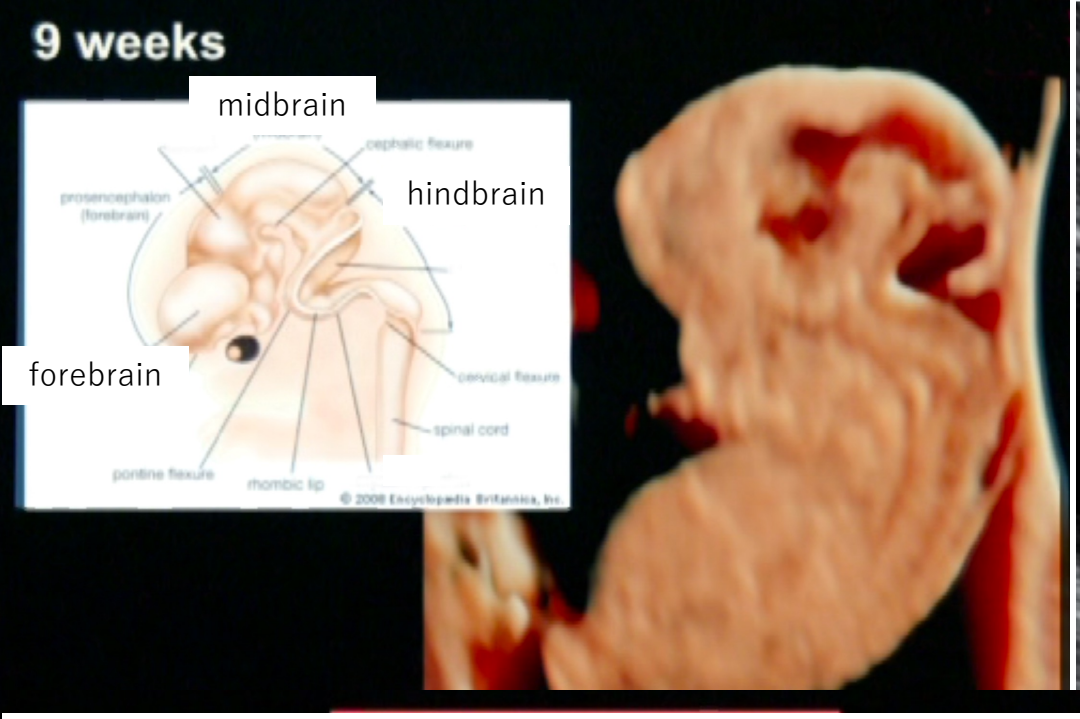
CRL:
9th weeks of gestation

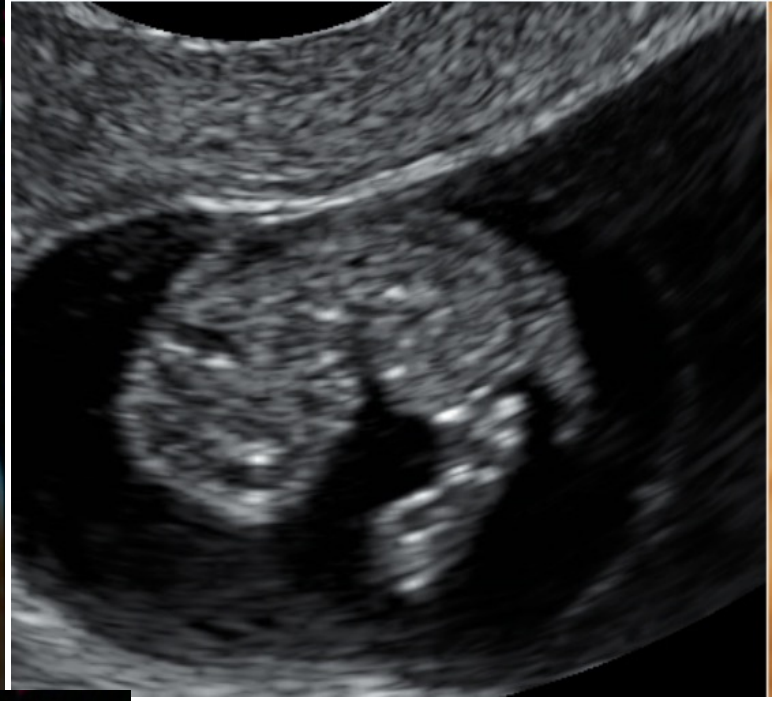
9th weeks of gestation

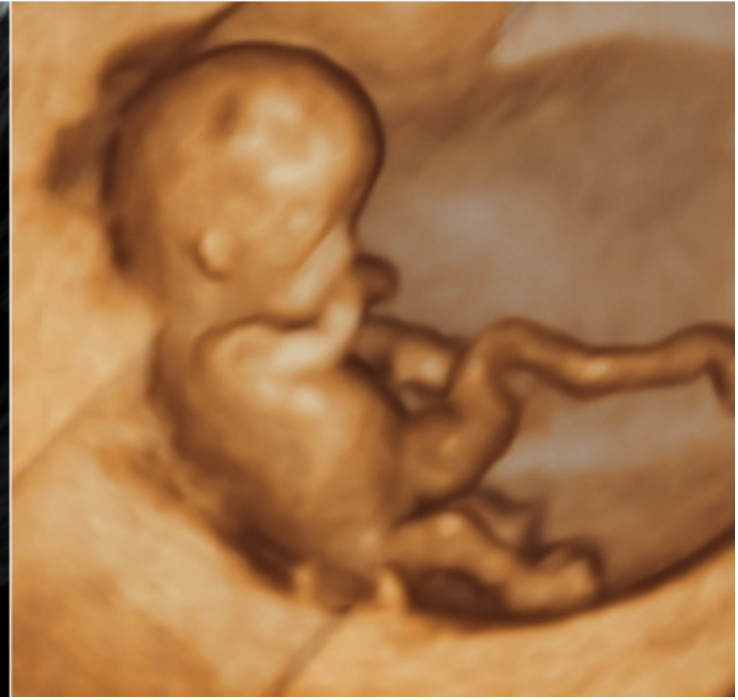


Ultrasonography at 7-8 weeks of gestation

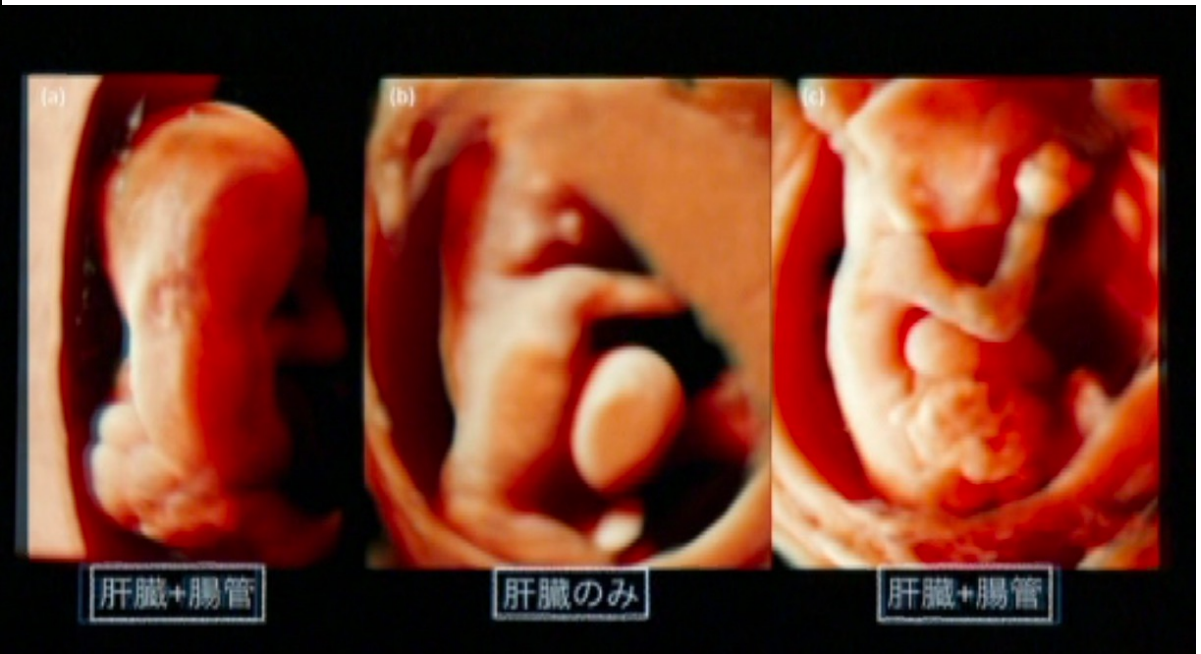
- 7th　　：brain vesicles in the cranium of embryo (→ p21)
- 8th　　：fetal movement
- end of 8th　：　physiological umbilical hernia　(diameter < 7 mm)
  (repaired by 12 weeks of gestation)
- luteal cyst：　Shrinking at 12-16 weeks of gestation
  (after placentation)







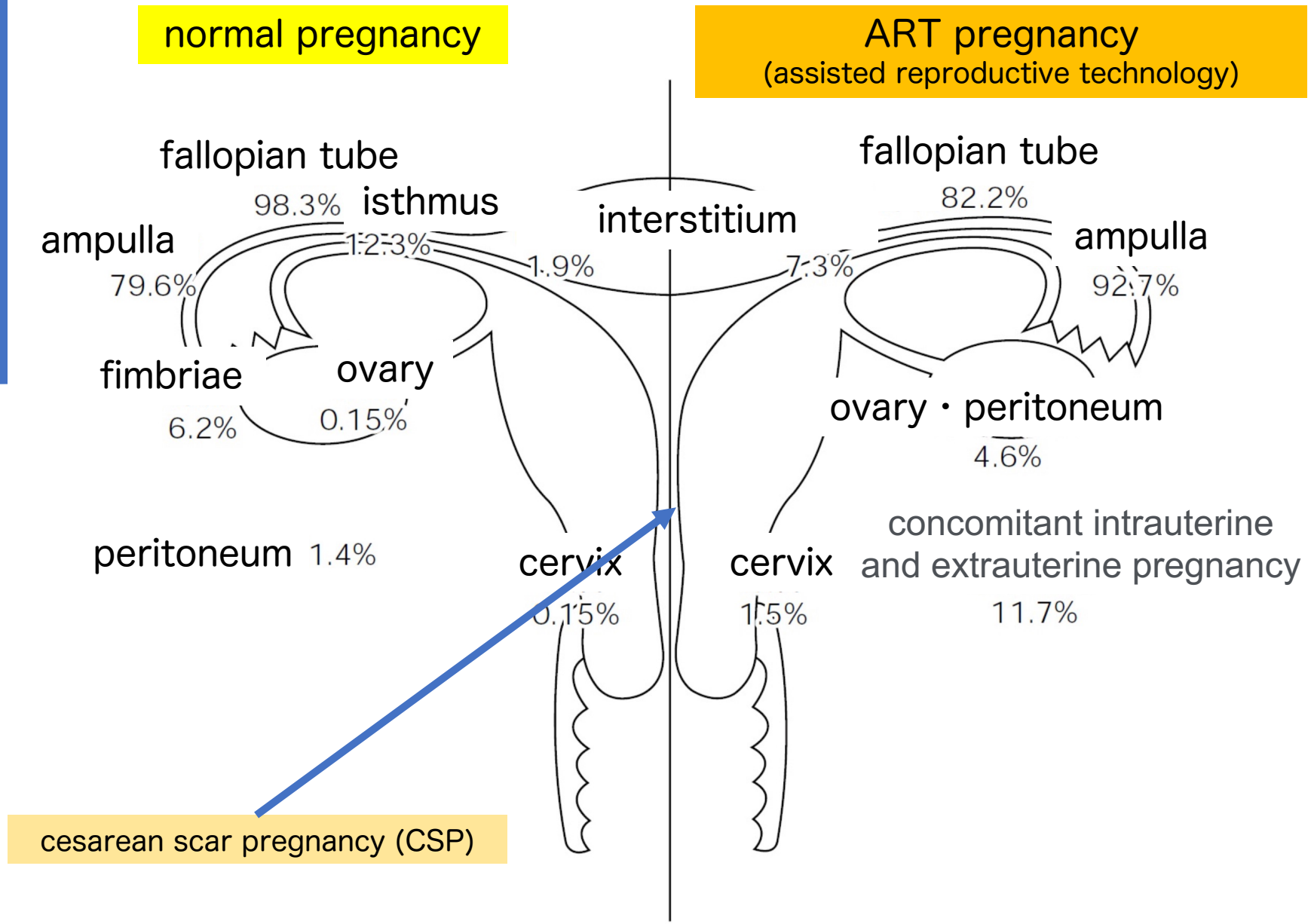
pathophysiological umbilical hernia (diameter ≧ 10 mm)



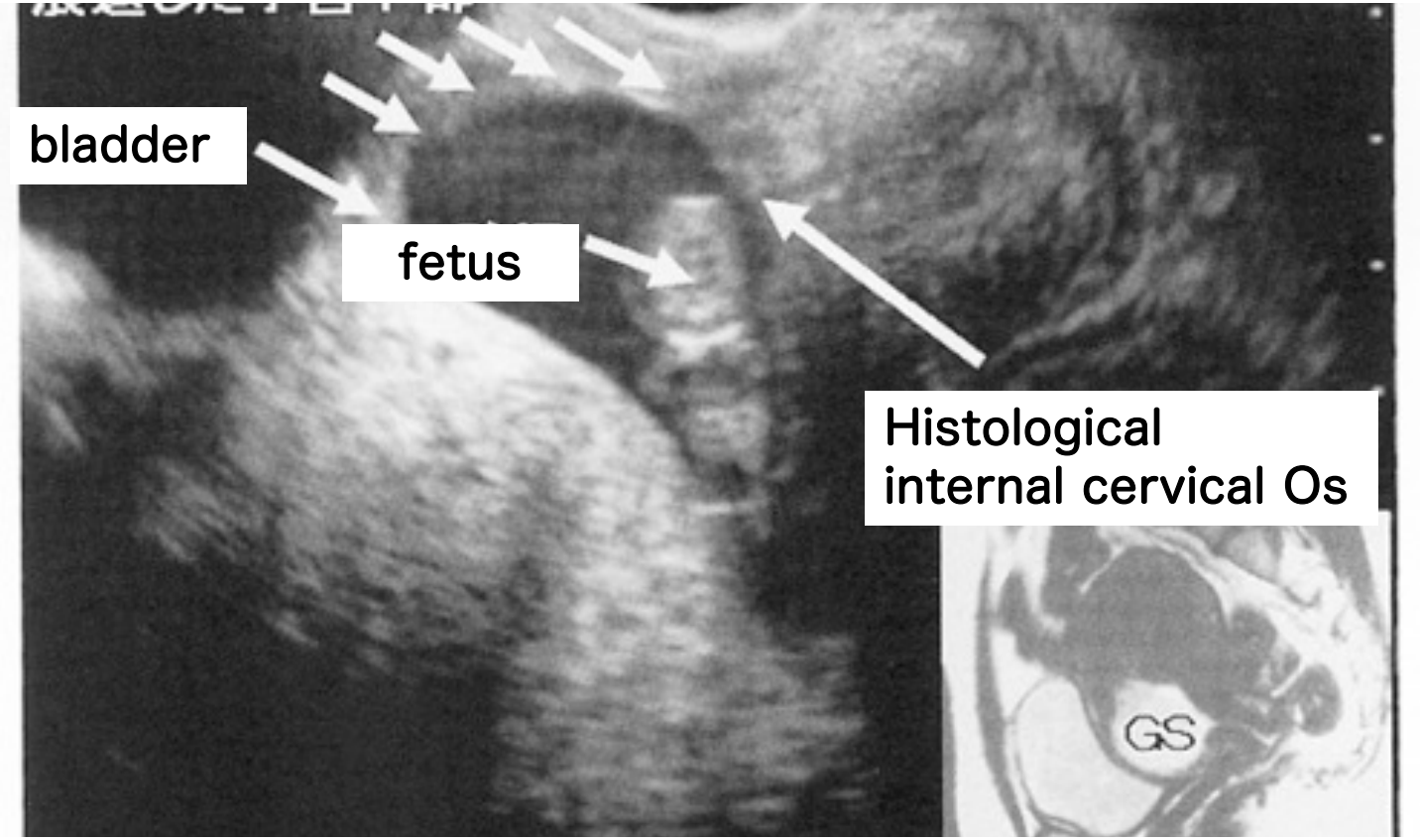
Points to be diagnosed in early pregnancy

- ectopic pregnancy
- myoma
- chromosomal anomaly

Ectopic pregnancy

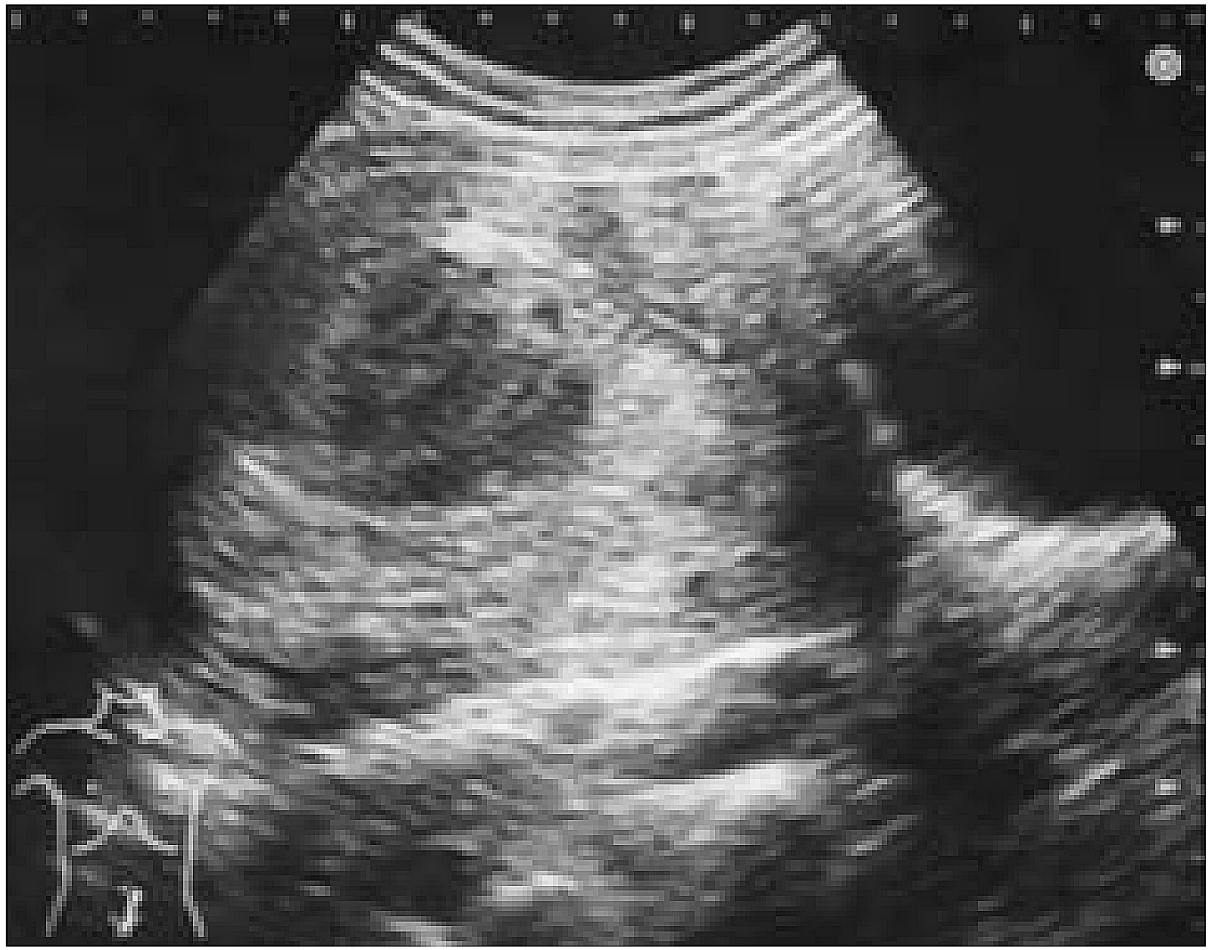


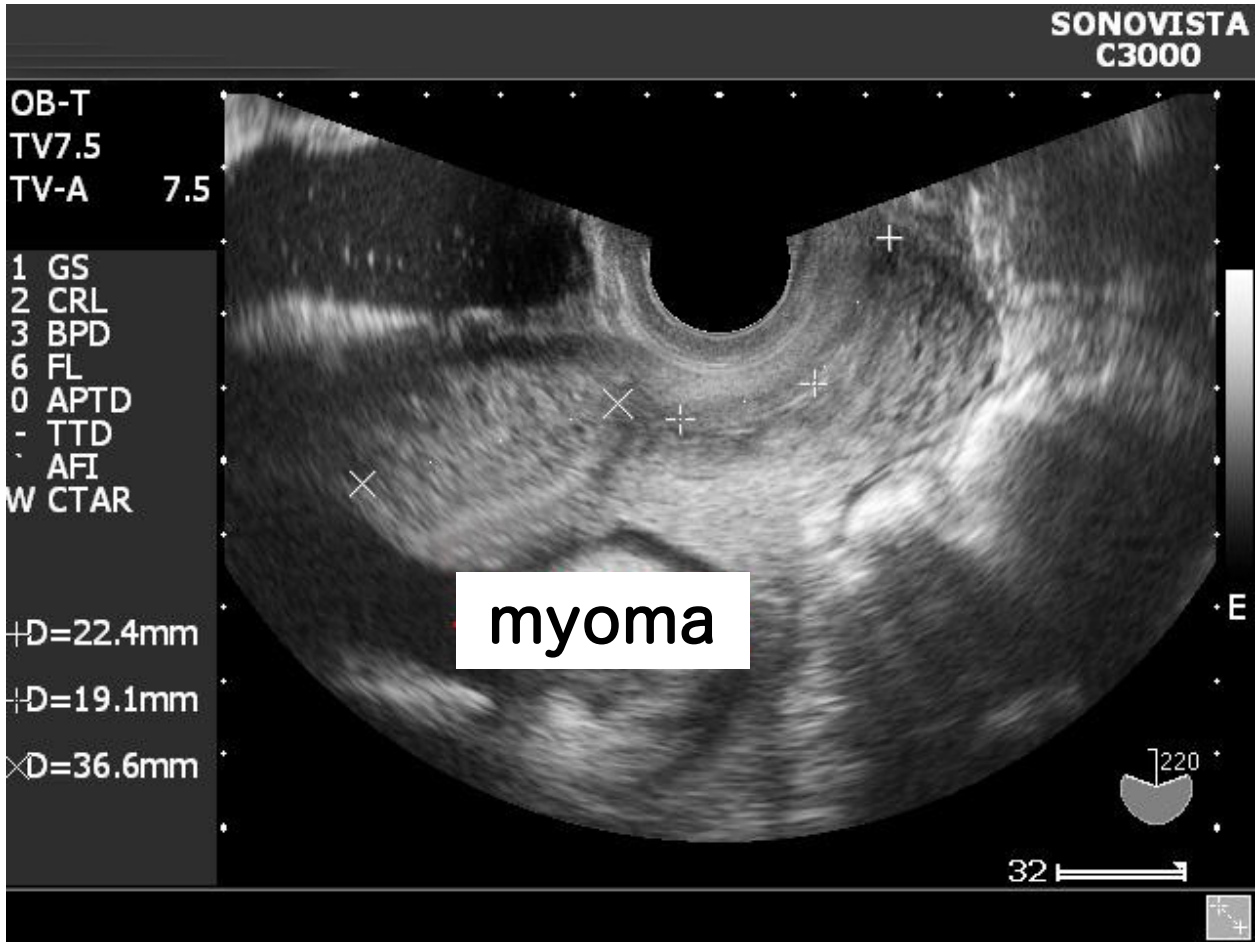
Cesarean scar pregnancy (CSP)



- The lower part of corpus uteri is expanded and a gestational sac is formed despite the early stage of pregnancy. The myometrium is very thin and in a state of silent rupture. If the uterus is to be preserved, conservative treatment is performed by anti-cancer drugs such as MTX (methotrexate) or UAE.

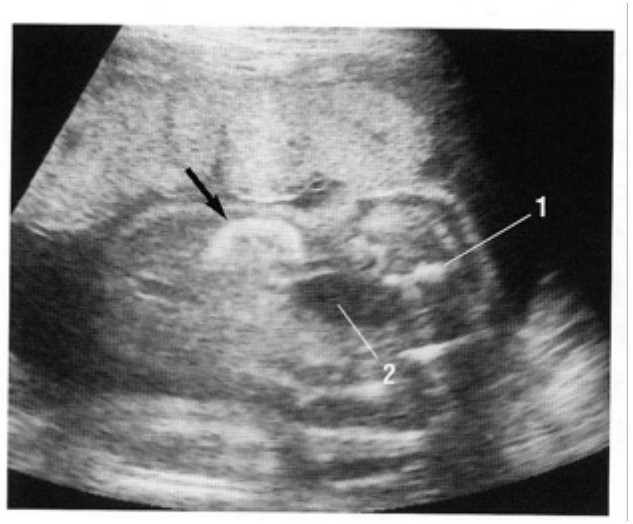
myoma uteri



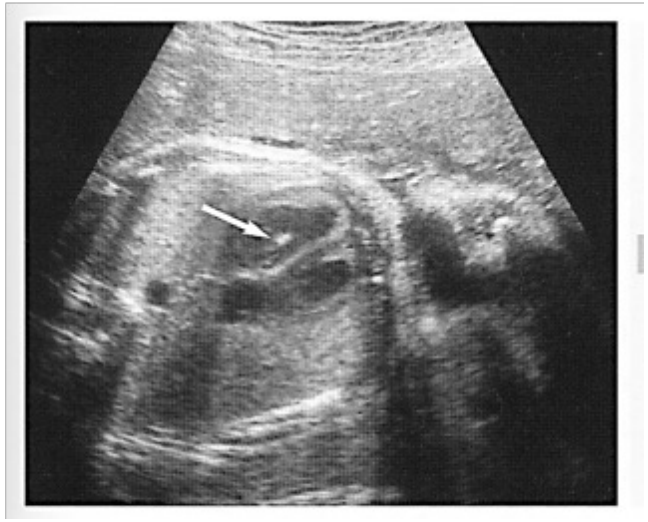


Early in pregnancy, physiological uterine contractions may look like myoma uteri. It is called as Braxton Hicks contractions.

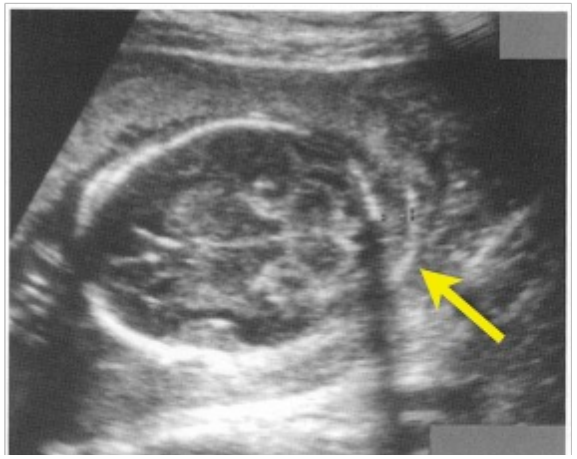
Soft marker for chromosomal anomaly



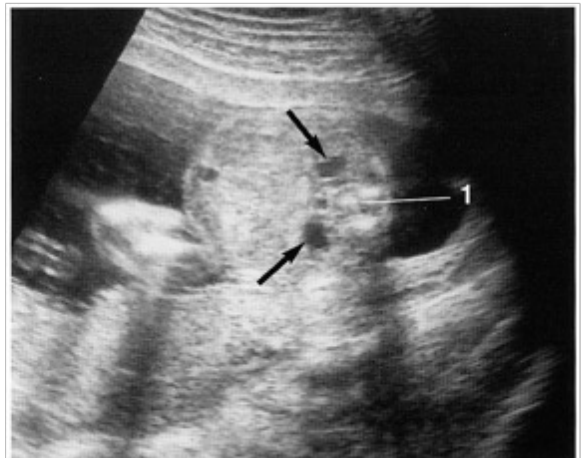
High Intensity Intestinal Echo



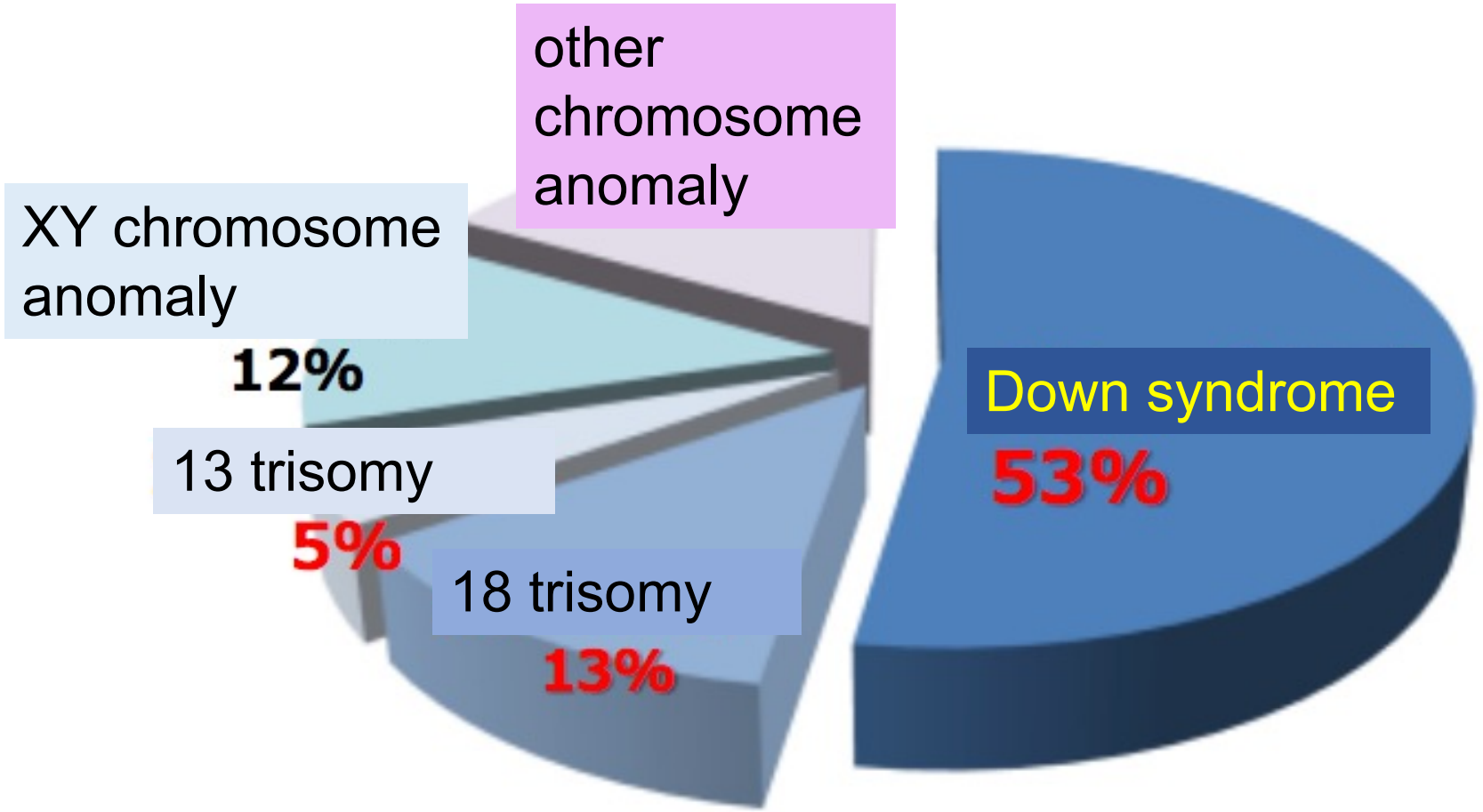
Intracardiac high intensity echo



nuchal translucency (NT)



mild pyelectasis

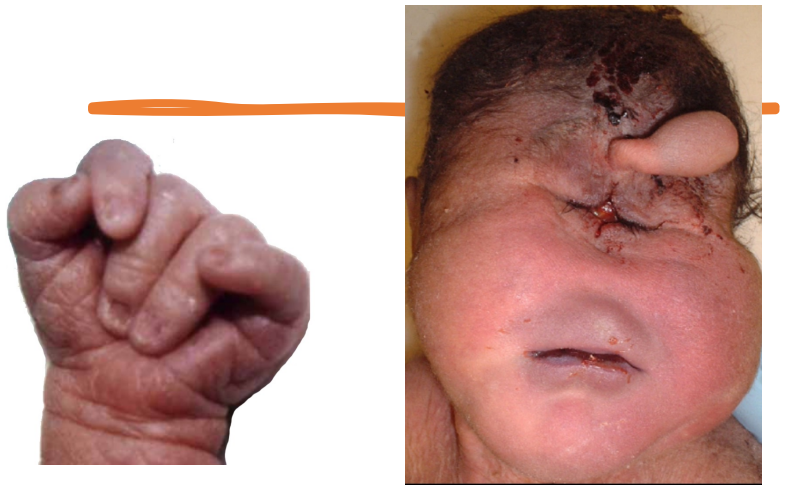


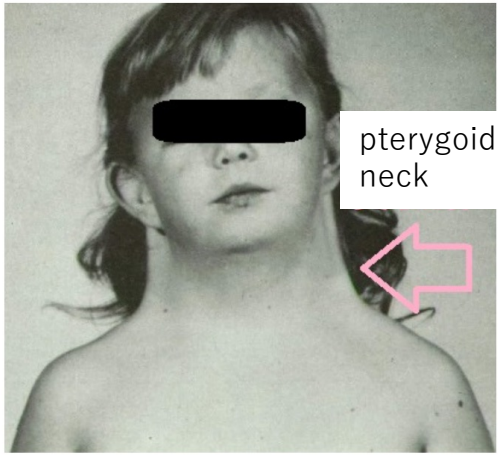
Congenital malformations occur in 3-5% of all pregnancies, 25% of which are caused by chromosomal abnormalities.

Frequency of Chromosomal Anomaly in Early Pregnancy

Data adapted from Wellesley, D, et al., Rare chromosome abnormalities, prevalence and prenatal diagnosis rates from population-based congenital anomaly registers in Europe(2000-2006). *Eur J of Hum Gen*, 11 January 2012.

