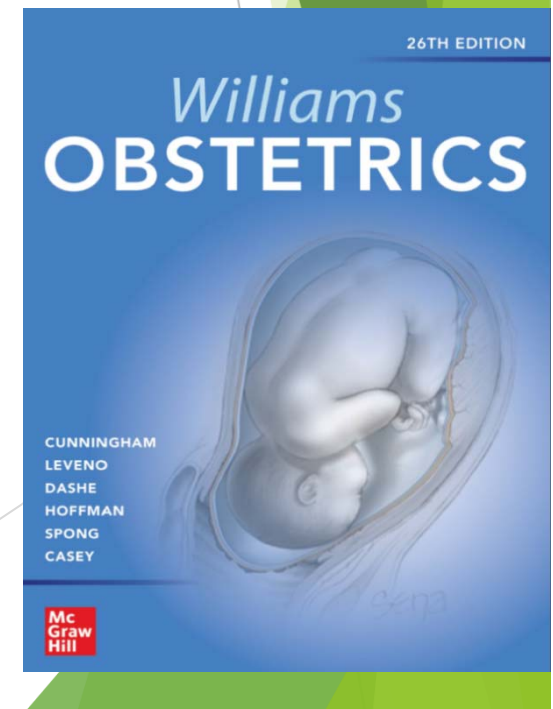


# CHAPTER 15

## Normal and Abnormal Fetal Anatomy

Presenter: R3 劉子榕



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## Standard Ultrasound

### Head, face, and neck

- Midline falx
- Cavum septum pellucidum
- Lateral ventricles
- Choroid plexus
- Cerebellum
- Cisterna magna
- Upper lip
- Nuchal skinfold measurement, 15–20 weeks

### Thorax and heart

- Situs
- Heart rate (M-mode)
- Four-chamber view of the heart
- Left ventricular outflow tract
- Right ventricular outflow tract
- 3-vessel view, if feasible
- 3-vessel trachea view, if feasible

## Detailed Ultrasound, Additional Components

### Head, face, and neck

- Cranial integrity and shape<sup>a</sup>
- Brain parenchyma<sup>a</sup>
- Lateral ventricle wall/lining and contour
- Third ventricle
- Fourth ventricle
- Corpus callosum
- Cerebellar vermis<sup>a</sup> and lobes
- Transverse cerebellar diameter
- Nasal bone measurement, 15–22 weeks<sup>a</sup>
- Profile<sup>a</sup>
- Coronal view of lenses, nose<sup>a</sup>, lips
- Orbits with measurement
- Maxilla<sup>a</sup>, mandible<sup>a</sup>, palate, tongue
- Ear position, size
- Neck<sup>a</sup>

### Thorax and heart

- Interventricular septum
- Superior/inferior venae cavae<sup>a</sup>
- Aortic arch<sup>a</sup>
- Ductal arch
- 3-vessel view<sup>a</sup>
- 3-vessel and trachea view<sup>a</sup>
- Lungs<sup>a</sup>
- Diaphragm integrity<sup>a</sup>
- Ribs

### **Abdomen**

- Stomach: presence, size, and situs
- Kidneys
- Urinary bladder
- Umbilical cord insertion into fetal abdomen
- Umbilical cord vessel number

### **Spine**

- Cervical, thoracic, lumbar, and sacral spine

### **Extremities (presence only)**

- Arms and legs
- Hands and feet

### **External genitalia**

- When indicated, e.g., multifetal gestation

### **Abdomen**

- Bowel, small and large
- Liver
- Gallbladder
- Spleen
- Renal arteries
- Adrenal glands
- Ventral wall integrity

### **Spine**

- Shape<sup>a</sup>, curvature<sup>a</sup>, conus medullaris
- Integrity of spine and overlying tissue<sup>a</sup>

### **Extremities**

- Architecture, position, number<sup>a</sup>
- Long-bone measurements
- Fingers and toes (number, position)<sup>a</sup>

### **External genitalia**

<sup>a</sup>In addition to all standard anatomy components, these detailed ultrasound components are required by the American Institute of Ultrasound in Medicine for normal cases submitted as part of the detailed ultrasound accreditation process. Modified from the American Institute of Ultrasound in Medicine, 2018, 2019, 2020a.

# BIOMETRY



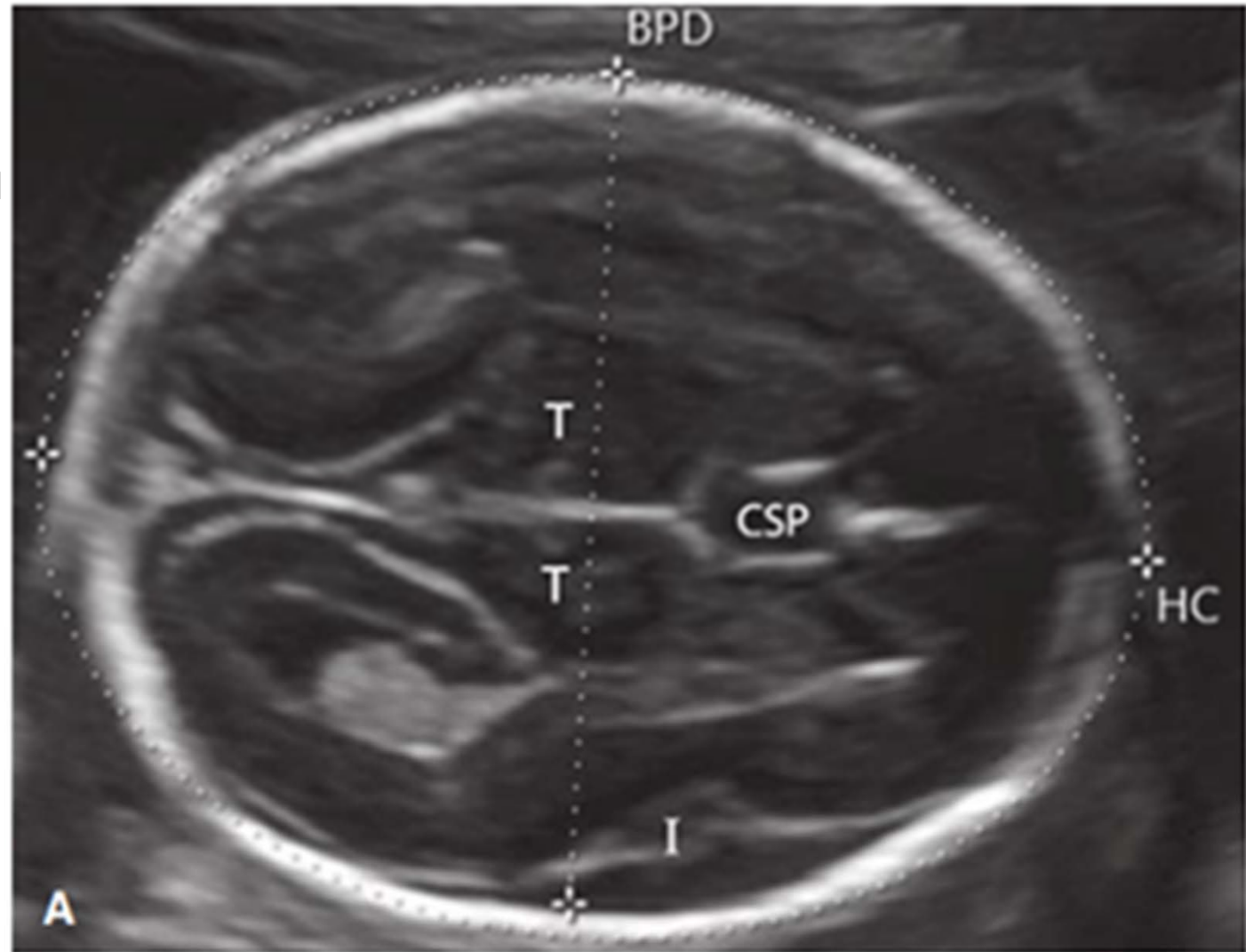
# Crown-rump length (CRL)



**FIGURE 15-1** The crown-rump length measures 61 mm in this 12-week, 4-day fetus.

# Biparietal diameter(BPD)

- ▶ Transthalamus
- ▶ Cerebellum should not be visible
- ▶ Out to in



# Abdominal circumference(AC)

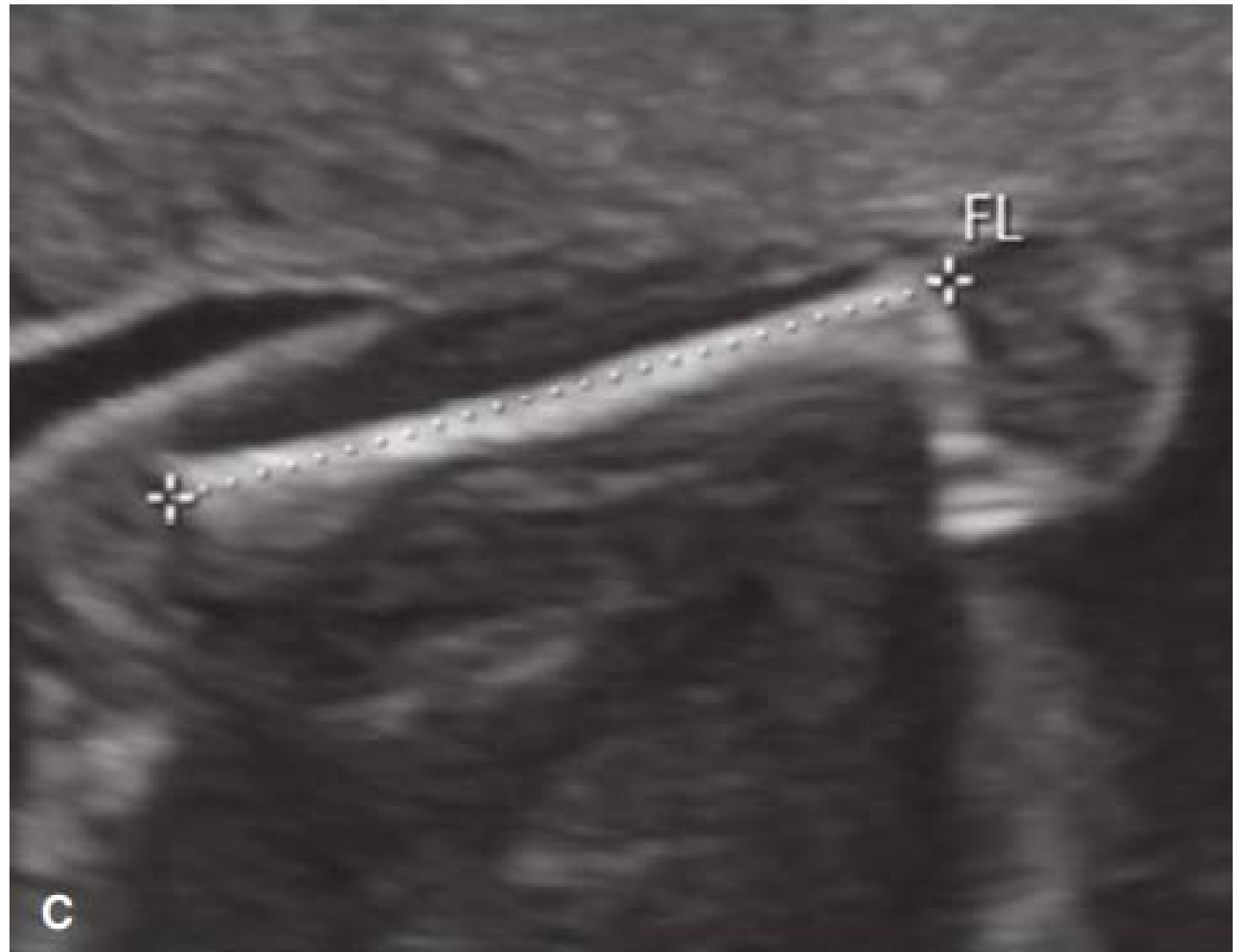
- ▶ J-shaped + portal sinus
- ▶ No more than 1 rib on either side
- ▶ Spine should be visible
- ▶ Kidneys should not be visible





## Femur length(FL)

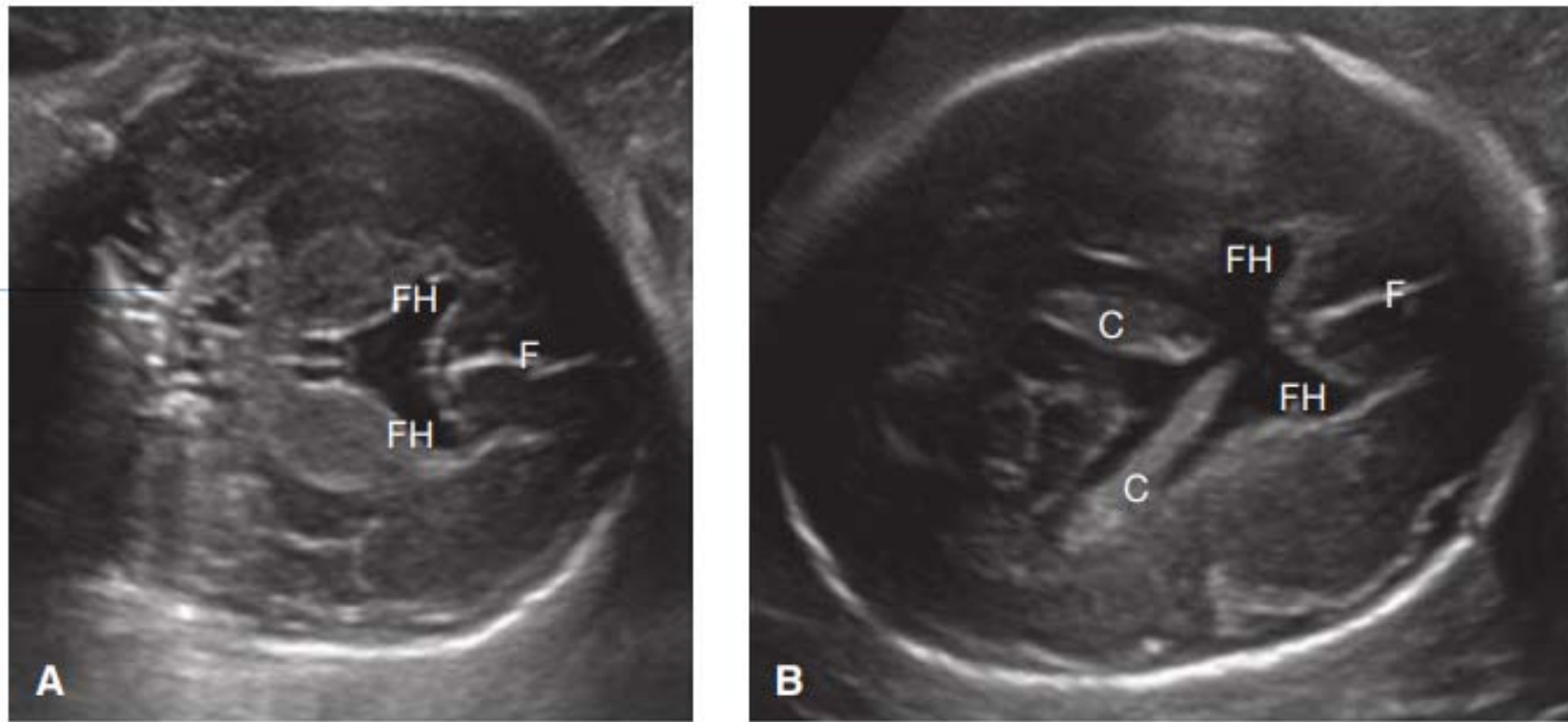
- ▶ Diaphysis to diaphysis
- ▶ FL/AC ratio: 20% to 24%
- ▶ FL/AC ratio Below 18% skeletal dysplasia



