THE FETAL FACE AND NECK

Chapter 59

Congenital Anomalies

- Anomalies of the face affect 1 in 600 births
- Examples of facial problems that may be found by ultrasound
  - Cleft lip
  - Hypotelorism
  - Hypertelorism
  - Micrognathia

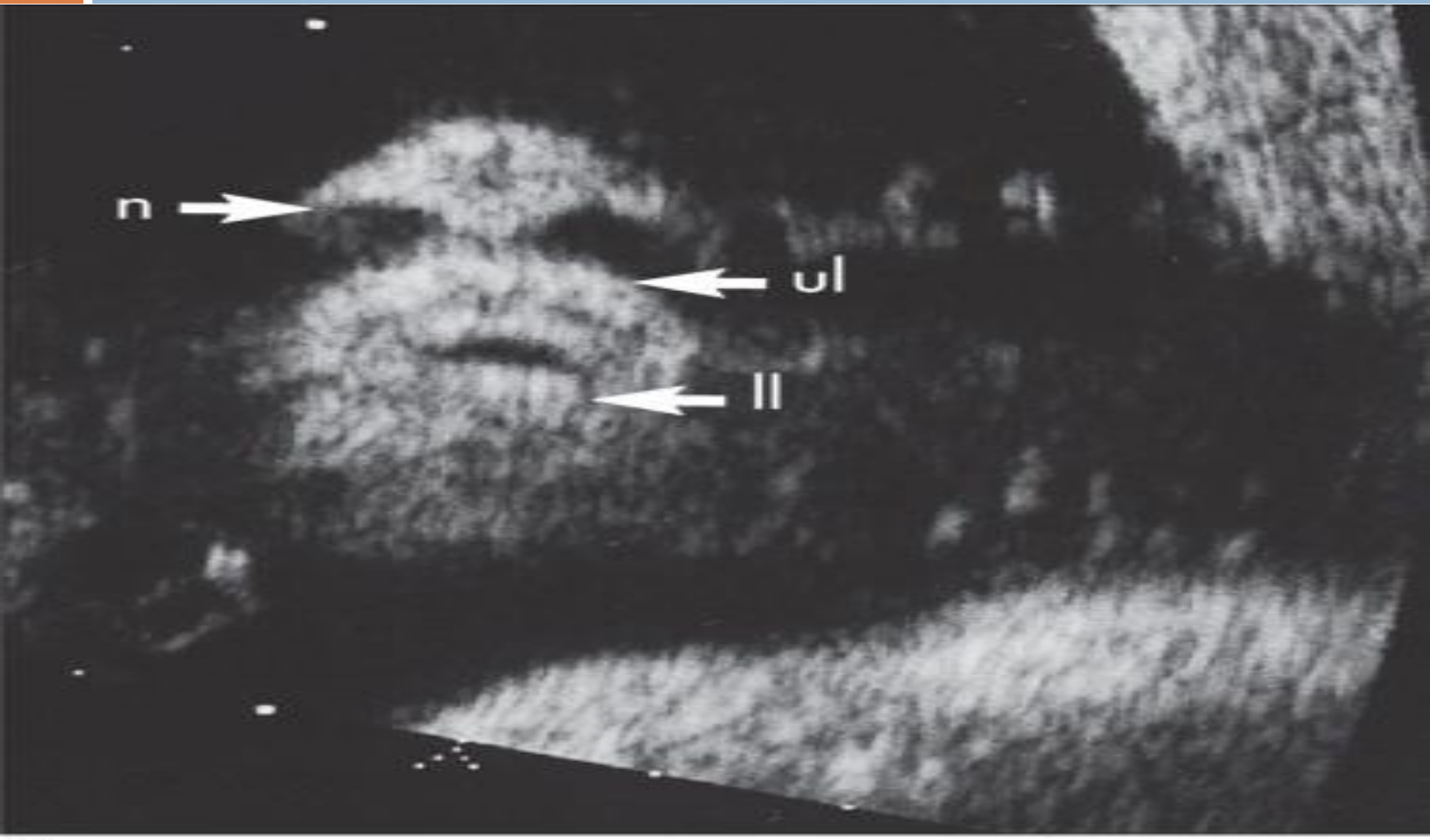
SONOGRAPHIC EVALUATION OF THE FETAL FACE

- Features of the fetal face can be identified at the end of the first trimester

- Fetal profile is well imaged with endovaginal beginning late first trimester to early second trimester
- Modified coronal view is best to image the cleft lip and palate
- Maxilla and orbits are well imaged in a true coronal plane
- Lens of the eye is seen as a small echogenic circle within the orbit