Major fetal malformations and chromosomal abnormalities





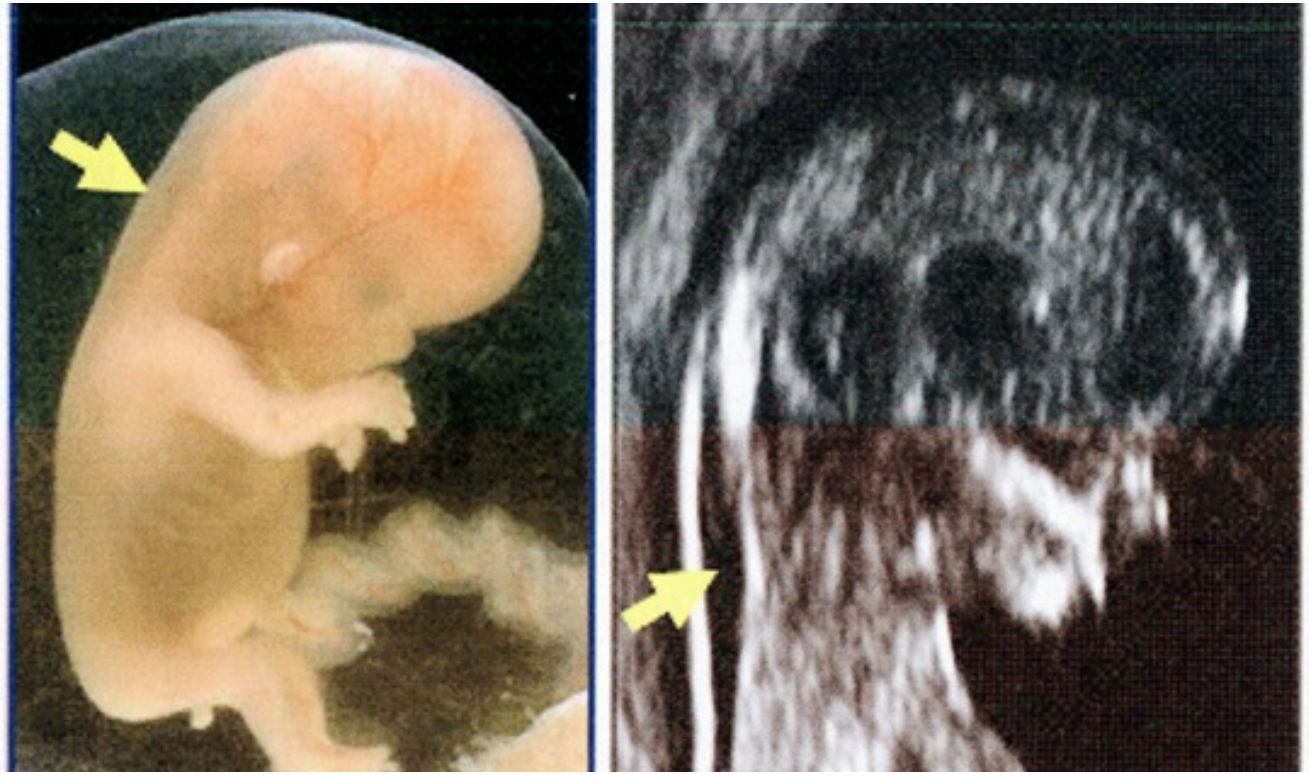
- **21 trisomy**：Low nasal bone formation or nasal bone defect, increased nuchal translucency (NT), cardiac anomaly, duodenal atresia
- **18 trisomy**：cardiac anomaly, esophageal atresia, fetal growth restriction, overlapping fingers
- **13 trisomy**：holoprosencephaly, microcephaly, facial deformity, cardiac anomaly
- **Turner syndrome (45XO)**：fetal hydrops, pleural effusion, ascites, cardiac anomaly, ductal dysplasia

- If a major malformation is found, recommend chromosome test such as amniocentesis.
- If compound malformations are observed: the possibility of chromosomal abnormalities increases.

Important soft marker

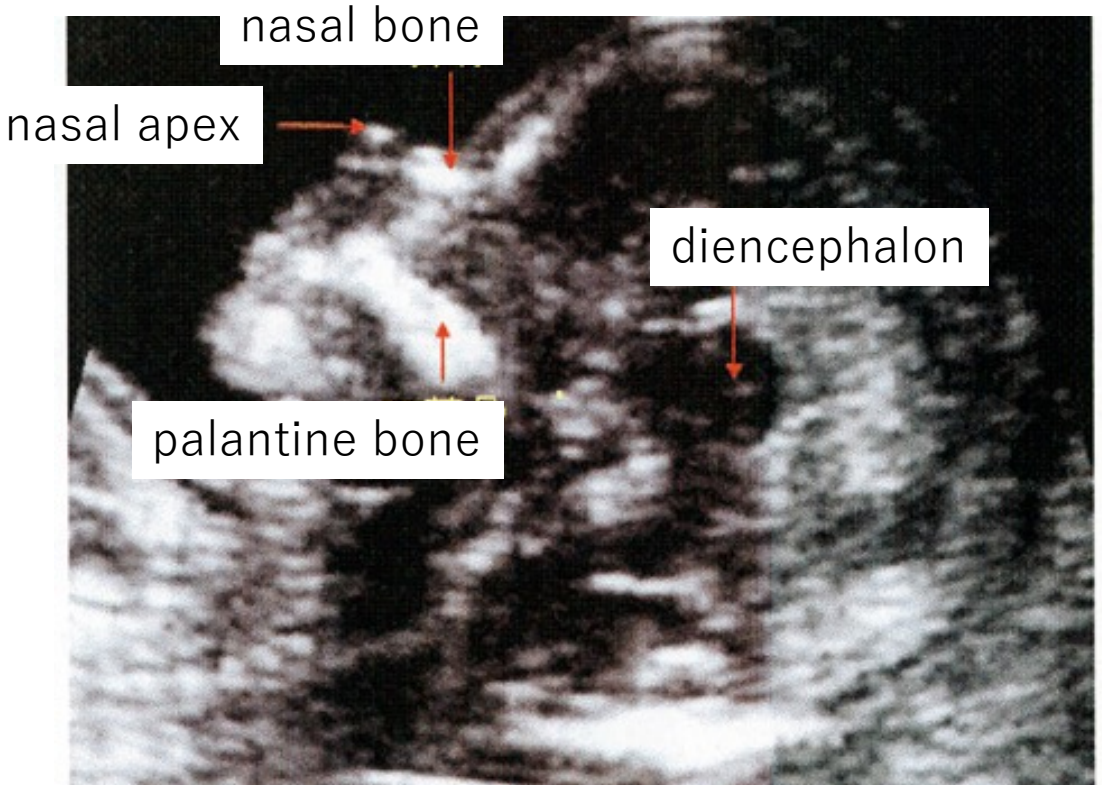
- nuchal translucency (NT), nasal bone defect
  - The likelihood ratio of 21trisomy in high NT (≧ 3.5mm) alone or nasal bone defect is 10-fold.

nuchal translucency (NT：11～13th)



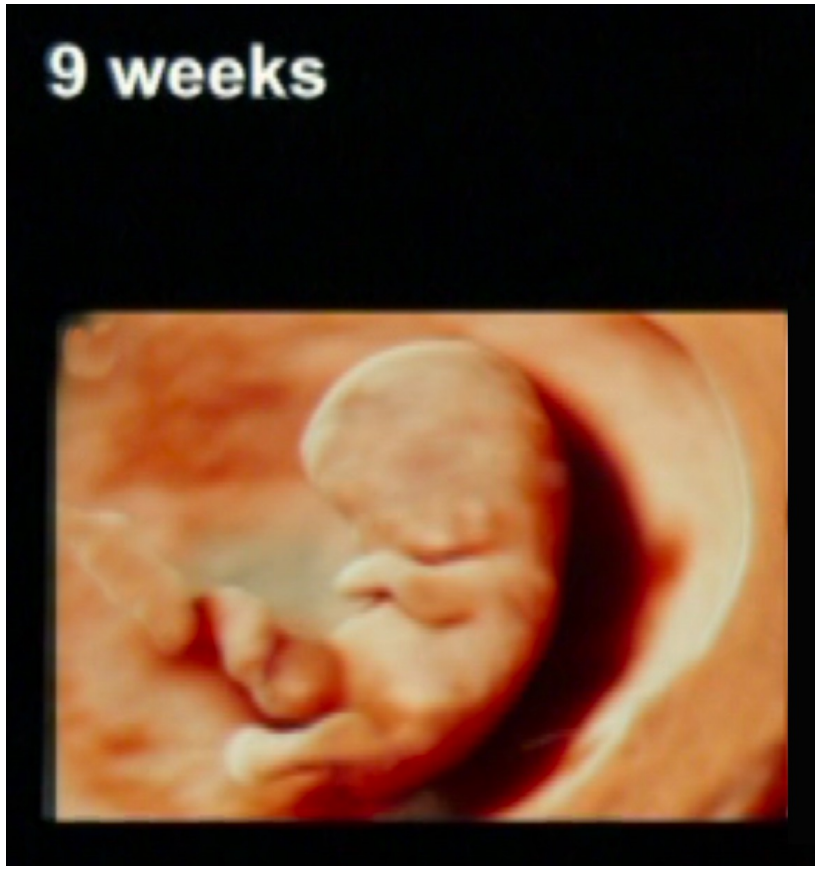
- Edema in the fetal neck, which may resolve spontaneously during the course of pregnancy.
- NT ≥ 3.5 mm (1% of all pregnancies): chromosome anomaly, cardiac malformation, impaired lymphatic return (45XO), fetal anemia (Parvovirus B19), fetal viral infection (Toxoplasma, Cytomegalovirus)

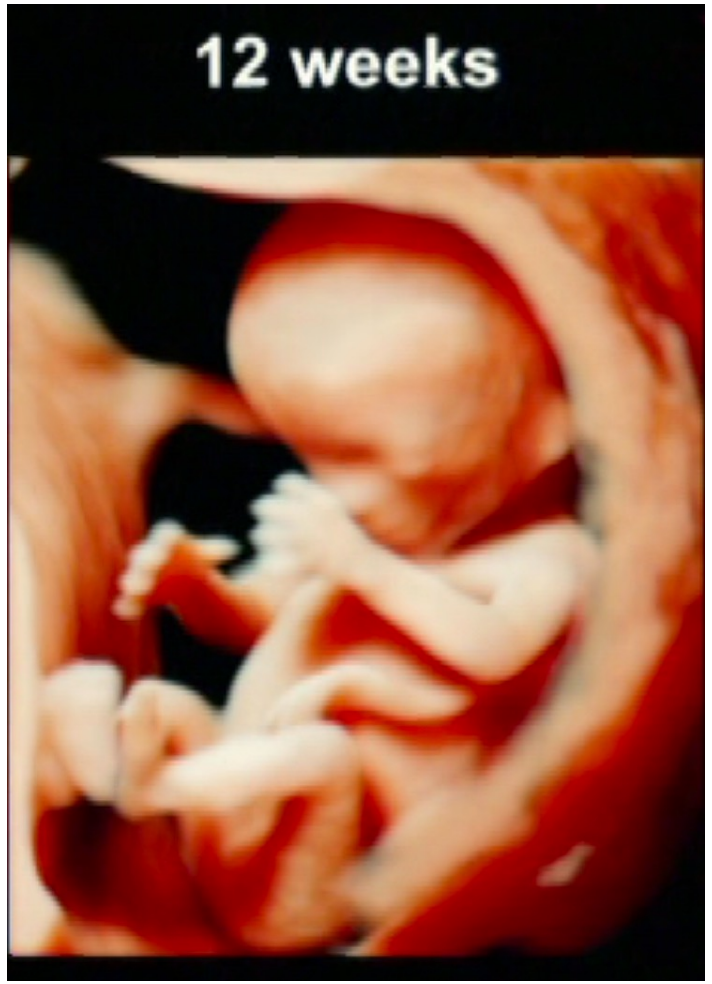
Nasal bone defect　(11～13th)

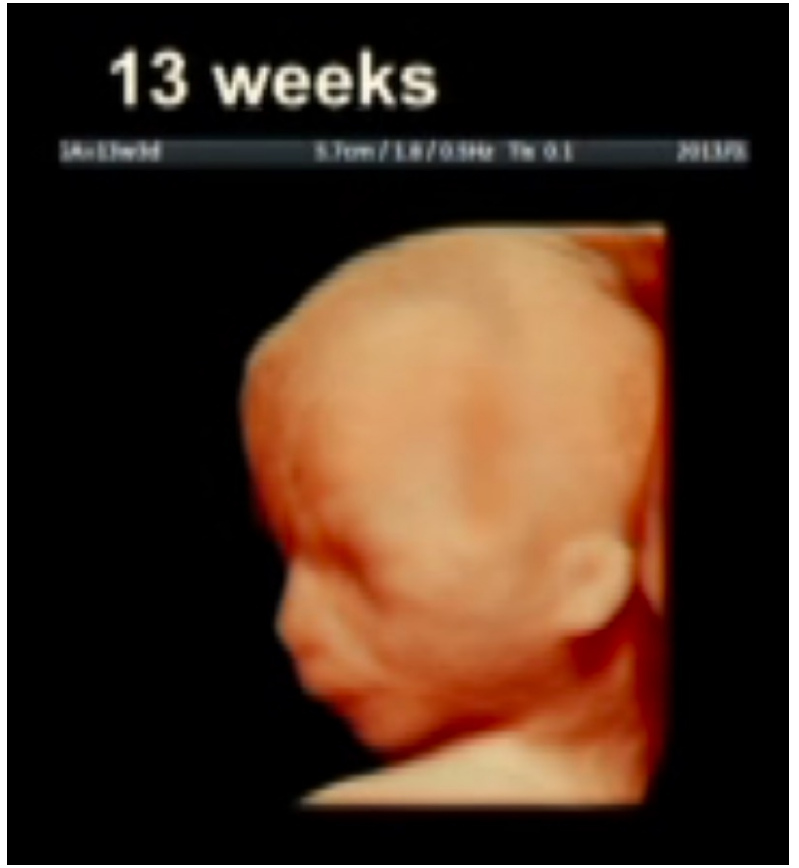


- Sagittal section: high-echo nasal apex and palatine bone are present.
- Hypoechoic diencephalon is seen.

- normal karyotype　: 1-3%
- 21 trisomy　: 60%
- 18 trisomy　: 50%
- 13 trisomy　: 40%


9 weeks


12 weeks


13 weeks

Examine the fetal body

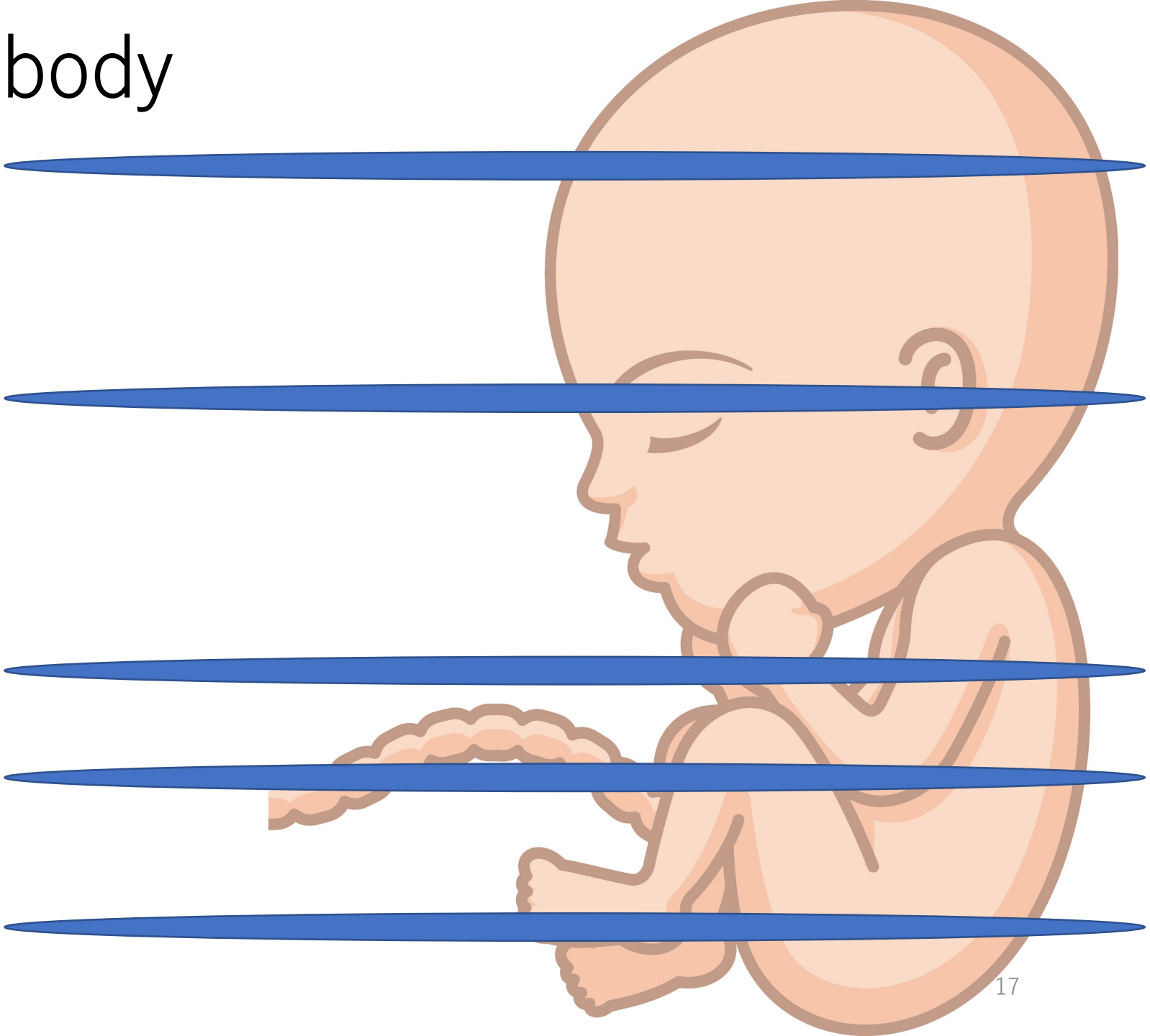
head

face

chest

abdomen

bone

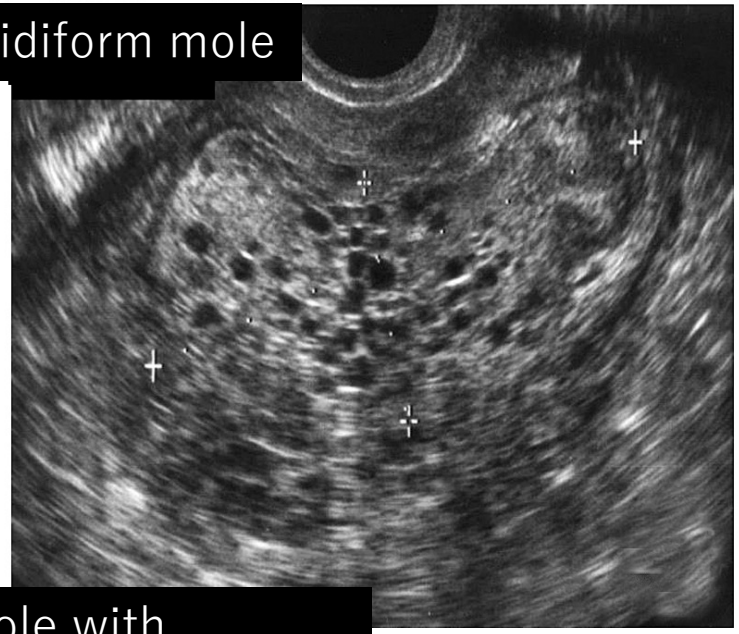


～15th week gestation

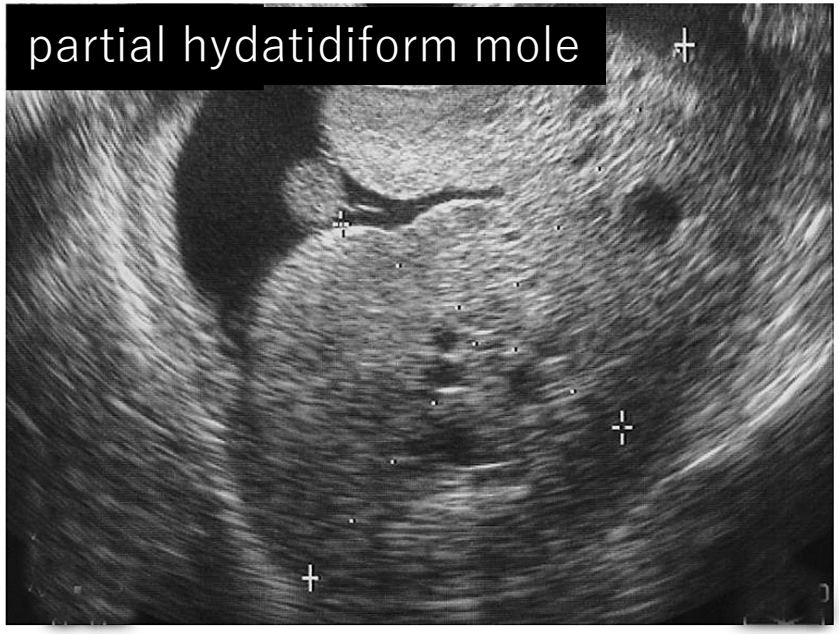
ultrasonographic findings	Diseases to speculate on
Abnormality of head (irregular skull)	Anencephaly, acrania (→p22)
single cerebral ventricle	Holoprosencephaly (13+) (→p22)
Defect of four extremities	
Rt deviated heart	Congenital diaphragmatic hernia (→p46)
Organs outside the abdominal cavity	Umbilical hernia, abdominal wall rupture (→p58)
Multicystic mass in utero	hydatidiform mole (→p19)
intramuscular myometrial mass	Myoma uteri
extrauterine mass	Ectopic pregnancy
Severe nuchal translucency (NT)	Down syndrome (21+)

hydatidiform mole

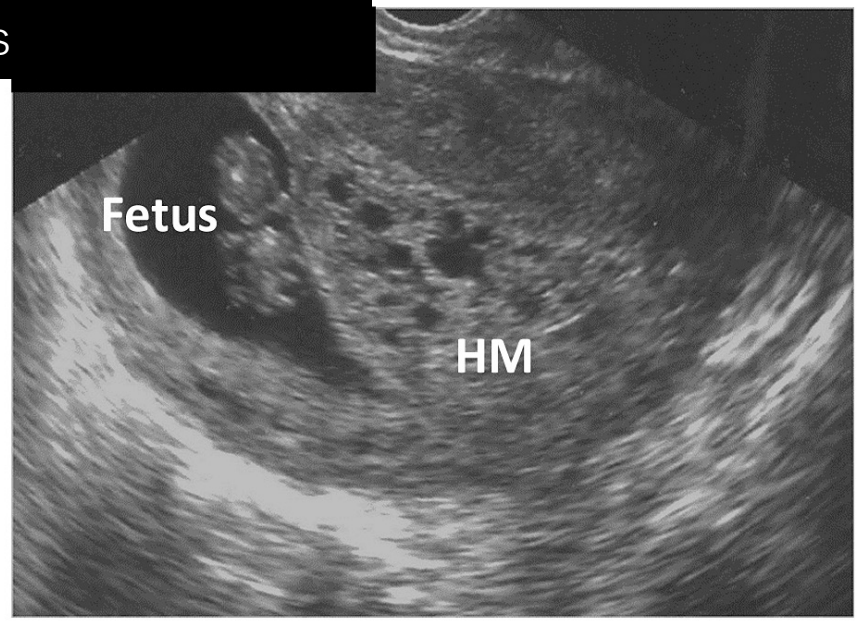
complete hydatidiform mole



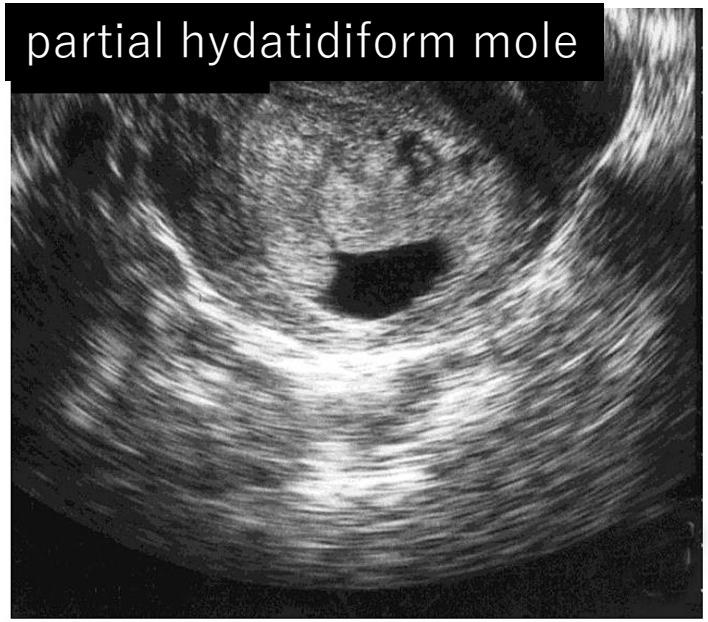
partial hydatidiform mole



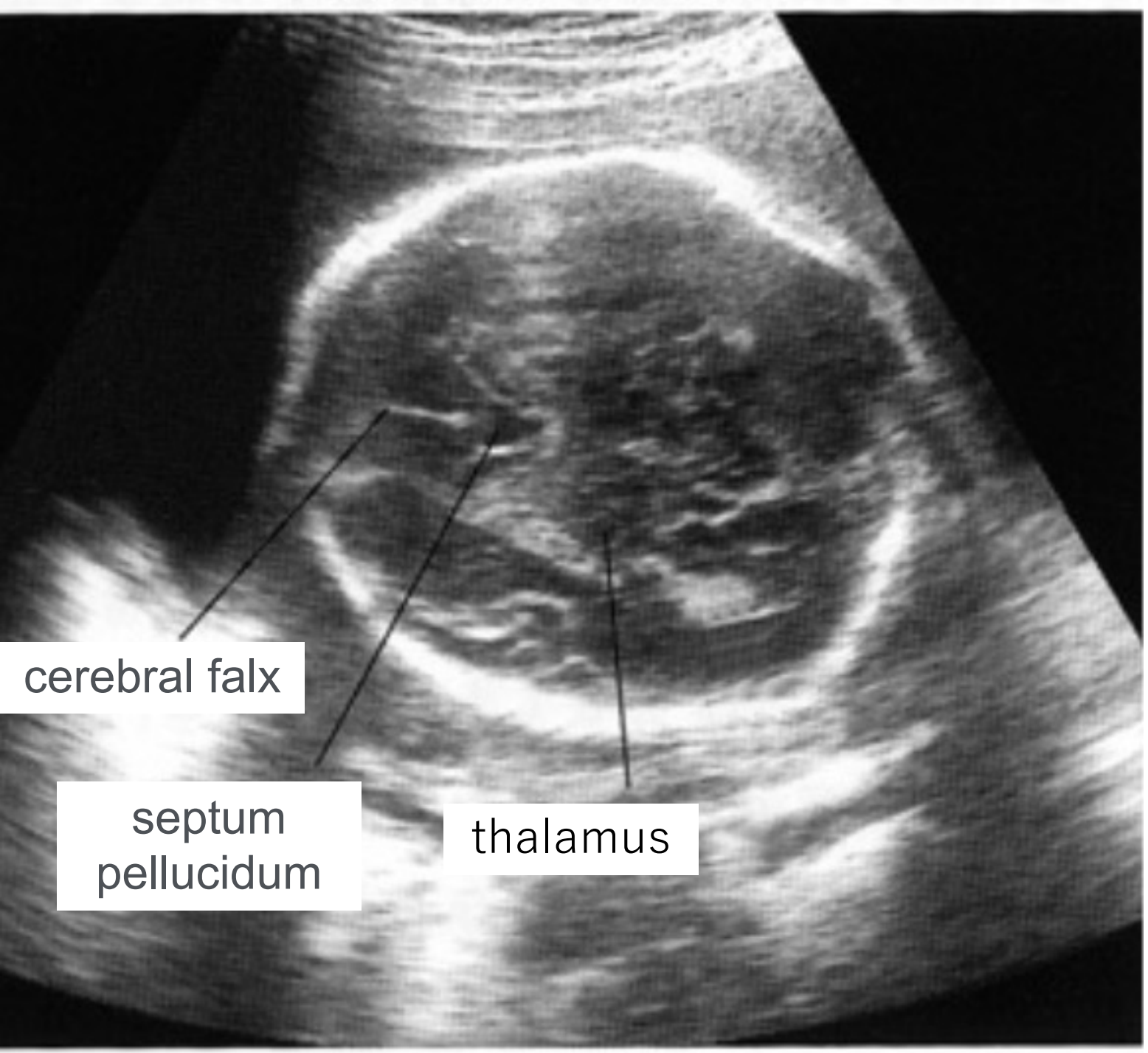
hydatidiform mole with coexistent fetus

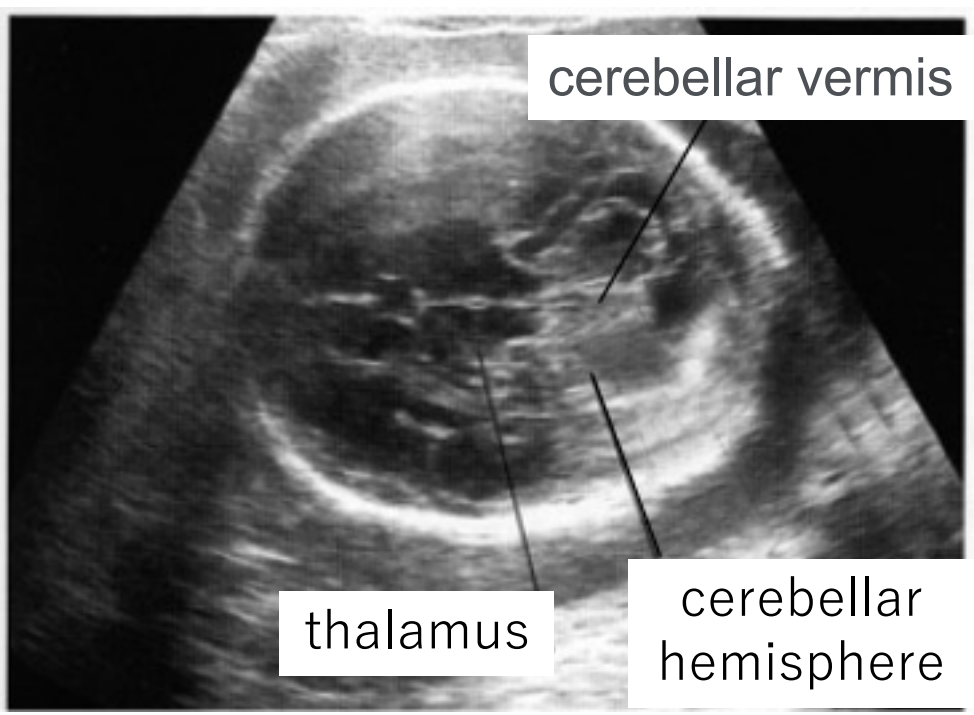


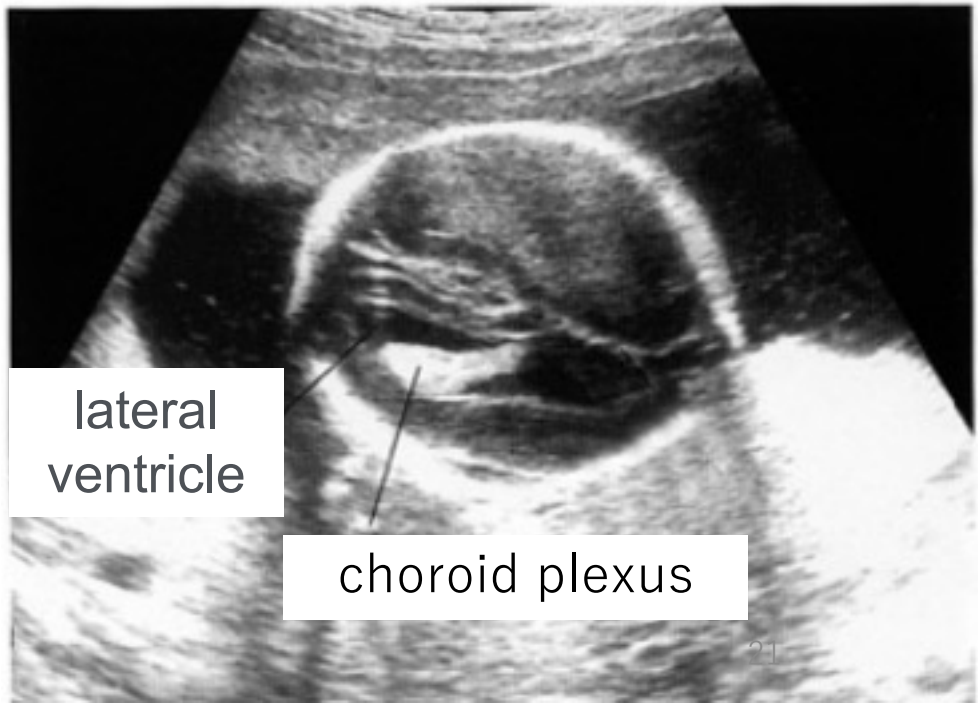
partial hydatidiform mole



Brain (head)

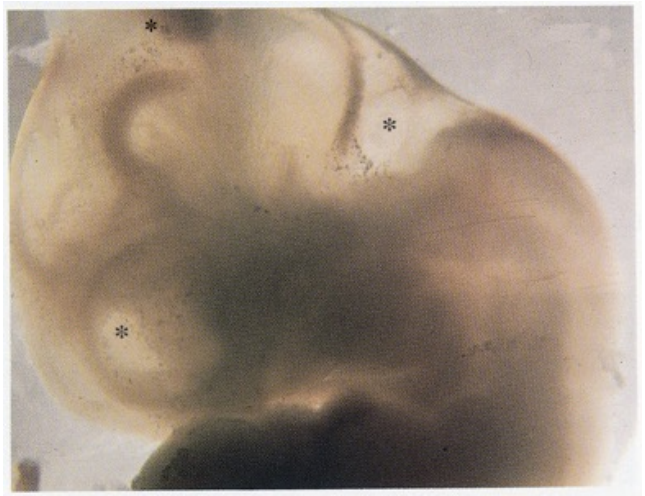

cerebral falx
septum pellucidum
thalamus

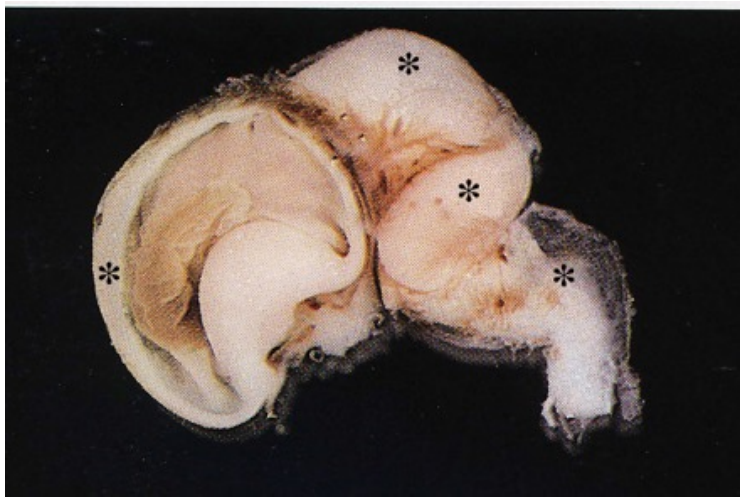

cerebellar vermis
thalamus
cerebellar hemisphere

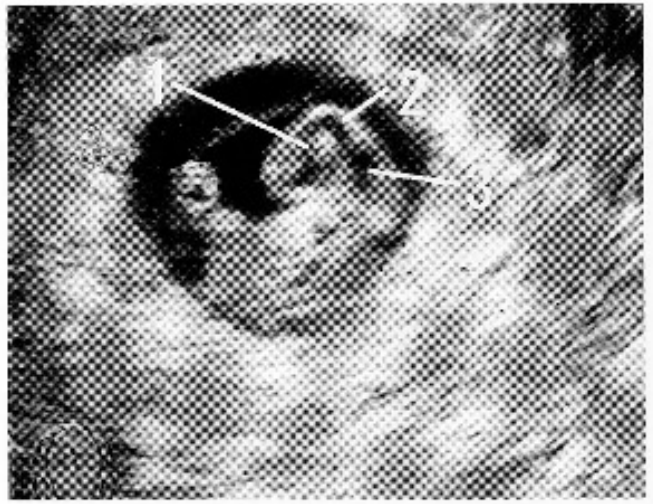

lateral ventricle
choroid plexus

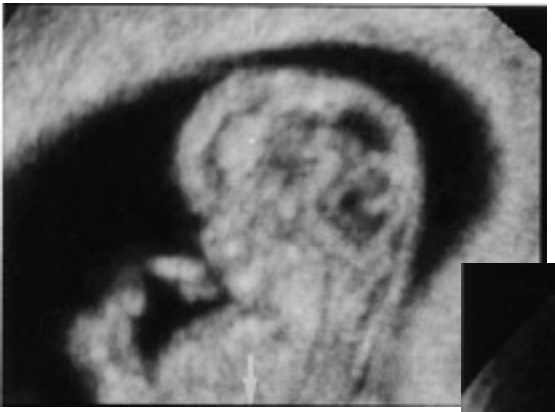
Embryology early in pregnancy (central nervous system)