# BRAIN AND SPINE

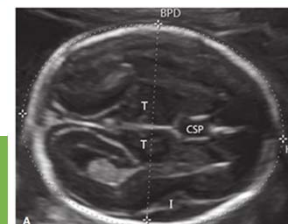


# Transthalamic view

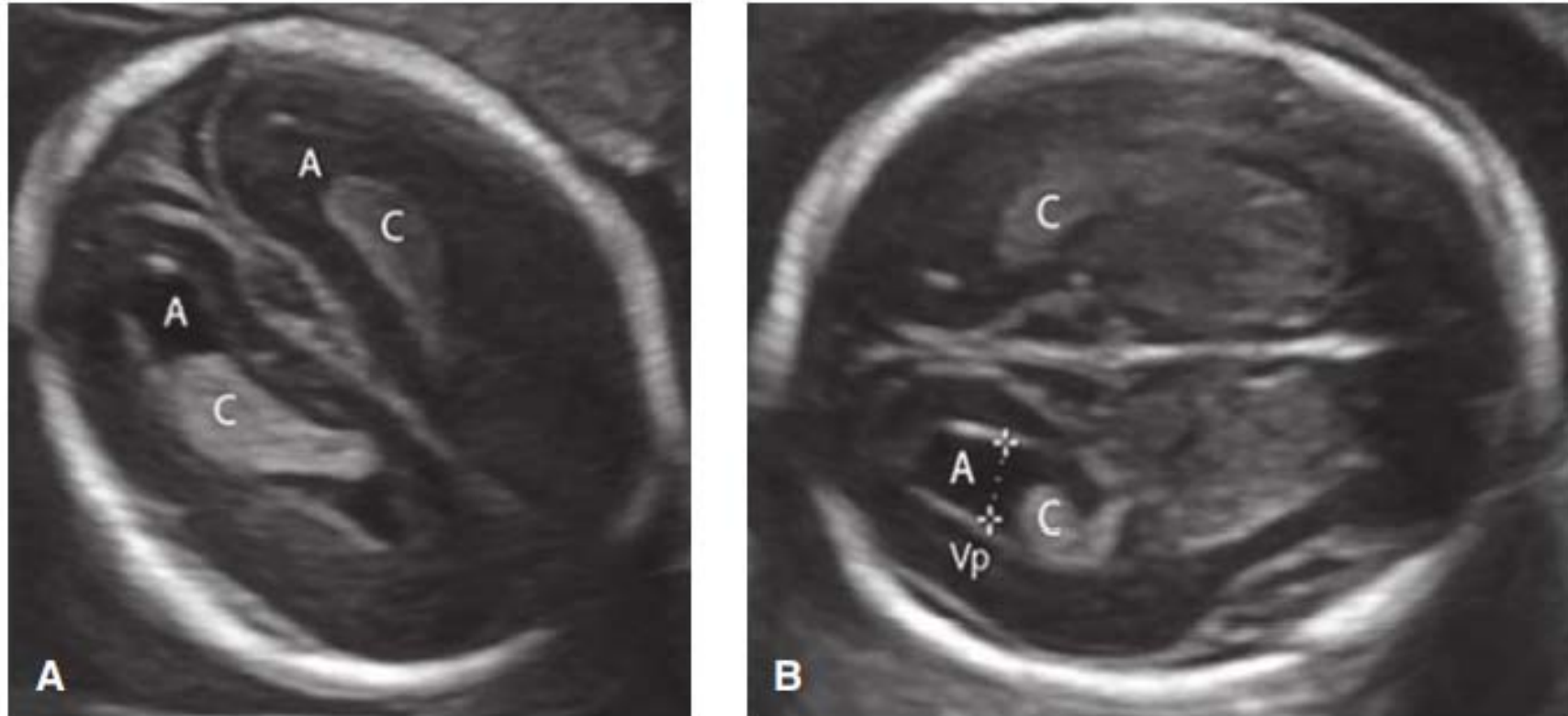


**FIGURE 15-3** Absence of the cavum septum pellucidum, with coronal (A) and transverse (B) images showing communication between the frontal horns (FH) of the lateral ventricles. This may be isolated but can occur in the setting of septo-optic dysplasia or lobar holoprosencephaly. C = choroid plexus; F = falx cerebri.

- 17~37weeks: abnormal CSP indicated a midline brain abnormality
- Septo-optic dysplasia · lobar holoprosencephaly, trisomy 18

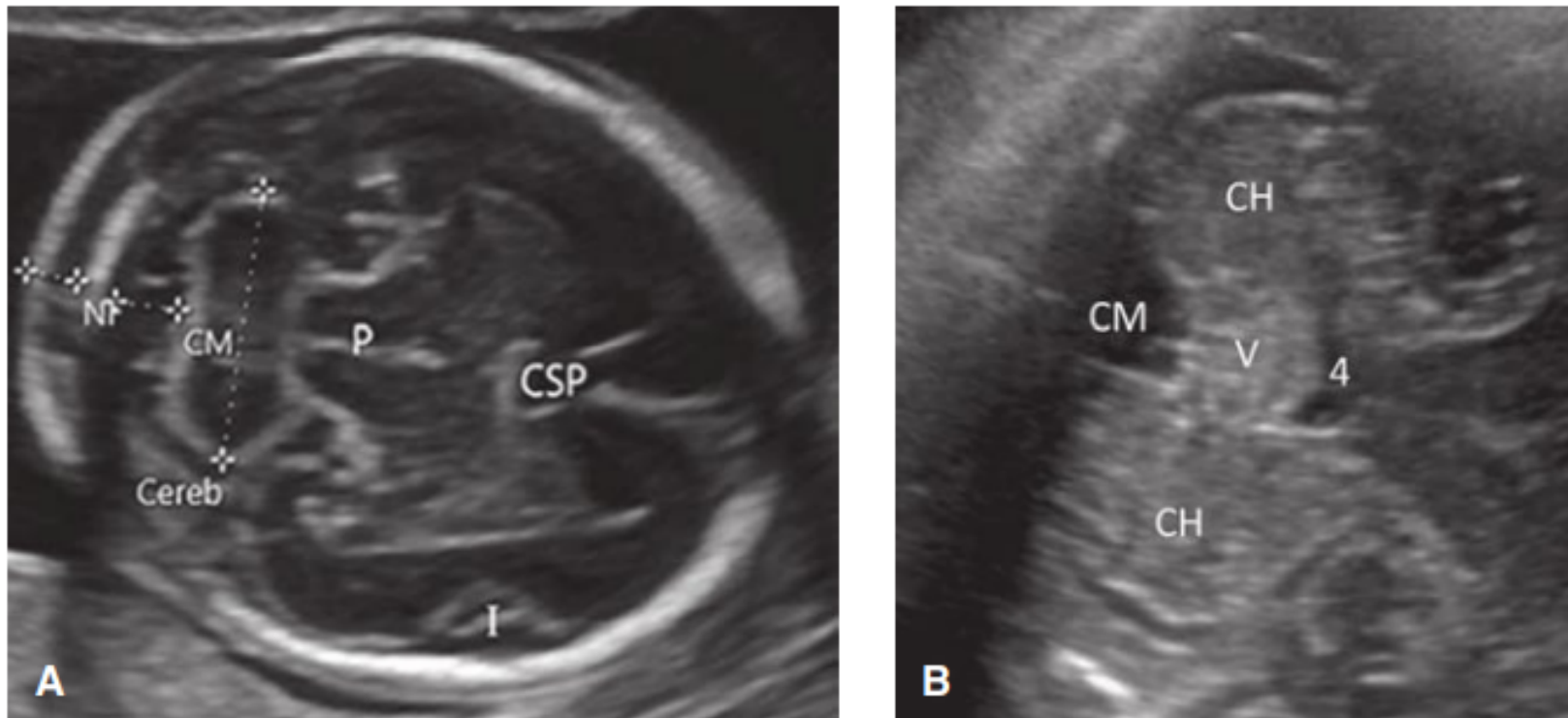


# Transventricular view



**FIGURE 15-4** Transventricular view. **A.** Transverse image of the lateral ventricles, which contain the choroid plexus (C). **B.** The ventricles are measured at the atria (A), the confluence of the temporal and occipital horns. The measurement is normally 5–9 mm. Vp = lateral ventricle. (Reproduced with permission from Rosa Robles, RDMS.)

# Transcerebellar view

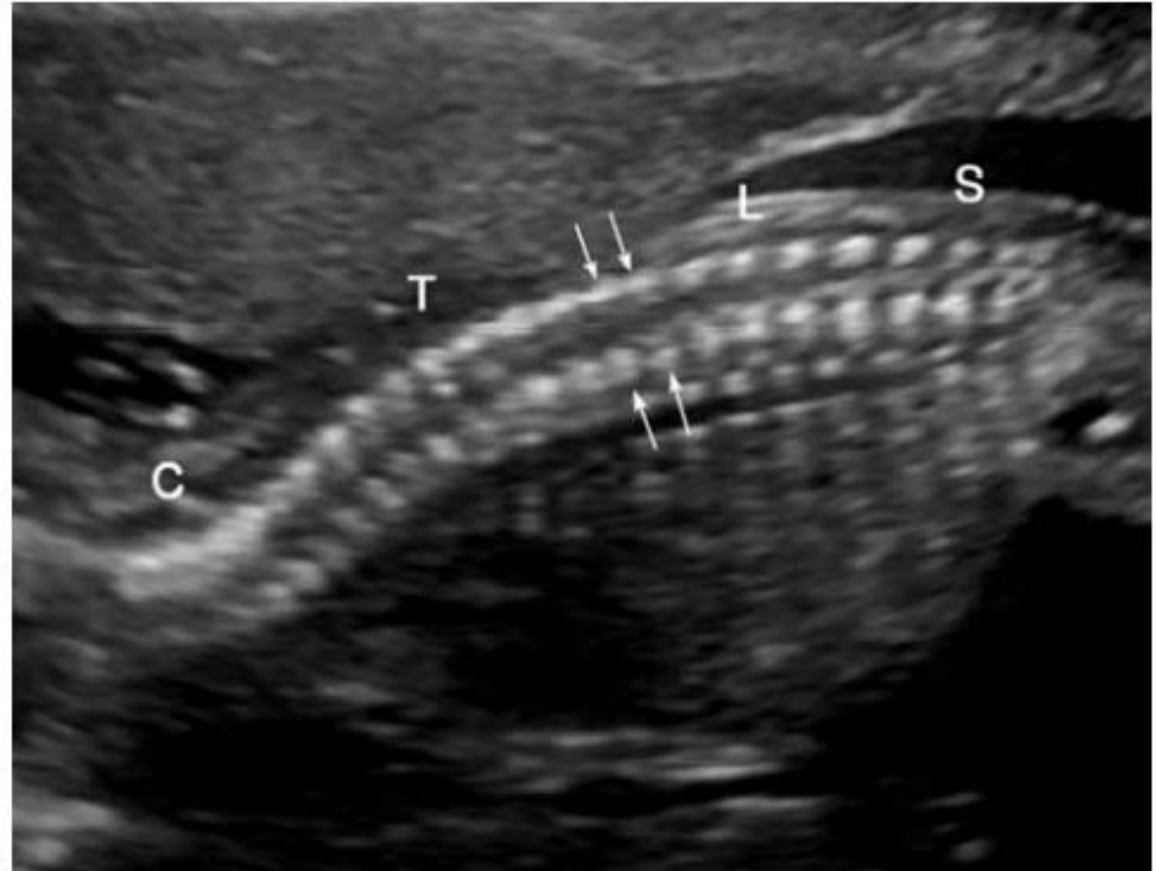
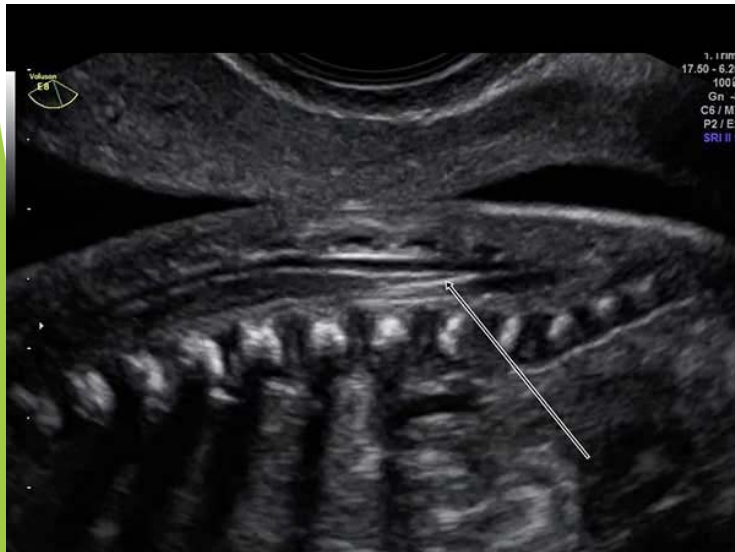


**FIGURE 15-5** Transcerebellar view. **A.** Transverse image of the posterior fossa showing measurement of the cerebellum (*Cereb*), cisterna magna (*CM*), and nuchal skinfold thickness (*NF*). **B.** Third-trimester image depicting the cerebellar hemispheres (*CH*) and cerebellar vermis (*V*). The fourth ventricle (*4*) is anterior to the vermis. *CSP* = cavum septum pellucidum; *I* = insula; *P* = cerebral peduncles.

- 15 and 20 weeks can measure nuchal skinfold thickness >>> Down syndrome
- 15 ~ 22 weeks, cerebellar diameter equivalent to the gestational age in weeks
- Cisterna magna 2 and 10 mm, enlarged absence of all or part of the vermis

# Spine

- Entire sacrum may not be visible **until 21 weeks**



**FIGURE 15-6** Normal fetal spine. This sagittal image depicts the cervical (C), thoracic (T), lumbar (L), and sacral spine (S). Arrows denote the parallel rows of paired posterior ossification centers, which represent the junction of vertebral lamina and pedicles.

# Neural tube Defects



**FIGURE 15-7** Anencephaly/acrania **A.** This transabdominal image at 11 weeks' gestation depicts relatively normal fetal development. **B.** A transvaginal image at 11 weeks demonstrates more clearly the protrusion of a disorganized mass of tissue that resembles a "shower cap." CRL = crown-rump length.