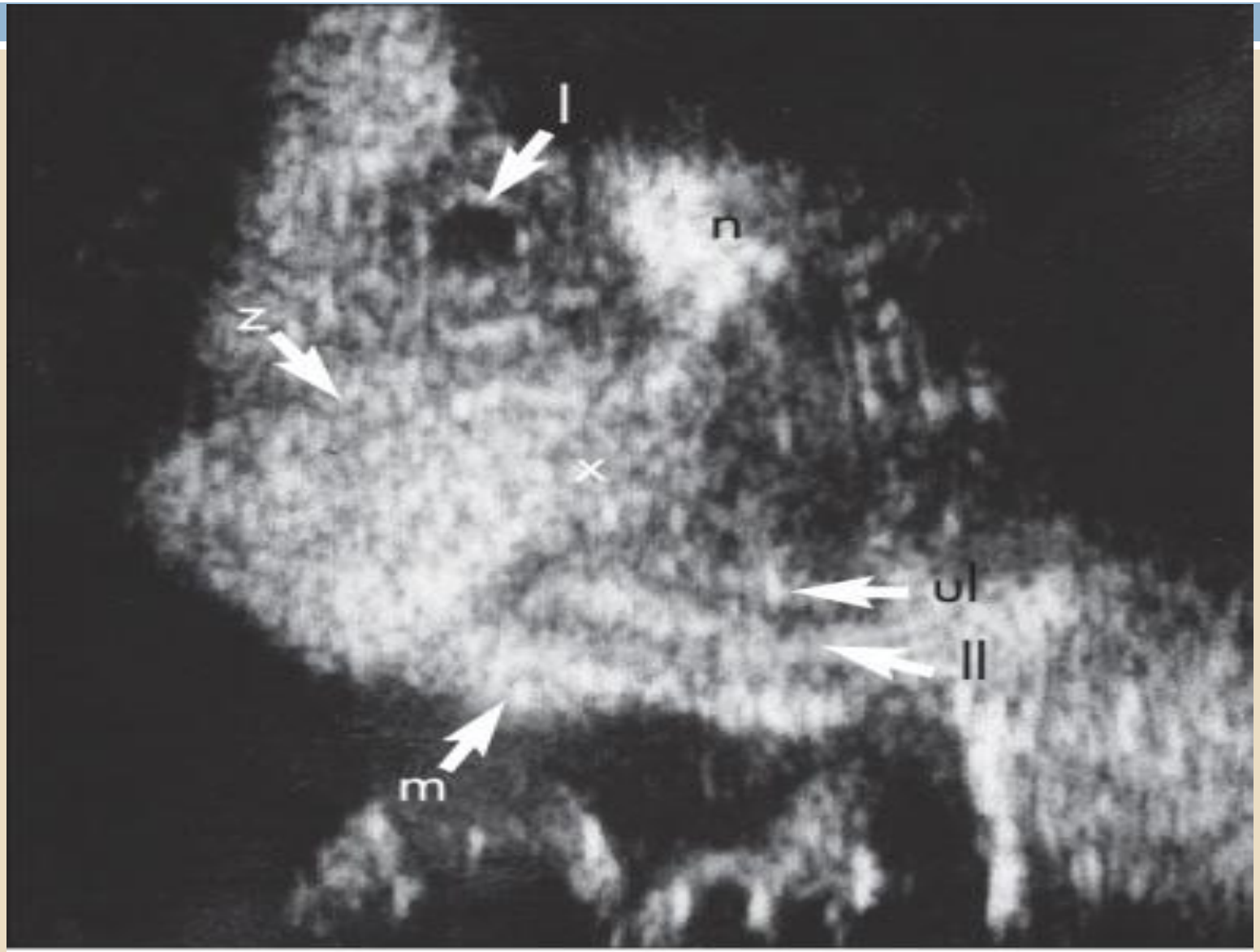
18 WEEK FETAL PROFILE

Modified Coronal View



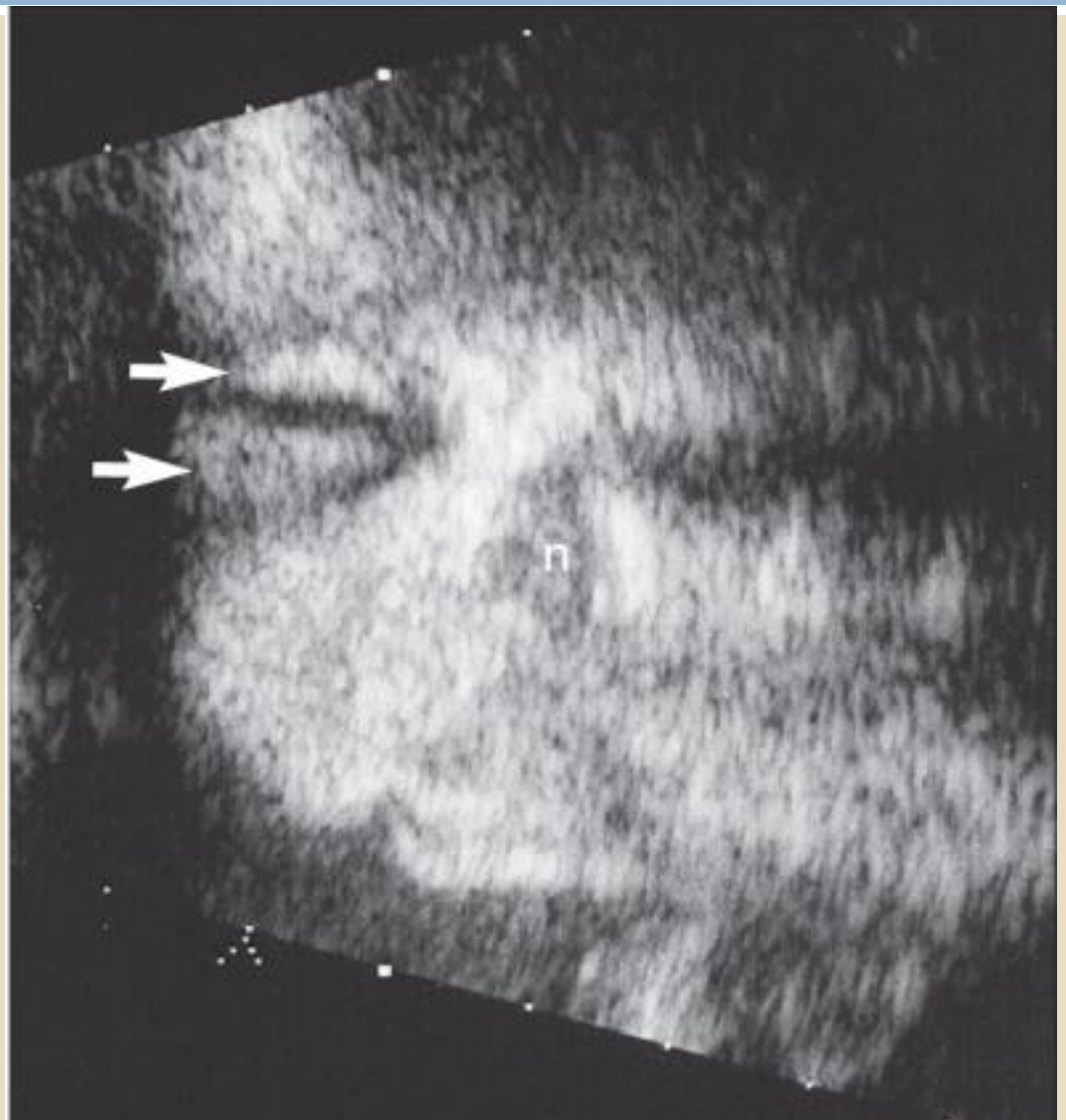
Modified Coronal View

Coronal View



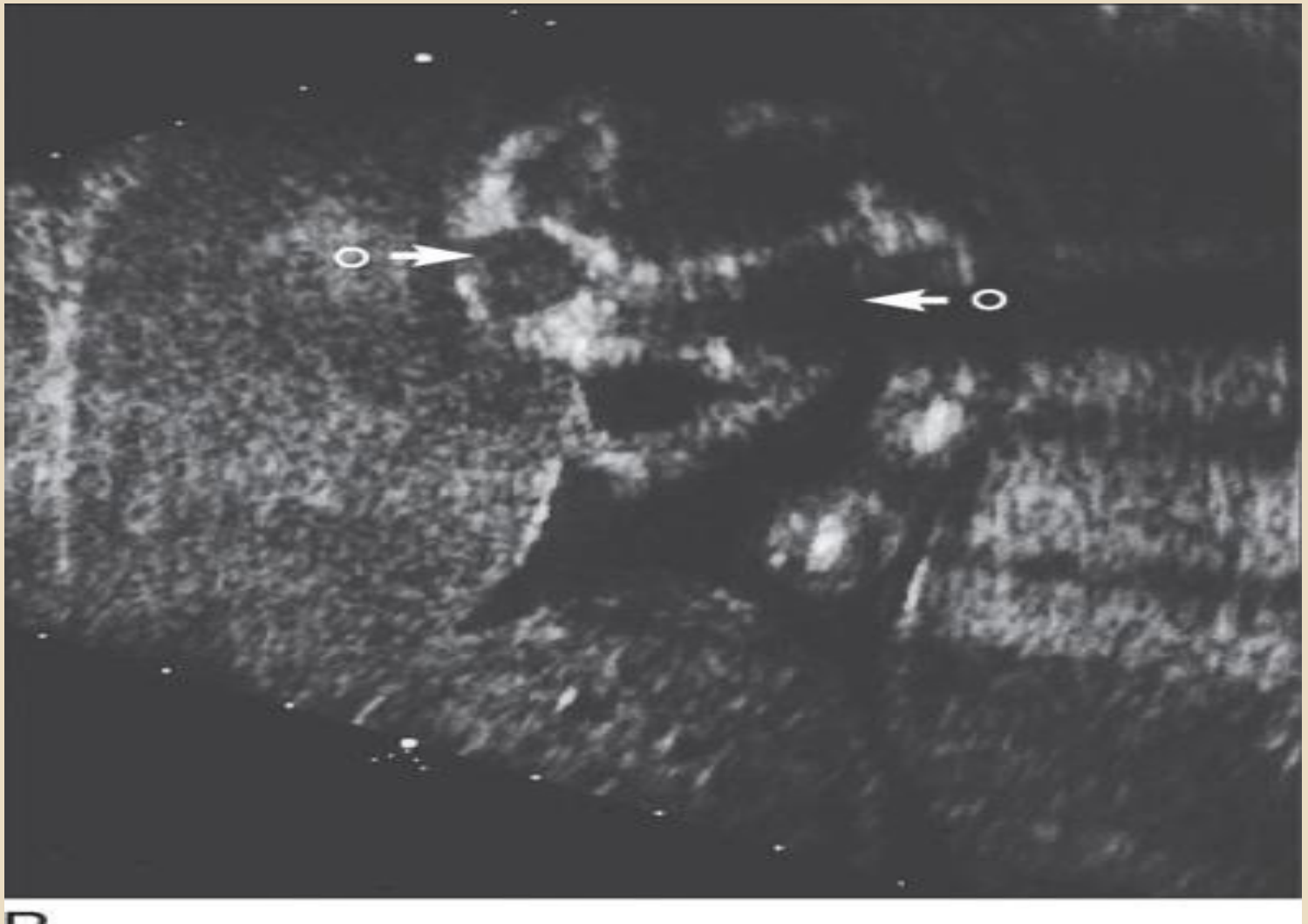
Coronal View

EYELIDS



More Anterior View

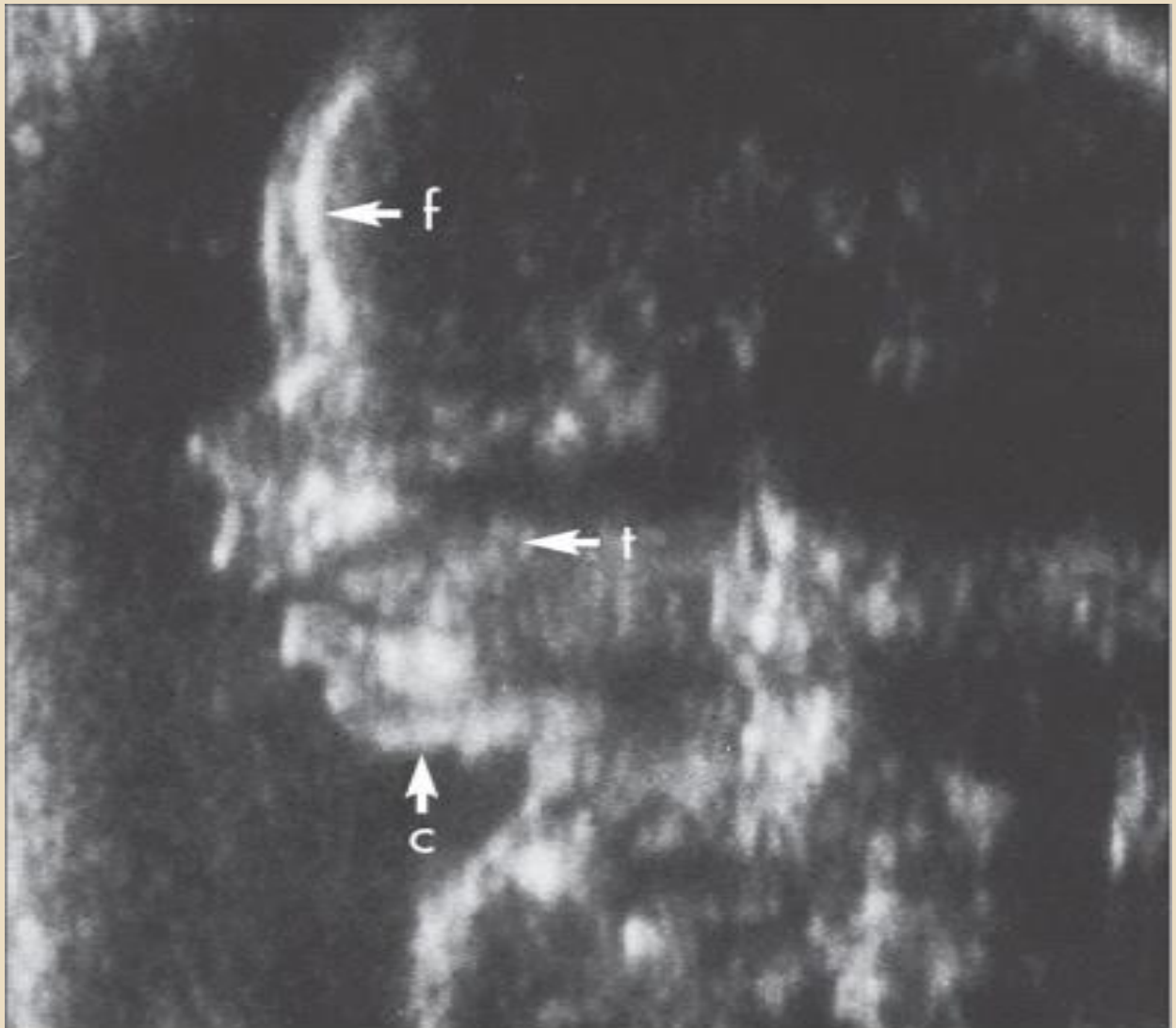
Coronal View



Longitudinal View

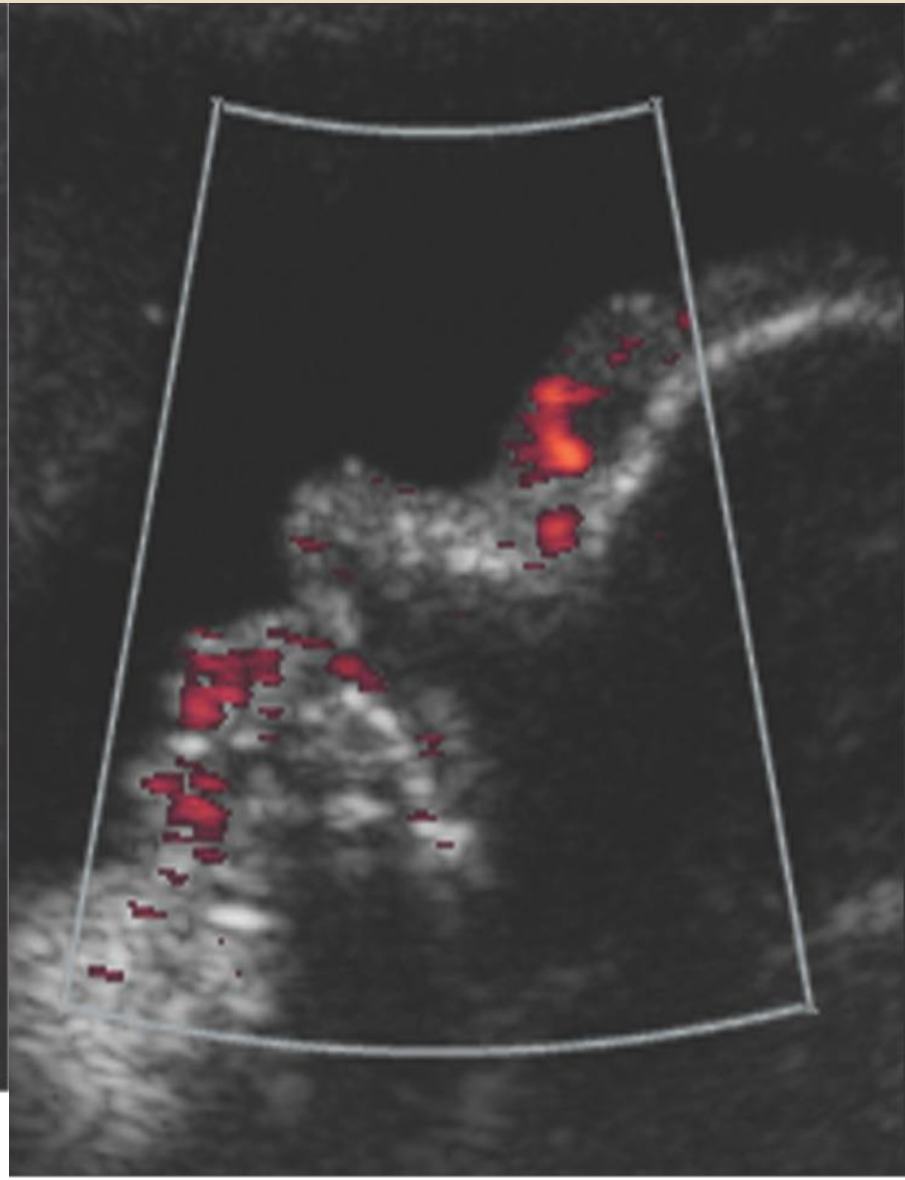
- Demonstrates
  - Nasal bones
  - Soft tissue
  - Mandible
  - Good to rule out:
    - Micrognathia
    - Anterior encephalocele