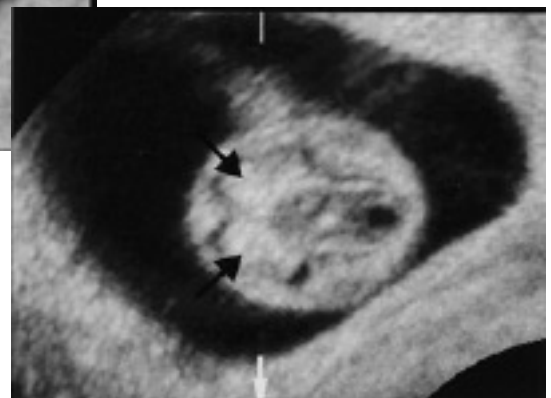
8th weeks of gestation







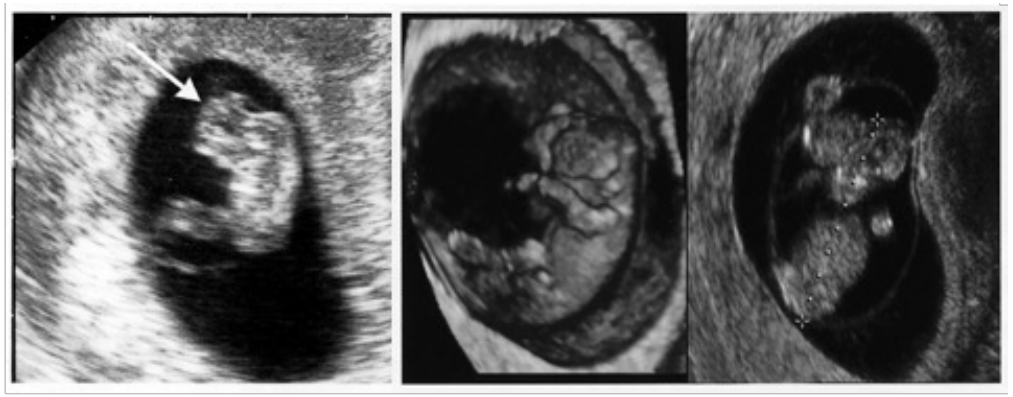




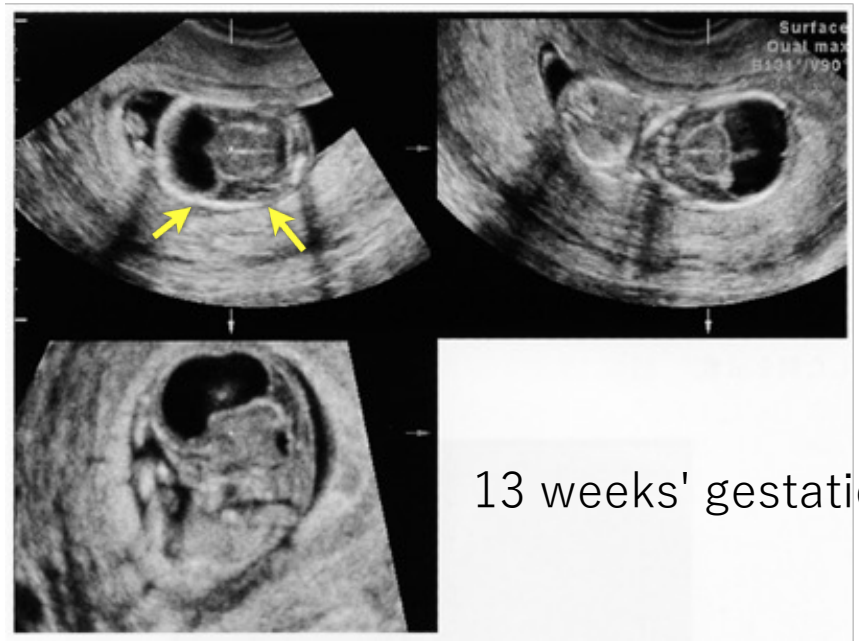
1: forebrain (lateral ventricle・third ventricle)
2: mesencephalon (mesencephalic aqueduct)
3: hindbrain (fourth ventricle)

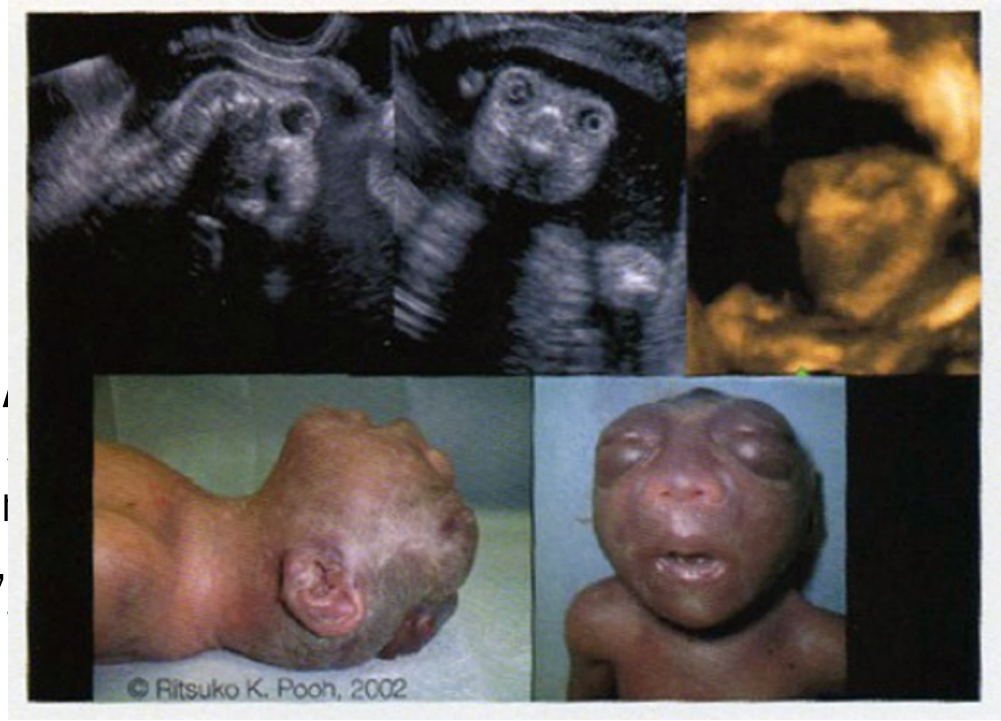
At 11th weeks gestation, the brain structure is almost completed, and the cerebellum can be observed.

Head abnormalities found in



7th week' gestation　10th week' gestation





- **Acrania~**
  the skull i
  into the a
  gestation,
  lost, resul

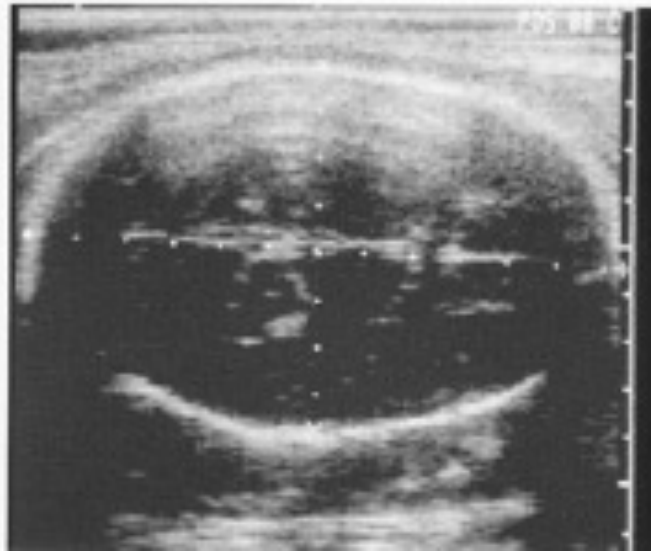
- **Holoprosencephaly**: Insufficient formation of the forebrain. Differentiation into the left and right lateral ventricles is impaired, forming a single ventricle. The thalamus is fused due to impaired differentiation of the mesencephalon. If facial malformations are present, 13 trisomy is mostly suspected.

18th～ week gestation

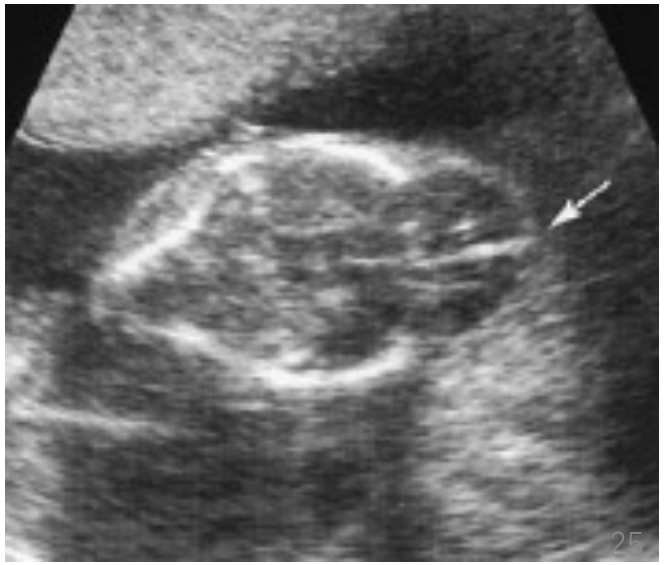
ultrasonographic findings	Diseases to speculate on
Systemic edema	Fetal hydrops (45XO) (→p74)
Small head (small BPD)	Fetal growth restriction (FGR) (→p25 & p76)
Abnormal heart structure	Congenital heart disease (→p49)
Ventriculomegaly	Agenesis of corpus callosum, Chiari type II Malformation, Dandy-Walker syndrome (→p27)
Lumbar back mass	Meningocele, sacrococcygeal teratoma (→p34)
Gastric bubble not visible on left side	esophageal atresia (→p55)
Abnormal cyst in the abdominal cavity	Ovarian cyst (→p61), duodenal atresia (→p57), hydronephrosis (→p62)
Oligohydramnion	Potter syndrome, MCDK
Short limb	Congenital bone disease (→p65)

Embryology early in pregnancy (central nervous system)

- **Dolichocephaly**：This is found in the pelvic position. When measuring estimated fetal weight, it should be corrected by head circumference (HC).

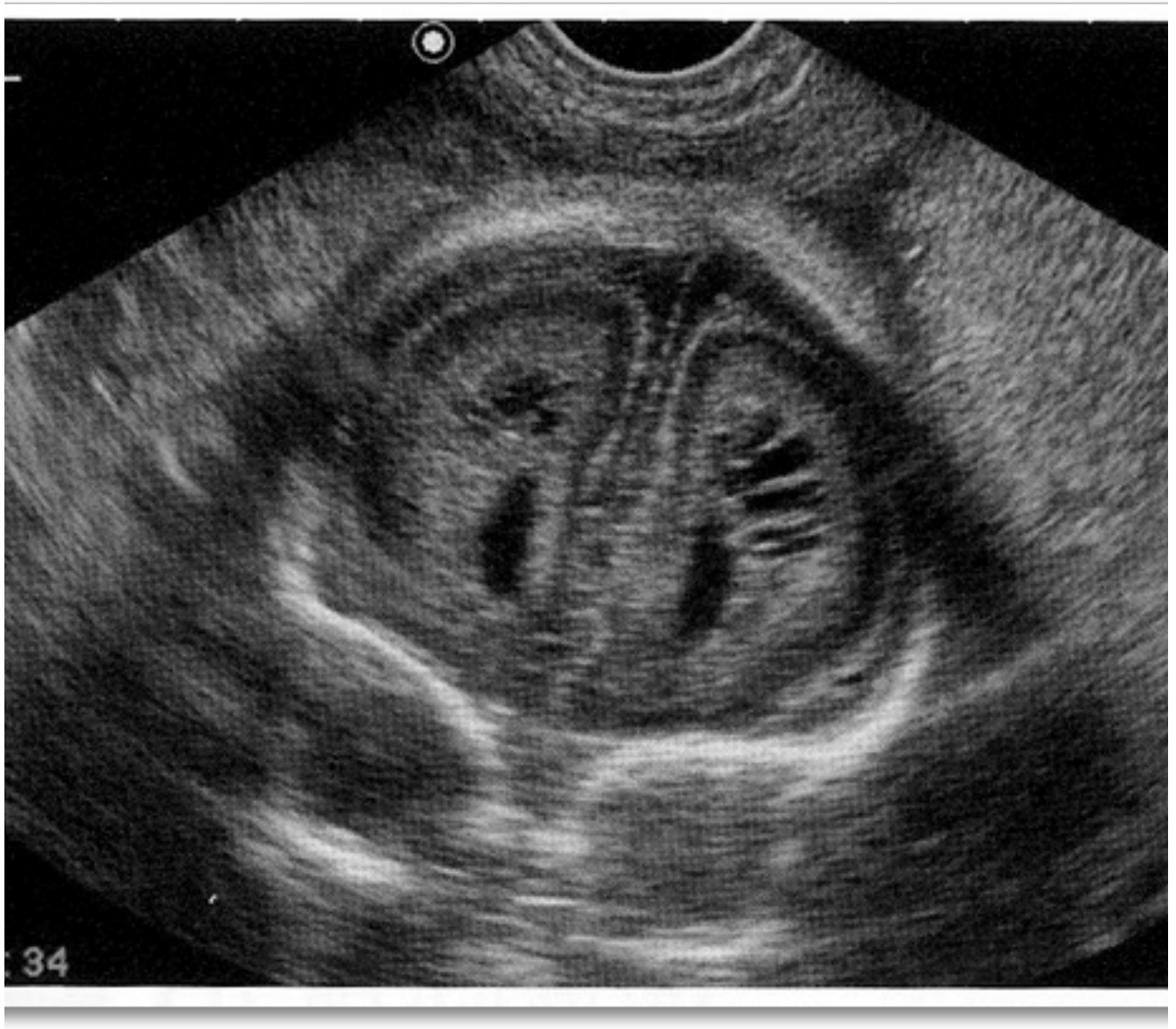


- **Microcephaly** : often based on impaired brain development due to fetal alcohol syndrome or other factors.

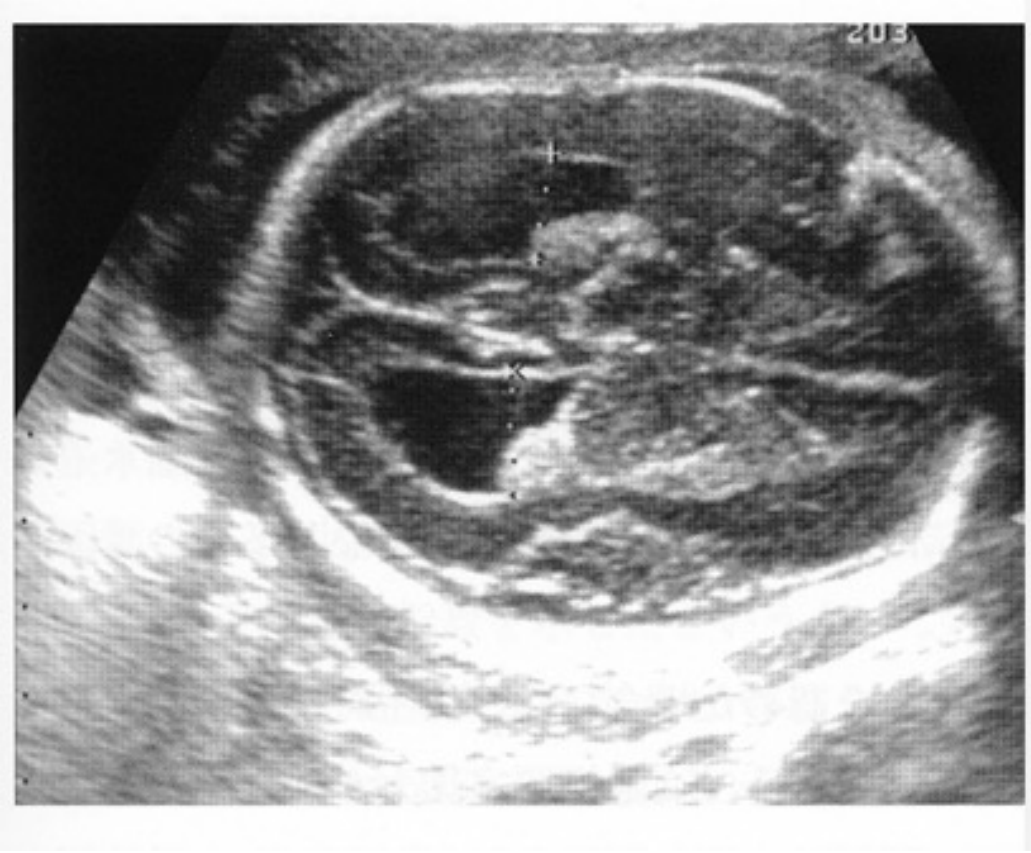


Fetal hypoxia →PVL (periventricular leukomalacia)

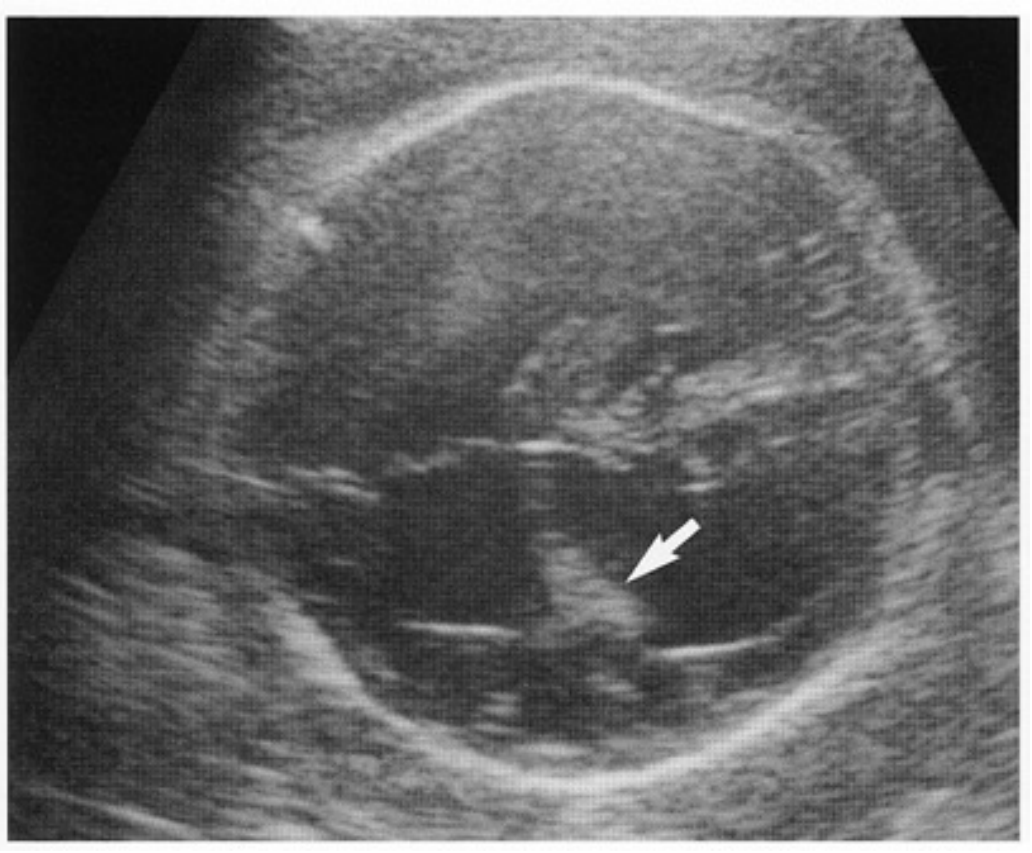
- Because the periventricular vessels are undeveloped in preterm low-birth-weight fetus, the blood supply to the periventricular area is inadequate during hypoxia and circulatory failure.
- 10-20% of cerebral palsy can result from hypoxemia from just before to just after delivery.



Ventriculomegaly

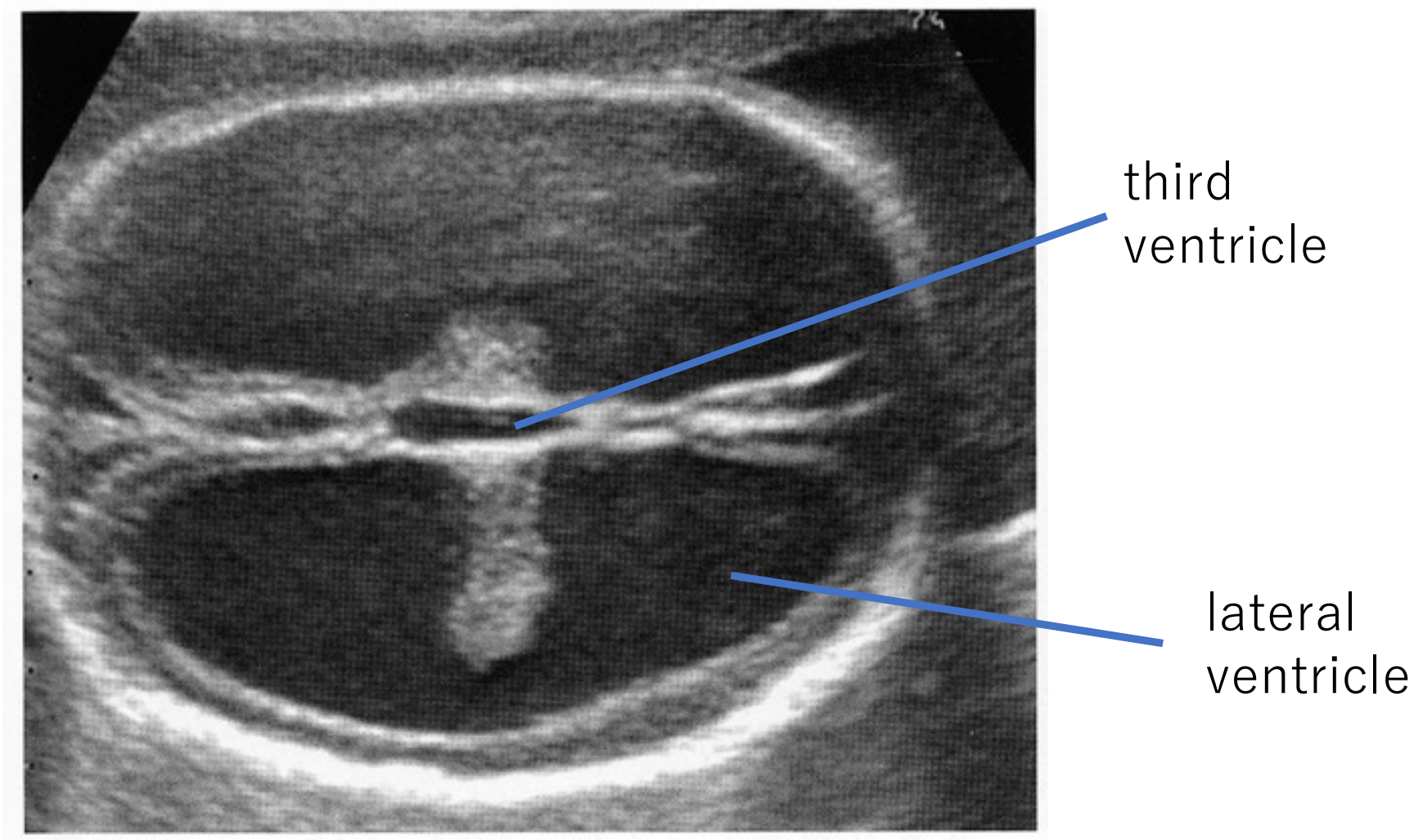


mild ventriculomegaly



dangling of choroid plexus

- diameter of lateral ventricle（Posterior Horn） ≧ 10mm
- **Cause**：chromosomal anomaly, cerebral hemorrhage, fetal viral infection (Toxoplasma, Cytomegalovirus)



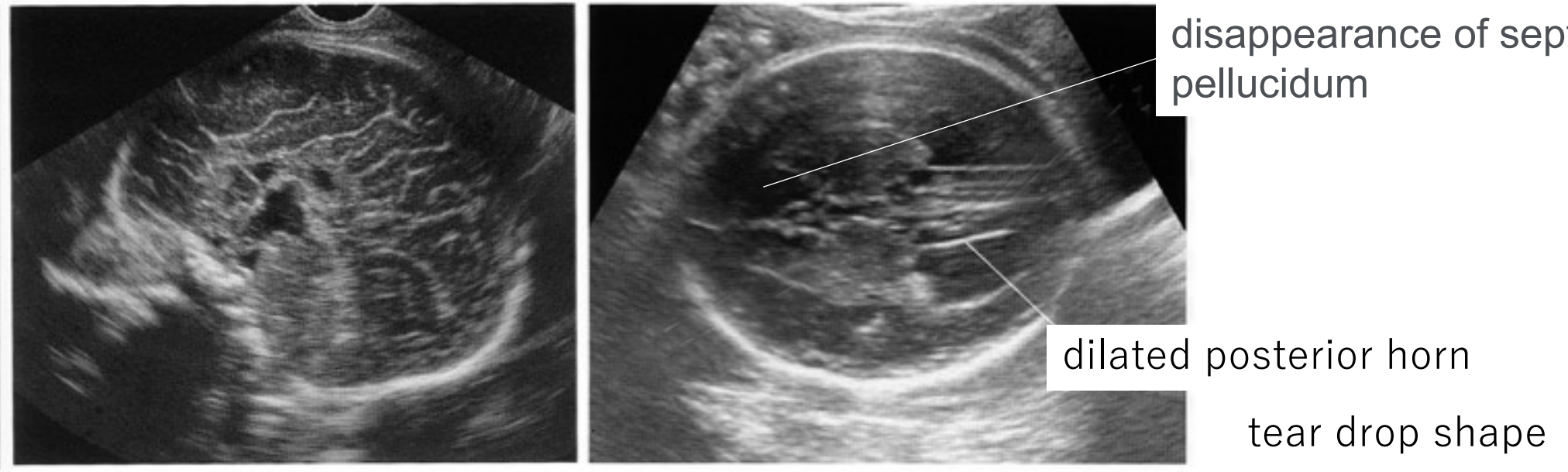
aqueductal stenosis

Causes of Fetal-specific ventriculomegaly

- Agenesis of corpus callosum
- Chiari type II Malformation
- Dandy-Walker syndrome

Agenesis of corpus callosum

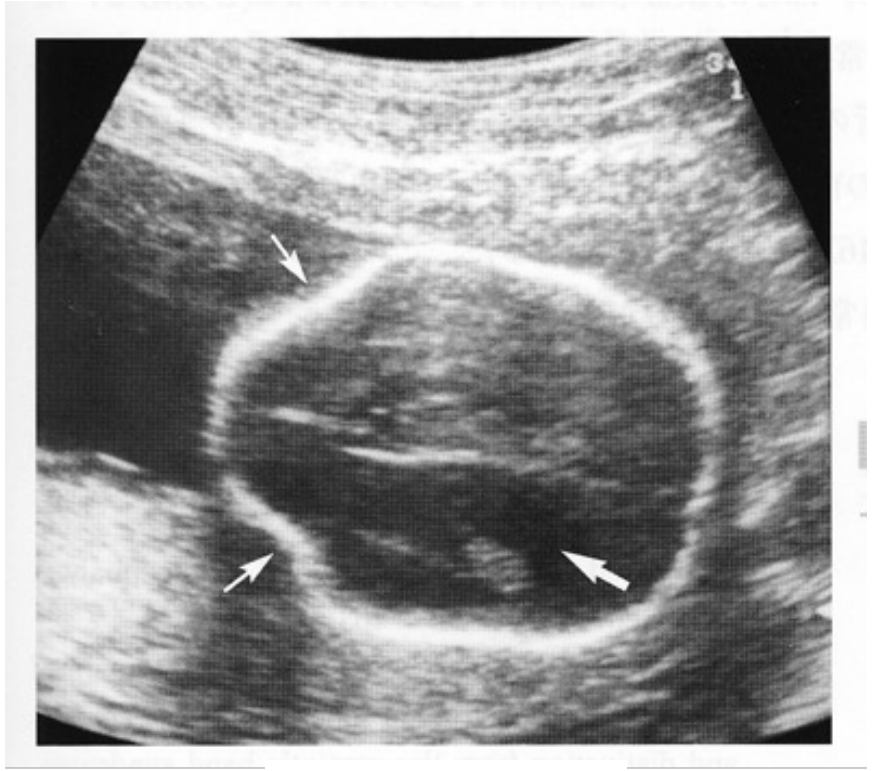
- Possible chromosomal abnormalities (8+, 13+, 18+). May be associated with various malformations. When it is an isolated case, neurological and other symptoms are rare.



37th weeks of gestation　31th weeks of gestation

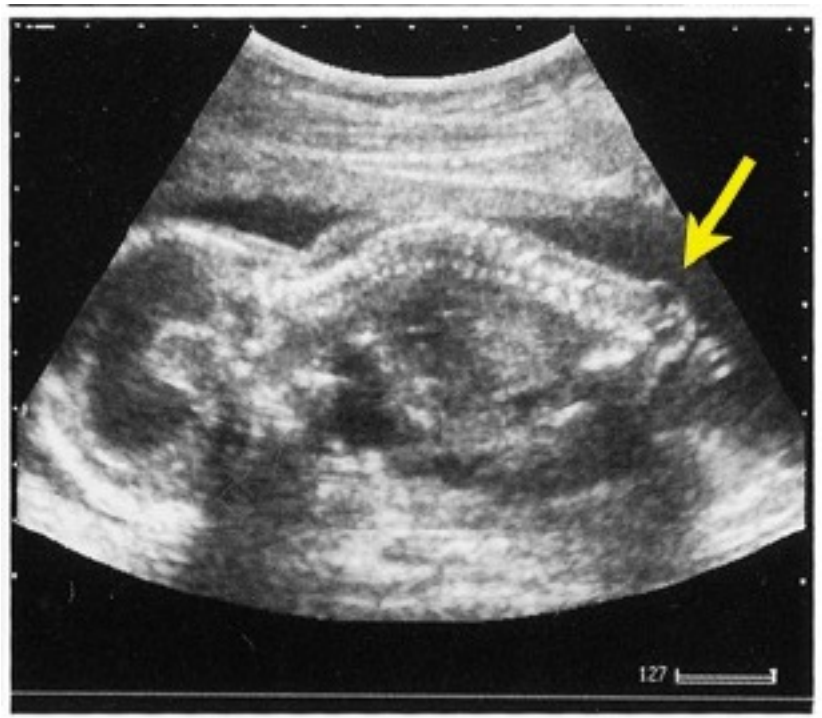
Chiari type II Malformation

- Cerebellum and other parts of the brain drop from the cranium into the spinal cavity.
- Chiari type 2 (Chiari malformation occurring in utero): In addition to the cerebellar tonsils, the cerebellar vermis and brainstem also drop down. Familial occurrence is seen with myelomeningocele. It is related to chromosomal abnormalities (18+, 13+, 21+)



lemon sign

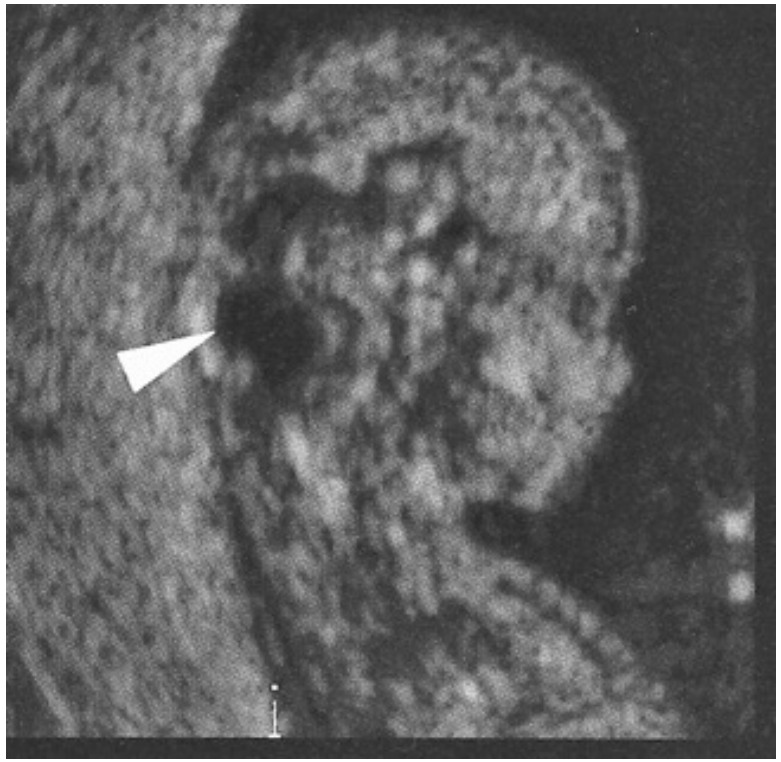
16th weeks of gestation



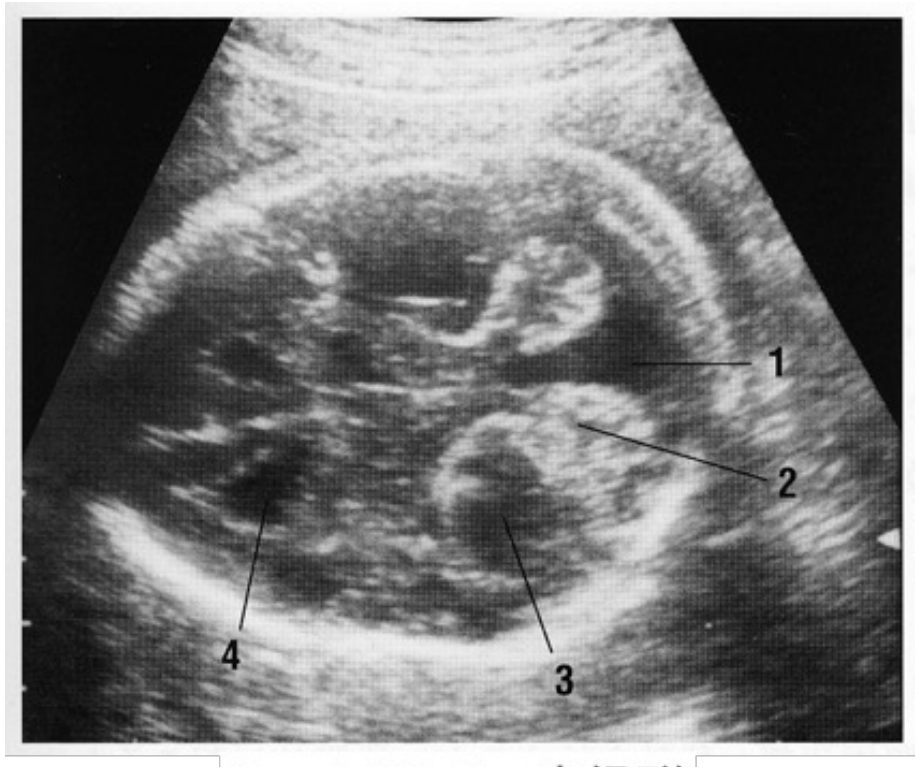
meningocele

Dandy Walker syndrome

- Cerebellar differentiation is completed by 11 weeks' gestation.
- **Causes of Dandy Walker syndrome**: DM, alcohol drinking, warfarin, CMV, rubella, chromosomal abnormalities