- Anencephaly is an absence of the cranium and telencephalon
- Neural tube closes by the embryonic age of **26 to 28 days**
- Neural-tube defects approximate 0.9 in 1000
- Between 15 and 20 weeks, **Maternal serum alpha-fetoprotein** threshold of 2.5 multiples of the median (MoM) is anticipated for neural tube defects



# Encephalocele

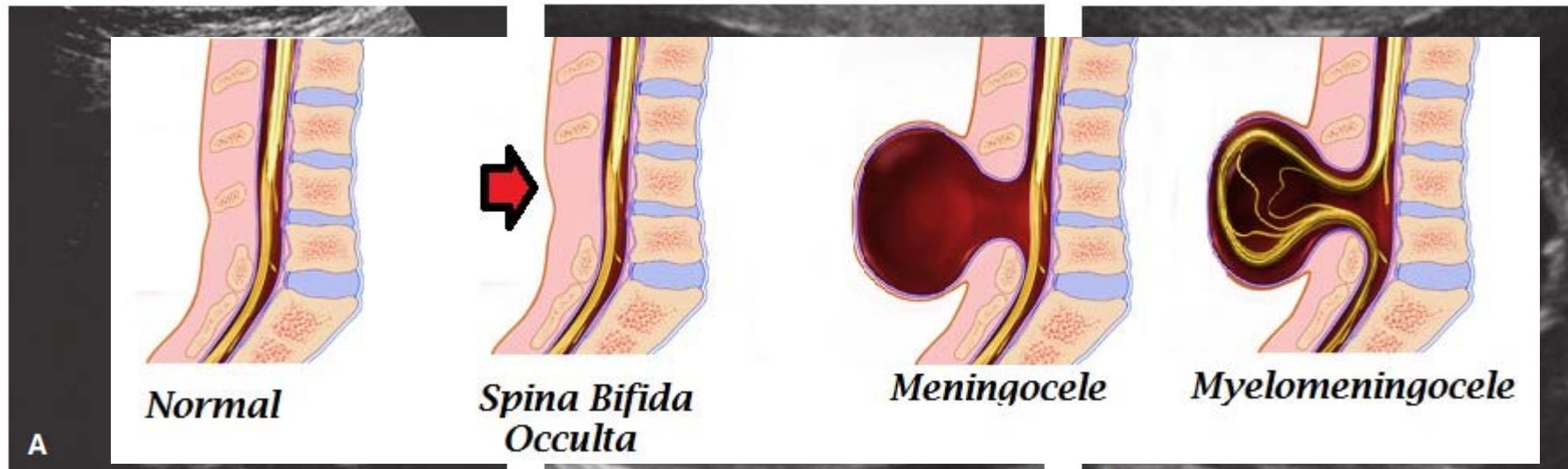


- Autosomal recessive Meckel-Gruber syndrome
- Chiari III malformation

**FIGURE 15-8** Encephalocele. This transverse image depicts a large defect in the occipital region of the cranium (*arrows*) through which meninges and brain tissue have herniated.



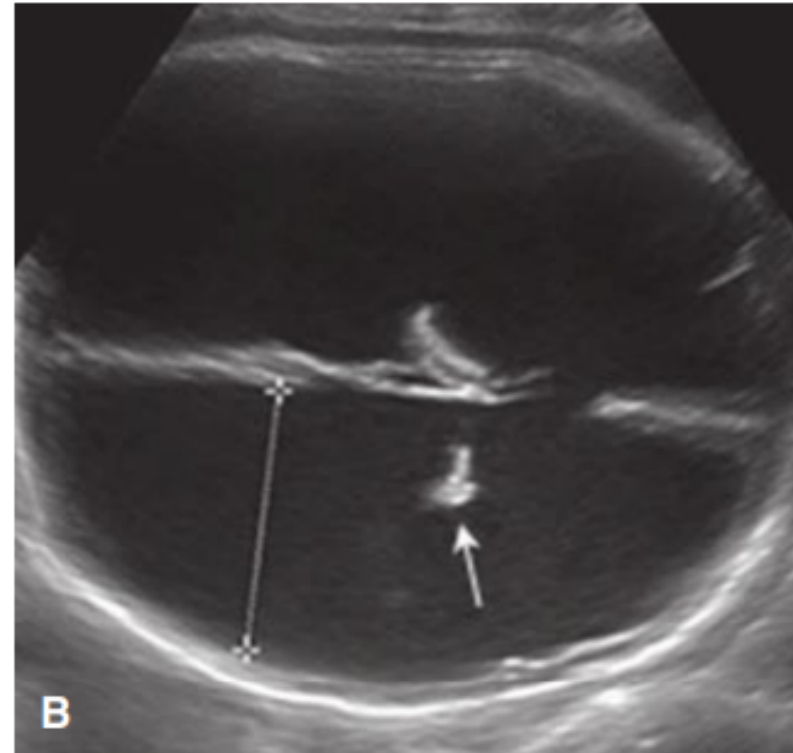
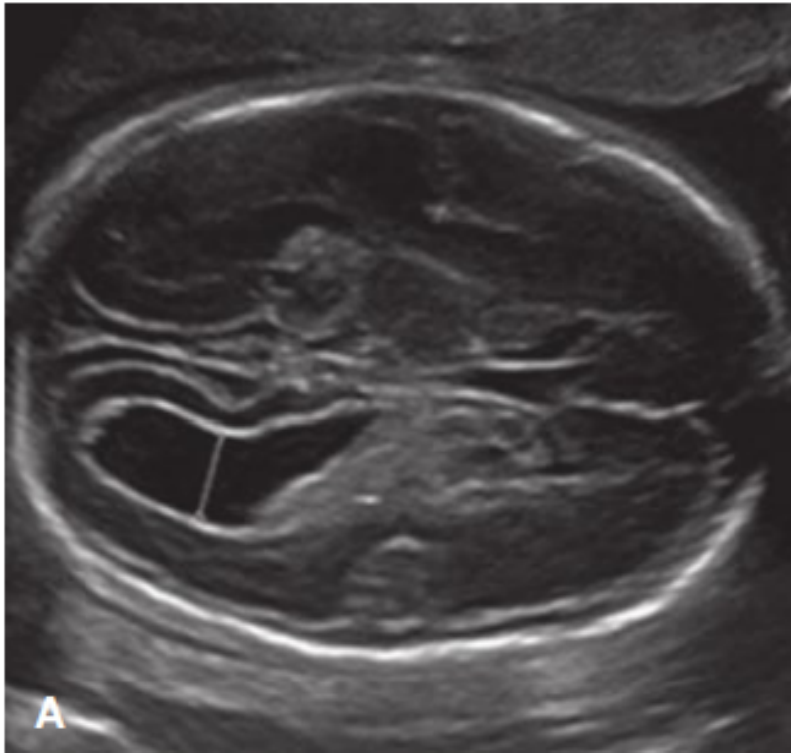
# Neural tube Defects



**FIGURE 15-9** Myelomeningocele. **A.** Sagittal image of a lumbosacral myelomeningocele. Arrowheads indicate nerve roots within the anechoic herniated sac. The overlying skin abruptly stops at the defect (*arrow*). **B.** Transthalamic image demonstrating flattening of the frontal bones (*arrows*)—**the lemon sign**. **C.** Transcerebellar image depicting **the banana sign**, an anterior curvature of the cerebellum (*arrows*) and effacement of the cisterna magna.



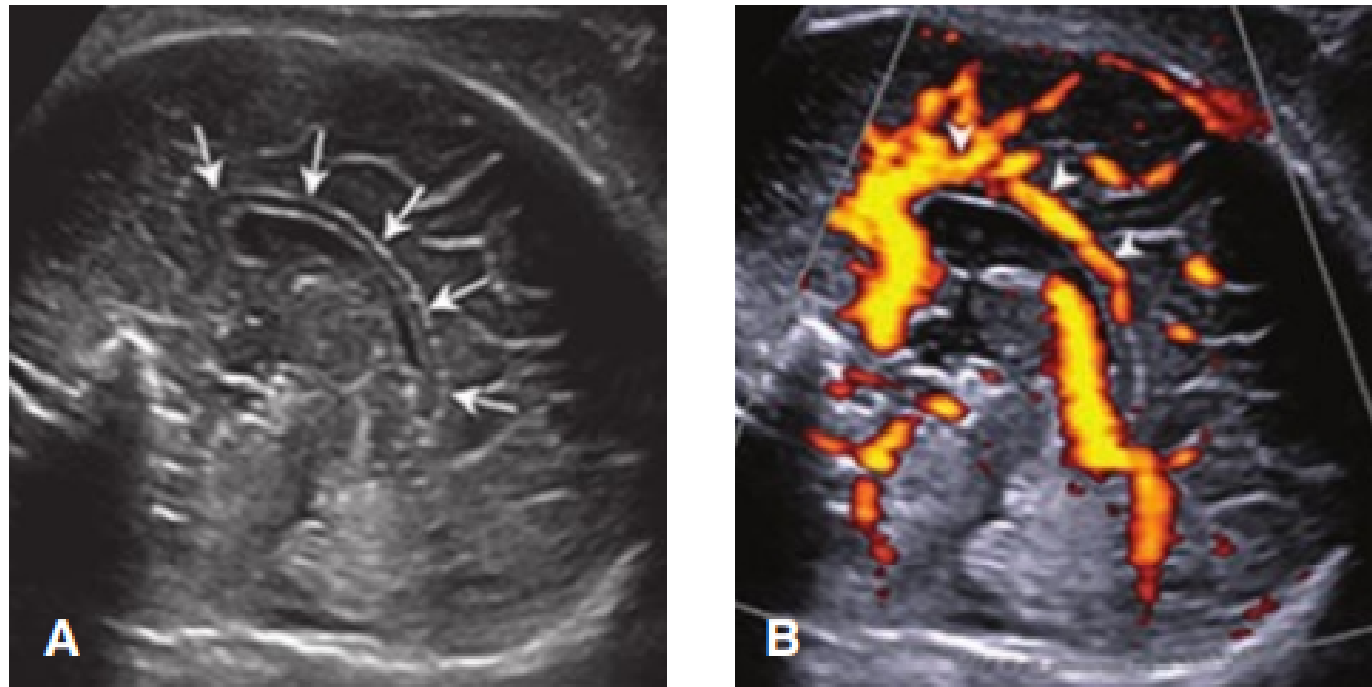
# Ventriculomegaly



**FIGURE 15-10** Ventriculomegaly. **A.** Mild ventriculomegaly. The atria measured 11 mm. No associated abnormality or underlying etiology was identified. **B.** Severe ventriculomegaly. In this fetus with aqueductal stenosis, the atria measured 45 mm. Arrow denotes the dangling choroid plexus.

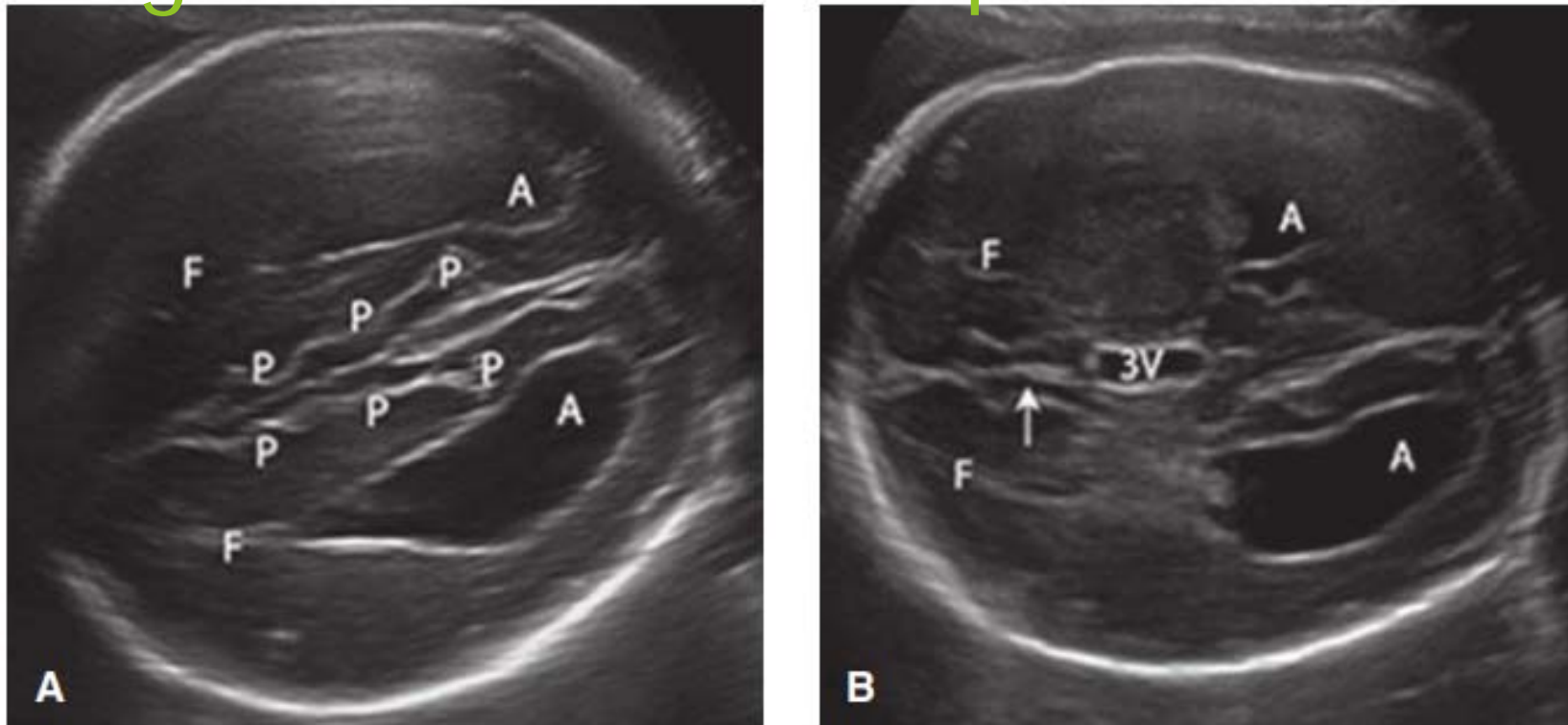
Mild ventriculomegaly: 10 to 12 mm, 90 percent normal  
Moderate, 13 to 15 mm, 75 percent  
Severe >15 mm

# Aggenesis of the Corpus Callosum



**FIGURE 15-11** Normal corpus callosum. **A.** Arrows point to the corpus callosum in this midsagittal image. **B.** Power Doppler image of the pericallosal artery (*arrowheads*).

# Agenesis of the Corpus Callosum

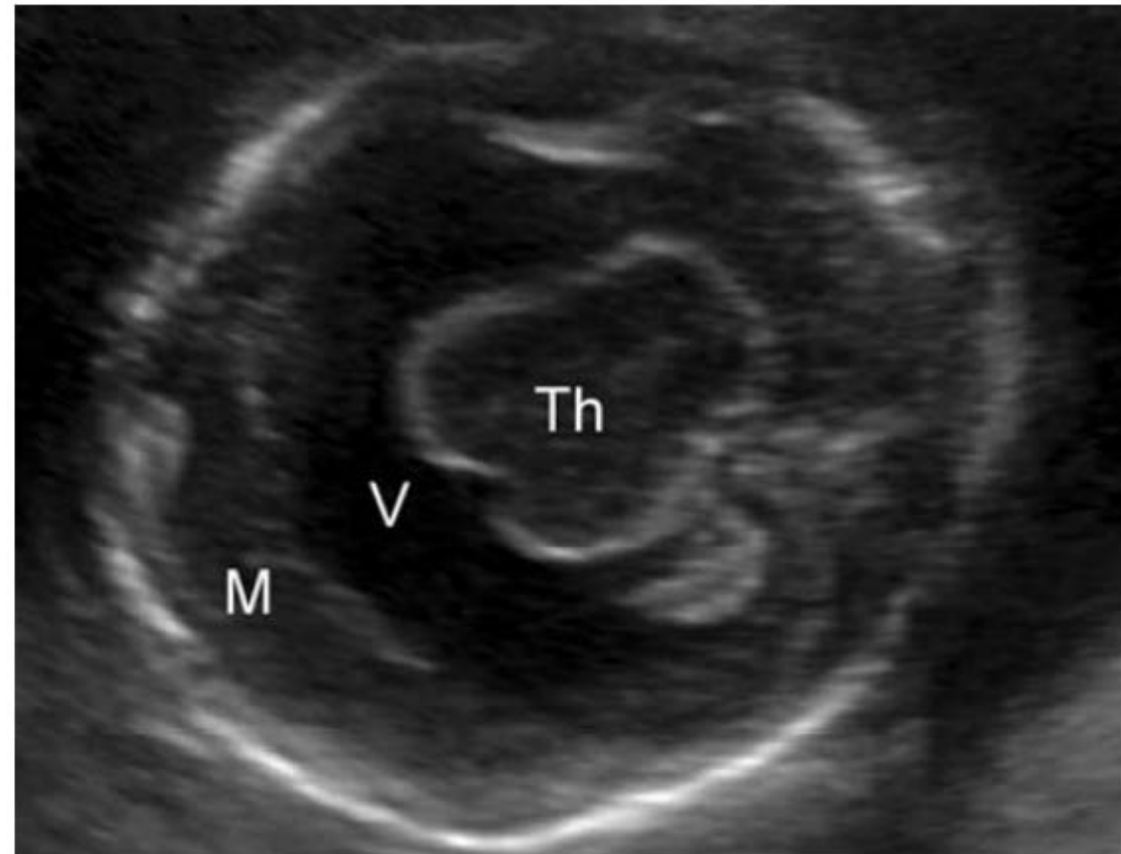
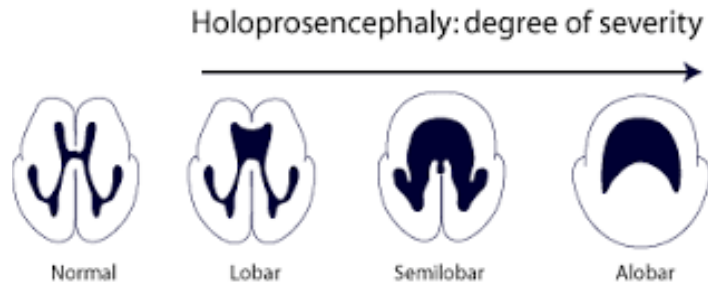


**FIGURE 15-12** Agenesis of the corpus callosum. **A.** This transverse image demonstrates a tear-drop-shaped ventricle. The frontal horns (F) are widely separated, no cavum septum pellucidum is visible, and bundles of Probst (P) line the midline. **B.** There is mild ventriculomegaly, no cavum septum pellucidum is visible (arrow), and the third ventricle (3V) is elevated and enlarged. A = atria.



Associated with other anomalies, aneuploidy, and more than 200 genetic syndromes

# Holoprosencephaly

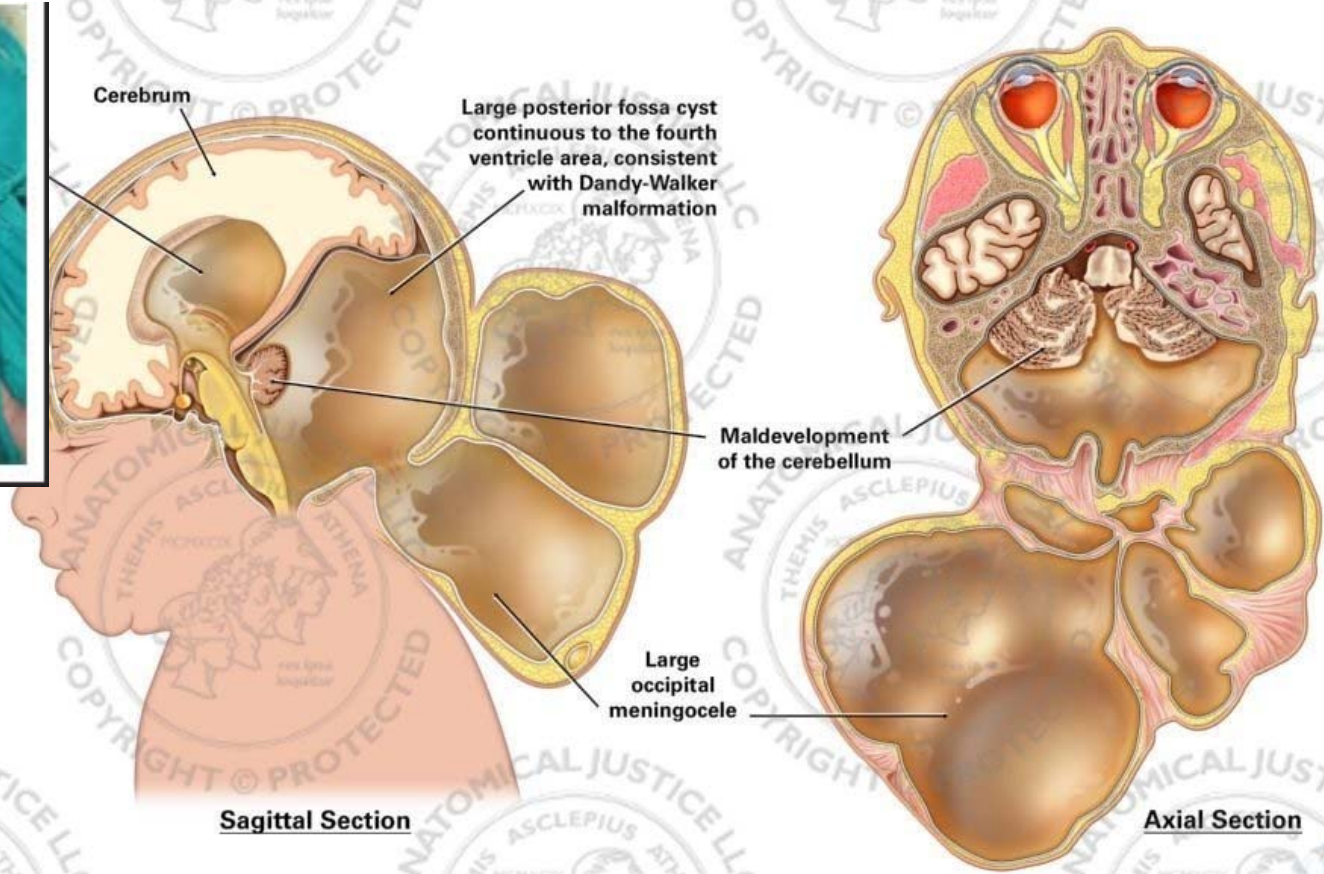


**FIGURE 15-13** Alobar holoprosencephaly. The thalami (*Th*) are fused and encircled by a monoventricle (*V*) with a covering mantle (*M*) of cortex. The midline falx is absent. (Reproduced with permission from Rafael Levy, RDMS.)

# Dandy-Walker Malformation

## Dandy-Walker Malformation

A rare congenital malformation involving the cerebellum and fourth ventricle

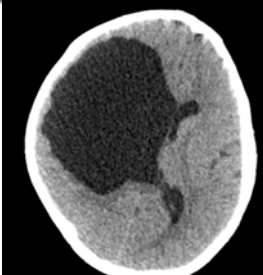
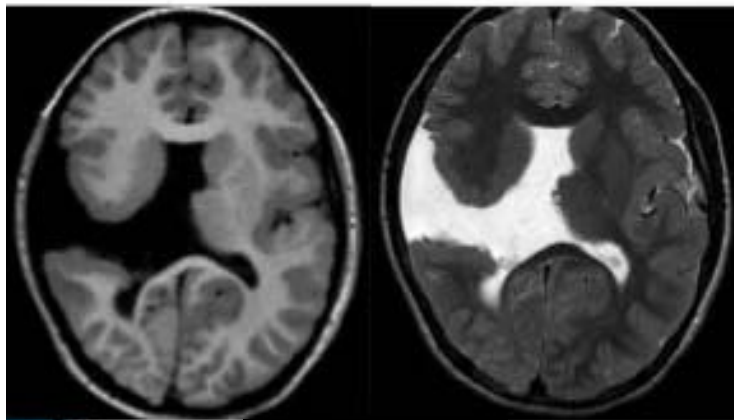
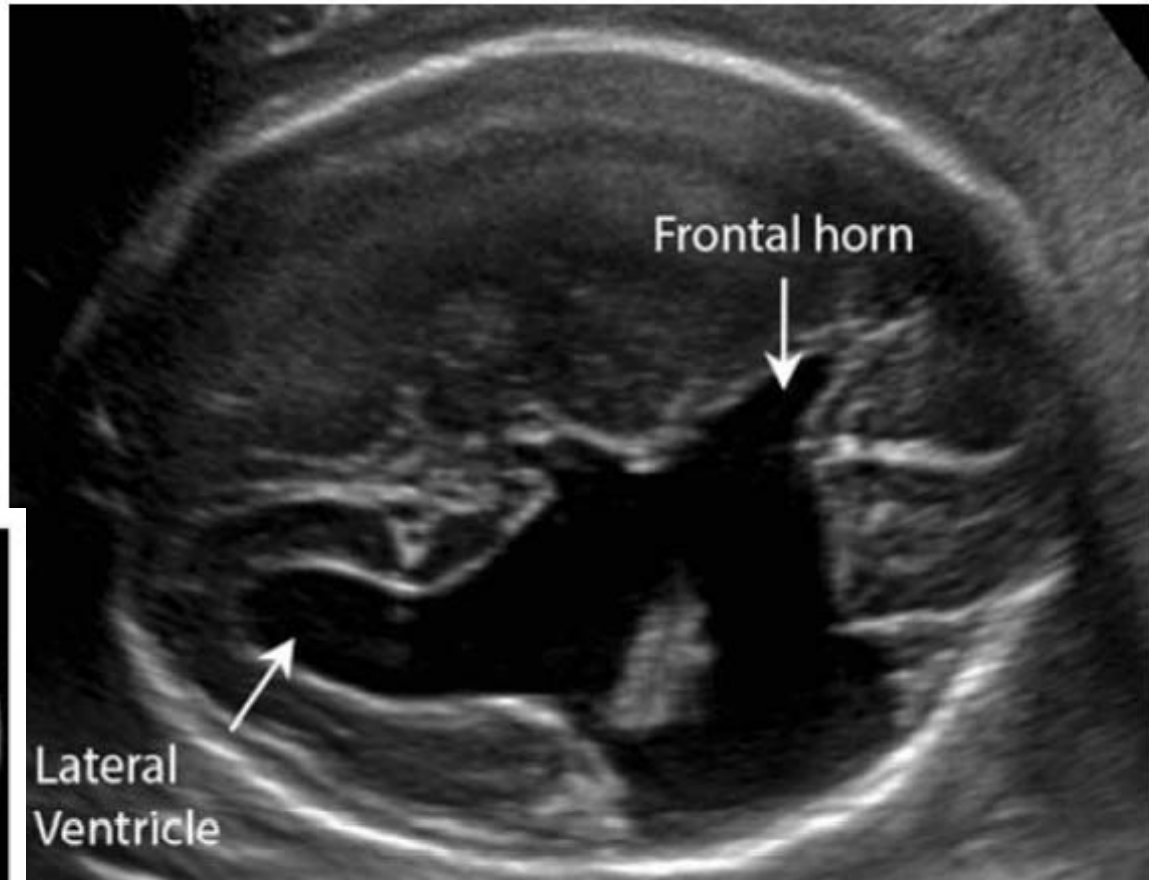


**Sagittal Section**

**Axial Section**

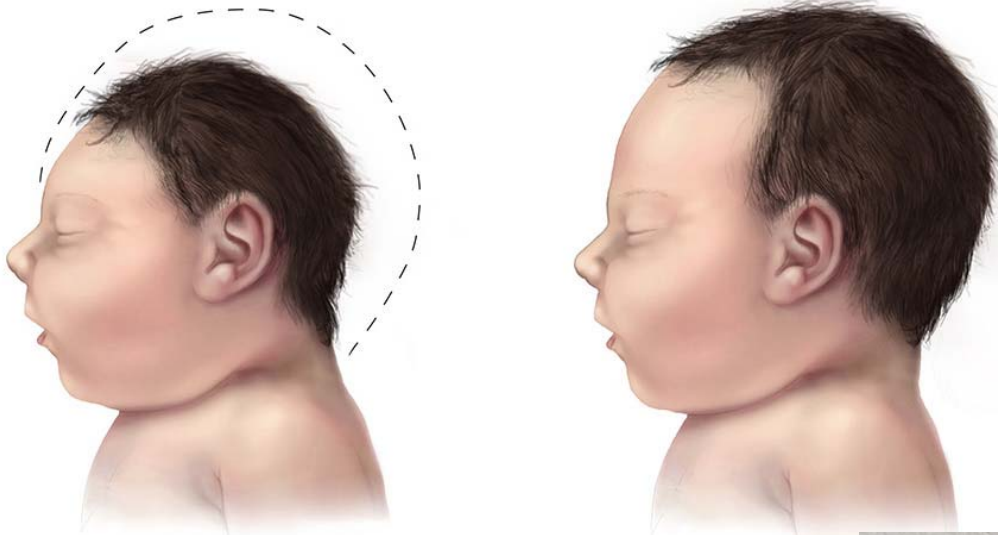
# Schizencephaly and Porencephaly

- An abnormality of neuronal migration
- Absence of the cavum Septum pellucidum and frontal horn communication
- COL4A-1 mutation



**FIGURE 15-15** Schizencephaly. This transverse image shows a large cleft extending from the right lateral ventricle through the cortex. Because the borders of the cleft are separate, the defect is termed *open-lipped*. (Reproduced with permission from Michael Davidson, RDMS.)

# Microcephaly

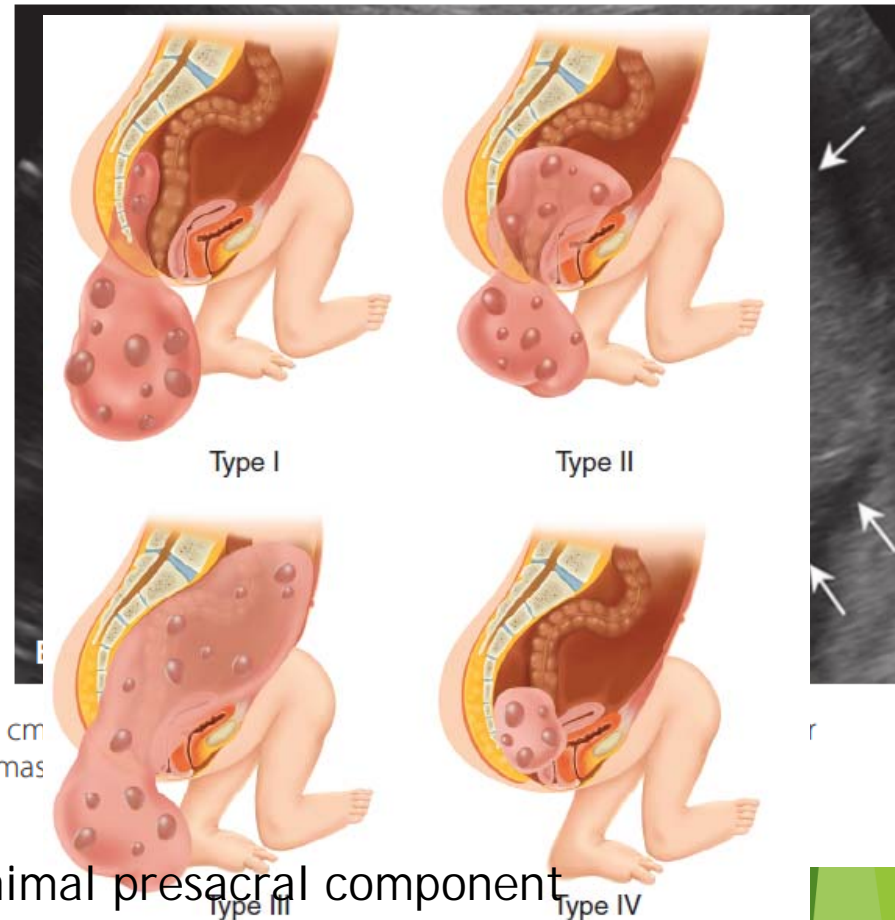


- Head circumference 3 standard deviations (SD) below the mean for gestational age
- Pathologic microcephaly 5SD





# Sacrococcygeal Teratoma



**FIGURE 15-16** Sacrococcygeal teratoma. This tumor enlarged from 3 cm during a 5-week period (B). Arrows depict the external borders of the mass.