11 weeks gestation



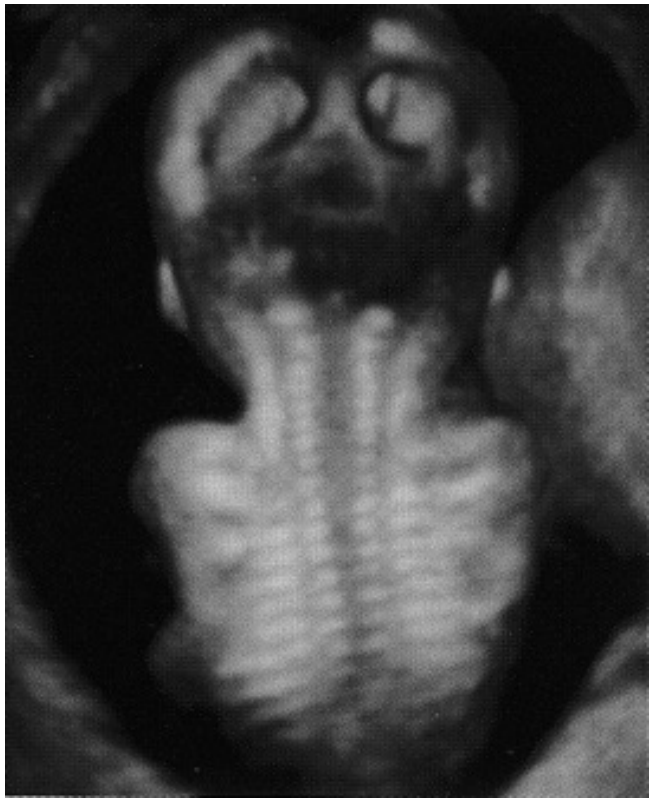
Dilated fourth ventricle: Cerebellar vermis hypoplasia and enlargement of the cisterna magna (18+, 13+)

Dandy Walker syndrome

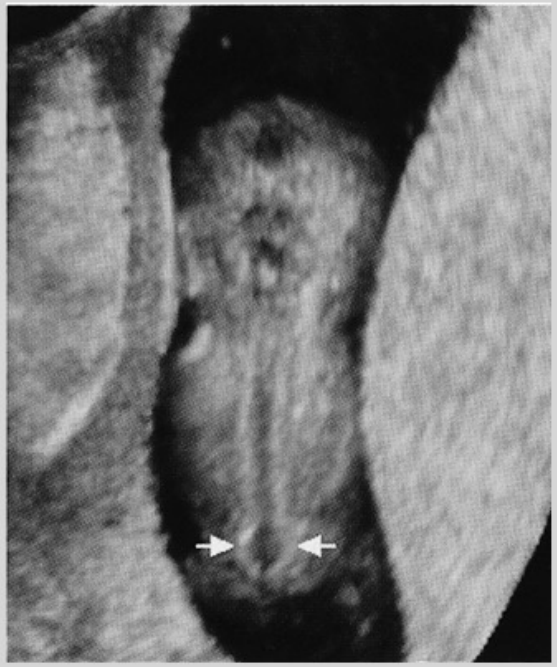
Spine

Embryology early in pregnancy (spine)

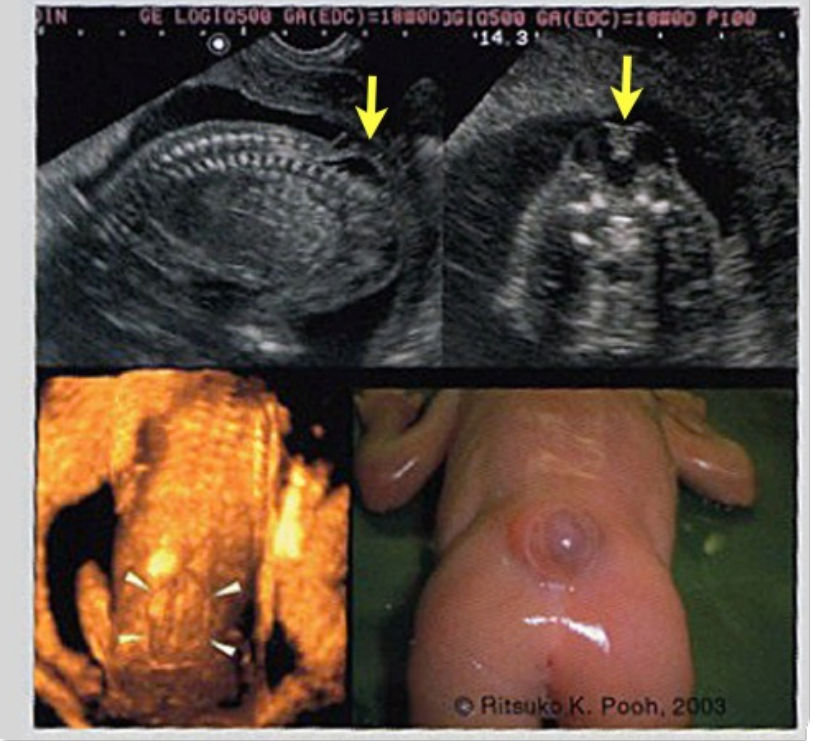
13th (normal spine)



9th spina bifida



18th spina bifida



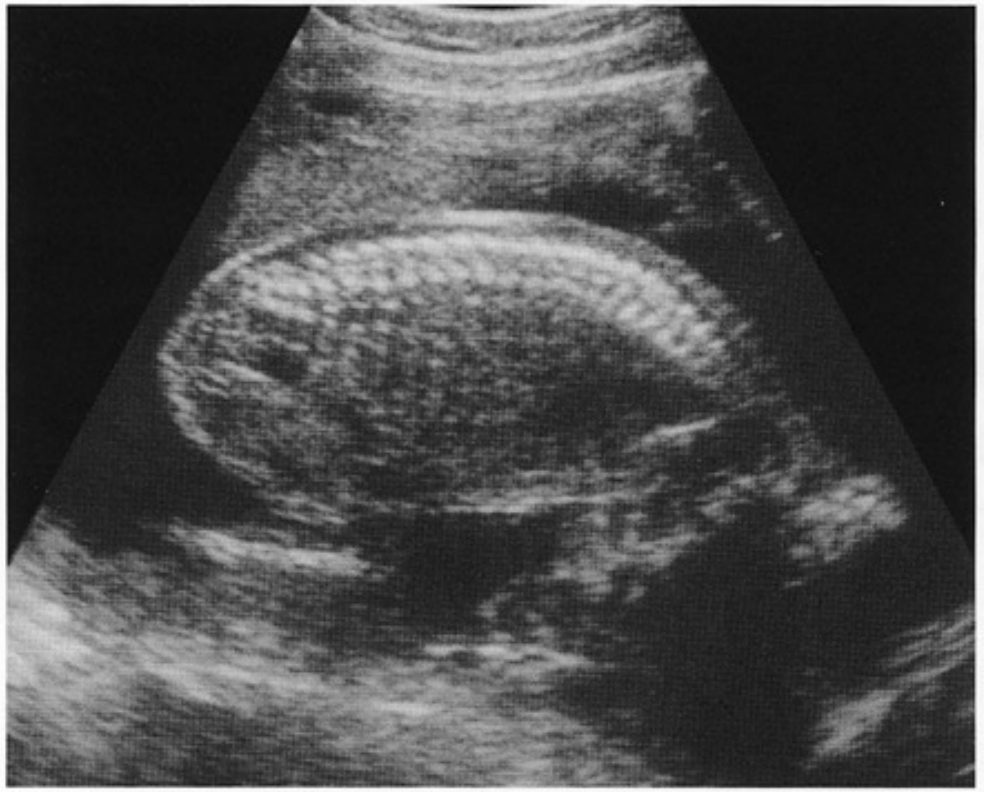
Spina Bifida

· Exposed nerves would be damaged.

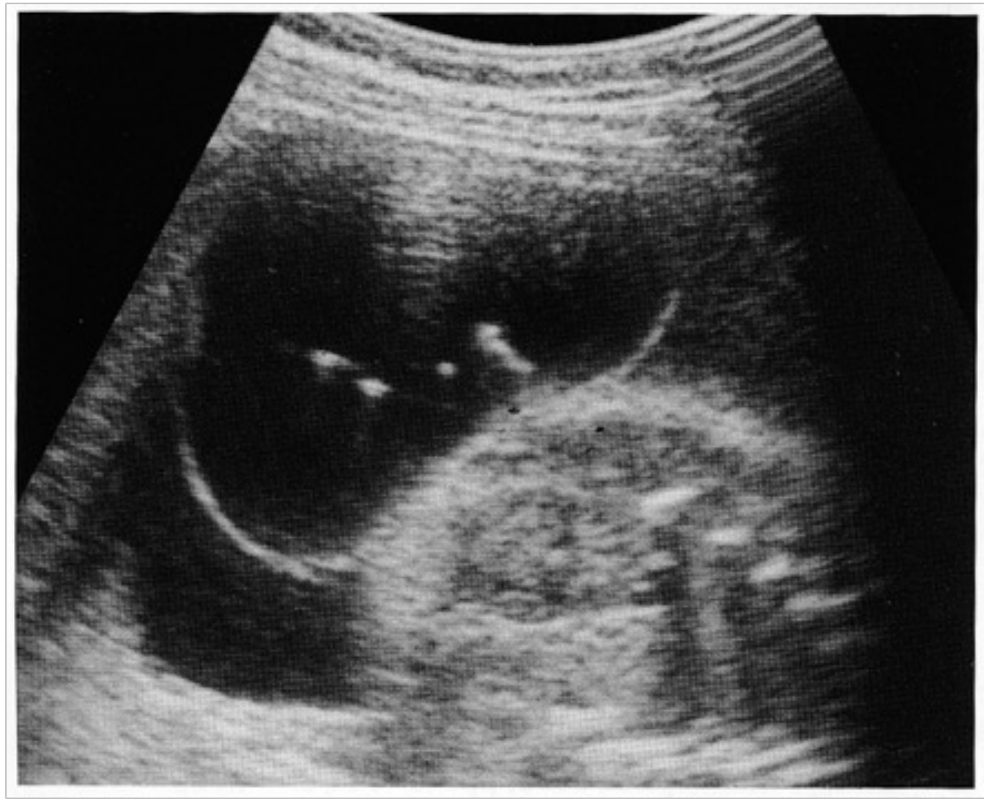
Causes: Chromosomal abnormalities, genetic abnormalities, diabetes, antiepileptic drugs

Adequate intake of folic acid supplements since before pregnancy can reduce the risk of spina bifida by 75%.

lumbosacral tumor



normal spine (21th)

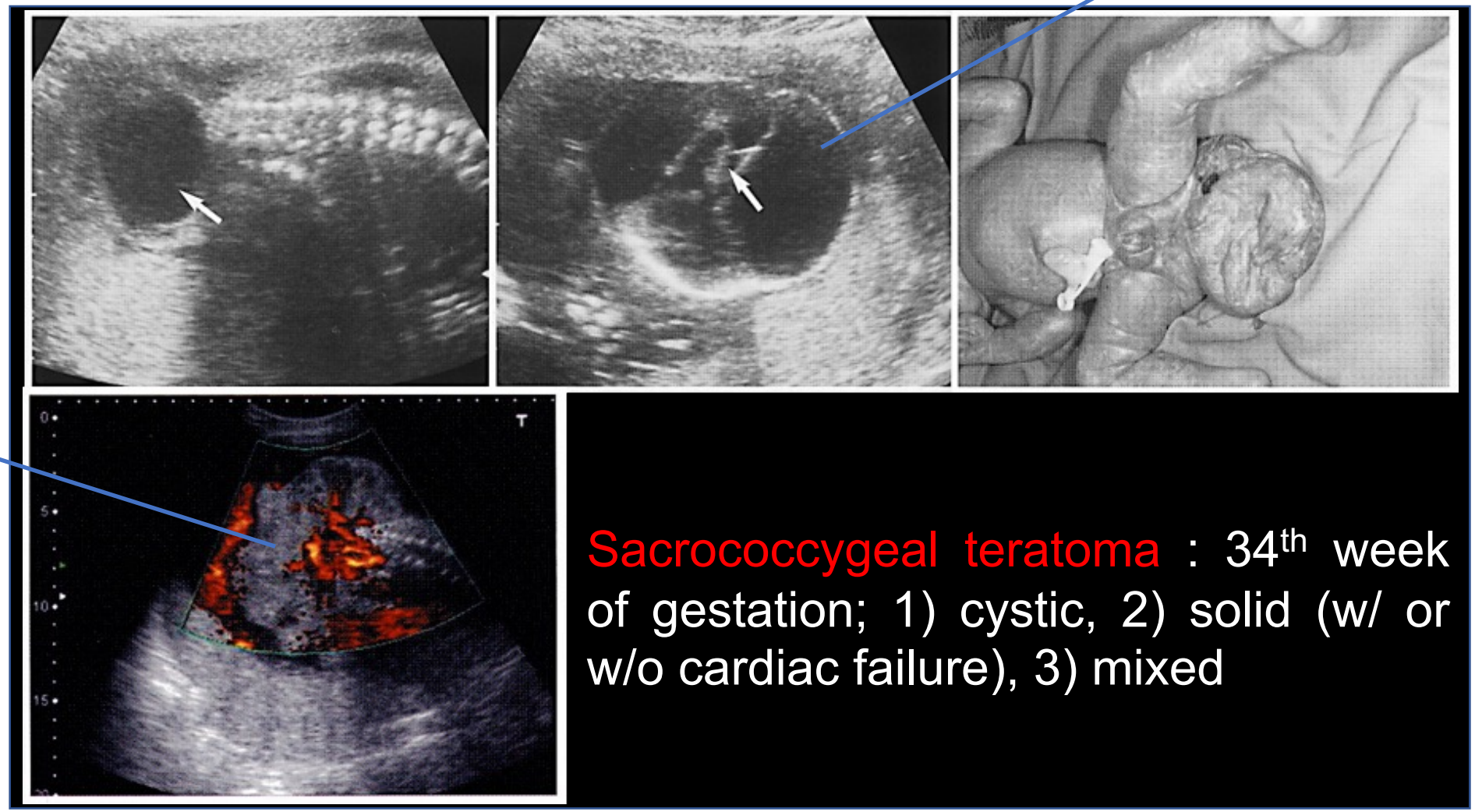


- Meningocele : cystic
- Myelomeningocele: The inside of the tumor appears complex because it contains nerve tissue. (Cauda equina syndrome) (28th)

lumbosacral teratoma

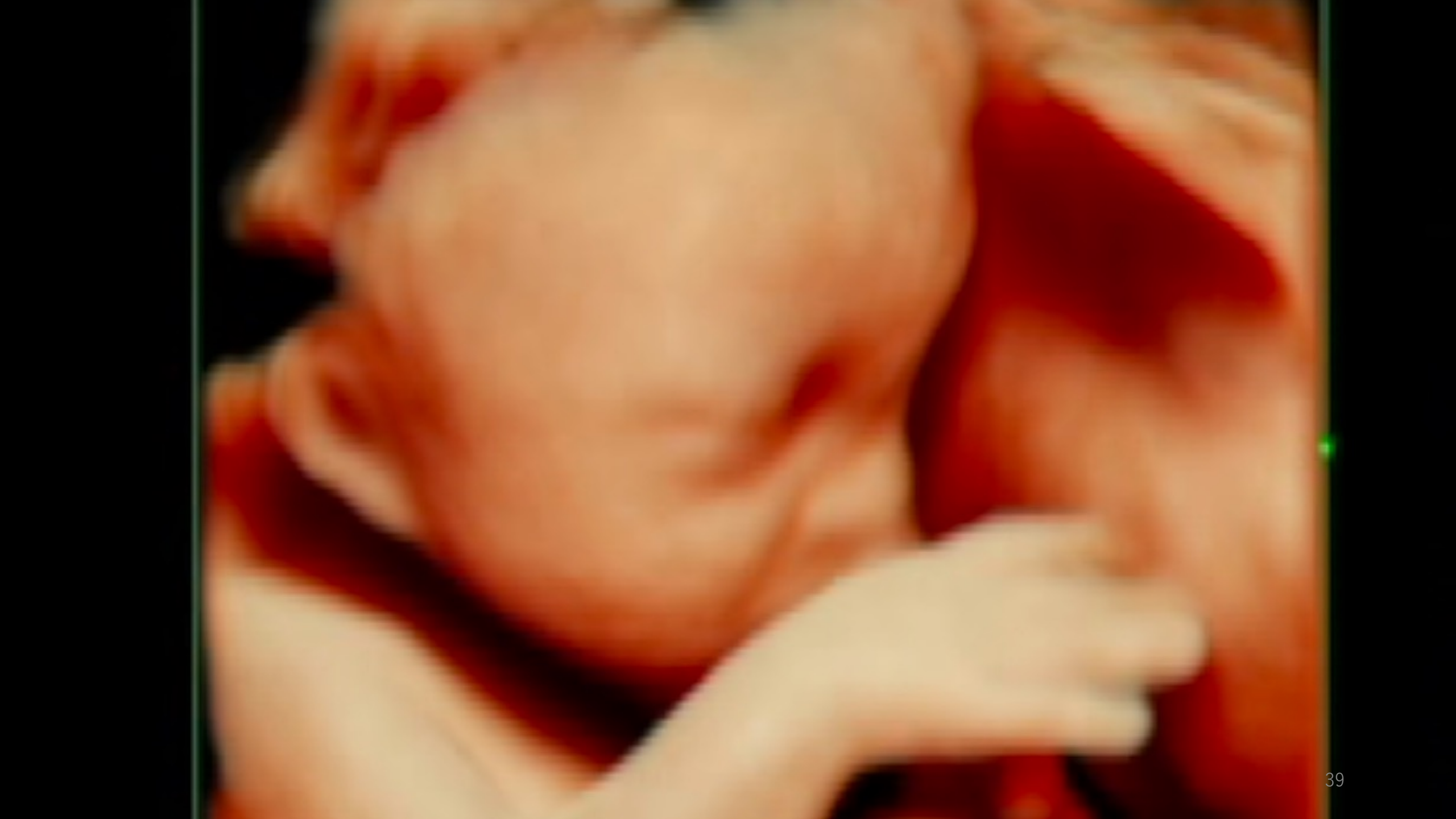
multicystic

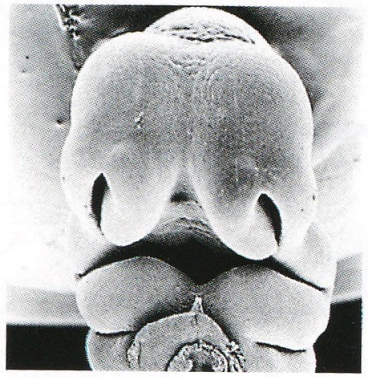
solid,
DIC



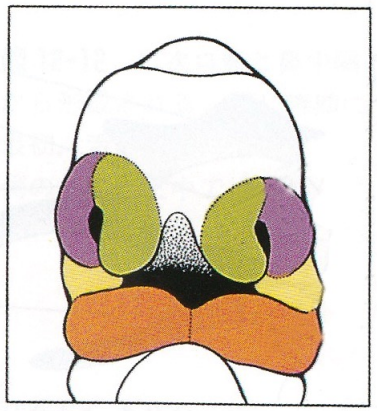
Sacrococcygeal teratoma : 34th week of gestation; 1) cystic, 2) solid (w/ or w/o cardiac failure), 3) mixed

Face

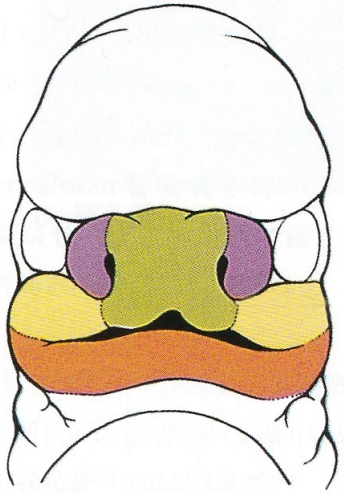




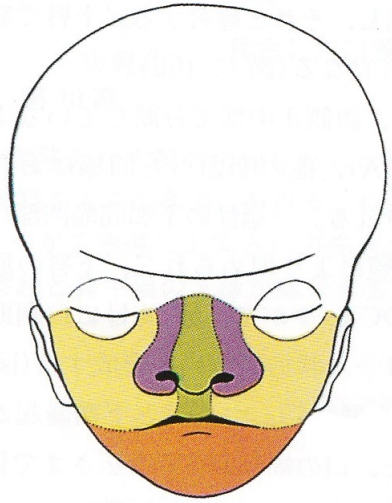
A 6th week of gestation



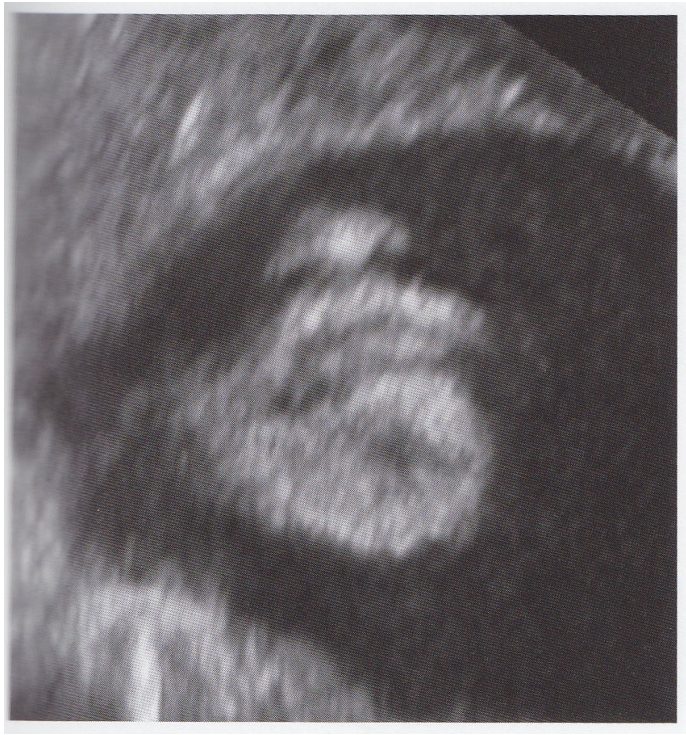
B 6th week of gestation



D 7th week of gestation



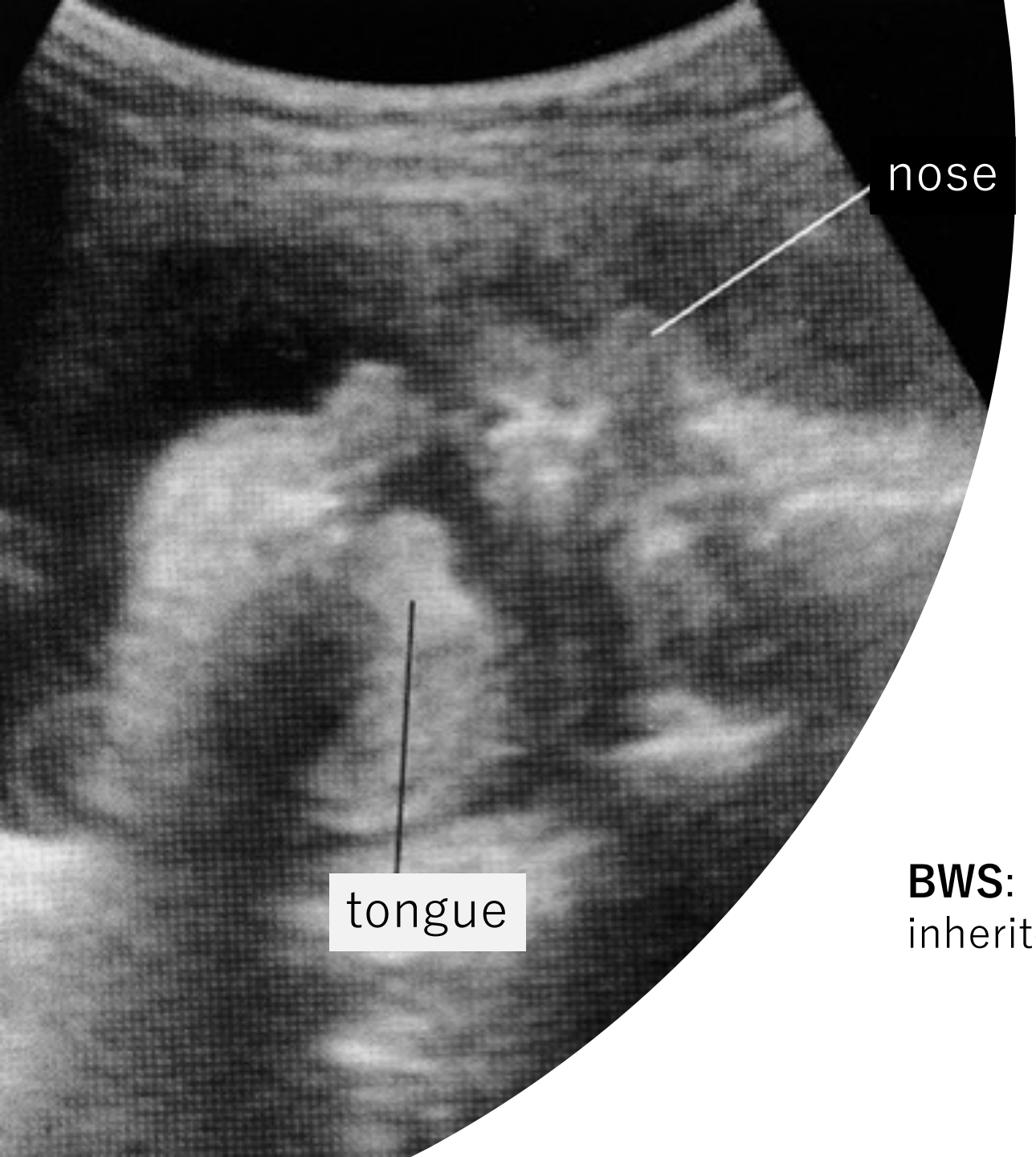
E 10th week of gestation



Some cases of cleft palate is caused by adrenal corticosteroid.

Folic acid is also effective to prevent the cleft palate.

The original facial tissues originate in pairs on the left and right sides, which develop and fuse in the center to form the lips and nose in the central part of the face. Failure of this fusion to occur and leaving a cleft in the center is believed to be the cause of cleft lip and palate.



Tongue

Macroglossia：

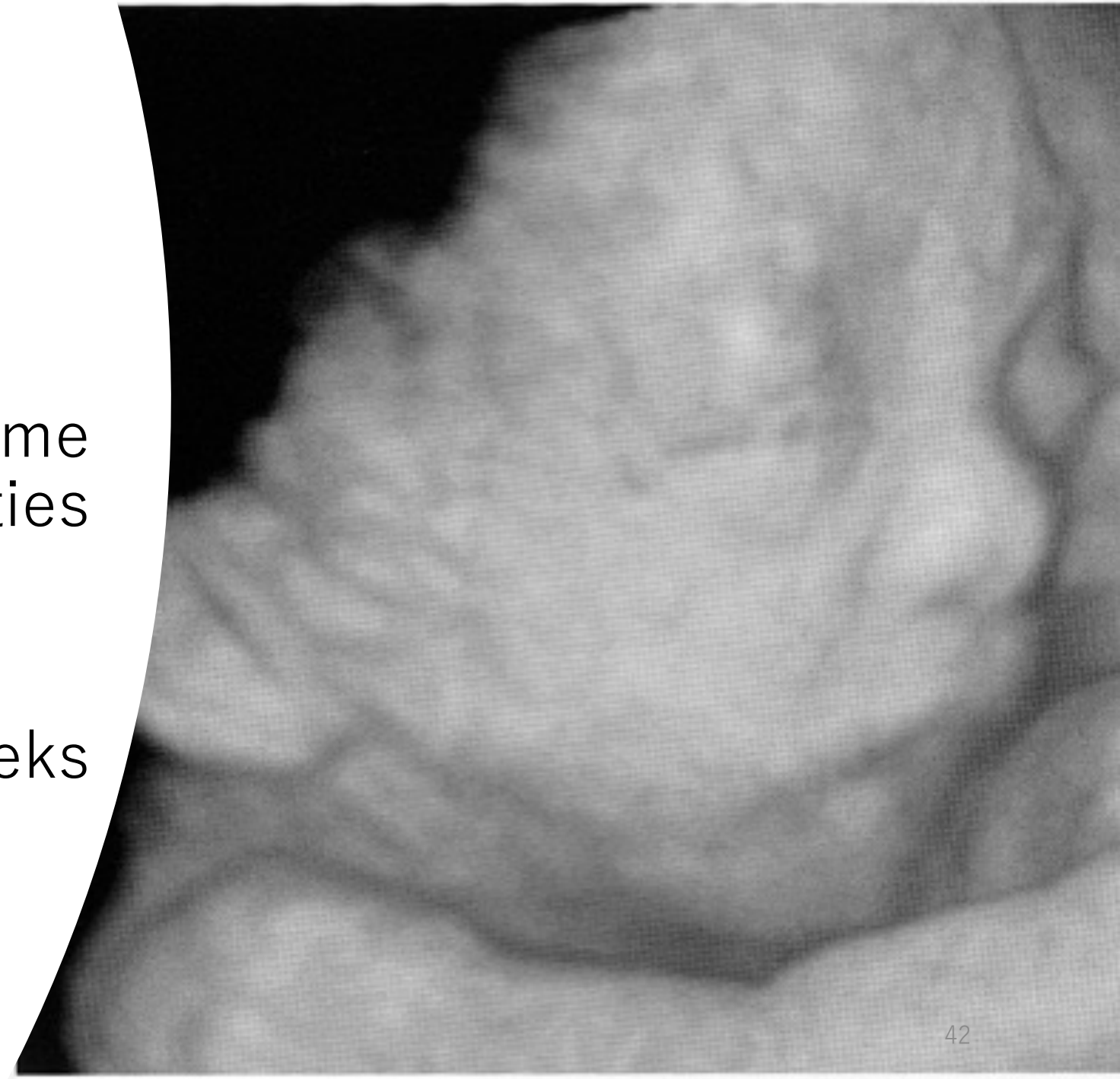
Beckwith-Wiedemann syndrome, 21trisomy

BWS: Growth-related genes in chromosome 11 are inherited only from the father. (Paternal Disomy)

Auricle

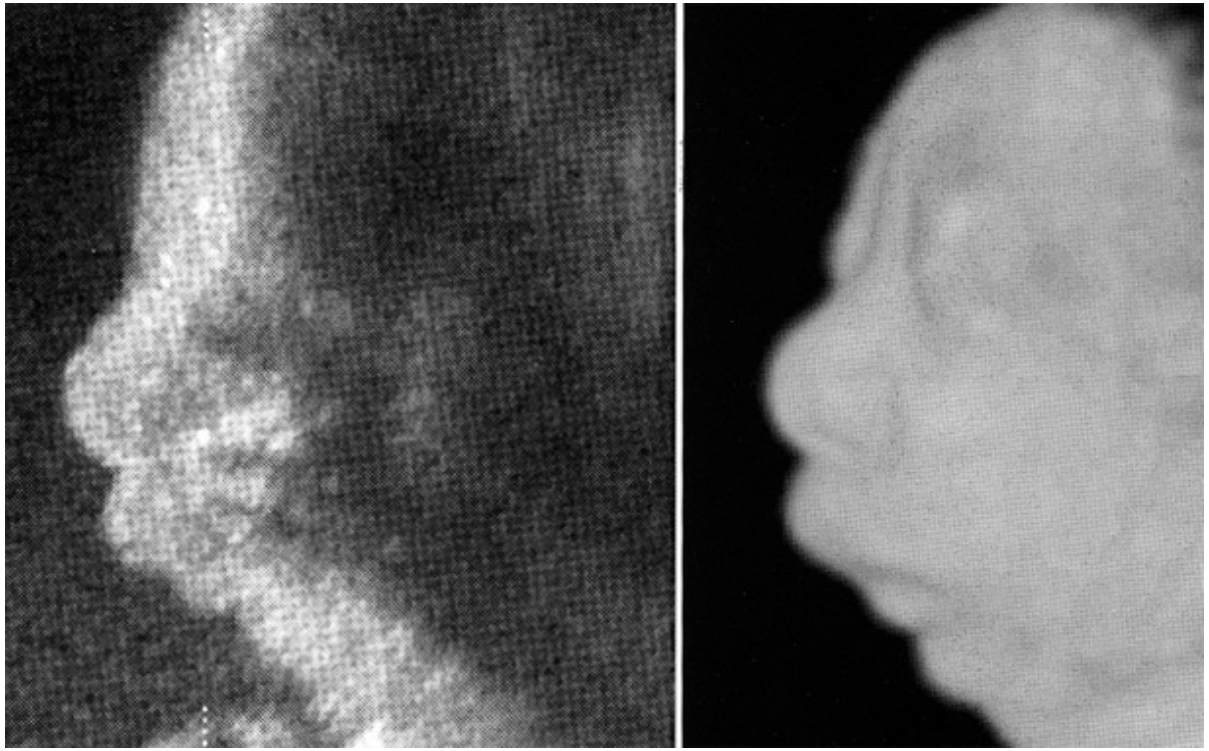
- **Microtia**: chromosome number abnormalities including 21+

- **Low set ears**:(28 weeks gestation; 18+, 21+)



Jaw

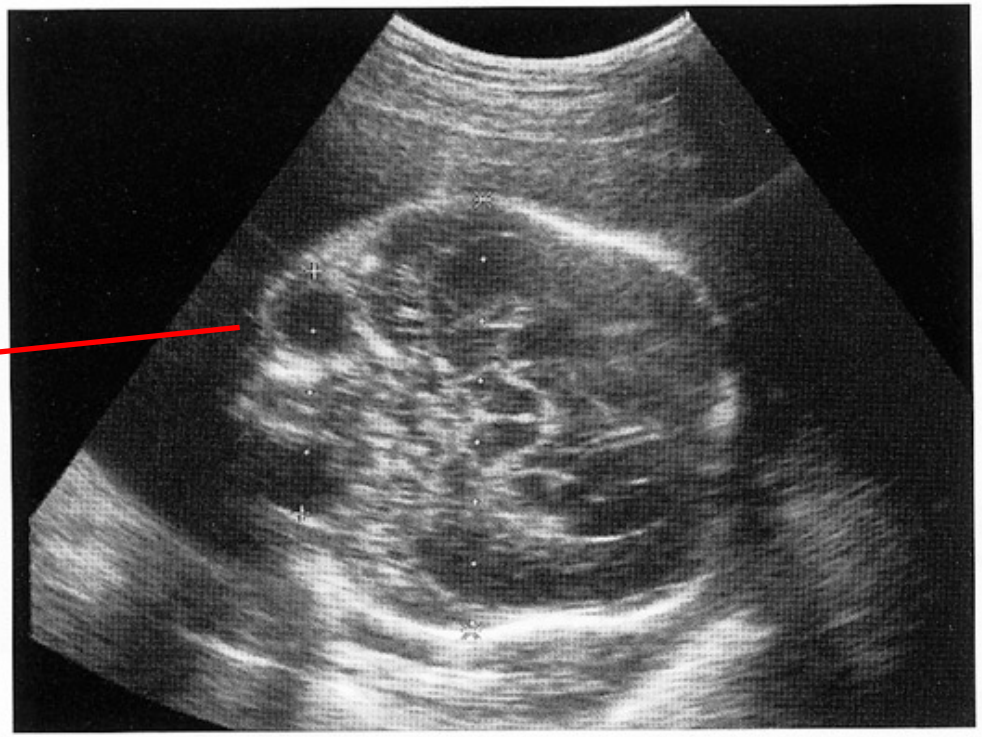
- **Micrognathia**：Pena-Shokar syndrome・Treacher-Collins syndrome・Pierre Robin症候群
- Micrognathia can cause polyhydramnion and respiratory failure.