Type 1 is predominantly external with a minimal presacral component

Type 2 is predominantly external but with a significant intrapelvic component;

Type 3 is predominantly internal and has abdominal extension;

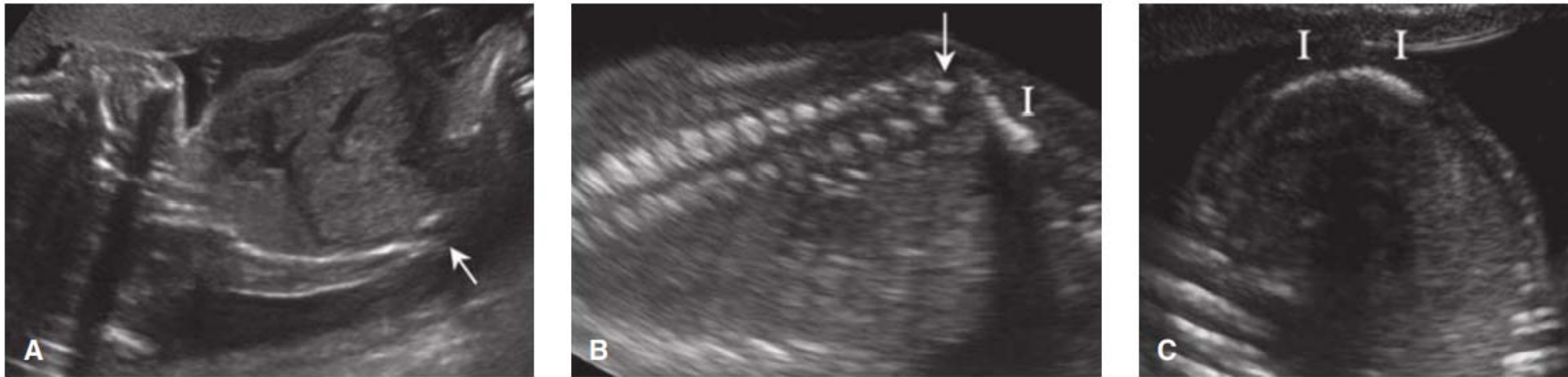
Type 4 is entirely internal with no external component

Histological type may be mature, immature, or malignant

Source: Diana Branch, M.D., M.P.H., *Fetal Pathology: Diagnosis and Management of the Fetal Pathologist*, www.obgyn.mhmedical.com, Copyright © McGraw-Hill Education. All rights reserved.



# Caudal Regression Sequence

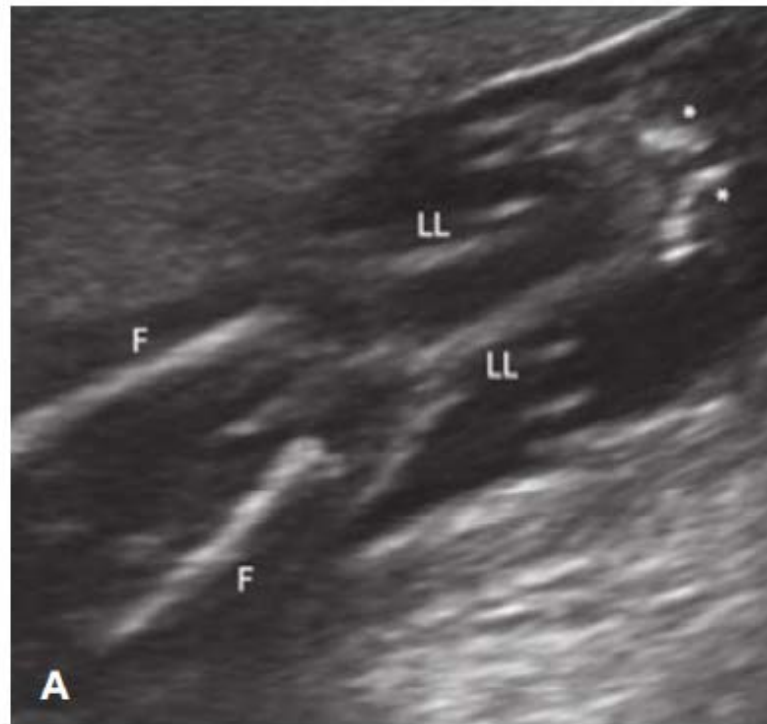


**FIGURE 15-17** Caudal regression sequence. **A.** The spine is markedly foreshortened. The arrow shows where it terminates. **B.** The spine ends abruptly above the level of the iliac wings (*l*). **C.** Without a vertebral body between the iliac wings (*l*), they assume a shield shape.

- Absence of the sacral spine
- **25 times** more prevalent in pregnancies complicated by **diabetes GDM**
- Genitourinary malformations and syndromes such as the VACTERL association



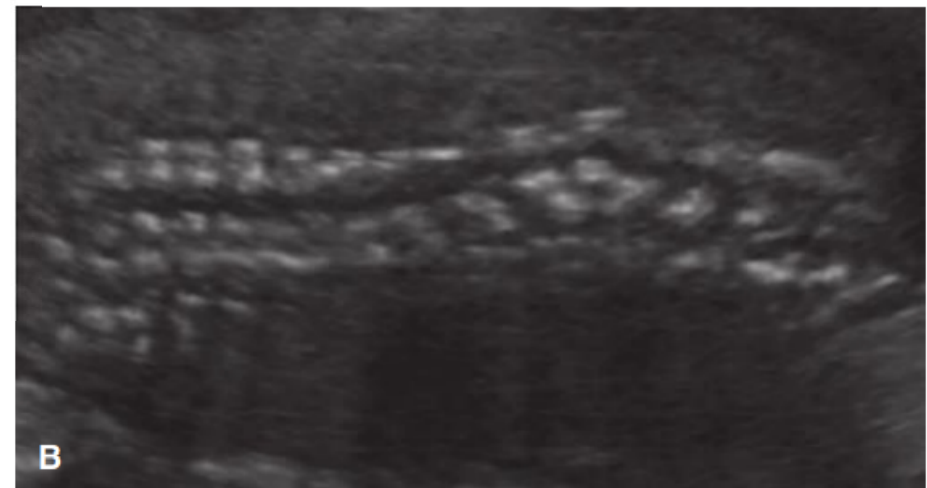
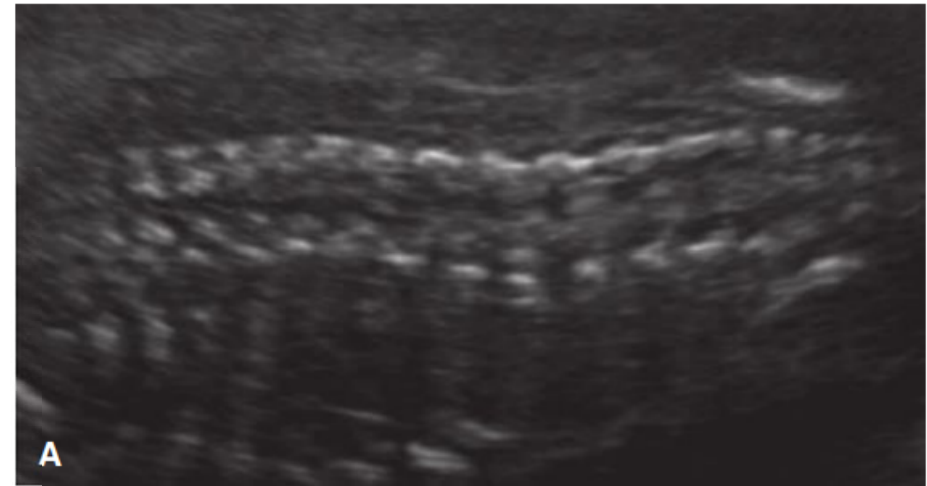
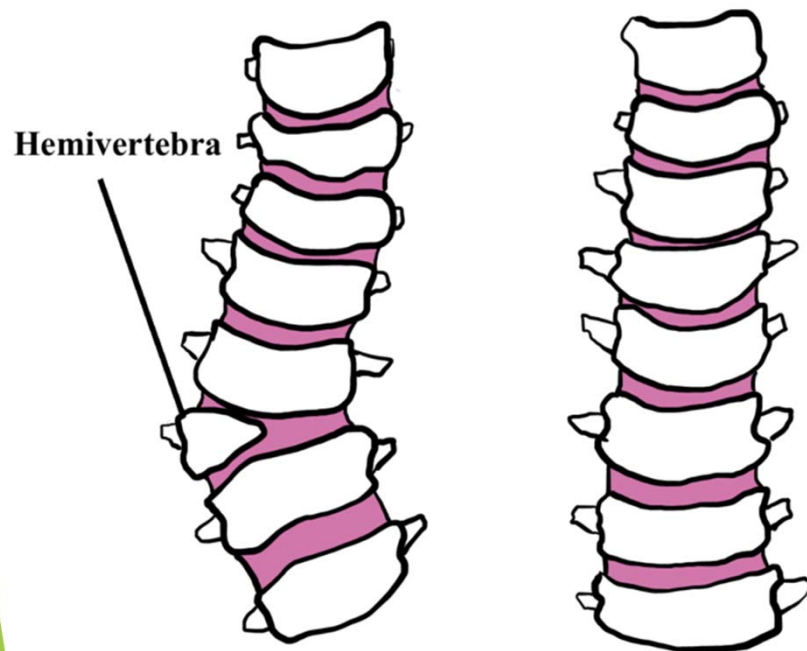
# Sirenomelia



**FIGURE 15-18** Sirenomelia. **A.** This single low lower leg bones (LL), and a fused foot with toes pointing outward (F). **B.** Arrows show the soft tissue outline of the lower extremity. Amniotic fluid is visible only because the gestational age is 17 weeks' gestation. By 18 weeks, absence of kidneys and bladder resulted in anhydramnios. (Reproduced with permission from Melissa Salvie, RDMS.)

- Single lower extremity in the midline and bilateral renal agenesis
- The extremity may contain one or two sets of bones and feet
- After 18 weeks' gestation, cause anhydramnios may complicate the diagnosis

# Hemivertebrae

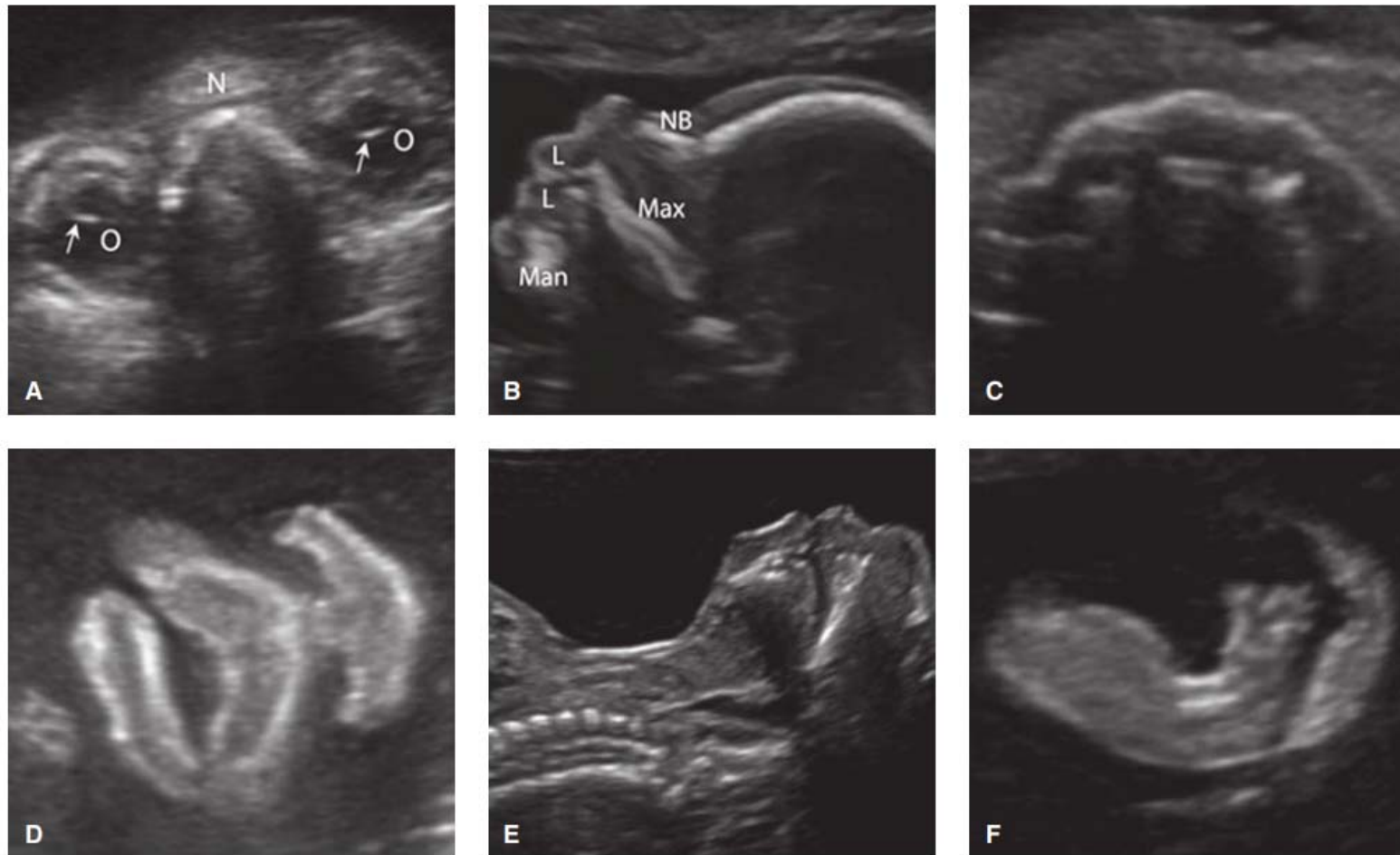


**FIGURE 15-19** Hemivertebrae result in abnormal spinal curvature in these coronal images. (Reproduced with permission from Rose Muli, RDMS.)