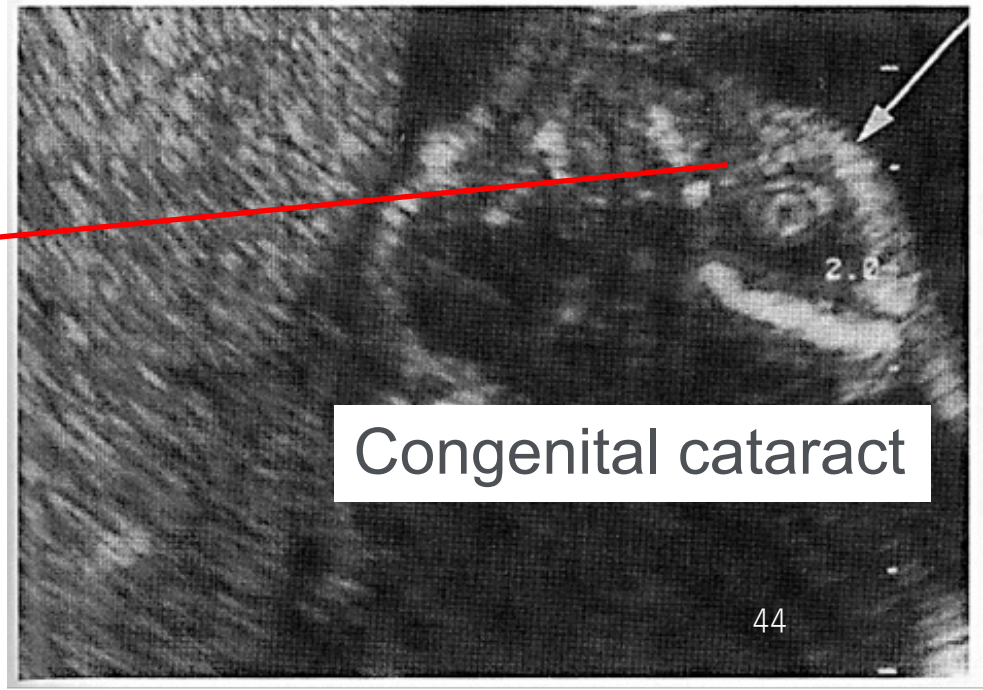
micrognathia（30 weeks gestation）

Eyes

- hypertelorism・hypotelorism：
  Malformation syndrome



- Microphthalmia・Anophthalmia：
  Infectious disease（CMV・Rubella・Toxo]
- Anophthalmia： 13+

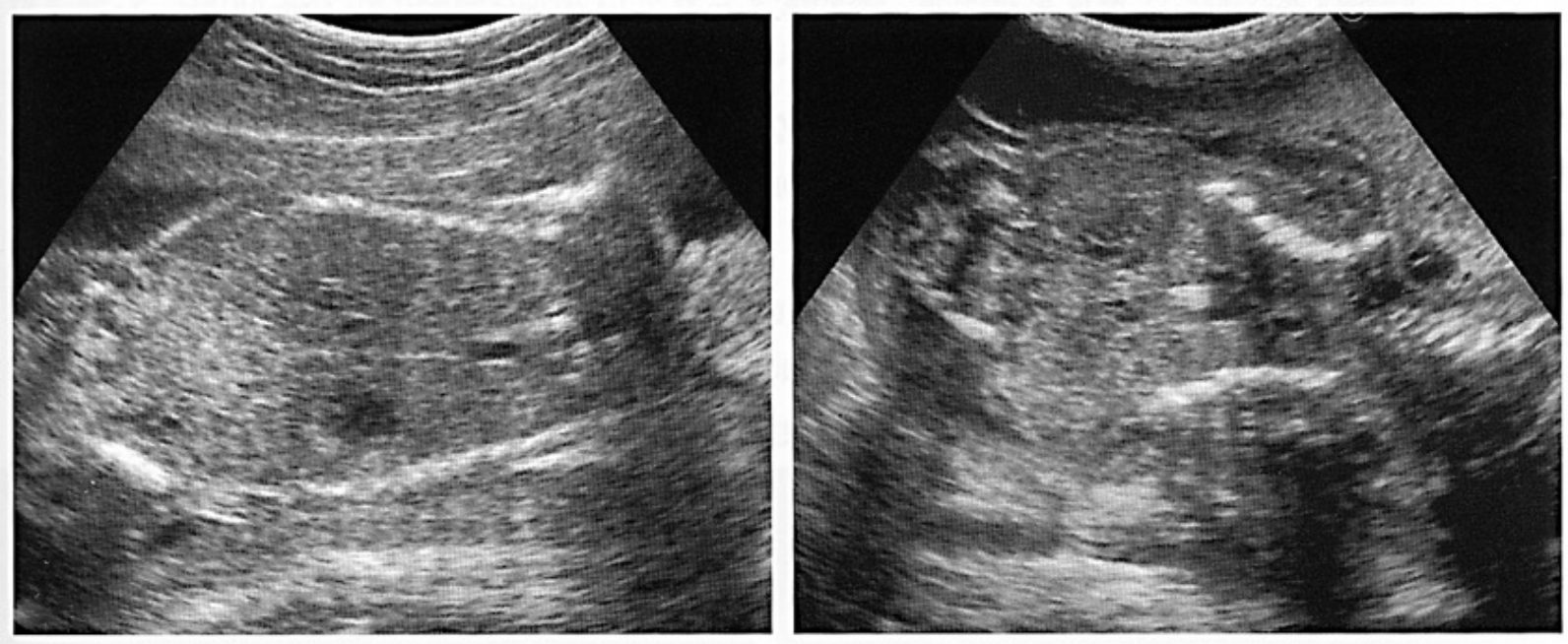


Chest

- It is important to prevent postnatal respiratory failure due to lung hypoplasia.
- The frequency of congenital cardiac diseases is about 1%.

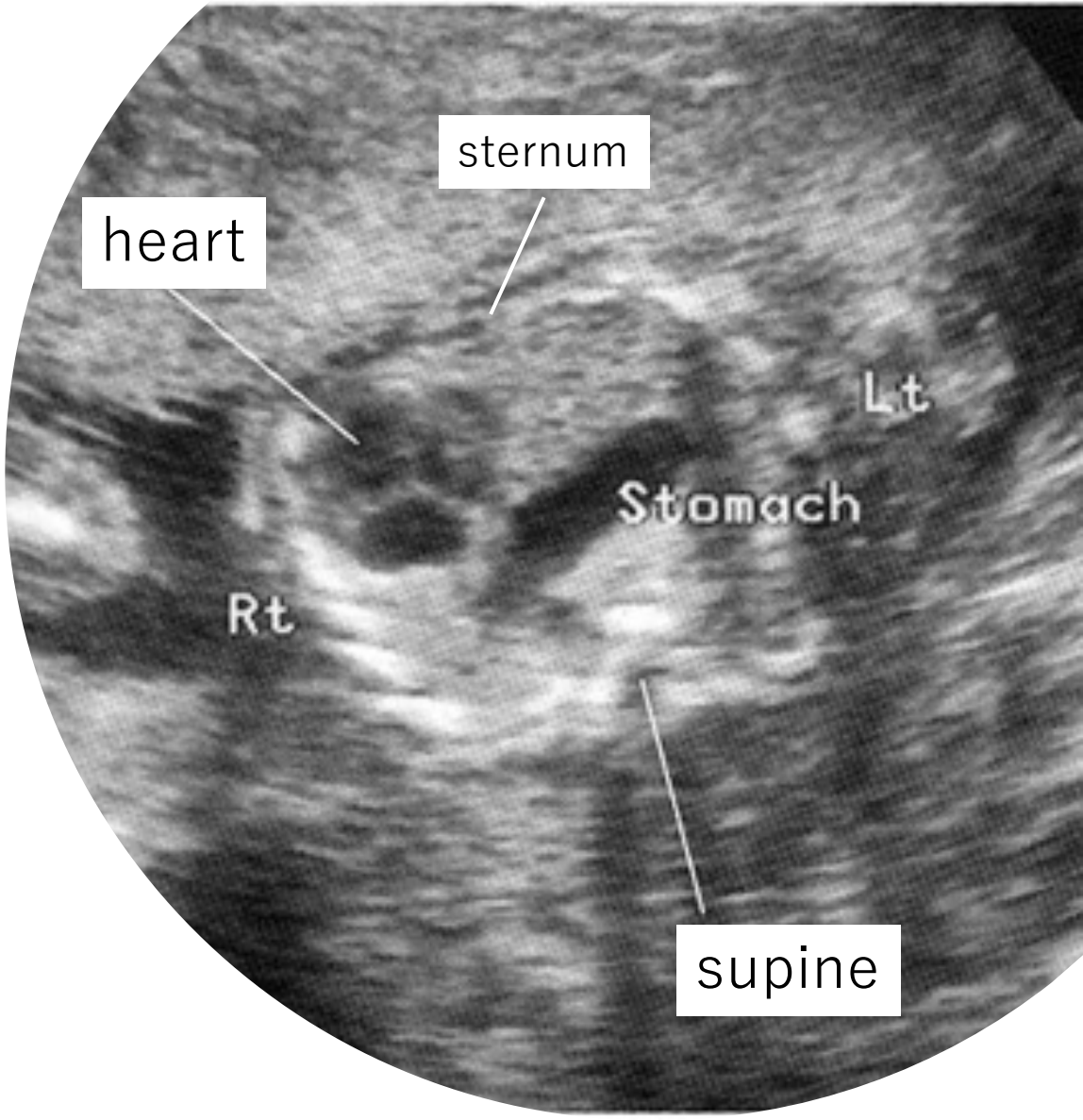
Embryology in early pregnancy (thorax)

- Pulmonary hypoplasia
  - Caused by oligohydramnion (PROM: premature rupture of membranes), tumor in the thorax, etc.
  - Diaphragmatic hernia, congenital cystic adenomatoid malformation, pulmonary sequestration, etc.



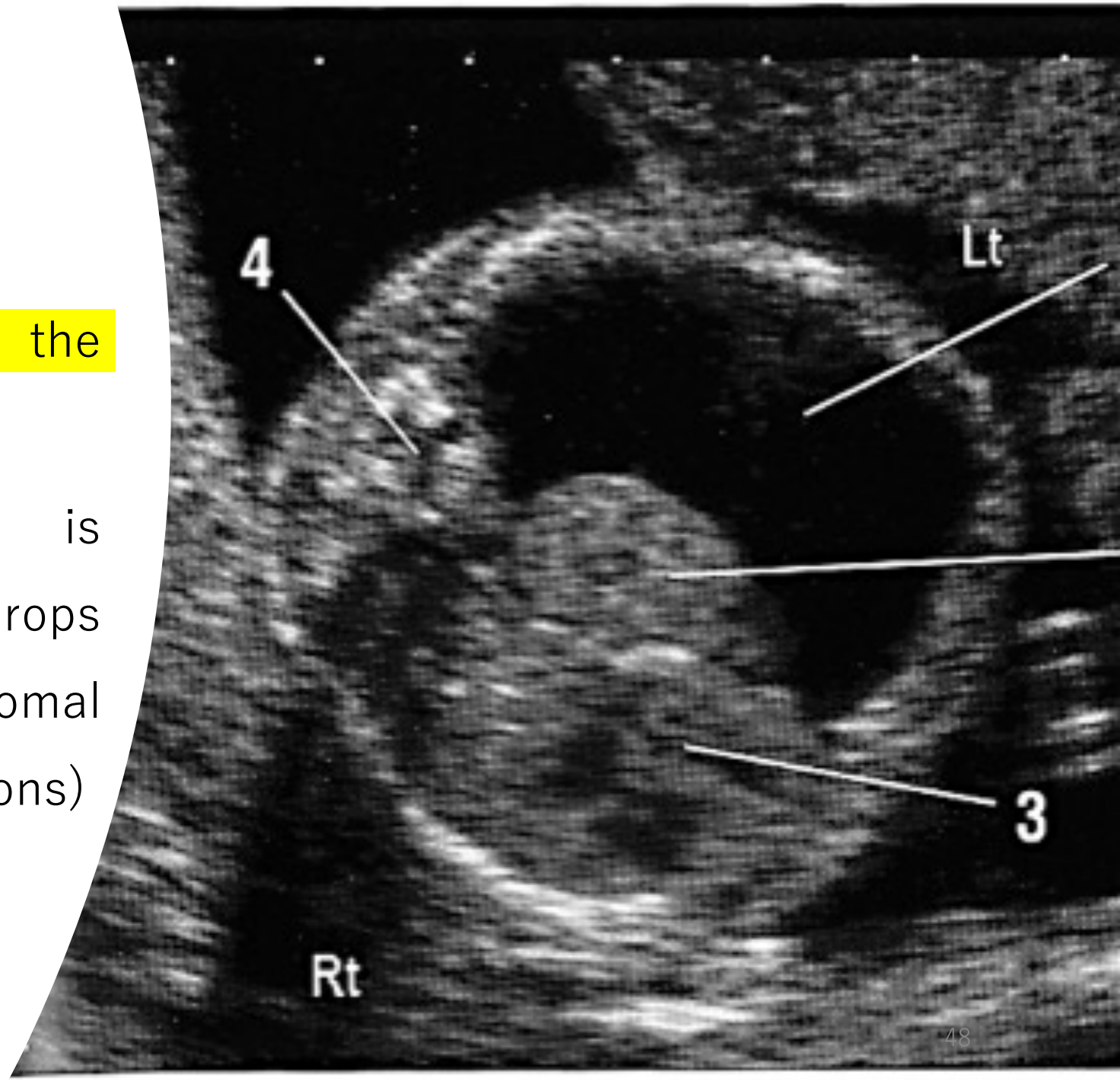
Diaphragmatic hernia

- When a physiologic umbilical hernia is refluxed, increased intra-abdominal pressure causes intrusion of abdominal organs into the thoracic cavity through the incomplete primitive diaphragm.
- Poor prognosis: early intrusion of abdominal organs into the thorax.
- Good prognosis: Intrusion of abdominal organs after the second trimester.
- Possible chromosomal abnormality (18+).



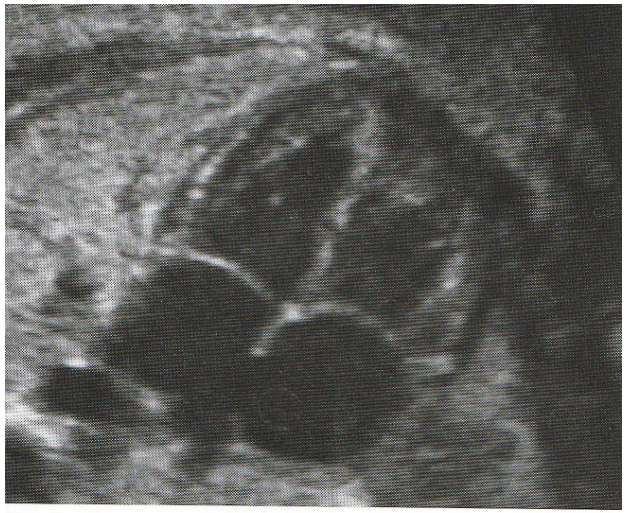
Pleural effusion

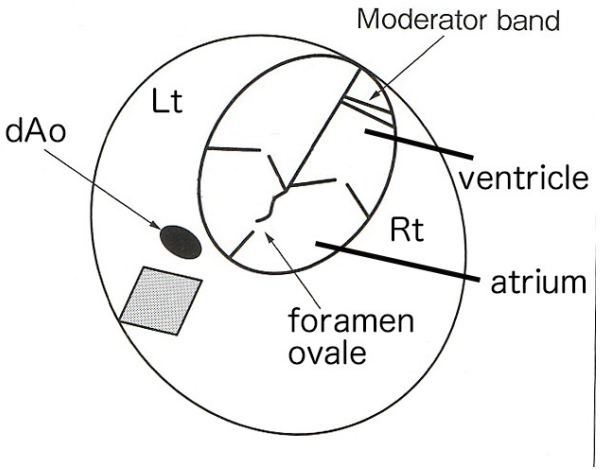
- If it is unilateral, chylothorax is the most common.
- Bilateral pleural effusions is associated with fetal hydrops (infection, anemia , chromosomal abnormalities, cardiac malformations)



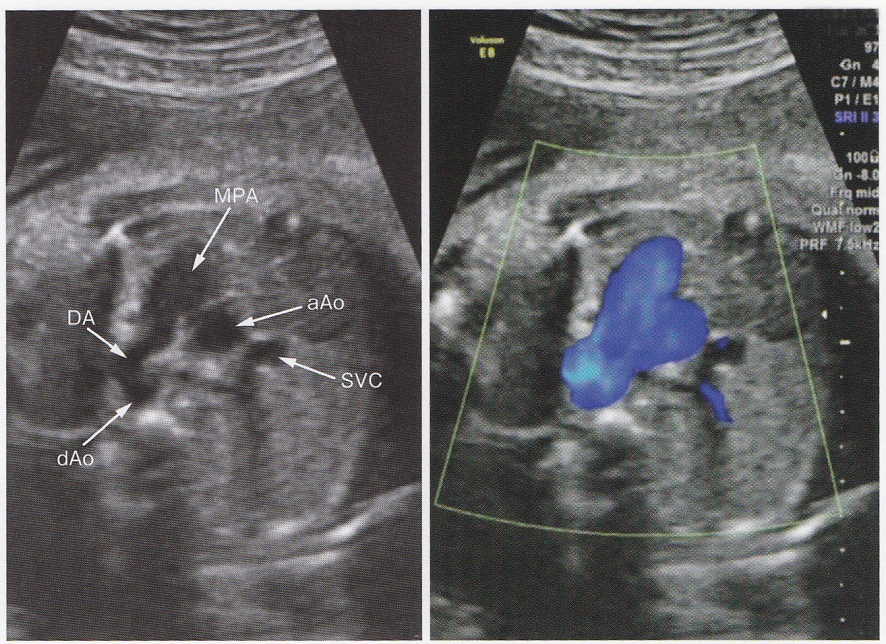
heart

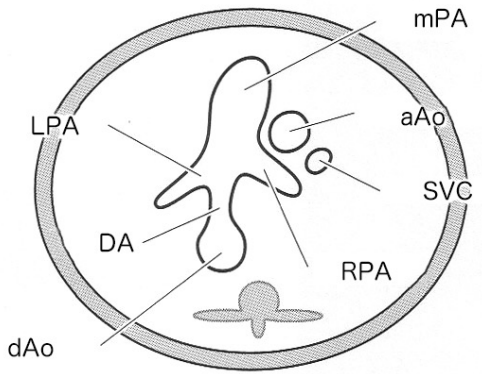
Screening of cardiac anomaly



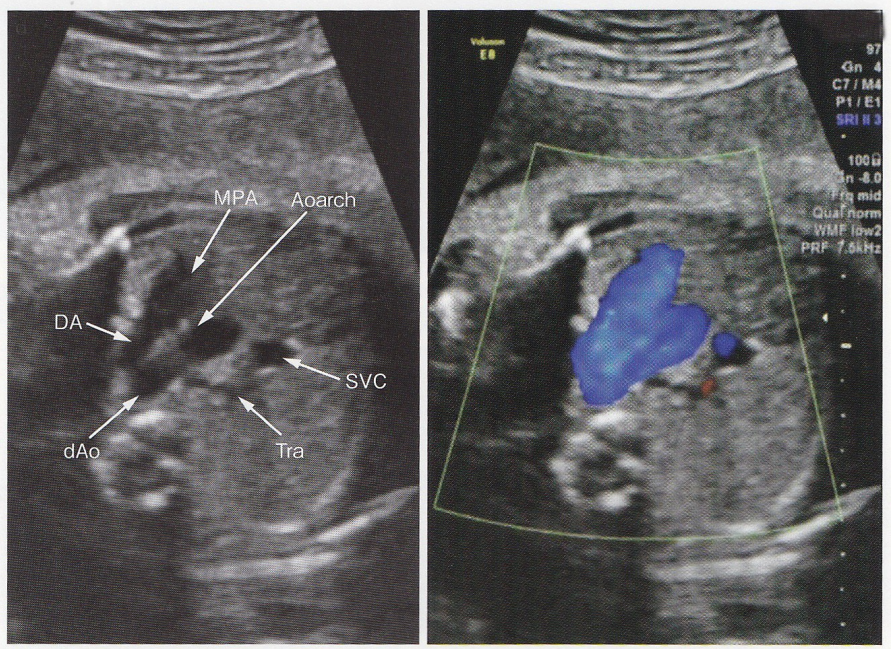


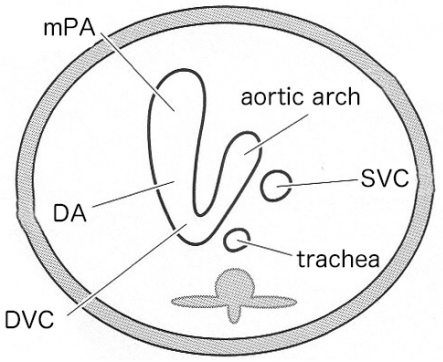
four chamber view





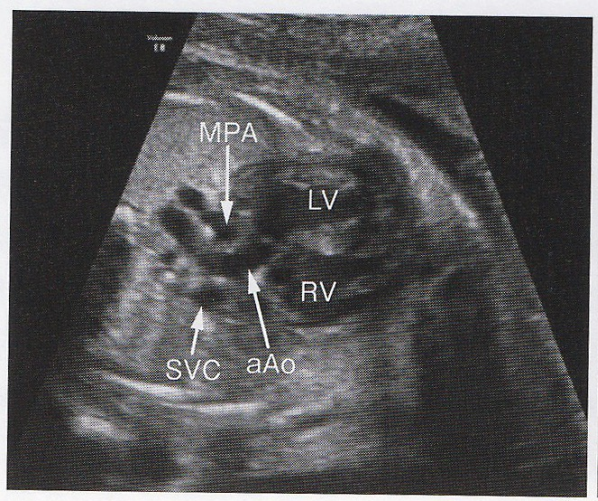
three vessel view

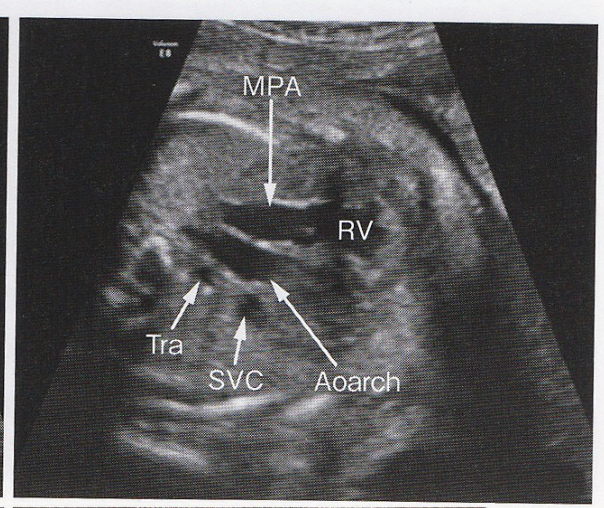


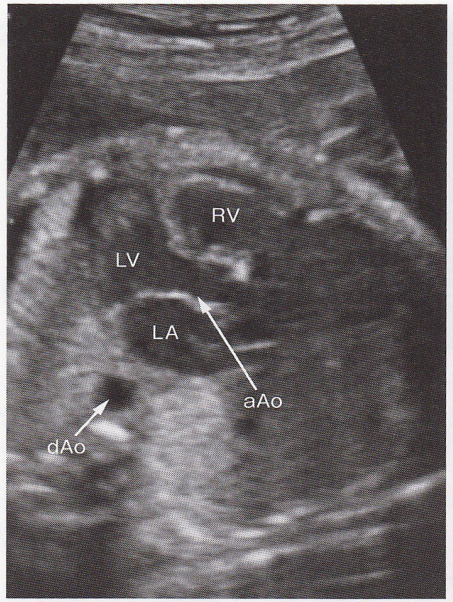


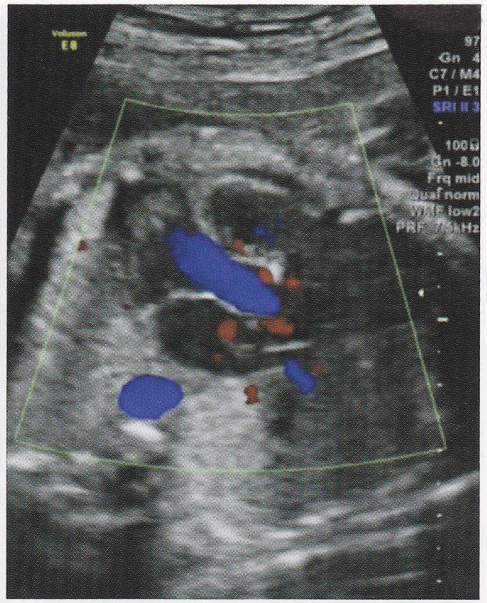
three vessel trachea view

Screening of cardiac anomaly

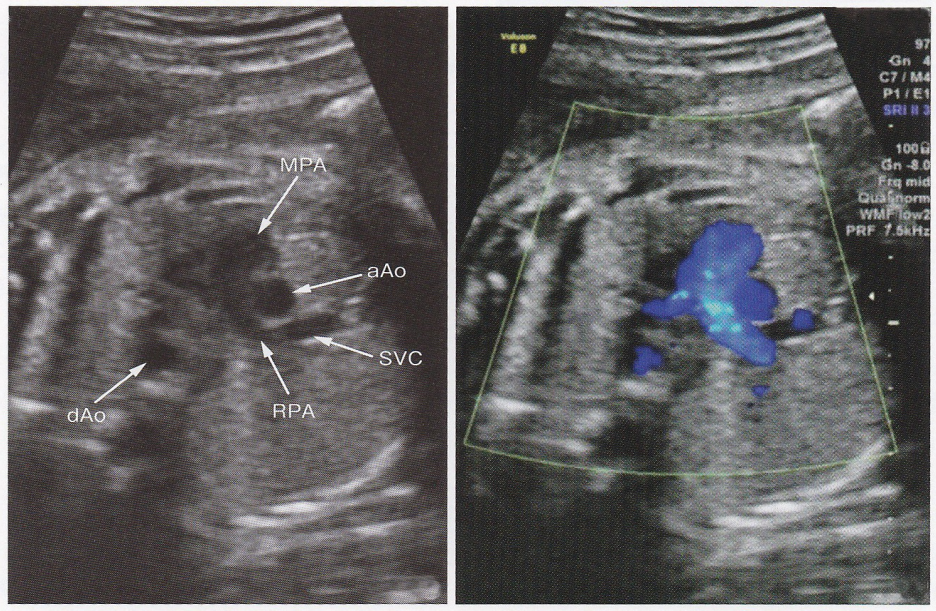


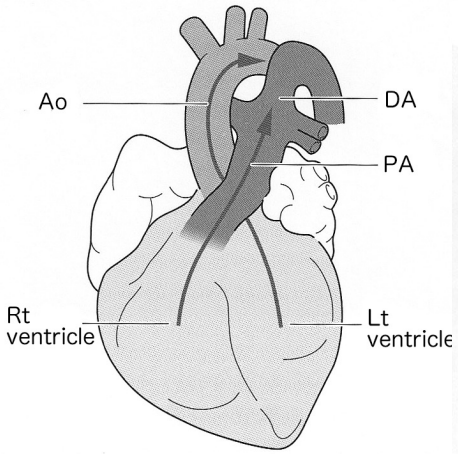


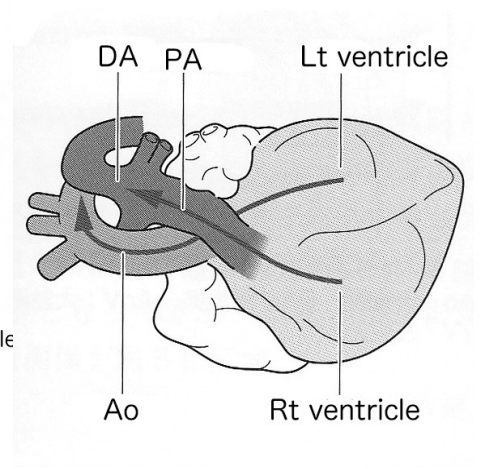




left ventricular outflow tract

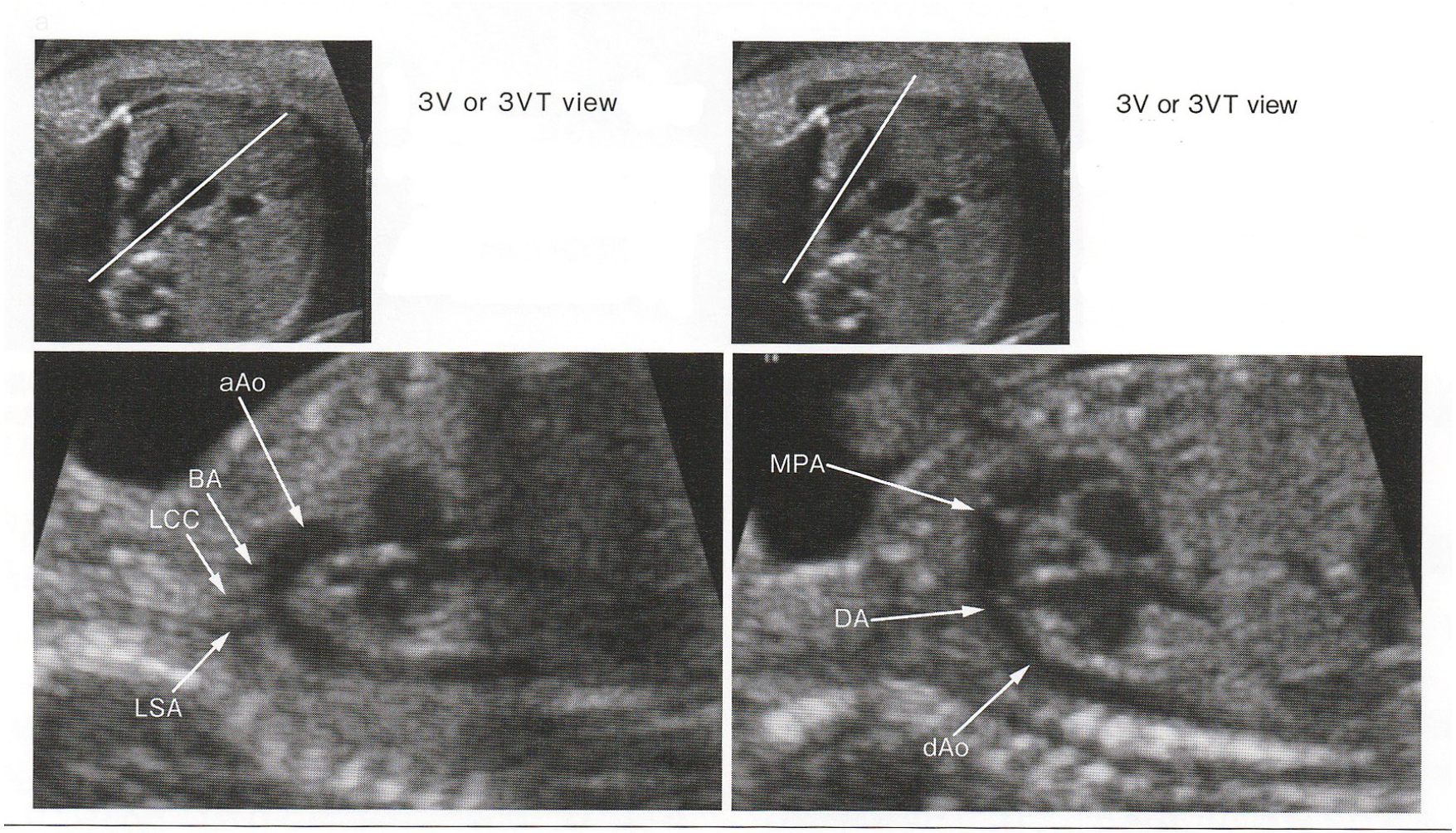




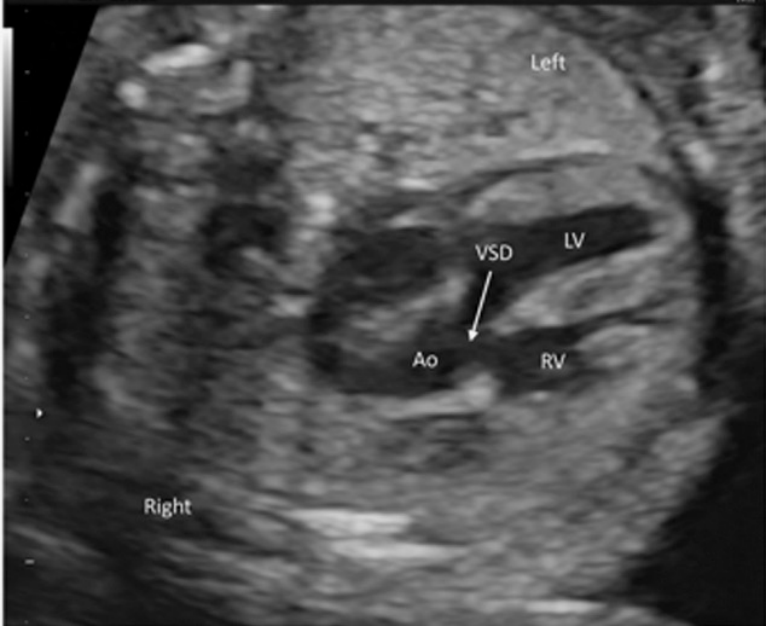


right ventricular outflow tract

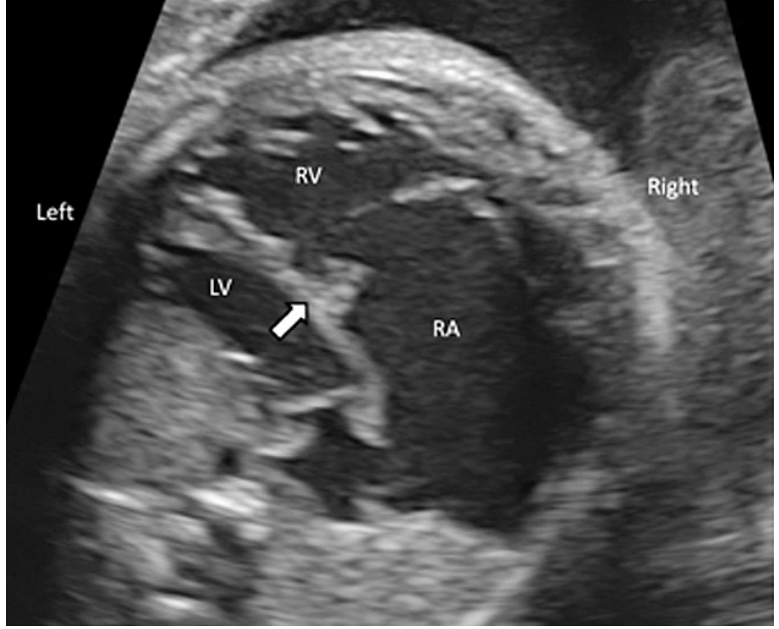
Screening of cardiac anomaly



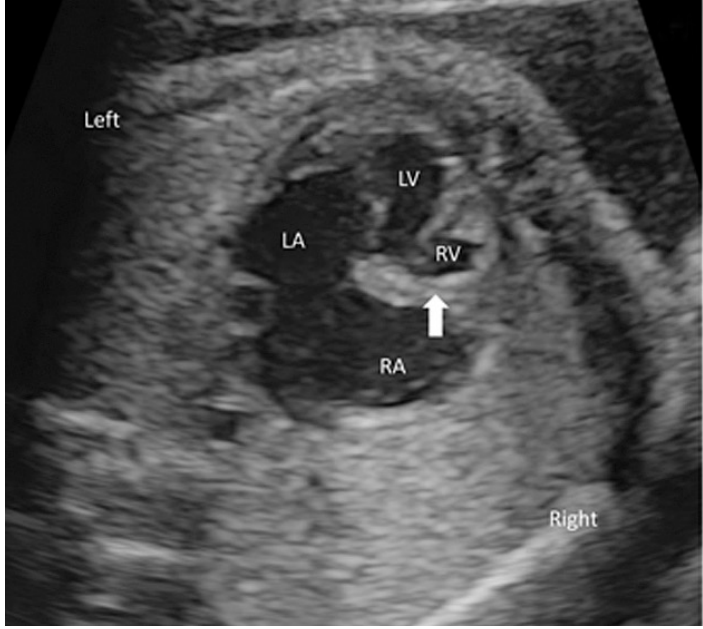
aortic Arch



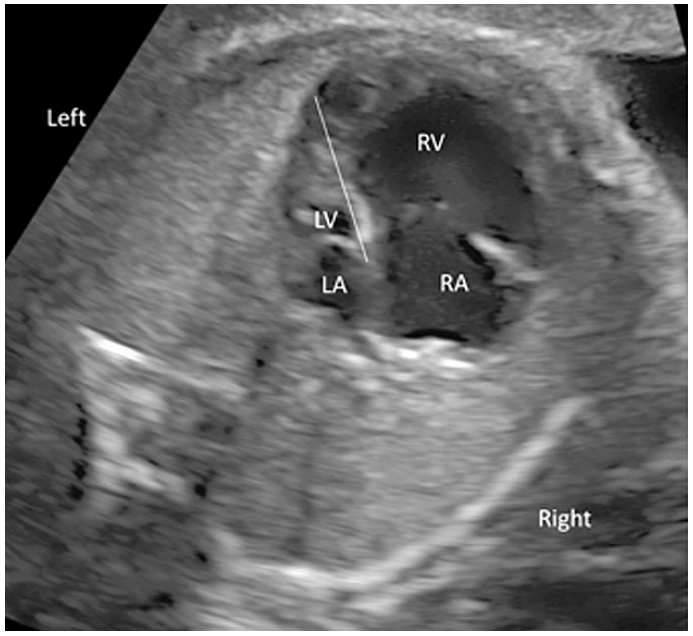
Tetralogy of Fallot (TOF)