# HEAD, FACE, AND NECK

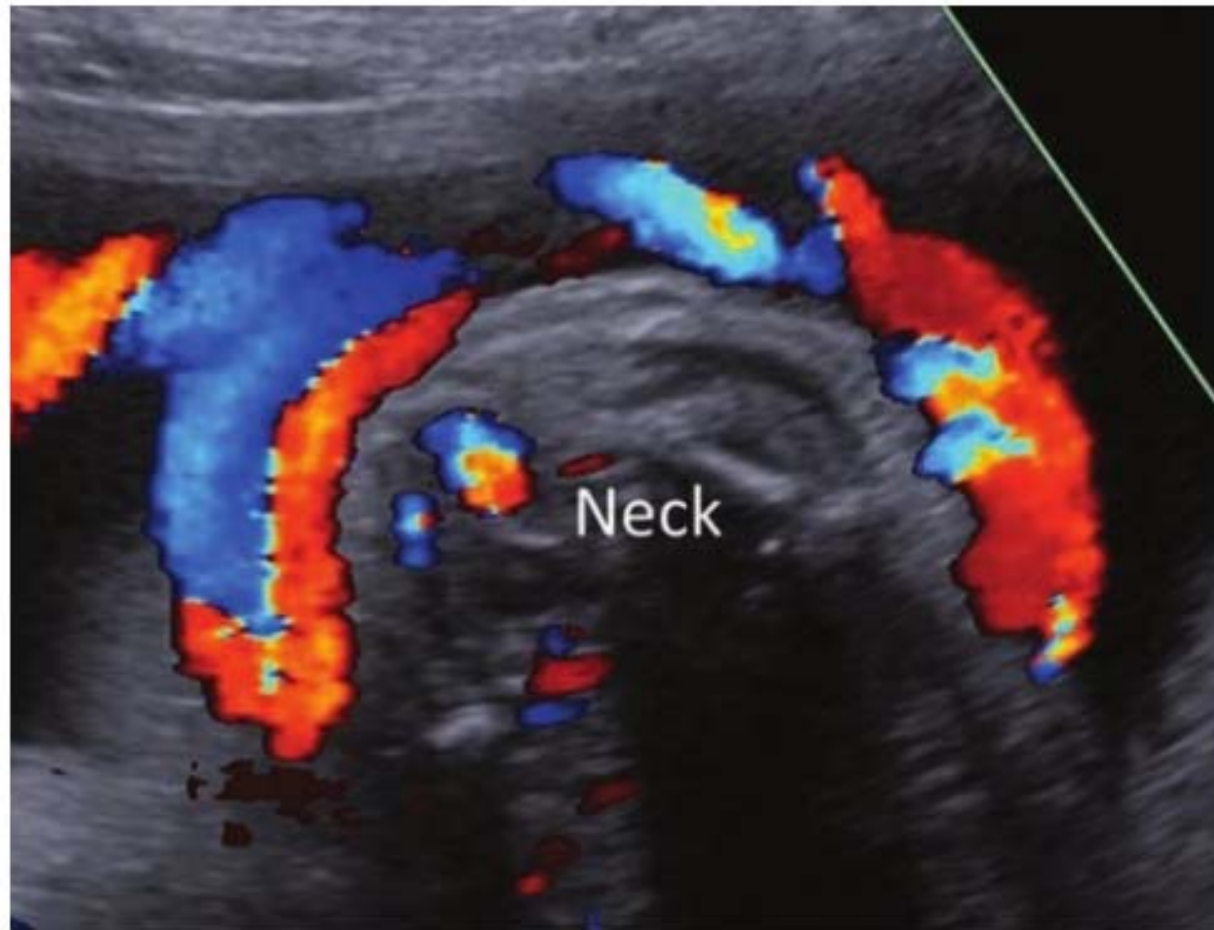


# Craniofacial anatomy



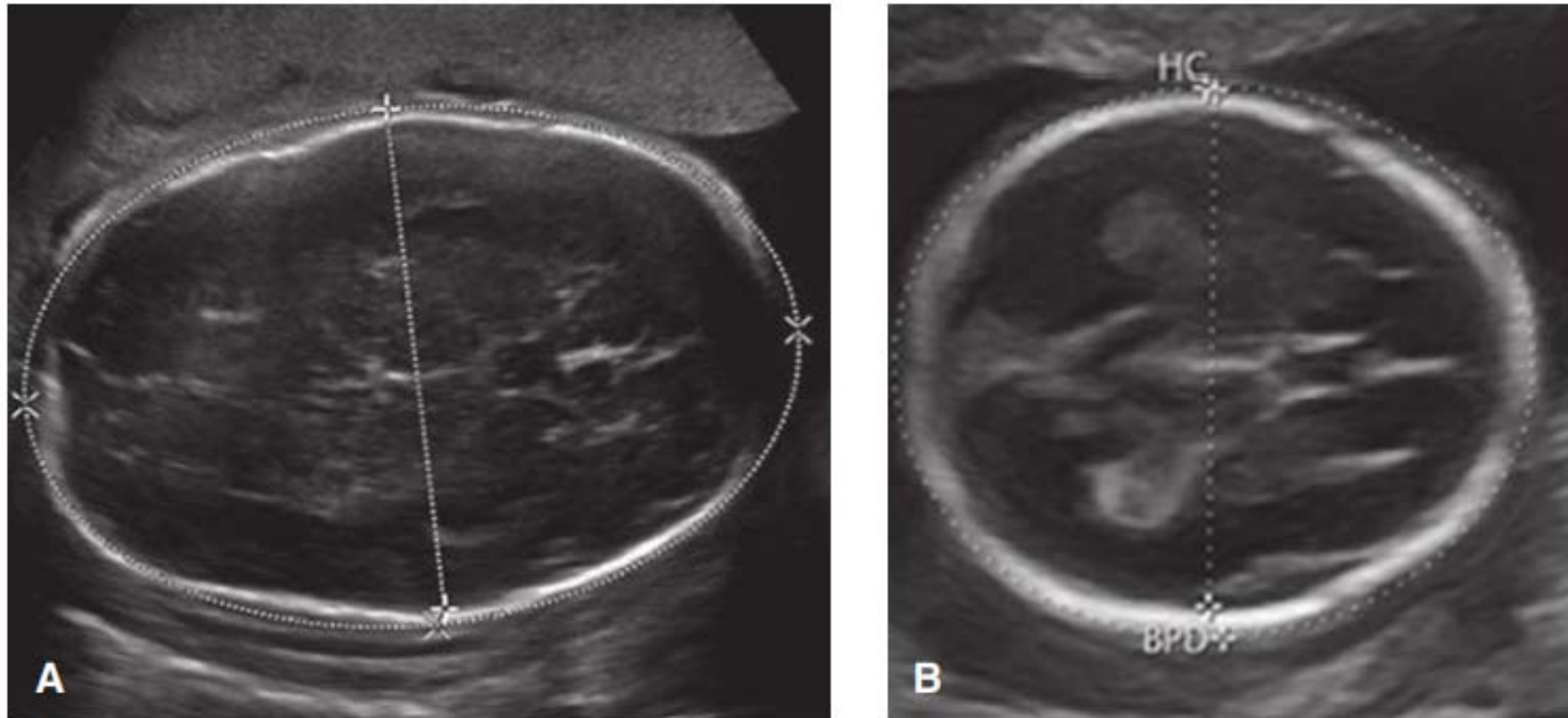
**FIGURE 15-20** Normal craniofacial and neck anatomy. **A.** Transverse images of orbits (*O*) and nose (*N*). The small circle within each orbit is the lens. The distance between the orbits roughly approximates the width of each orbit. **B.** Sagittal view of the face, depicting the nasal bone (*NB*), lips (*L*), maxilla (*Max*), and mandible (*Man*). **C.** Transverse image of the alveolar ridge. **D.** Coronal view of the nose, upper lip, and lower lip. **E.** Sagittal image of the neck. **F.** Image of the ear. (Reproduced with permission from Devi Nanandhan, RDMS.)

# Nuchal cord



**FIGURE 15-21** Nuchal cord incidentally noted with color Doppler in a transverse image of the fetal neck at 34 weeks' gestation.

# Dolichocephaly and Brachycephaly

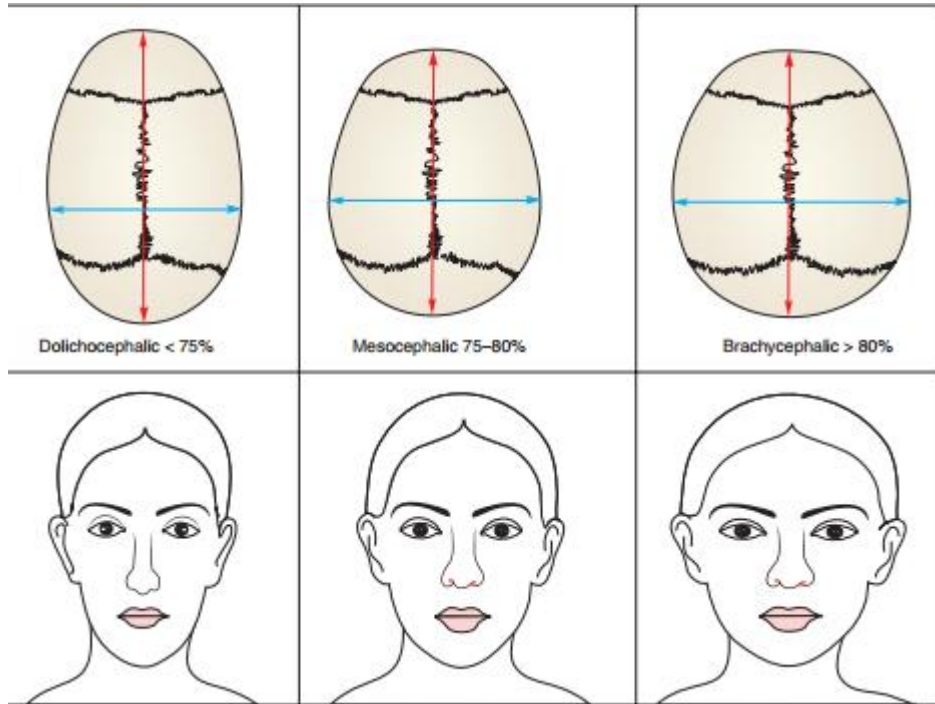


**FIGURE 15-22** Transthalamic images demonstrating dolichocephaly (A) and brachycephaly (B). The biparietal diameter (BPD) and head circumference (HC) are measured in each image.

- Dolichocephaly can occur with neural-tube defects
- Brachycephaly may be seen in fetuses with Down syndrome

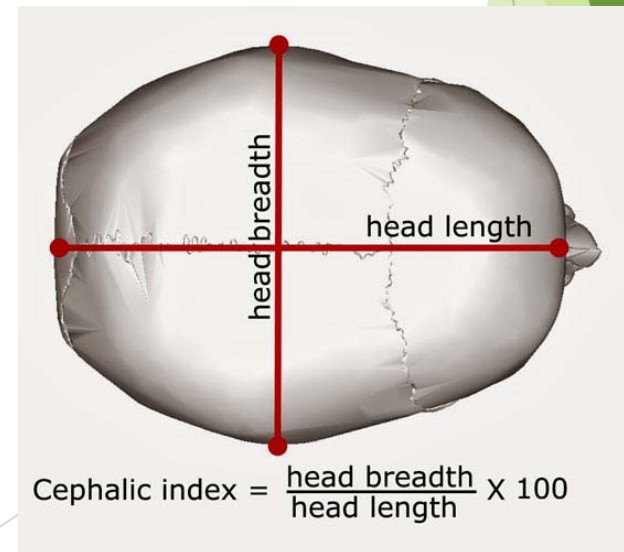


# Cephalic index

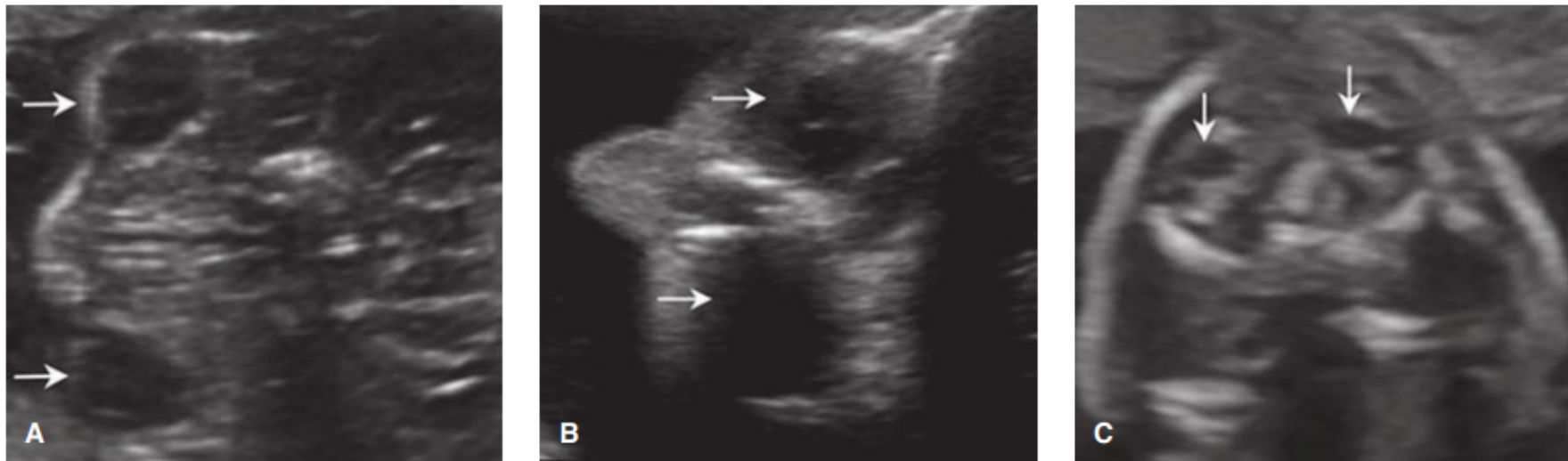


Classification	
Hyperdolichocephalic	65.5-69.9
Dolichocephalic	70.0-74.9
Mesocephalic	75.0-79.9
Brachycephalic	80.0-84.9
Hyperbrachycephalic	85.0-89.9
Ultrabrachycephalic	90.0->90

$$\text{Cephalic index} = \frac{\text{Maximum skull width}}{\text{Maximum skull length}} \times 100$$



# Abnormalities of Orbits and Nose



**FIGURE 15-23** Abnormalities of the orbits. **A.** Hypertelorism in a fetus with trisomy 18. **B.** Hypotelorism in a fetus with trisomy 13 and alobar holoprosencephaly. **C.** Microphthalmia. This fetus also had trisomy 13. Arrows point to the eyes.