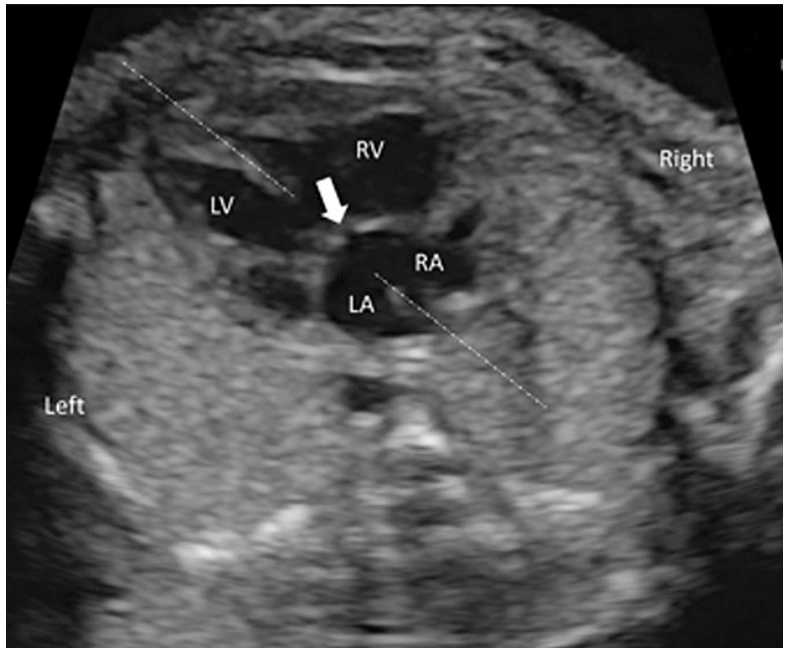
Ebstein anomaly



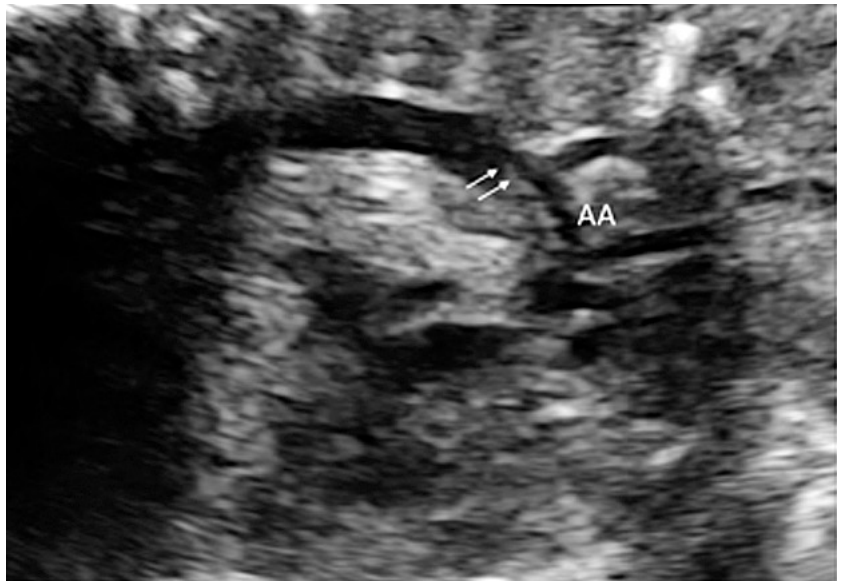
tricuspid atresia



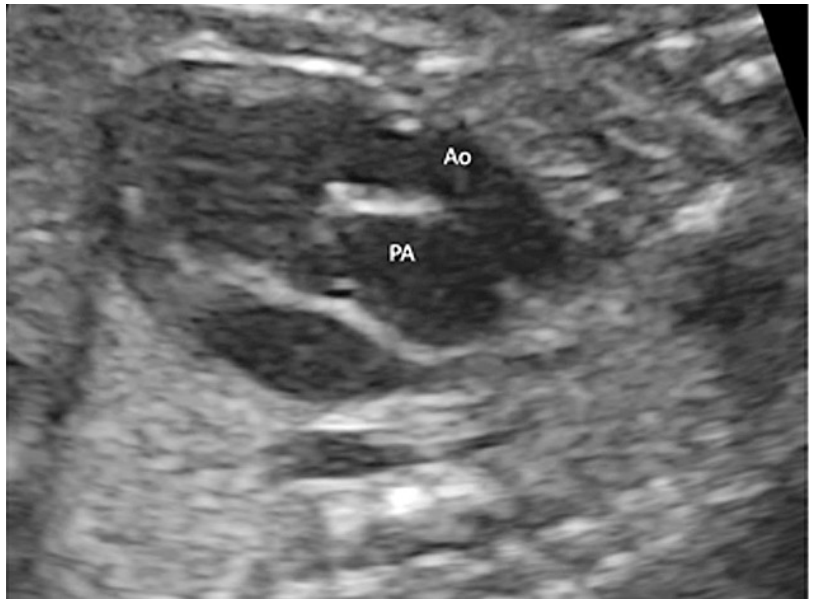
hypoplastic left heart syndrome (HLHS)



atrioventricular septal defect (ASD)



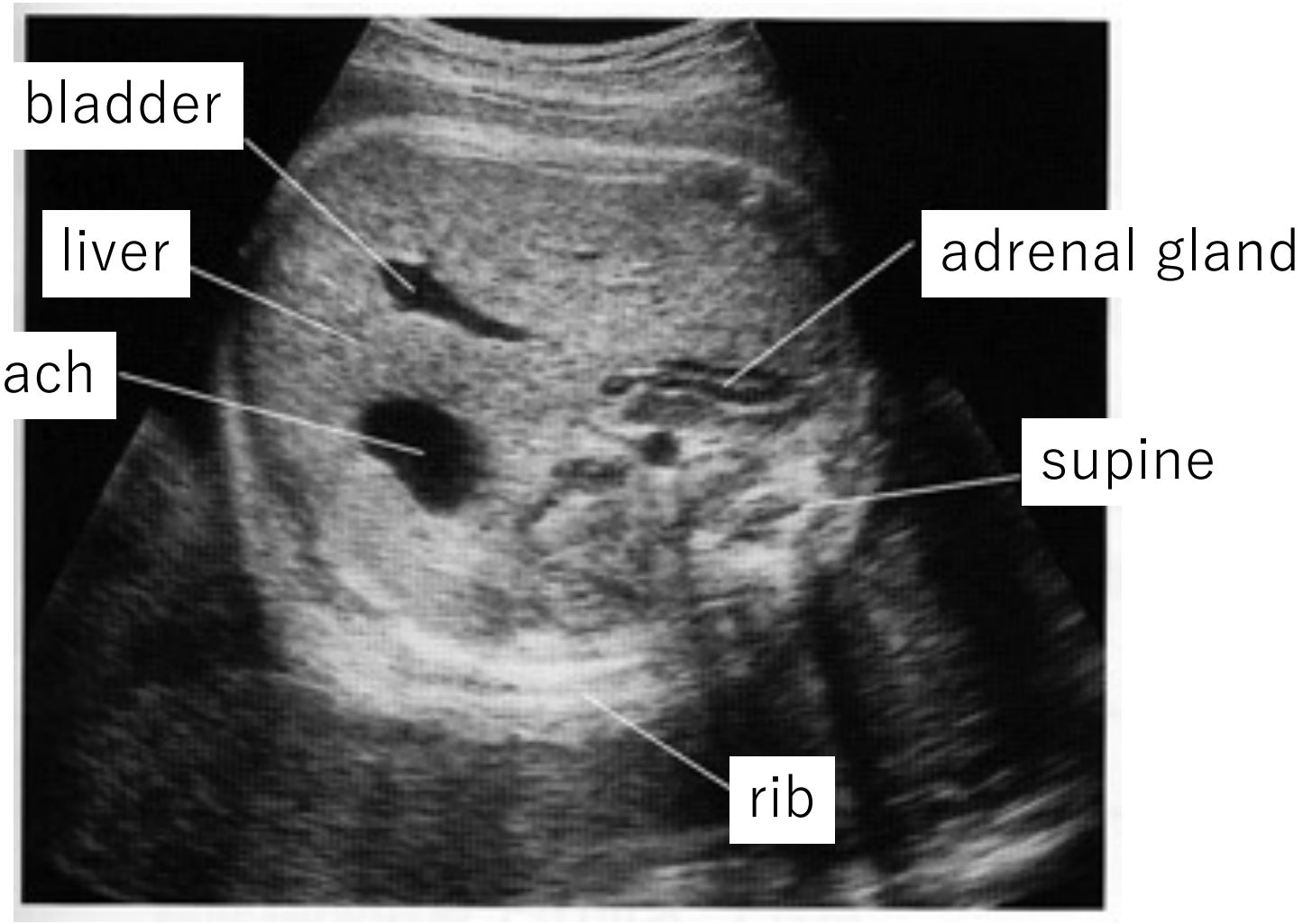
coarctation of aorta (CoA)



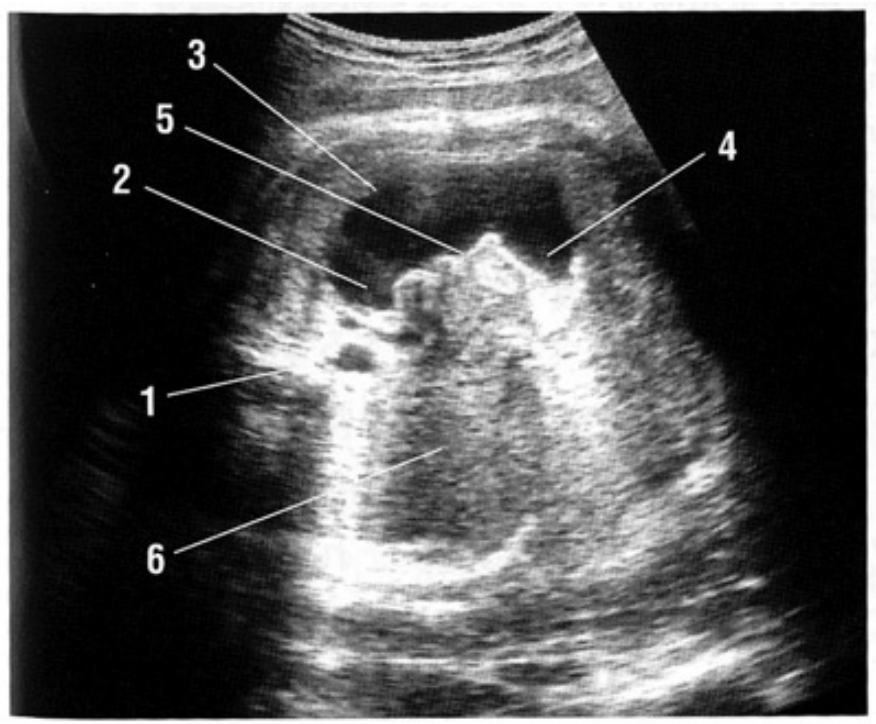
complete transposition of great arteries (TGA)

Abdomen

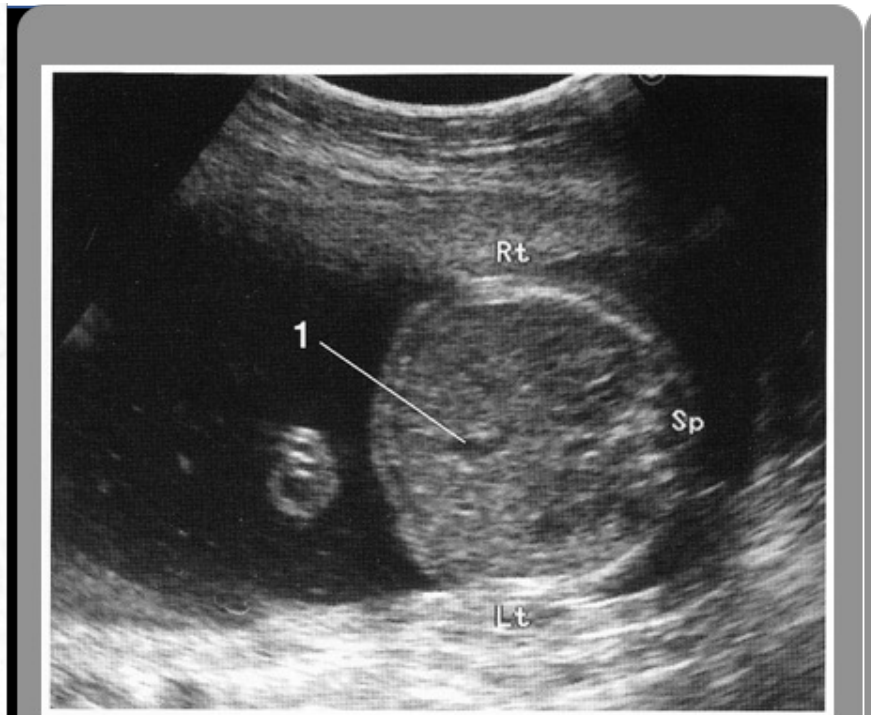
Upper abdomen (40th weeks gestation)



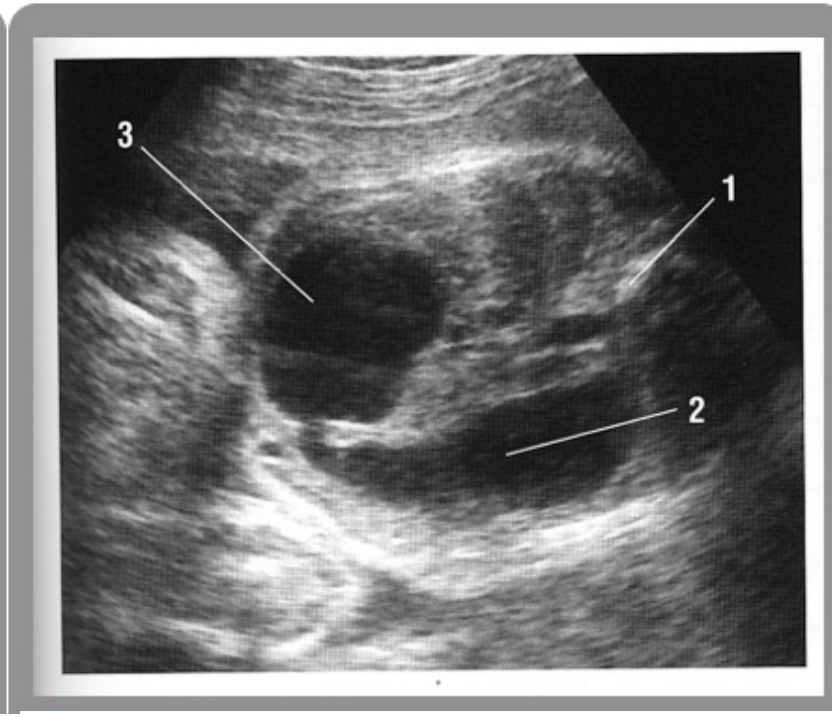
Enlargement of gastrointestinal tract：mid-term (>15mm) and late-term (>20mm)



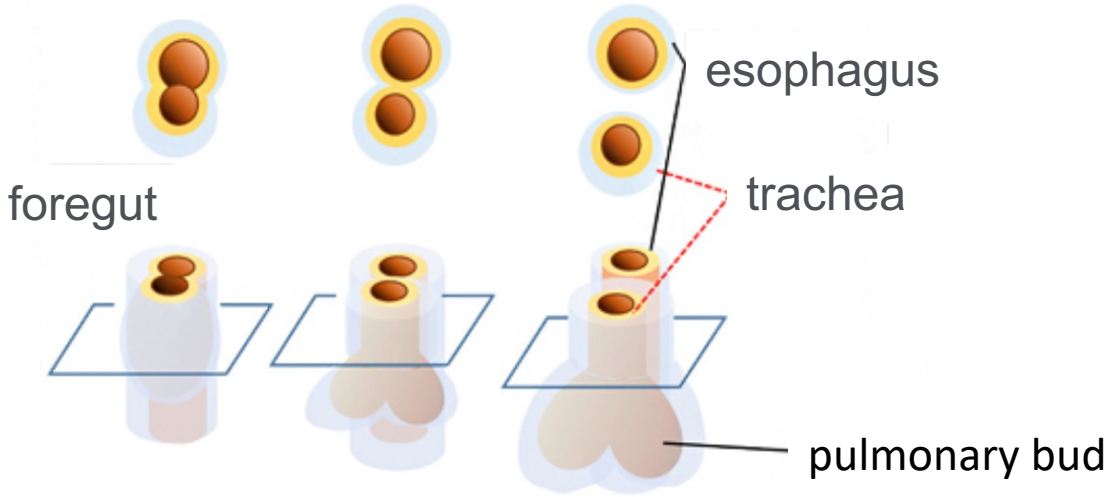
abdominal echo of normal fetus (38th)
2: gastroduodenal region
4: gastropyloric region, 6: liver

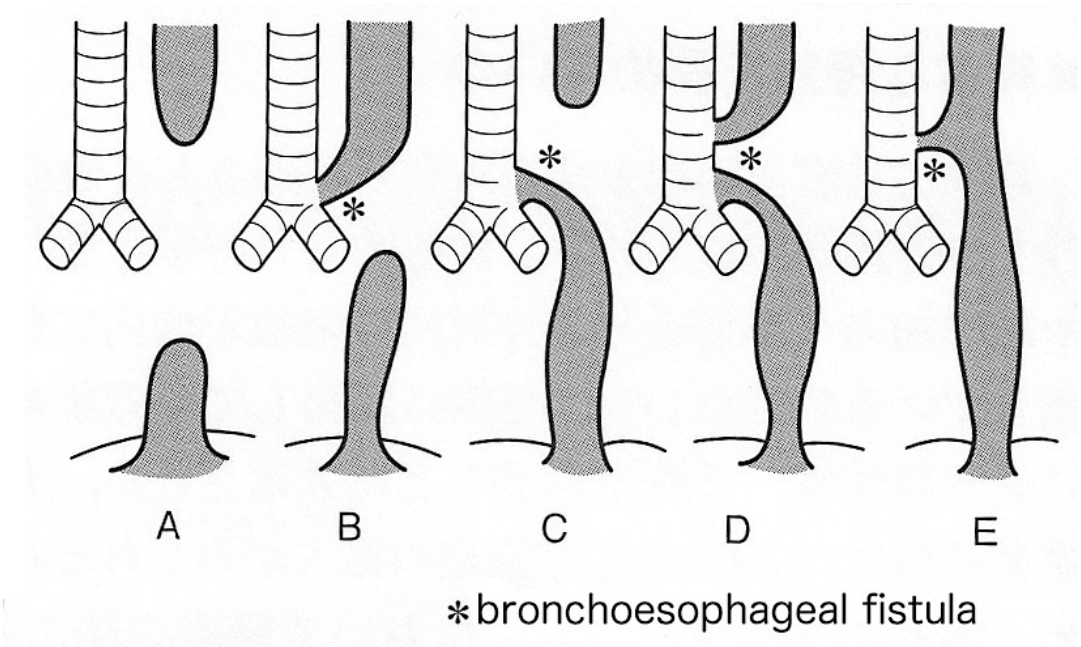


Esophageal atresia (20th): extremely small gastric vesicle, polyhydramnion, FGR (21+)

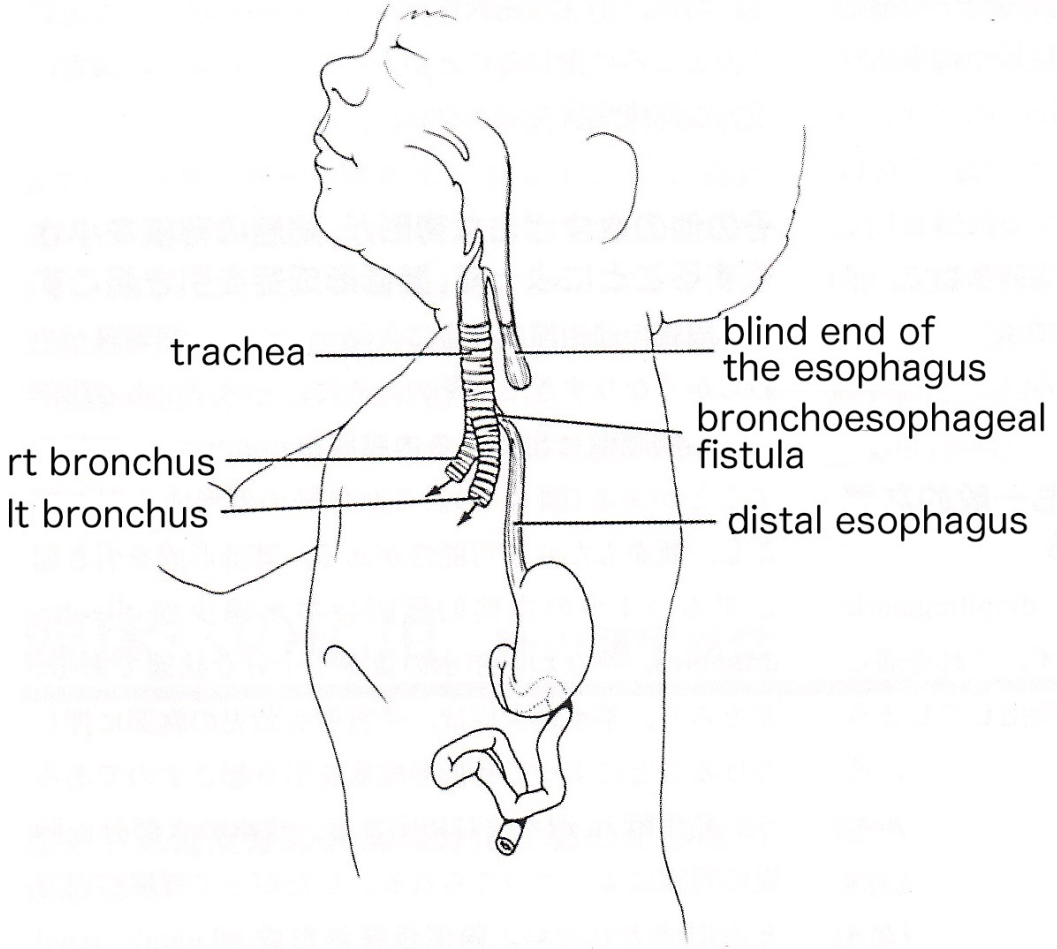


Duodenal atresia (32th): double bubble sign (gastric vesicle and duodenal enlargement) (21+)

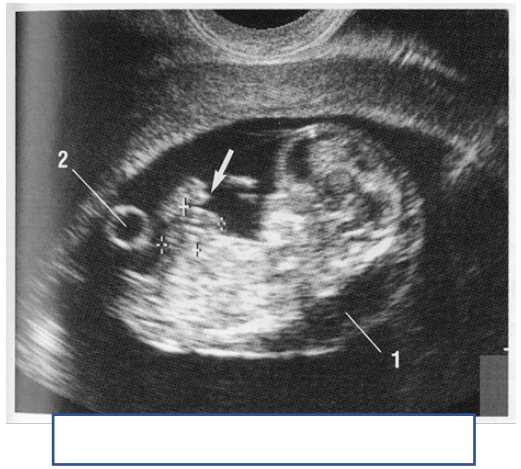


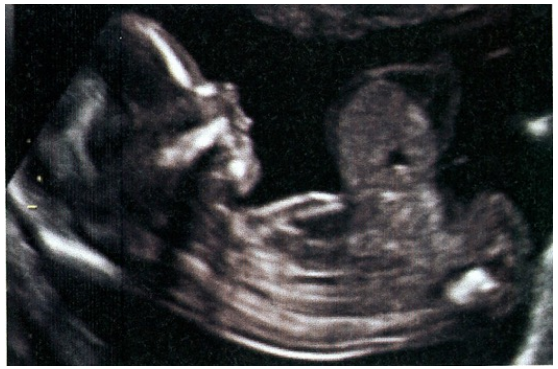


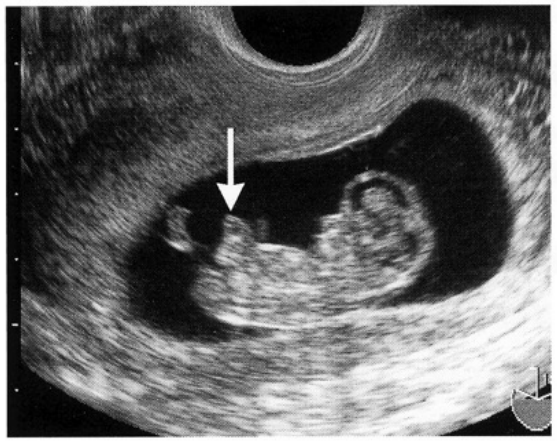
Gross classification of esophageal atresia

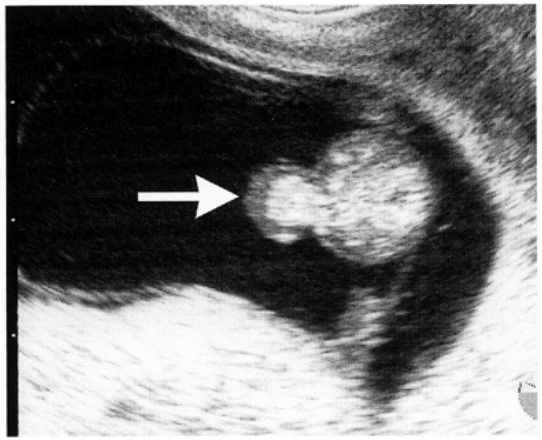


Esophageal atresia of Gross C







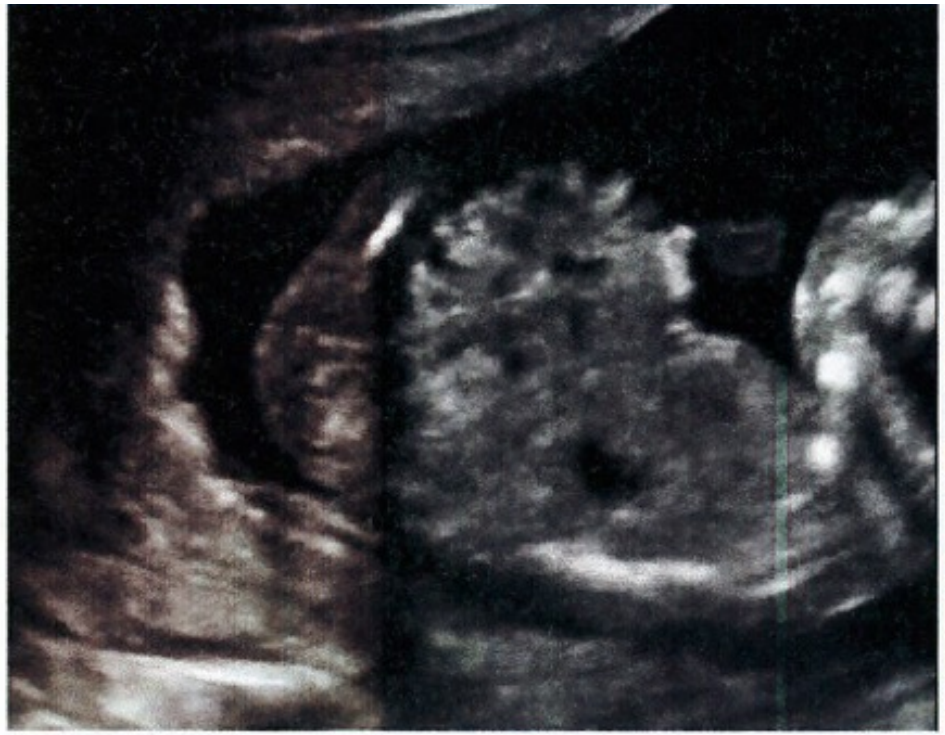


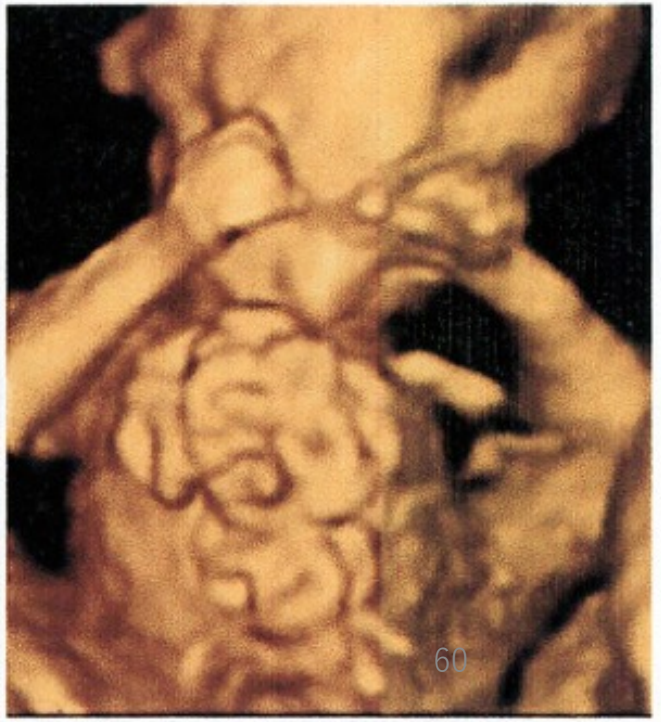
umbilical hernia (12th)

- Lt upper figure: Umbilical hernia (11 weeks' gestation): The hernia sac is more than 10 mm and NT 7 mm, so 21 trisomy is suspected.
- Association with 18+, 13+: If only the intestinal tract migrates into the hernia sac, the risk of chromosomal abnormality is decreased compared to when the liver is included.
- Physiologic hernia sacs are never more than 7 mm.

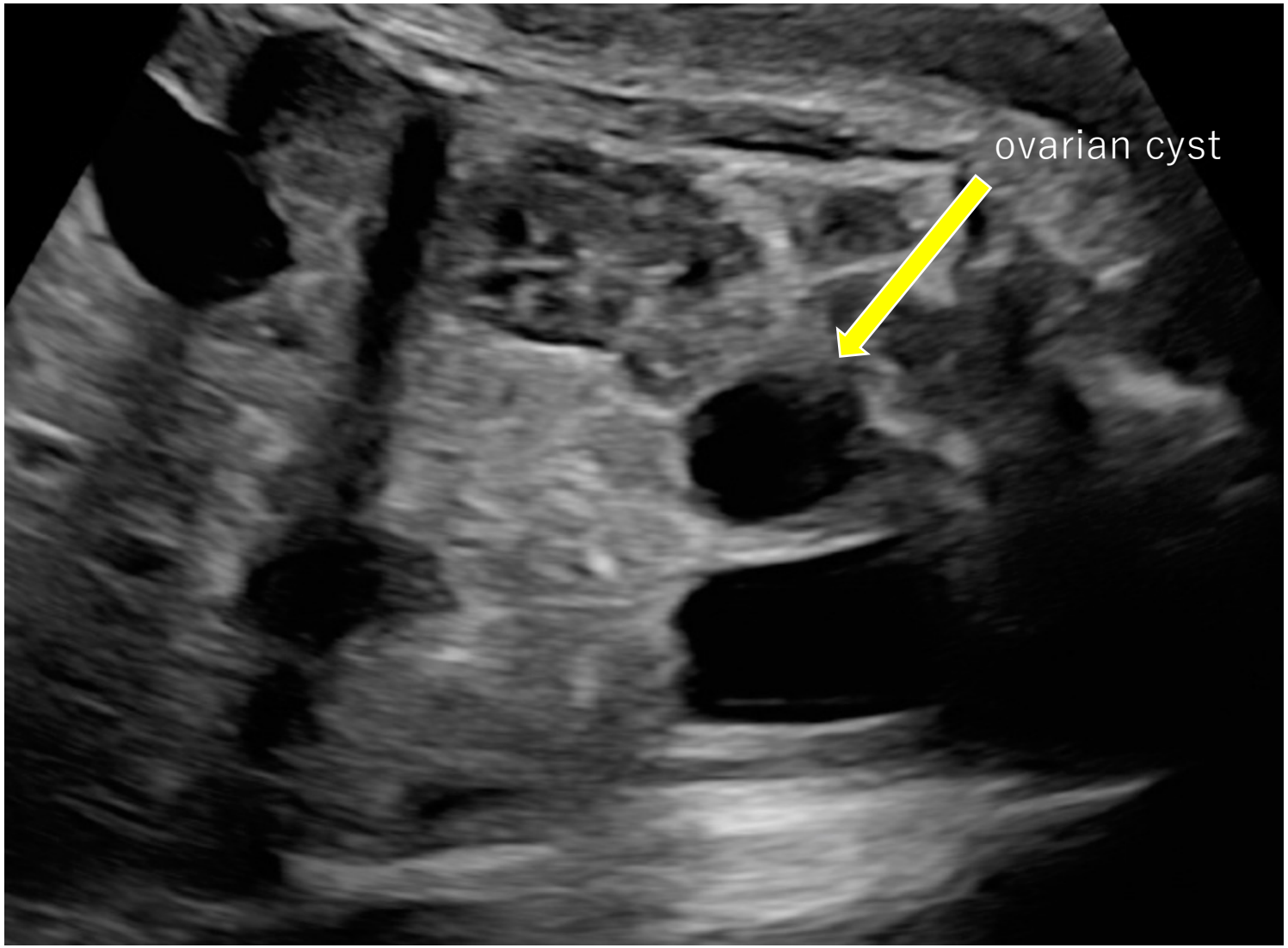
Gastroschisis (12th week of gestation)

- Chromosomal abnormalities are rare.
- The umbilical cord is normal and the intestinal tract go from a nearby defect in the abdominal wall.
- The intestinal tract becomes inflamed and thickened by amniotic fluid, causing postnatal intestinal obstruction.



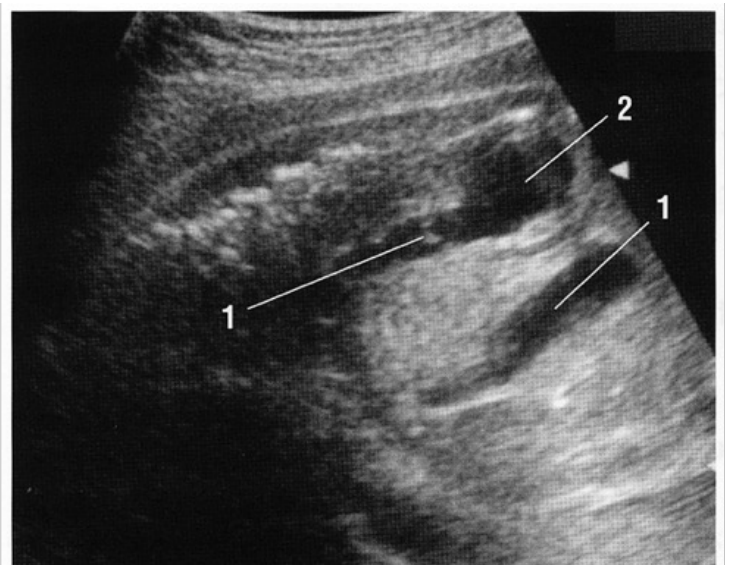


Ovarian cyst



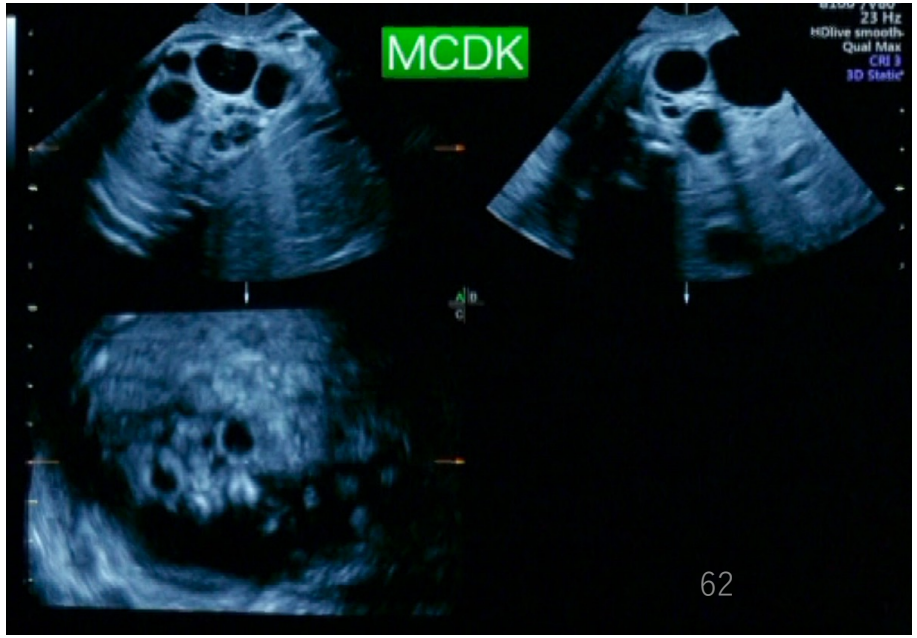
Hydronephrosis

- Cause:
  - ➢Ureteropelvic junction stenosis (prognosis is good).
  - ➢Vesicoureteral junction stenosis (prognosis depends on the ureteral diameter)



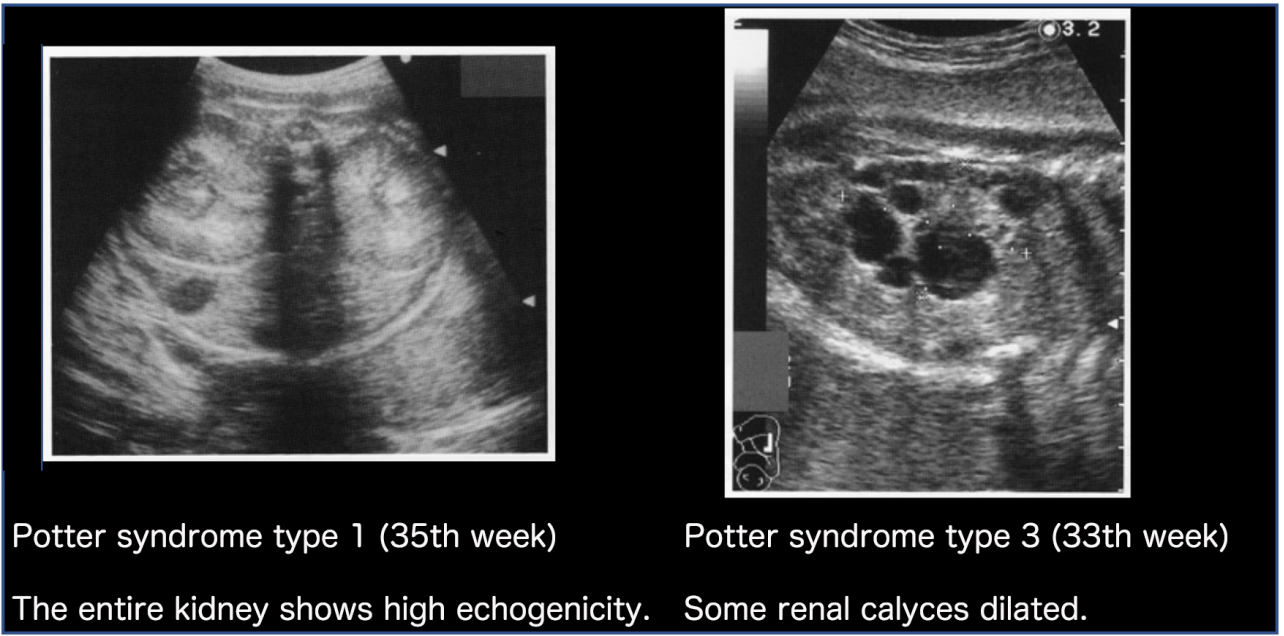
Vesicoureteral junction stenosis
29 weeks of gestation

- However, the differential diagnosis of multiple dysplastic kidney (MCDK) is important because the prognosis of MCDK is poor.



multiple dysplastic kidney (MCDK) (Potter syndrome)

- **Potter type 1**: Symmetrical enlargement of the entire kidney due to microcystic dilation of the renal tubules. The prognosis is very poor if there is a family history (autosomal recessive inheritance) or if there is oligohydramnion.
- **Potter type 3** (MCDK): Kidney is occupied by cysts of various sizes. Unilateral cases have a good prognosis; however, bilateral cases are fatal because of renal failure (oligohydramnion). (Autosomal dominant inheritance)



Potter syndrome type 1 (35th week)

The entire kidney shows high echogenicity.

Potter syndrome type 3 (33th week)

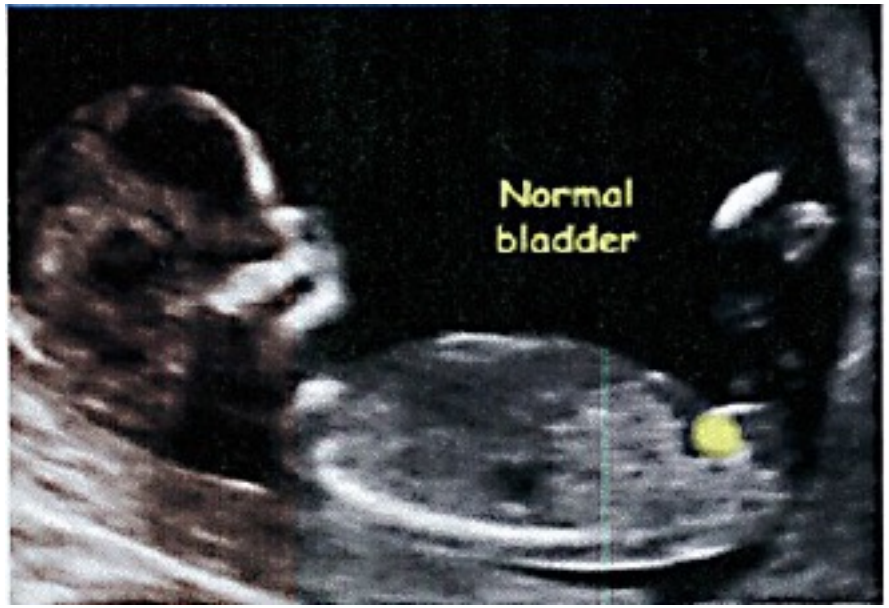
Some renal calyces dilated.

Potter type 1	infantile polycystic kidney disease (IPKD) autosomal recessive polycystic disease (ARPKD)
Potter type 2	multicystic dysplastic kidney (MCDK) sporadic
Potter type 3	adult polycystic kidney disease (APKD) autosomal dominant polycystic disease (ADPKD)

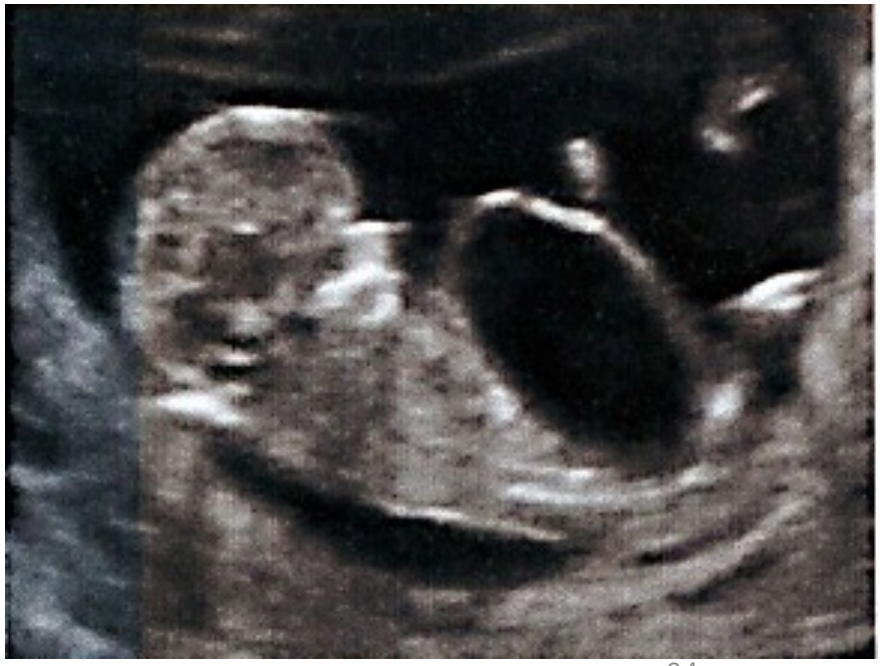
Giant bladder (11 weeks gestation)

Prune-Berry syndrome

- Fetal bladder diameter at 13 weeks gestation is less than 6 mm; amniotic membrane produces amniotic fluid until 15 weeks.
- If the bladder diameter is 7-15 mm, 20% have a chromosomal abnormality. (13 and 18 trisomies) In the case of normal chromosomes, 90% improve spontaneously. (8% need cystoamniotic shunt.)
- If the bladder length is greater than 15 mm, chromosomal abnormalities are present in 10% of cases, and if the chromosomes are normal, obstructive uropathy is the most likely cause.



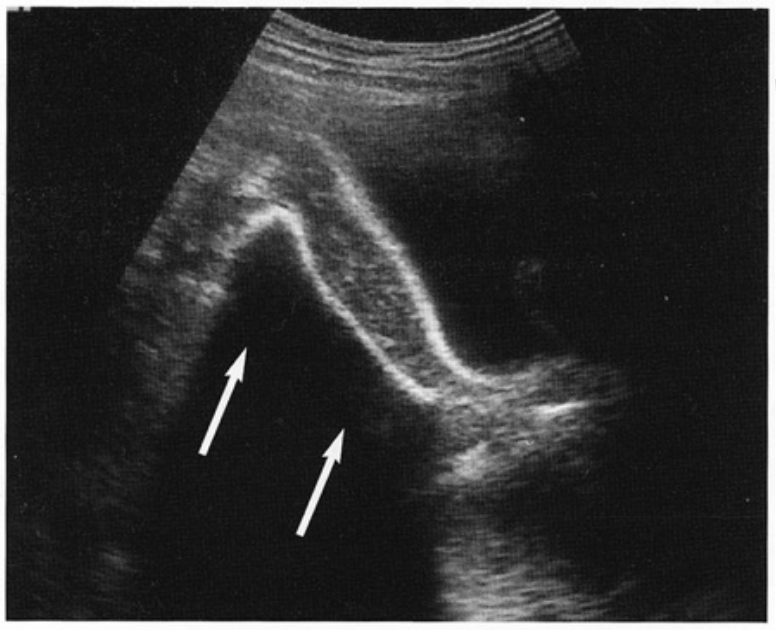
normal bladder



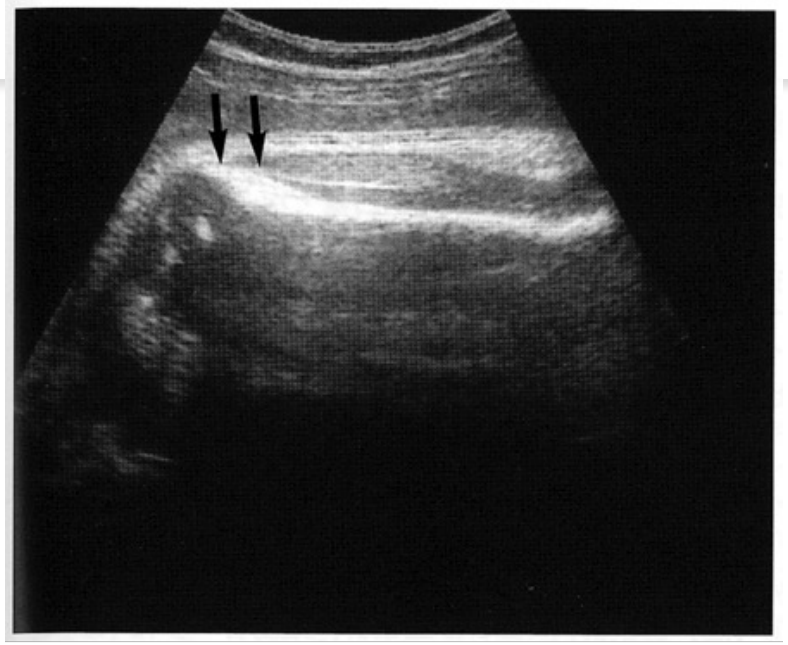
megacystis

Bone

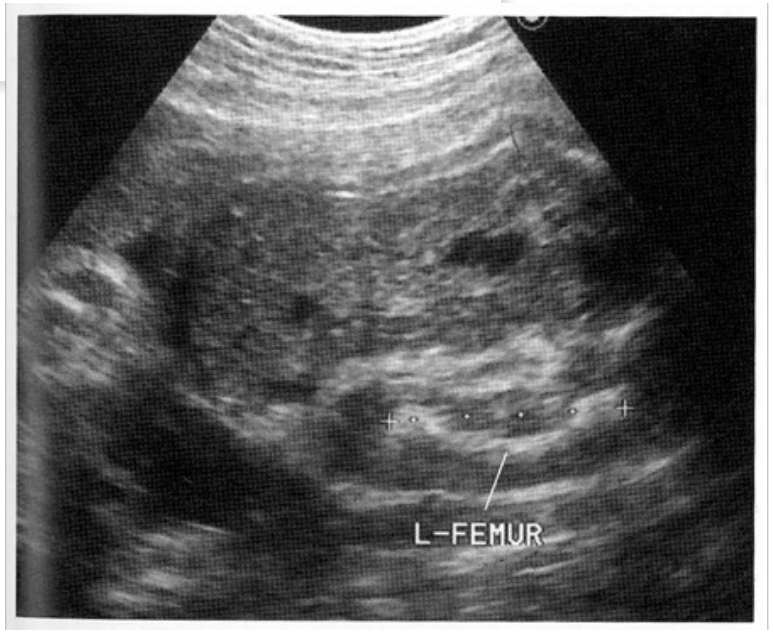
Skeletal development and abnormalities



long bone (26th)



long bone and cartilage (37th)



Thanatophoric dysplasia type 1(31th)
short and curved femur, bilateral enlarged epiphysis

As ossification progresses, only the bone surface is visualized and not the entire bone. It should be taken not to include the cartilage surface echoes at the epiphysis when measuring femoral length.

If there is shortening or kyphosis of the long bones, congenital bone disease such as osteogenesis imperfecta should be considered.

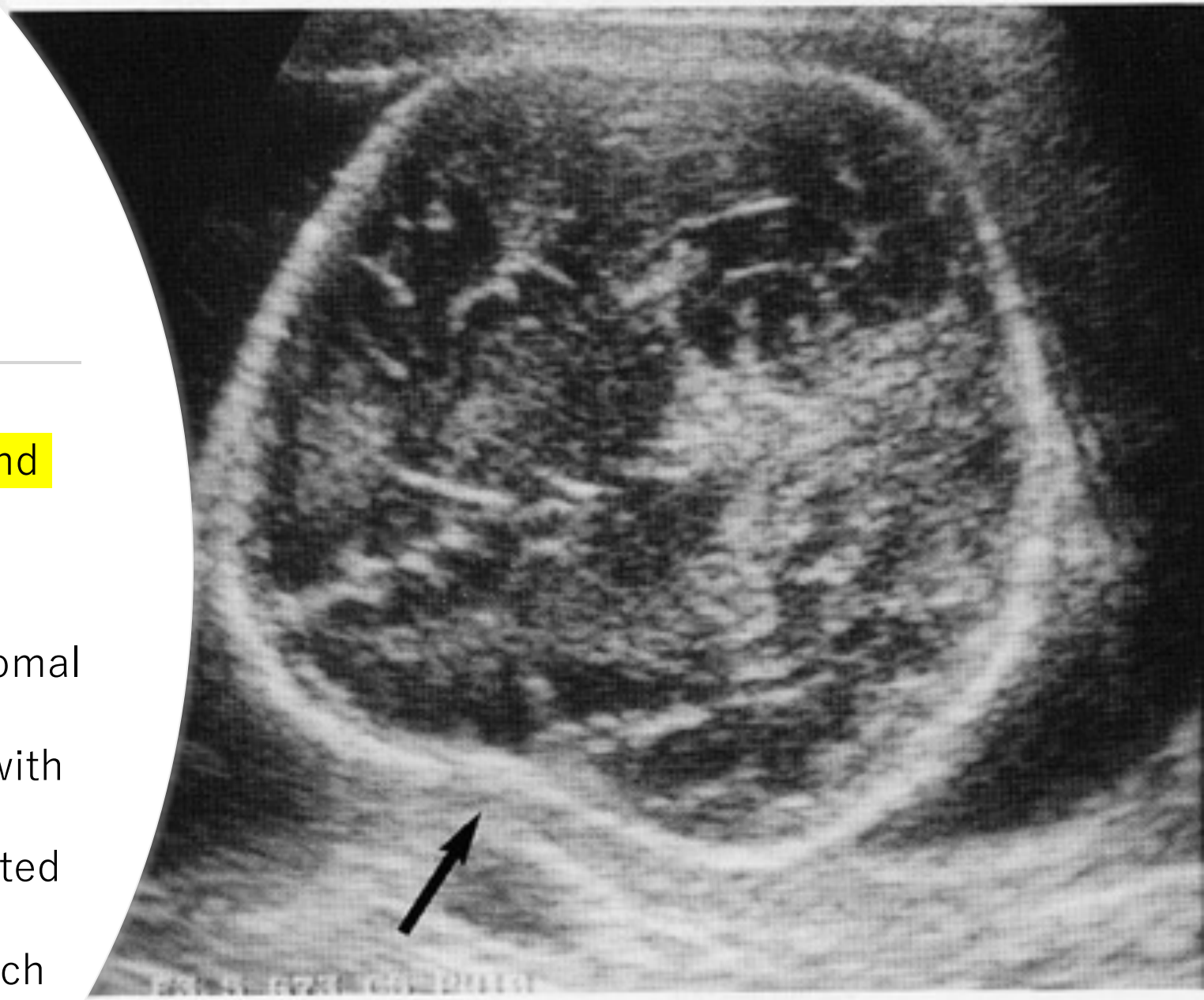
Hypophosphatasia

- Ossification failure

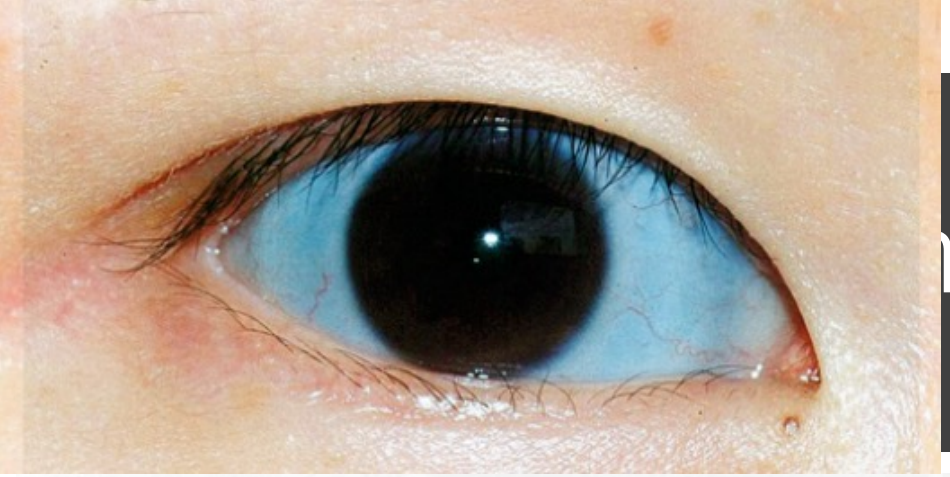
  → hypophosphatasia: skull is thin and easily deformed.

  → It is characterized by low serum alkaline phosphatase (ALP) level.

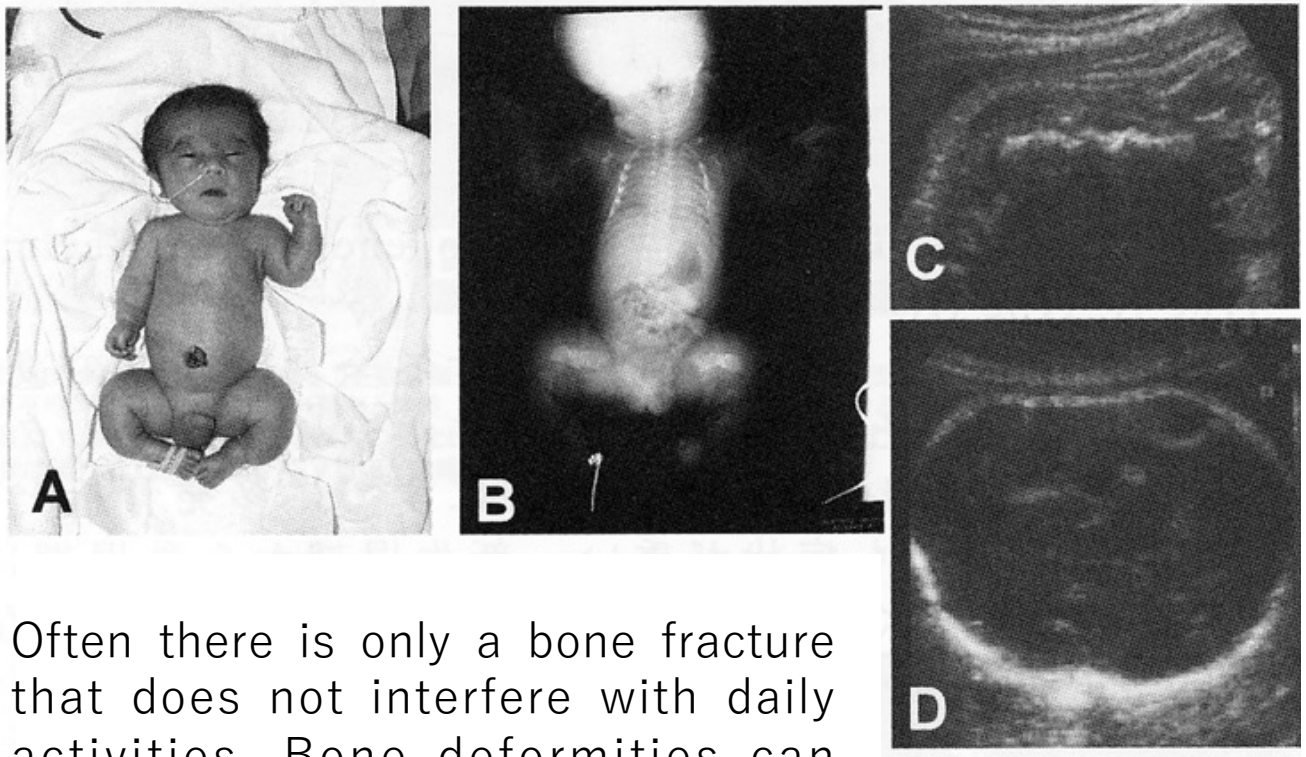
  → Most severe patients have autosomal recessive inheritance.
- Bone dysplasia is often associated with lung hypoplasia.
- Prognosis is poor if they are associated with lung hypoplasia.
- Enzyme replacement therapy, in which ALP is replenished in the body by venous injection, is effective.



33th gestational week



nperfecta congenita type 2・3

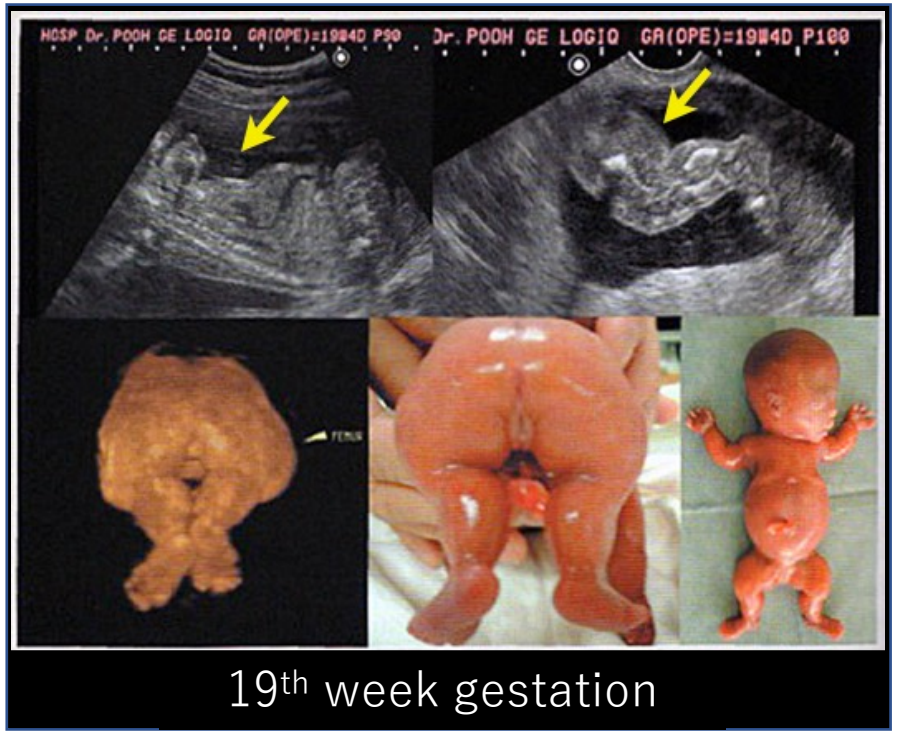


Often there is only a bone fracture that does not interfere with daily activities. Bone deformities can cause varying degrees of gait disturbance.

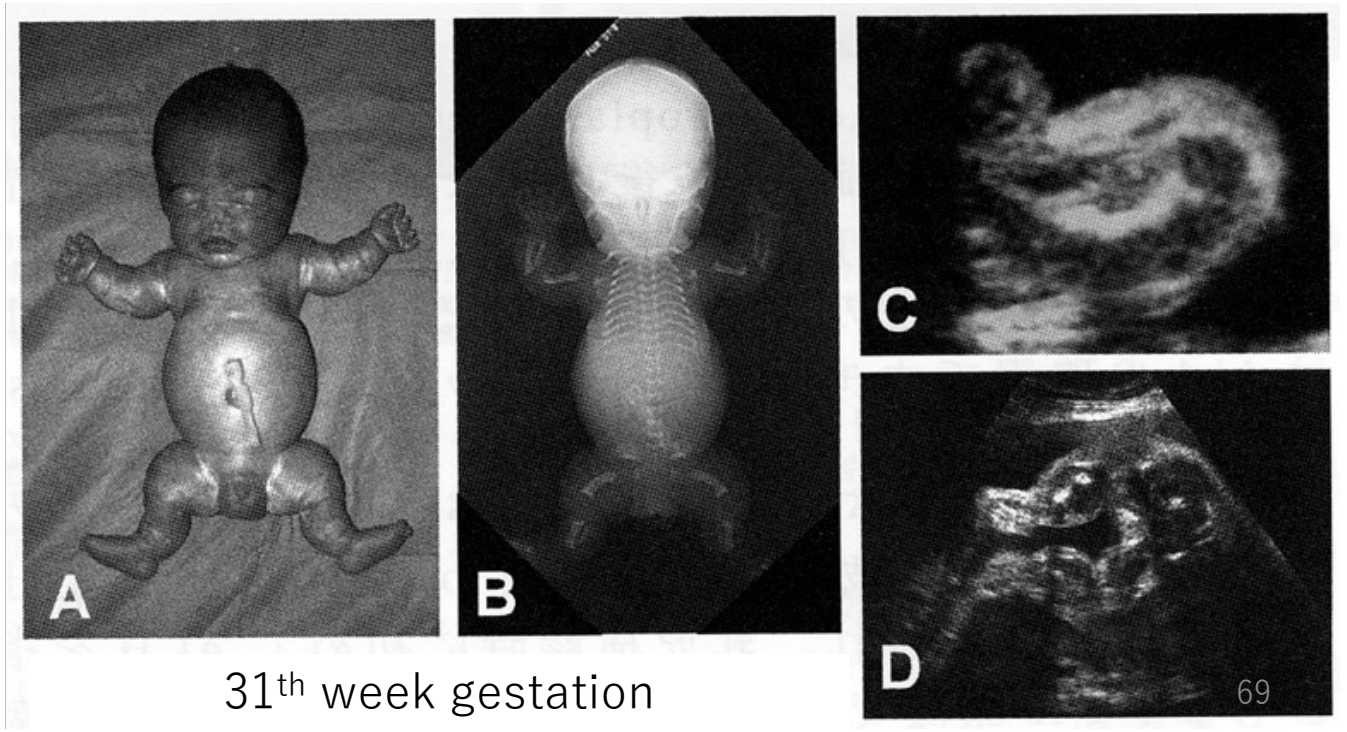
- genetic mutation of type I collagen
- **Type 2**: dead around the birth because of lung hypoplasia
- **Type 3**: Severe and often requires a wheelchair
- **Type 1, 4**: milder than others
- **symptoms**: easily fractured, blue sclera, hearing loss
- Wide, short long bones with multiple fractures, and marked deformity
- Bell-shaped thoracic hypoplasia due to multiple fractures of the ribs.
- Skull is membranous and soft due to osteogenesis imperfecta.

Thanatophoric dysplasia

- Caused by point mutations in the fibroblast growth factor receptor 3 (FGFR3) gene resulting in chondrocyte abnormalities.
- The femur is deformed like an old telephone receiver, the thorax is small, and the head is large in relation to the trunk. The abdomen is distended and the limbs are always extended. Polyhydramnion is caused by thorax dysplasia. Respiratory failure.



19th week gestation



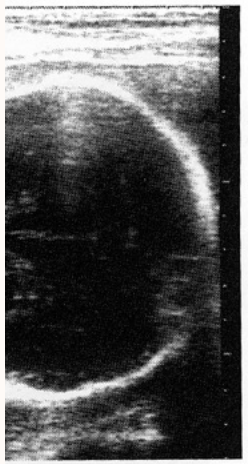
31th week gestation

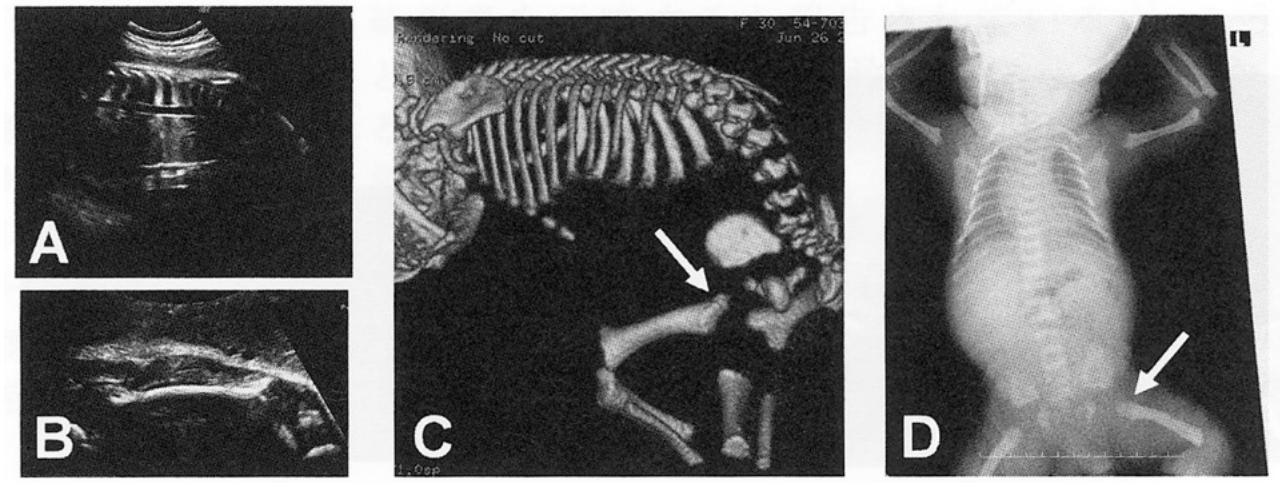
ophia foetalis (37th week)

mutation in FGFR3 resulted in depressed cartilage growth.

c facial features (relatively large cerebral cranium,
d depressed nasal root).