- Hypertelorism common finding in trisomy 18



# Abnormalities



**A** Median cleft lip and palate



**B** Cebocephaly



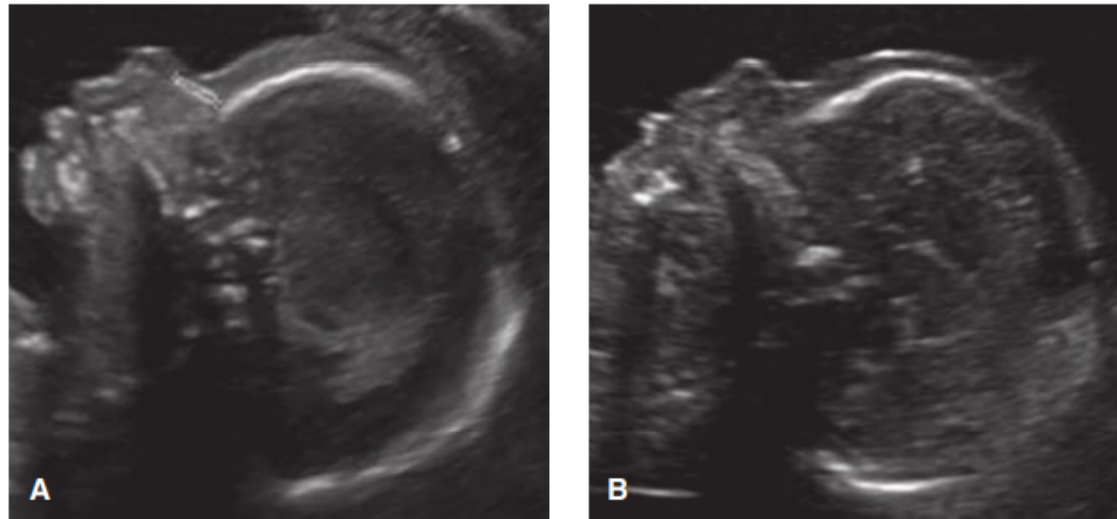
**C** Ethmocephaly



**D** Cyclopia

**FIGURE 15-24** Nasal abnormalities associated with holoprosencephaly. **A.** Coronal image demonstrating the proboscis protruding a single nostril (cebocephaly). **B.** Photograph of a

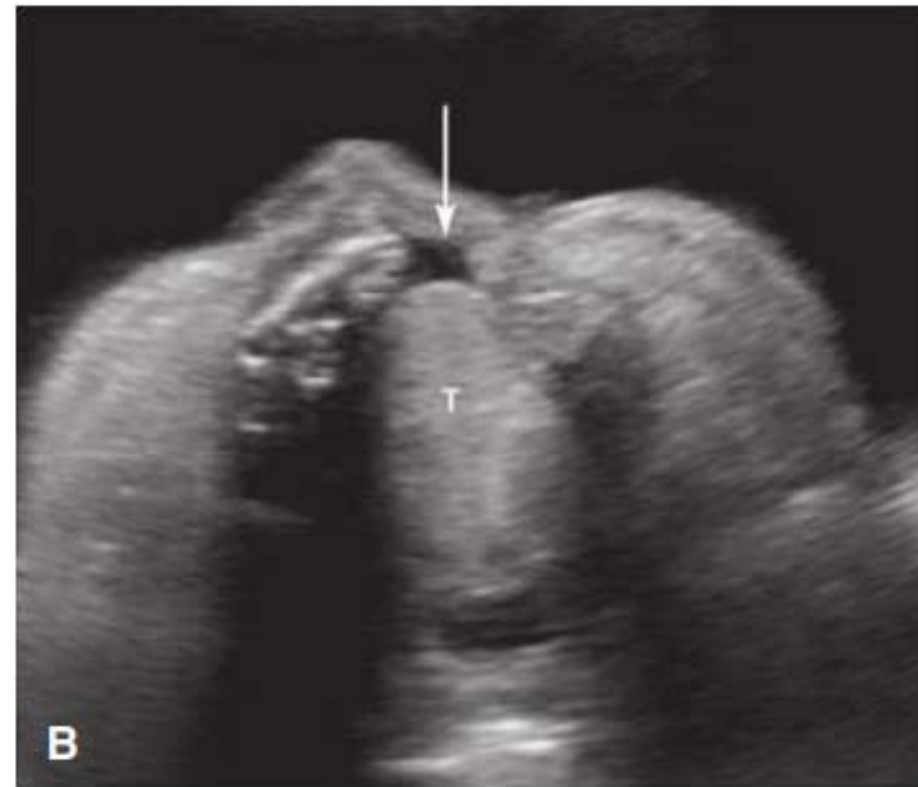
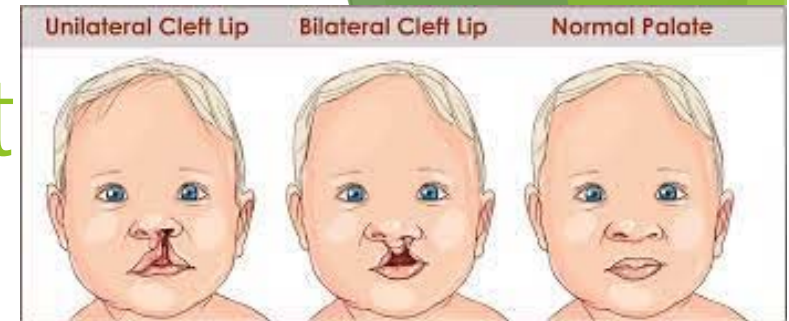
# Abnormalities of Orbits and Nose



**FIGURE 15-25** Nasal bone (and its absence). **A.** Sagittal image of the profile showing measurement of a normal nasal bone at 19 weeks. **B.** Fetus with trisomy 21, also at 19 weeks, with no visible nasal bone. (Reproduced with permission from Jason McWhirt, RDMS.)

- An aneuploidy marker that confers increased risk for fetal Down syndrome

# Facial Cleft

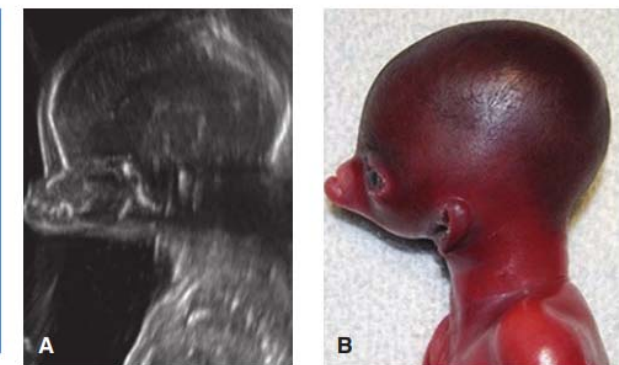


**FIGURE 15-26** Cleft lip/palate. **A.** This fetus has a prominent unilateral (left-sided) cleft lip. **B.** Transverse view of the palate in the same fetus demonstrates a defect in the alveolar ridge (*arrow*). The tongue (*T*) is also visible.

# Micrognathia

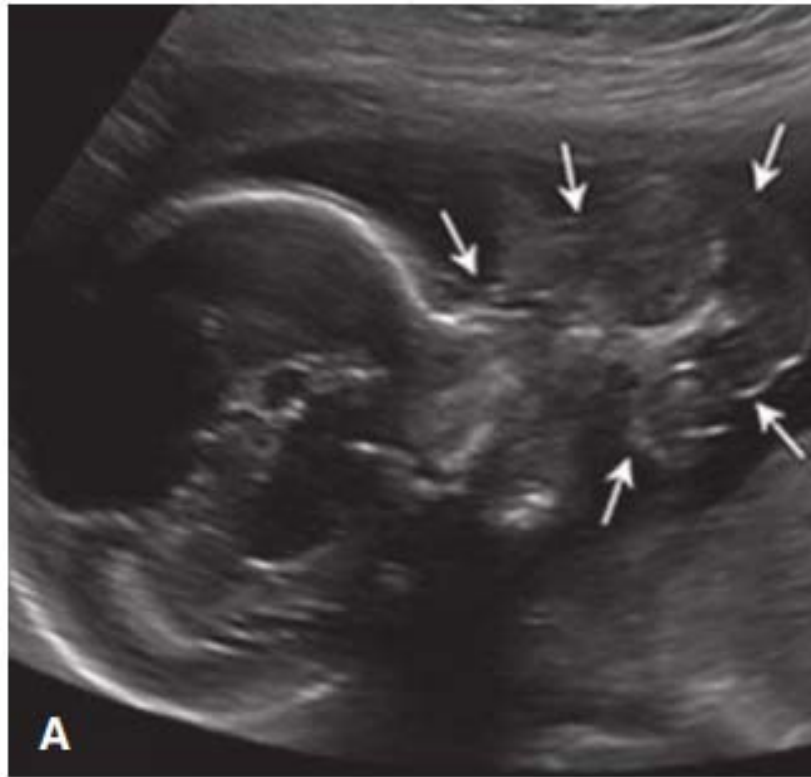


**FIGURE 15-27** Micrognathia. **A.** Sagittal image of a fetus with severe micrognathia. **B.** 3-dimensional ultrasound rendering depicts the recessed chin and downslanting palpebral fissures. **C.** A transverse image of the mandible was used to calculate a jaw index for this fetus.



**FIGURE 15-28** Agnathia-otocephaly, ultrasound (**A**) and post-delivery (**B**) images. With this rare, lethal anomaly the mandible fails to develop, and the ears are inferiorly displaced and may be fused in the midline.

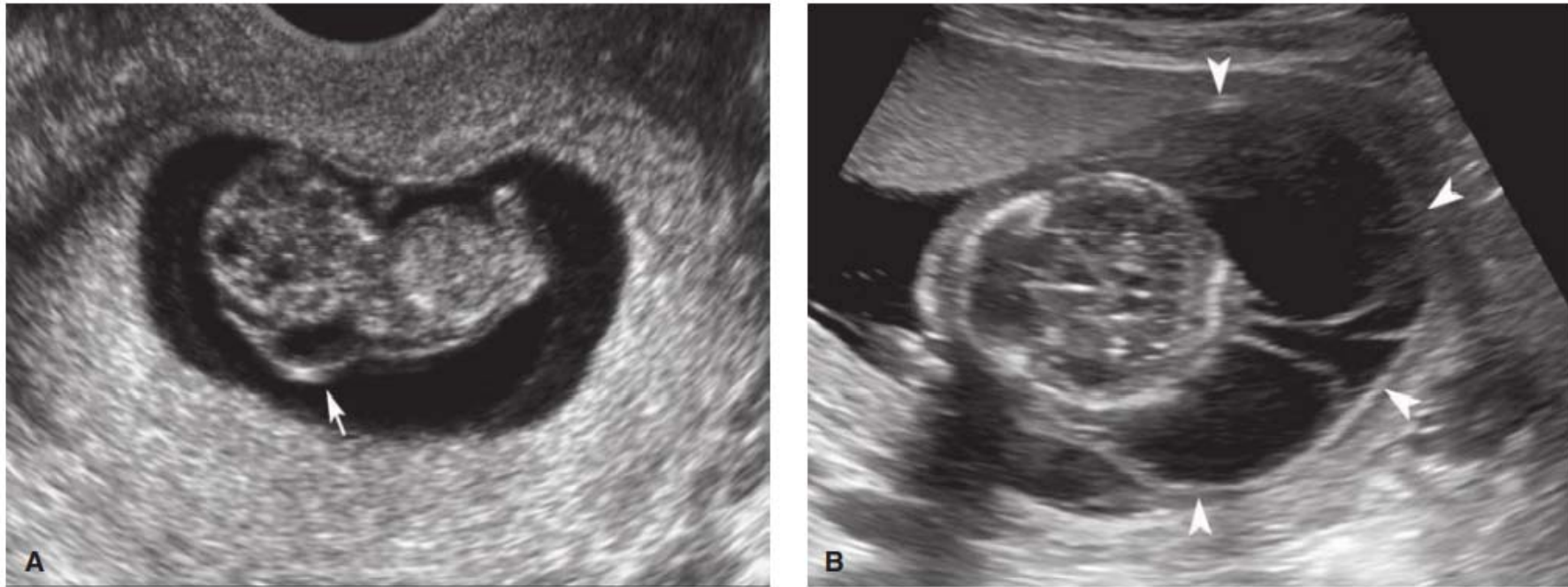
# Epignathus



**FIGURE 15-29** Epignathus, ultrasound (A) and postdelivery (B) from the oral cavity or pharynx and may grow outward from the and into the brain, as in this fetus (*arrowhead*). Arrows depict the (Reproduced with permission from Halima Abdirahman, RDMS.)

**Fig. 1**

# Cystic Hygroma



**FIGURE 15-30** Cystic hygromas. **A.** This 9-week fetus with a cystic hygroma (*arrow*) was later found to have Noonan syndrome. **B.** Massive multiseptated hygromas (*arrowheads*) in the setting of hydrops fetalis at 15 weeks' gestation.

- Impaired lymphatic drainage from the head into the jugular vein leads to an accumulation of fluid in jugular lymphatic sacs
- First-trimester fetuses with cystic hygromas are **five times** more likely to be aneuploid

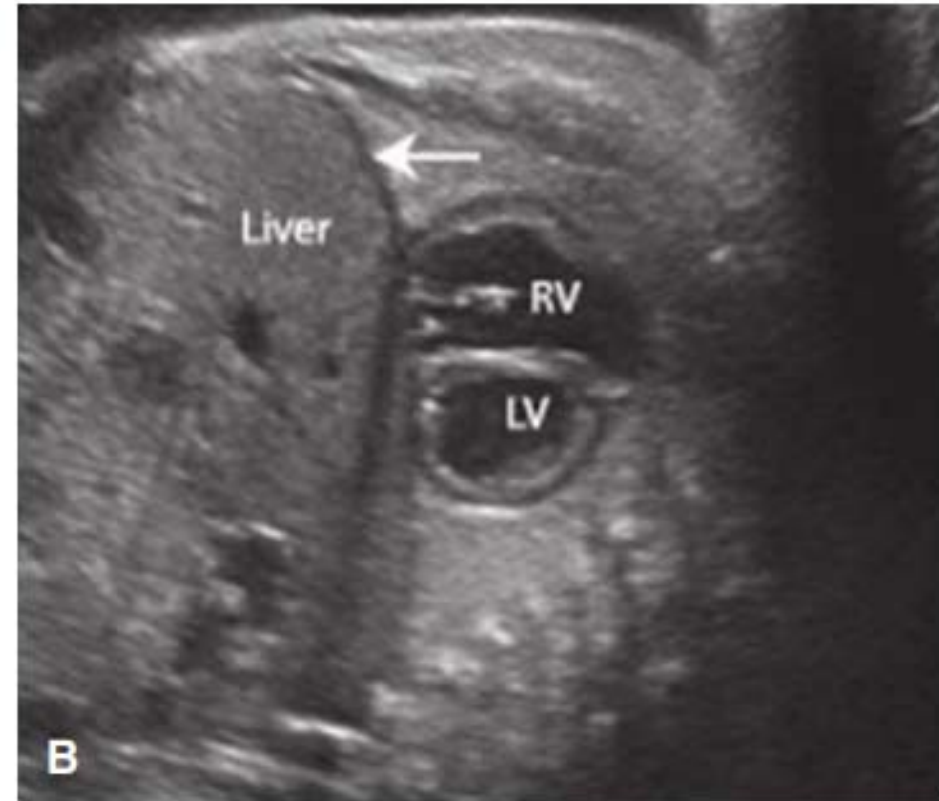
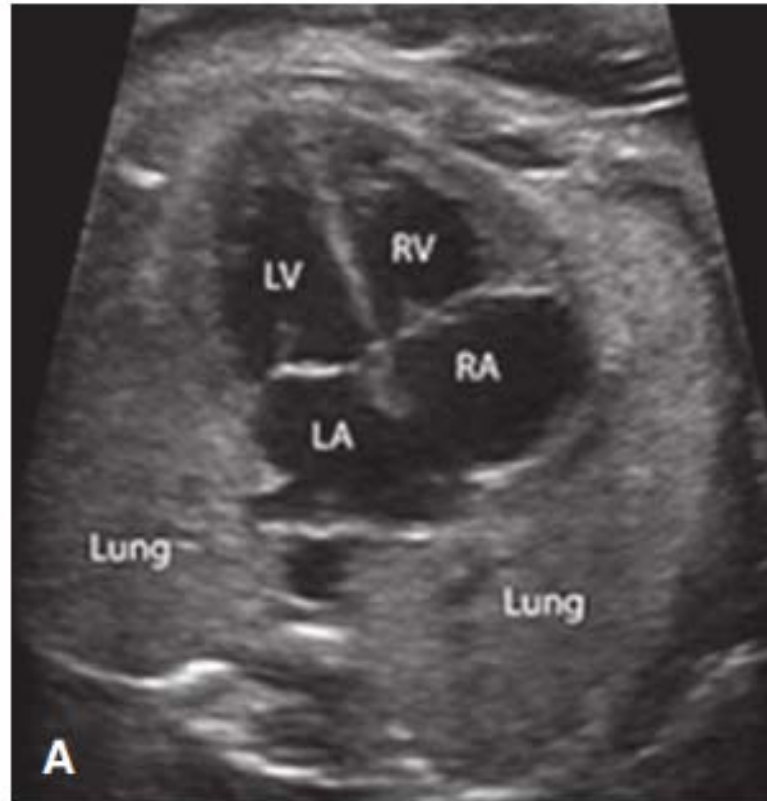