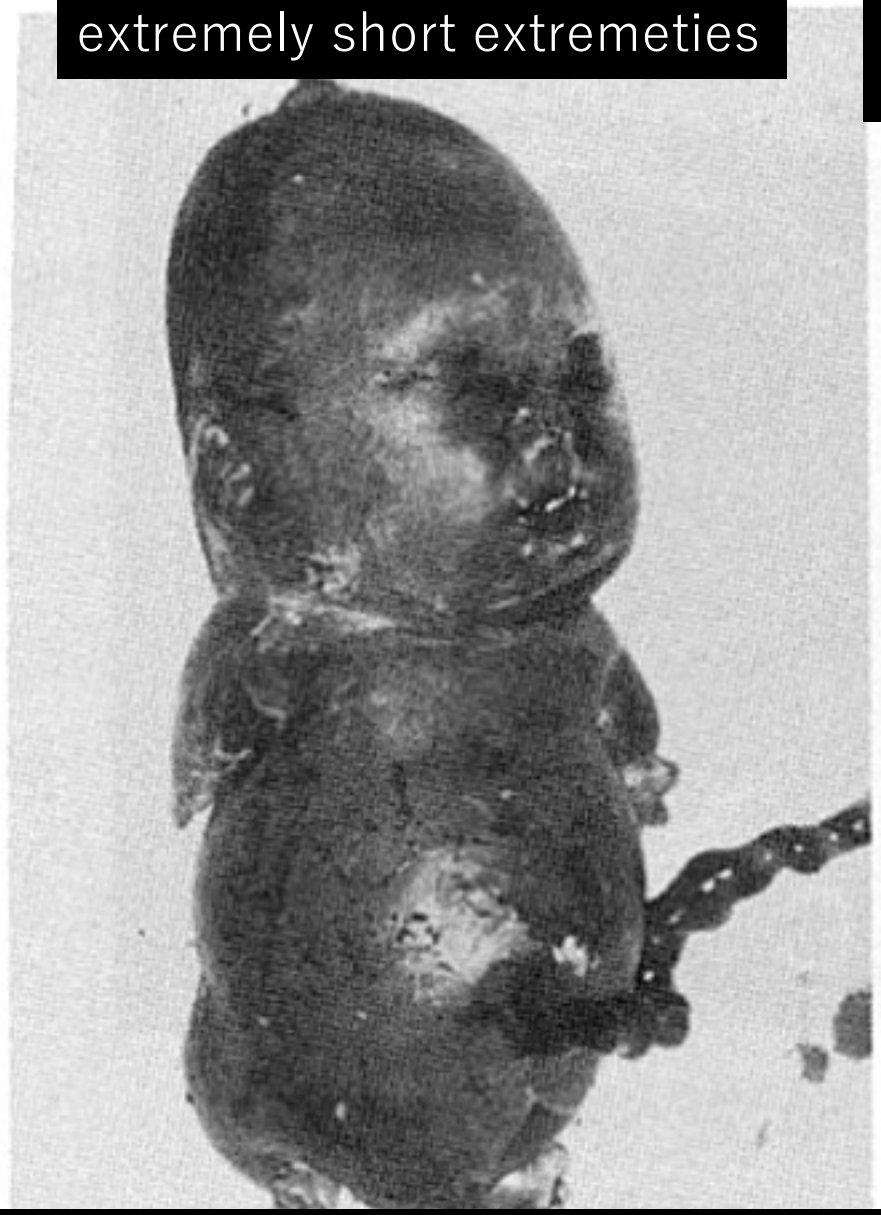
onography are weak.(B) Prognosis is good.

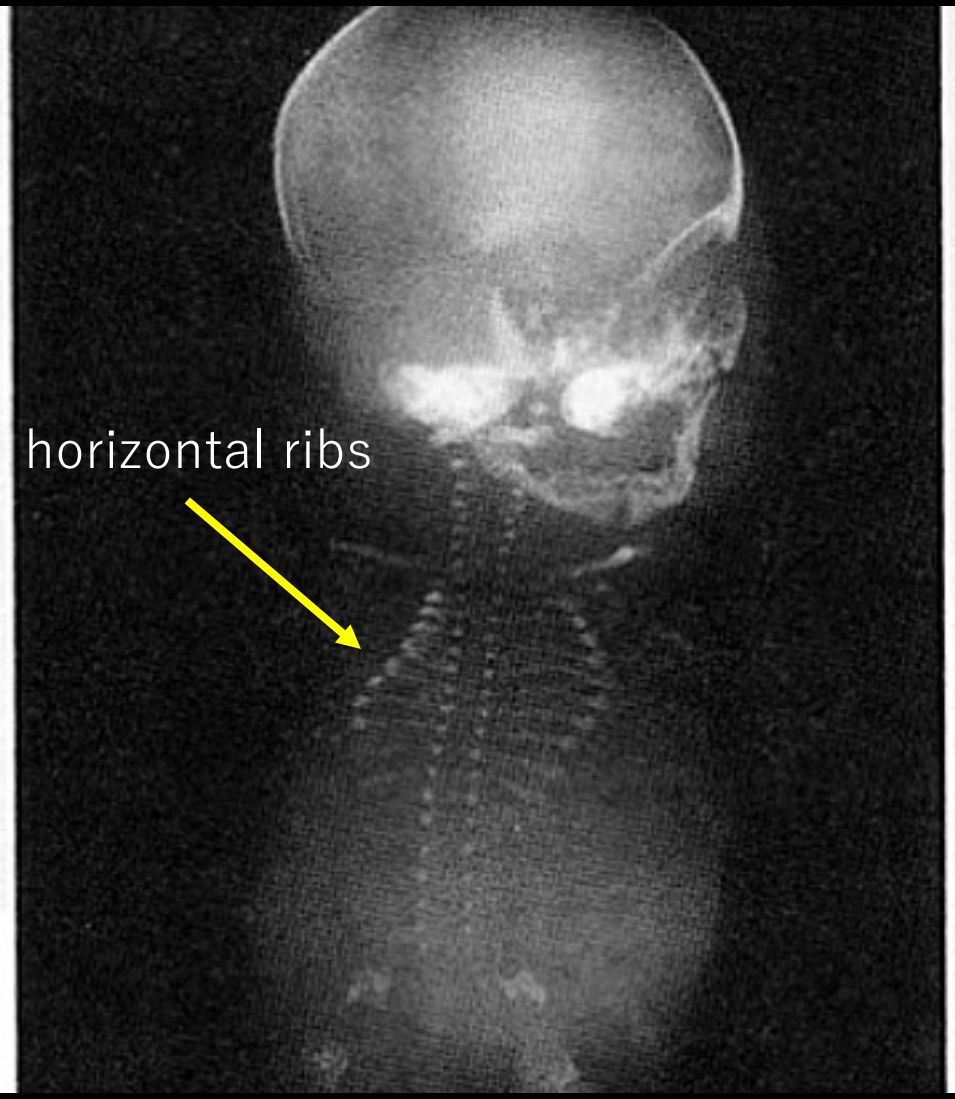


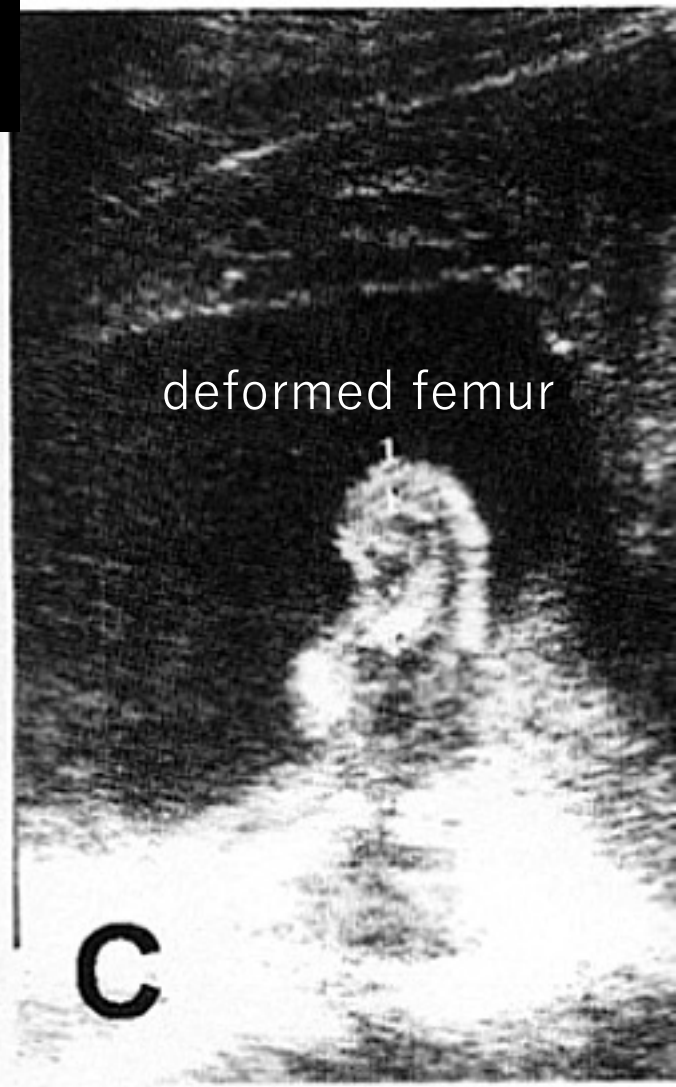


A:軽度の胸郭低形成 B:大腿骨の軽度の弯曲と短縮 C:cupping D:帯状の透亮像

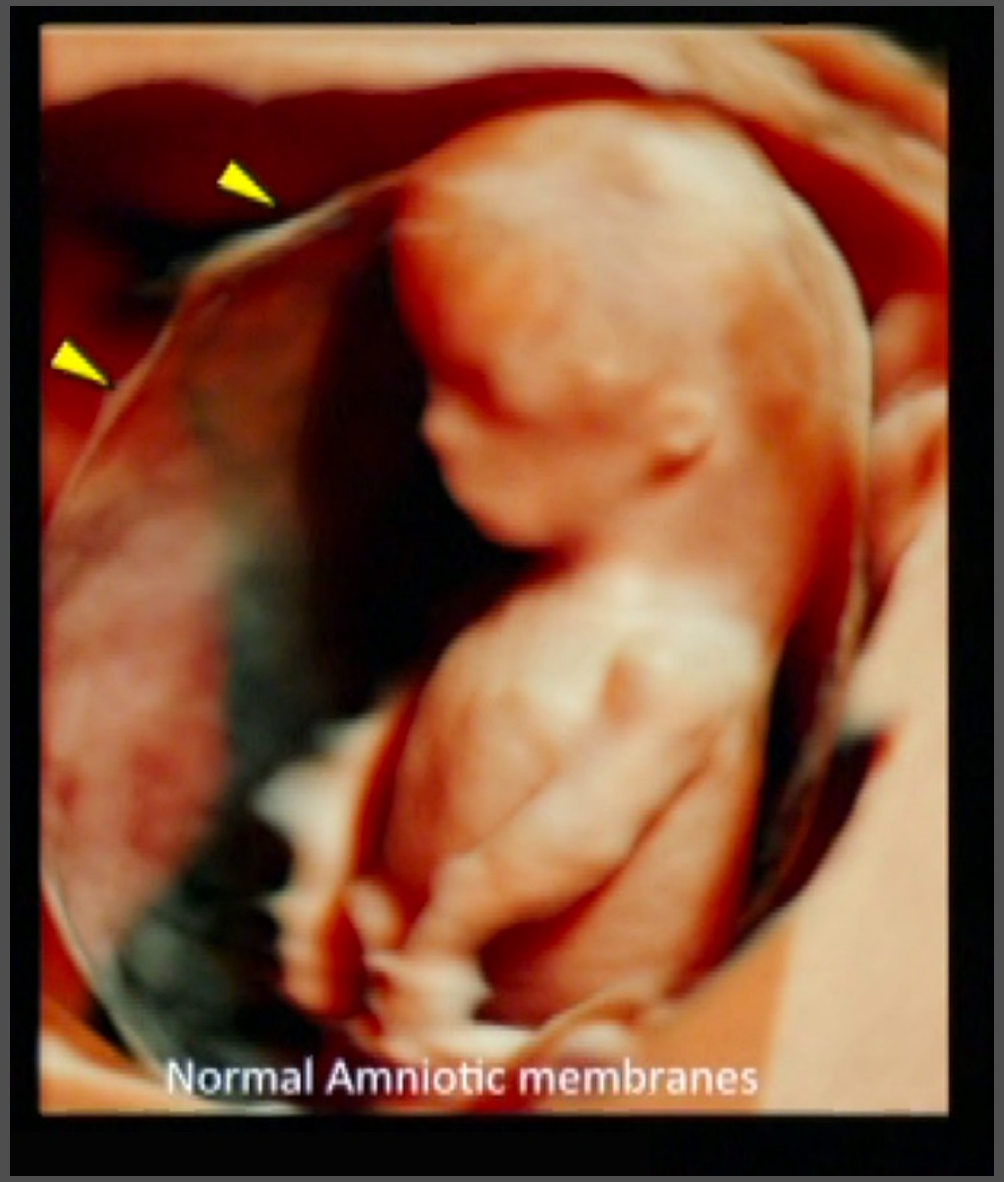
Achondrogenesis

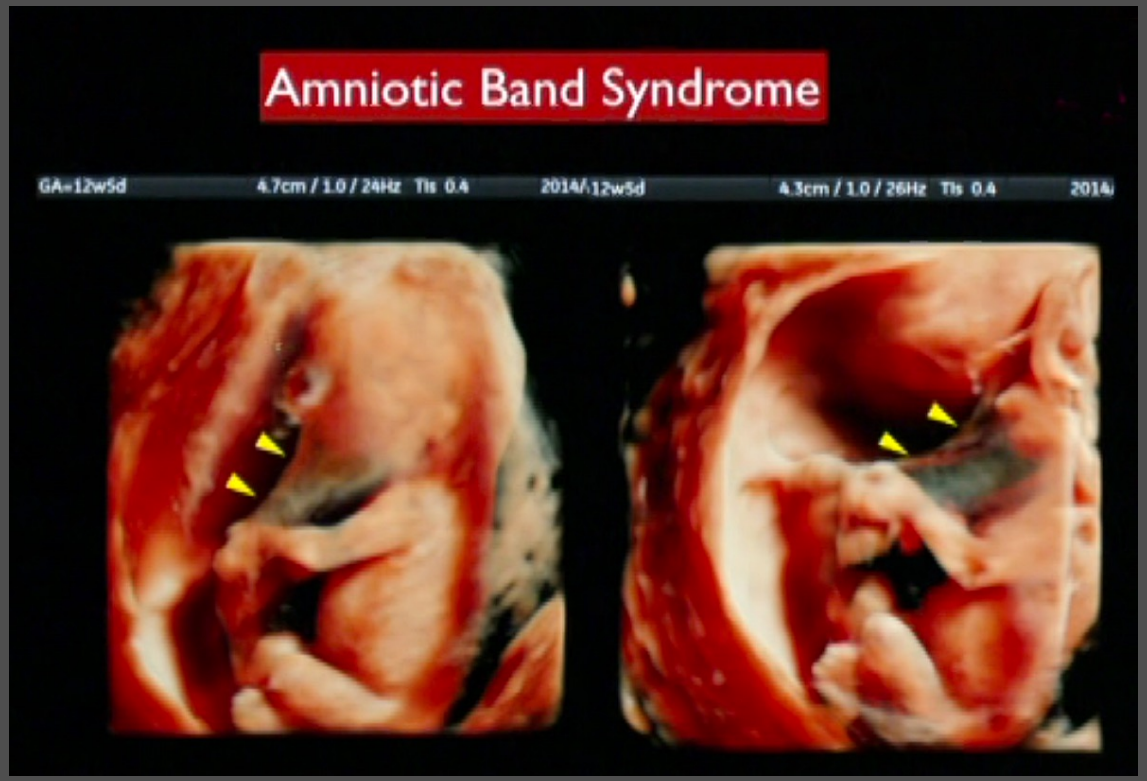




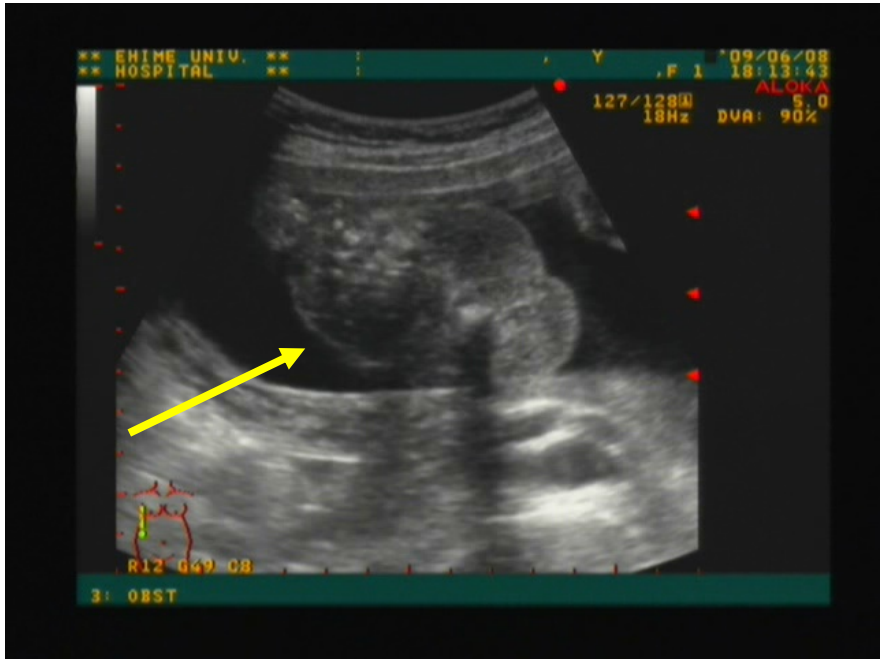


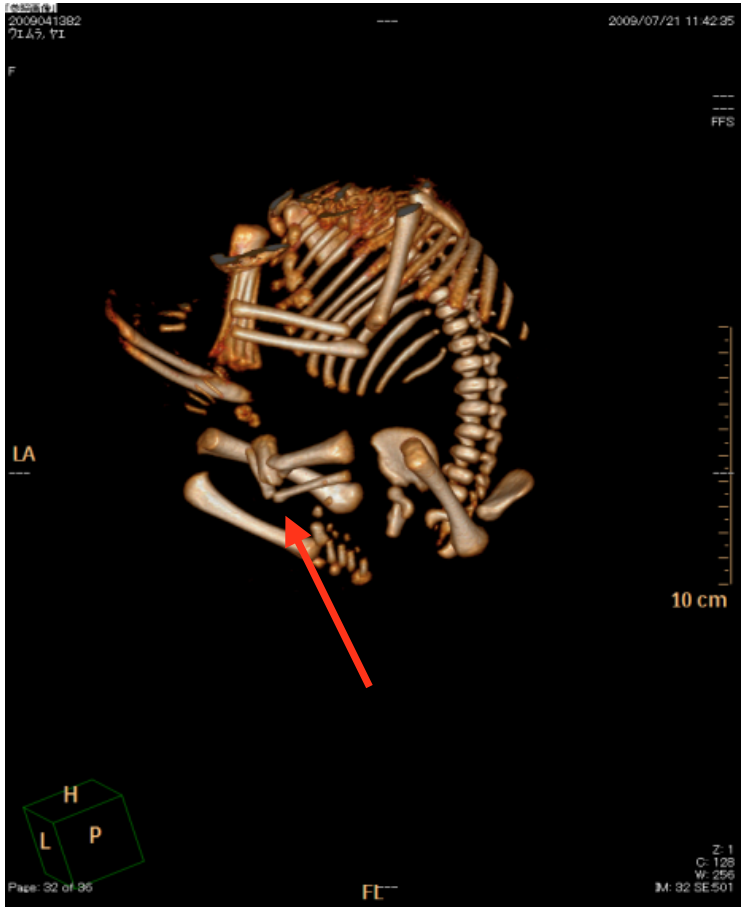
Ossification failure of the spine and pelvis and hypoplasia of the thorax to hypoplasia of the lungs, resulting in the appearance of polyhydramnion. Severe limb shortening and deformity. Large head. Short trunk and abdominal distention. The prognosis is the poorest of all skeletal dysplasia.


Normal Amniotic membranes


Amniotic Band Syndrome
GA=12w5d
4.7cm / 1.0 / 24Hz TIs 0.4
2014/12w5d
4.3cm / 1.0 / 26Hz TIs 0.4
2014

constriction band syndrome
(A part of amniotic band syndrome)





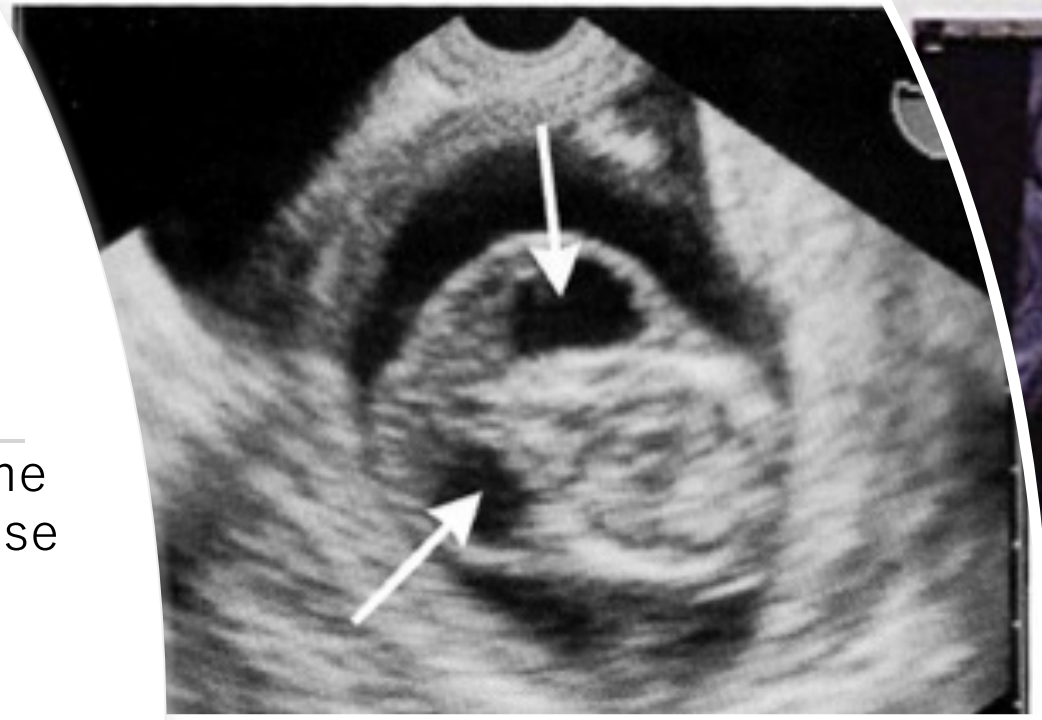
Fetal hydrops

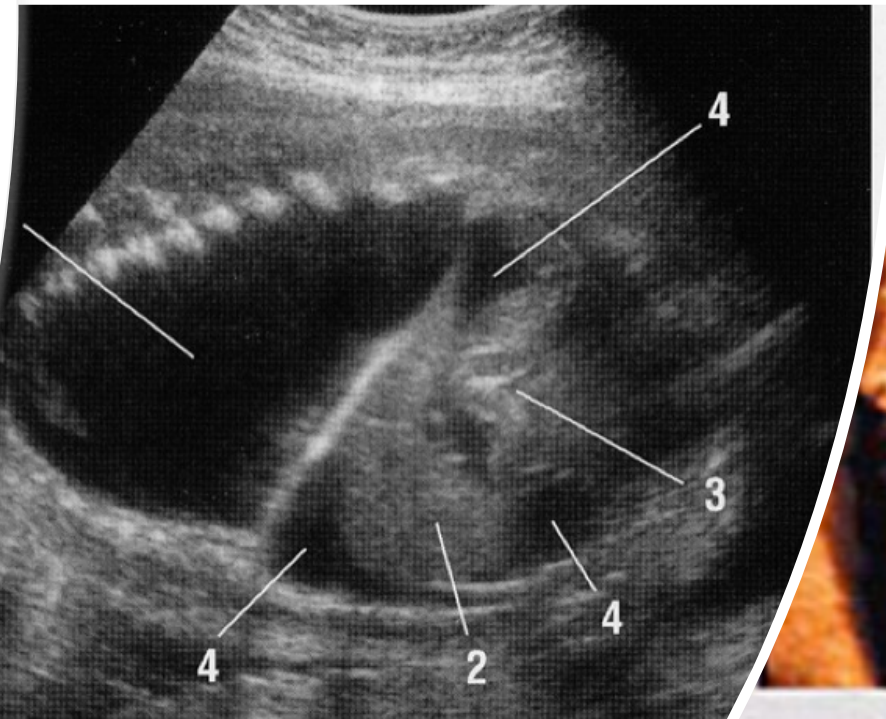
Fetal hydrops

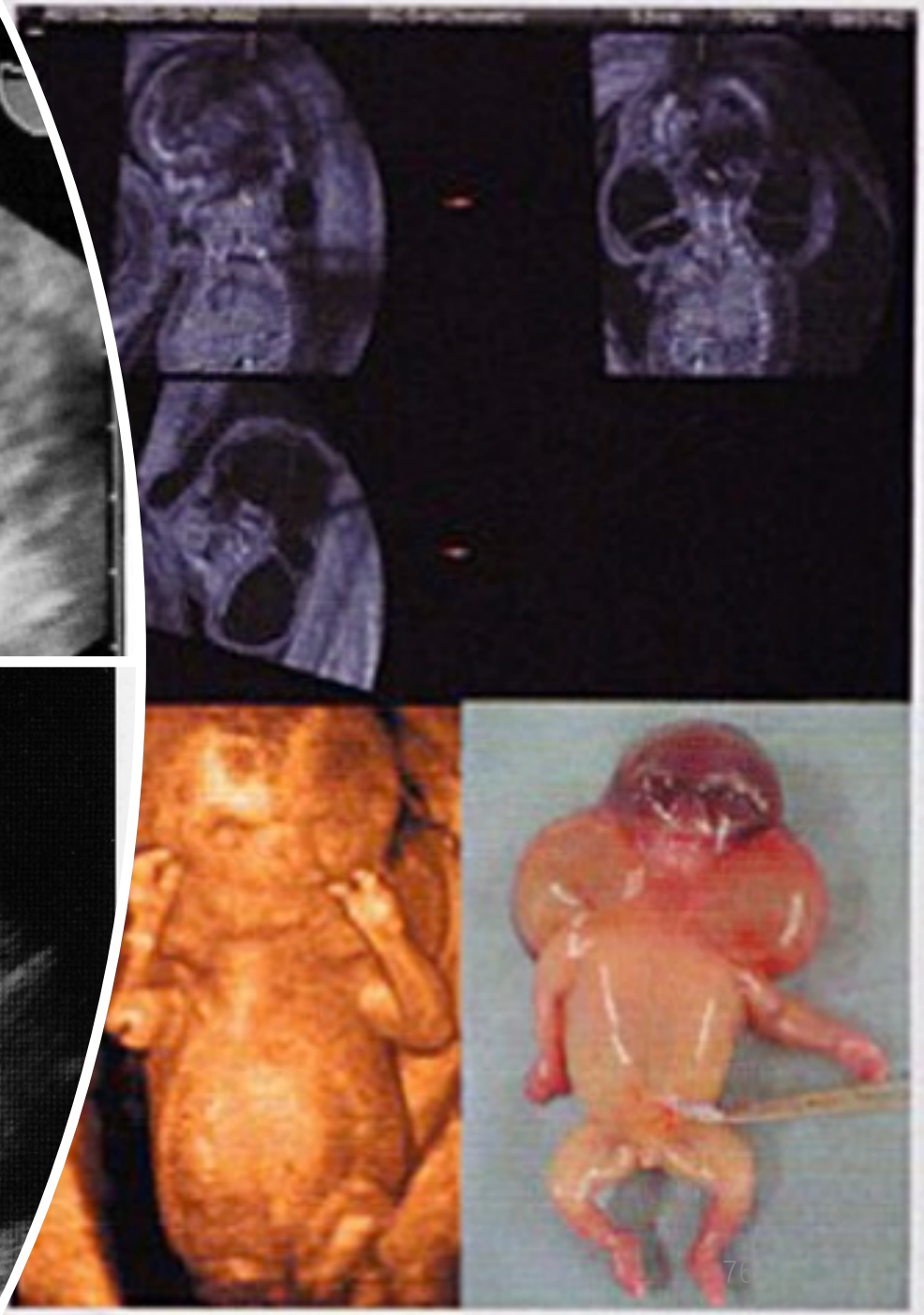
- Fetus has pleural and ascites effusions and subcutaneous edema.
- Sometimes, anemia can be diagnosed by an increased maximum systolic velocity of the middle cerebral artery (MCA). → fetal transfusion is necessary
- **Immune fetal hydrops**: Rh incompatibility is frequent. When fetal hydrops appears, Hb is already less than 5 g/dL. Polyhydramnion and edematous placenta are observed.
- **Non-immune fetal hydrops**: 90% of fetal hydrops. (Causes) Chromosomal abnormalities (45XO, etc.), intrauterine infection (parvovirus B19, cytomegalovirus), TTTS, cardiac malformation, fetal arrhythmia, CCAM (Congenital cystic adenomatoid malformation), diaphragmatic hernia. 40% of patients are found to have morphological abnormalities. Mortality rate is 70%.

Fetal hydrops

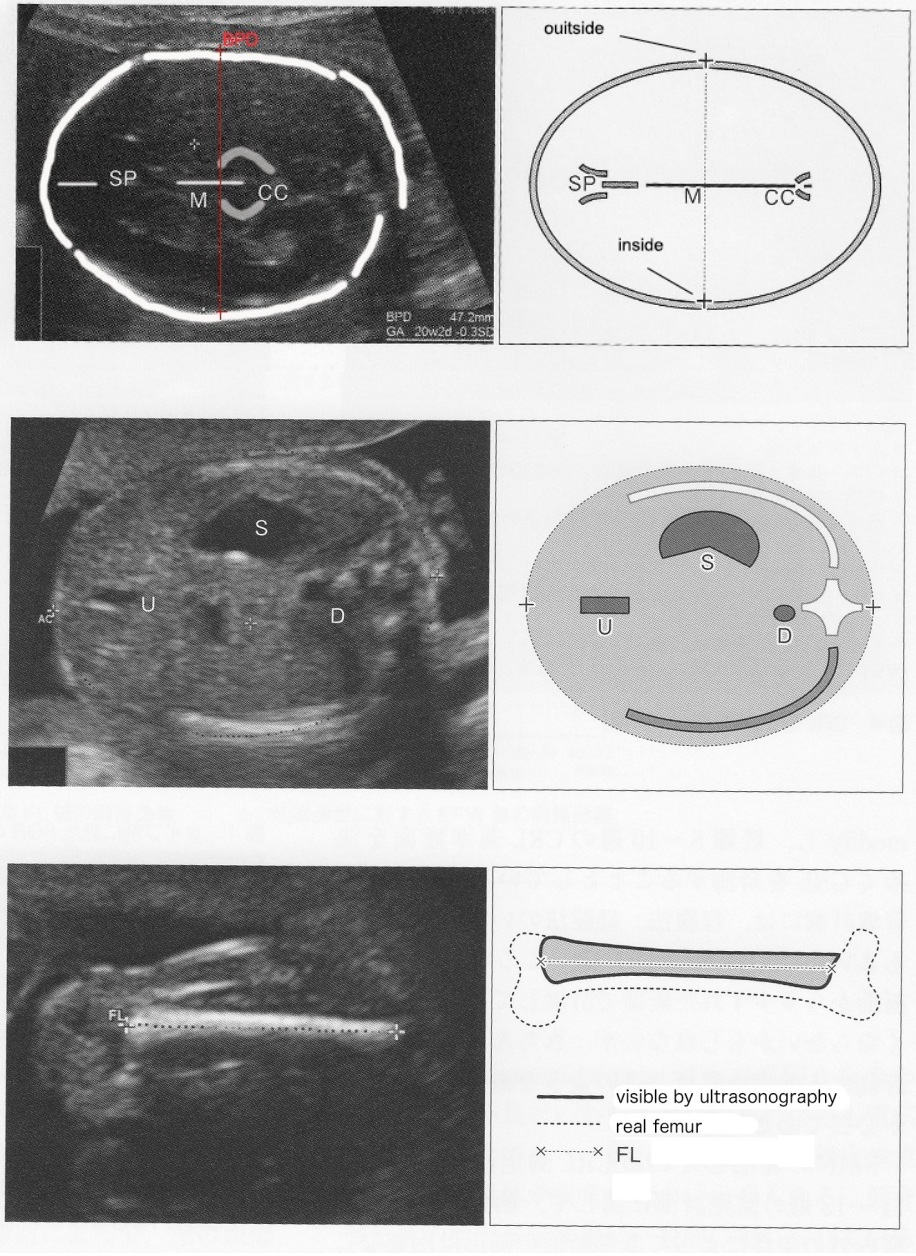
- **Cause**: XO, 18+, 13+. The prognosis is poor for those with large cysts or fetal hydrops.
- **cystic hygroma**: Malformation of the lymphatic system in the neck, axilla, thorax, and lower extremities. It may resolve spontaneously during pregnancy. Often associated with chromosomal abnormalities (21+, 45X)
- The prognosis is also poor in early pregnancy.







Fetal Measurement



CRL （Crown Rump Length） 8th week+0～12th week+0
BPD （Biparietal diameter） 12th week+0～16th week+0

FGR

Low birth weight: less than 2,500 g, very low birth weight: less than 1,500 g, very low birth weight: less than 1,000 g.

Susceptible to complications such as intracranial hemorrhage, RDS, and infection.

Causes: **Smoking** symmetrical FGR (placental dysfunction due to vasoconstriction caused by nicotine and CO, cellular damage caused by cyanide), **Alcohol** (aldehyde, a metabolite of ethanol, etc., impairs organ differentiation.), alcohol (alcohol passes through the placenta for long periods of time and impairs fetal cell proliferation, etc.), **HDP** asymmetrical FGR(impaired blood flow)

BPD : The midline is bilateral symmetry and the septum pellucidum and quadrigeminal cistern are depicted.