# THORAX

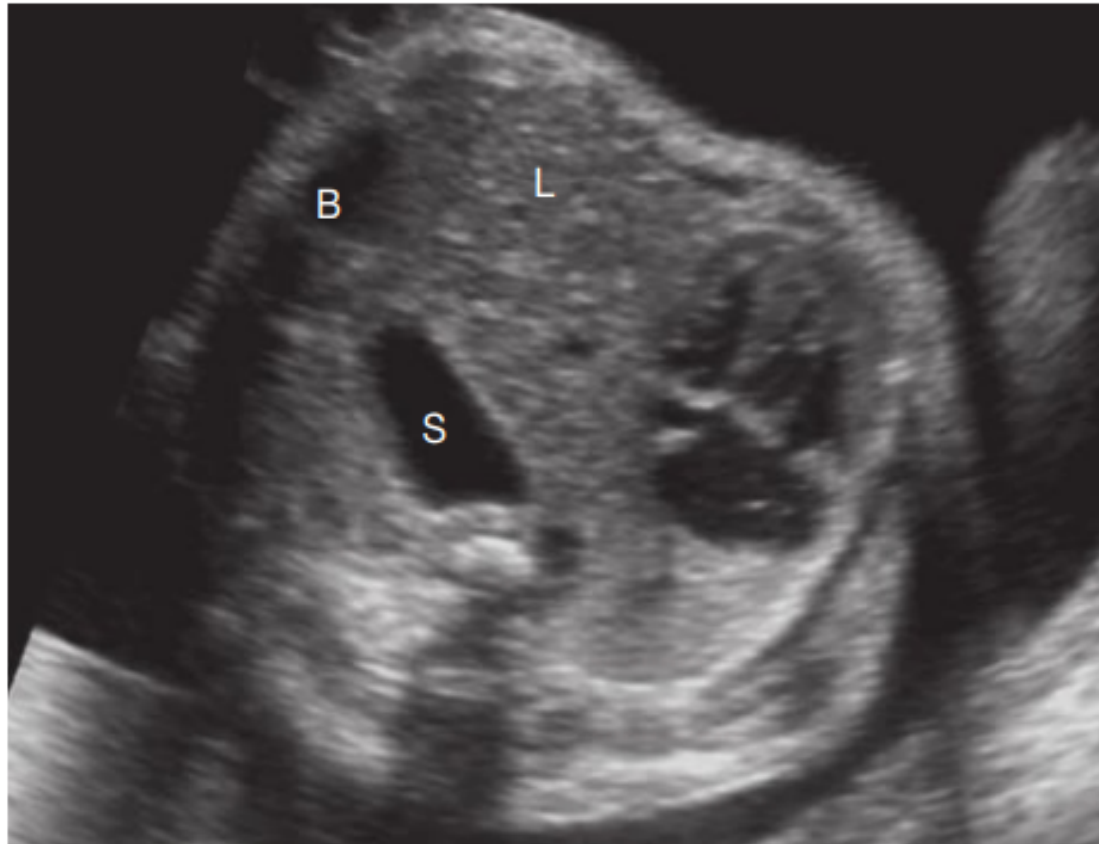


# Toracic anatomy



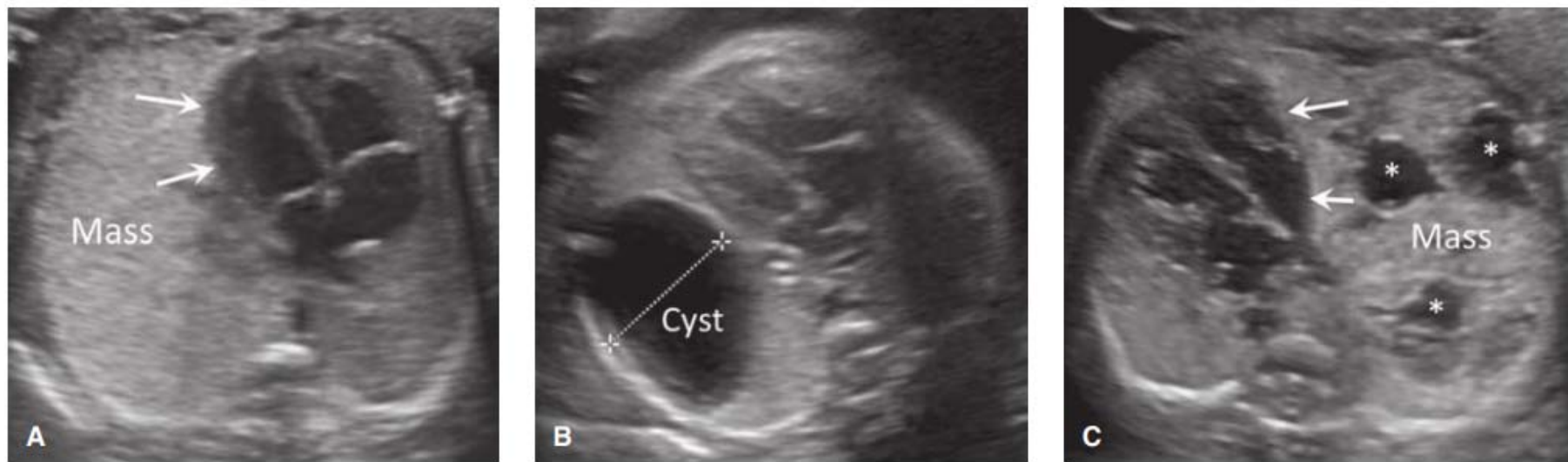
**FIGURE 15-31** Normal thoracic anatomy. **A.** The lungs each occupy one third of the area in the four-chamber view of the heart. **B.** The diaphragm (*arrow*) appears as a hypoechoic line in between the lung and liver in this parasagittal view. LA = left atrium; LV = left ventricle; RA = right atrium; RV = right ventricle.

# Diaphragmatic Hernia



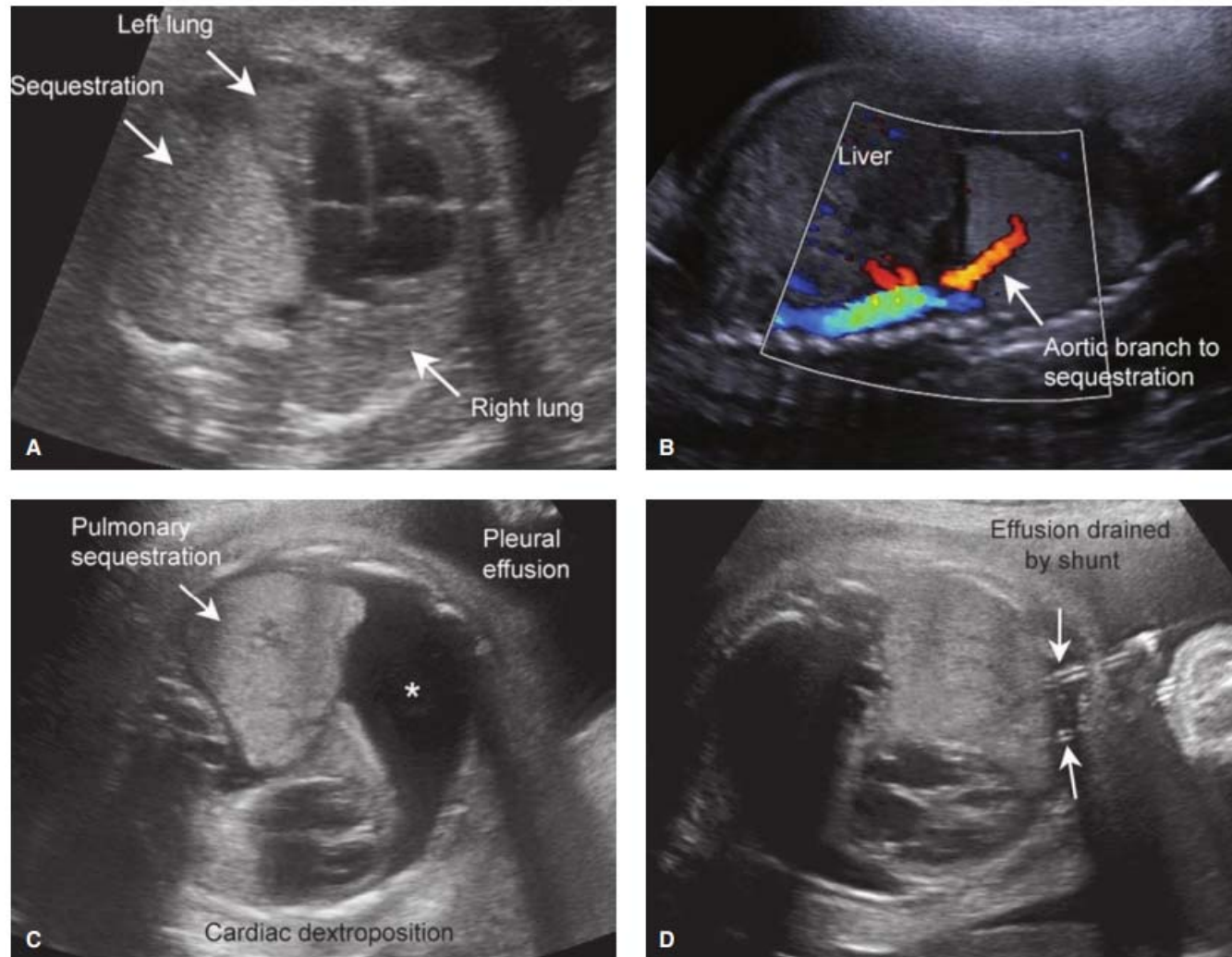
**FIGURE 15-32** Congenital diaphragmatic hernia. In this transverse view of the thorax, the heart is shifted to the right side of the chest by a left-sided diaphragmatic hernia containing stomach (S), liver (L), and bowel (B).

# Congenital Cystic Adenomatoid Malformation



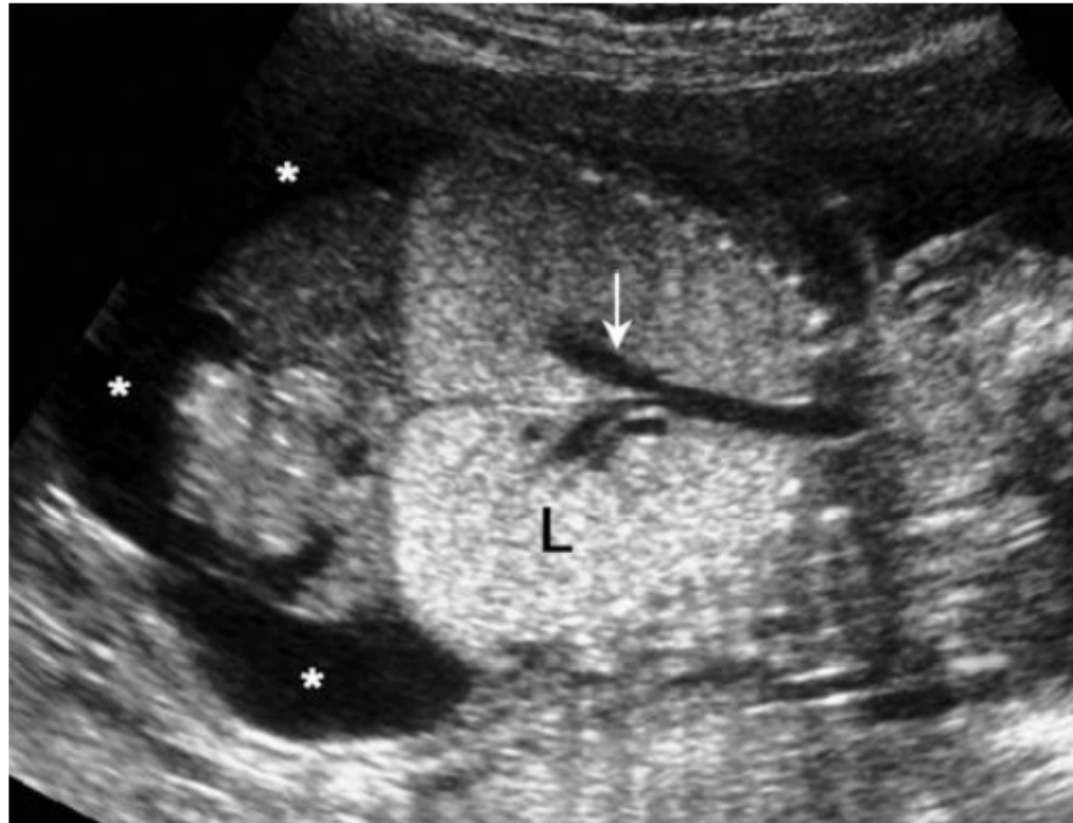
**FIGURE 15-33** Congenital cystic adenomatoid malformation (CCAM). **A.** This left-sided microcystic CCAM is an echogenic mass that fills the left hemithorax and causes mediastinal shift, which displaces the heart (*arrows*) to the right side of the chest. **B.** This left-sided macrocystic CCAM contains a cyst as large as the heart and also displaces the heart to the right. **C.** This right-sided CCAM contains multiple cysts of varying size (\*) and displaces the heart farther to the left side of the chest (*arrows*).

# Pulmonary Sequestration



**FIGURE 15-34** Pulmonary sequestration. **A.** Transverse image of the thorax depicts a left lower lobe pulmonary sequestration (PS) in this 25-week fetus. **B.** Sagittal image showing that blood supply to the mass is from a branch of the abdominal aorta, which confirms the diagnosis. **C.** Over the next 3 weeks, a large ipsilateral pleural effusion develops (*asterisk*), resulting in mediastinal shift and dextroposition of the heart to the far-right thorax. **D.** After placement of a double-pigtail shunt through the chest wall, which drains the effusion into the amniotic fluid, the lung significantly reexpanded. Arrows point to coils of the pigtail shunt. (Reproduced with permission from Dr. Elaine Duryea.)

# Congenital High Airway Obstruction Sequence



**FIGURE 15-35** Congenital high airway obstruction sequence (CHAOS). The lungs (*L*) appear brightly echogenic, and the bronchi (*arrow*) are dilated with fluid. Flattening and eversion of the diaphragm is common, as is ascites (*asterisks*).

# Take home message

- ▶ ELECTRONIC FETAL MONITORING
- ▶ NONREASSURING FETAL STATUS