    - Nasal bridge defects
    - Examine upper lip

Longitudinal View



Longitudinal View

A



Transverse View

- Demonstrates
  - Orbital abnormalities
  - Intraorbital distances
  - Good to evaluate
    - Maxilla
    - Mandible
    - Tongue

Fetal Facial Evaluation

- Not routinely included in a basic fetal scan
- Family history of craniofacial malformation or another congenital anomaly
  - Face should be screened for a coexisting facial malformation
- Many fetuses with a facial defect also have chromosomal abnormalities

Fetal Facial Evaluation

- Extensive facial screening may be hindered by:
  - Bone shadowing
  - Poor fetal positioning
  - Oligohydramnios
  - Maternal obesity
- Facial anomalies often indicate a specific syndrome or condition
  - Orbital fusion and a proboscis
    - Suggest alobar holoprosencephaly

Fetal Facial Evaluation

- Facial anomalies are heterogeneous
  - Occur as:
    - Isolated defects
    - Part of a syndrome
- FX of a facial anomaly may prompt a targeted study
  - Although recurrence risks are relatively low
    - Less than 5%

ABNORMALITIES OF THE FACE AND NECK

Abnormalities of the Facial Profile

- Questions When Evaluating Facial Profile
  - Are the orbits normally spaced?
  - Are the nose and nasal bridge clearly imaged: Is a proboscis or cebocephaly present?
  - Are any periorbital masses apparent?
  - Is the upper lip intact?
  - Is the tongue normal size?
  - Is the chin abnormally small?
  - Are the ears normal size and position?

Sonography of the Facial Profile

- Mid-sagittal scans through the face appear as curvilinear surface with differentiation of forehead, nose, lips, chin
  - Cloverleaf skull - Misshaped skull with cloverleaf appearance
  - Frontal bossing - Lemon-shaped skull or absent, depressed nasal bridge
  - Strawberry-shaped cranium - Bulging of frontal bones
  - Masses of nose and upper lip - Distortion of facial profile (look for cleft lip)

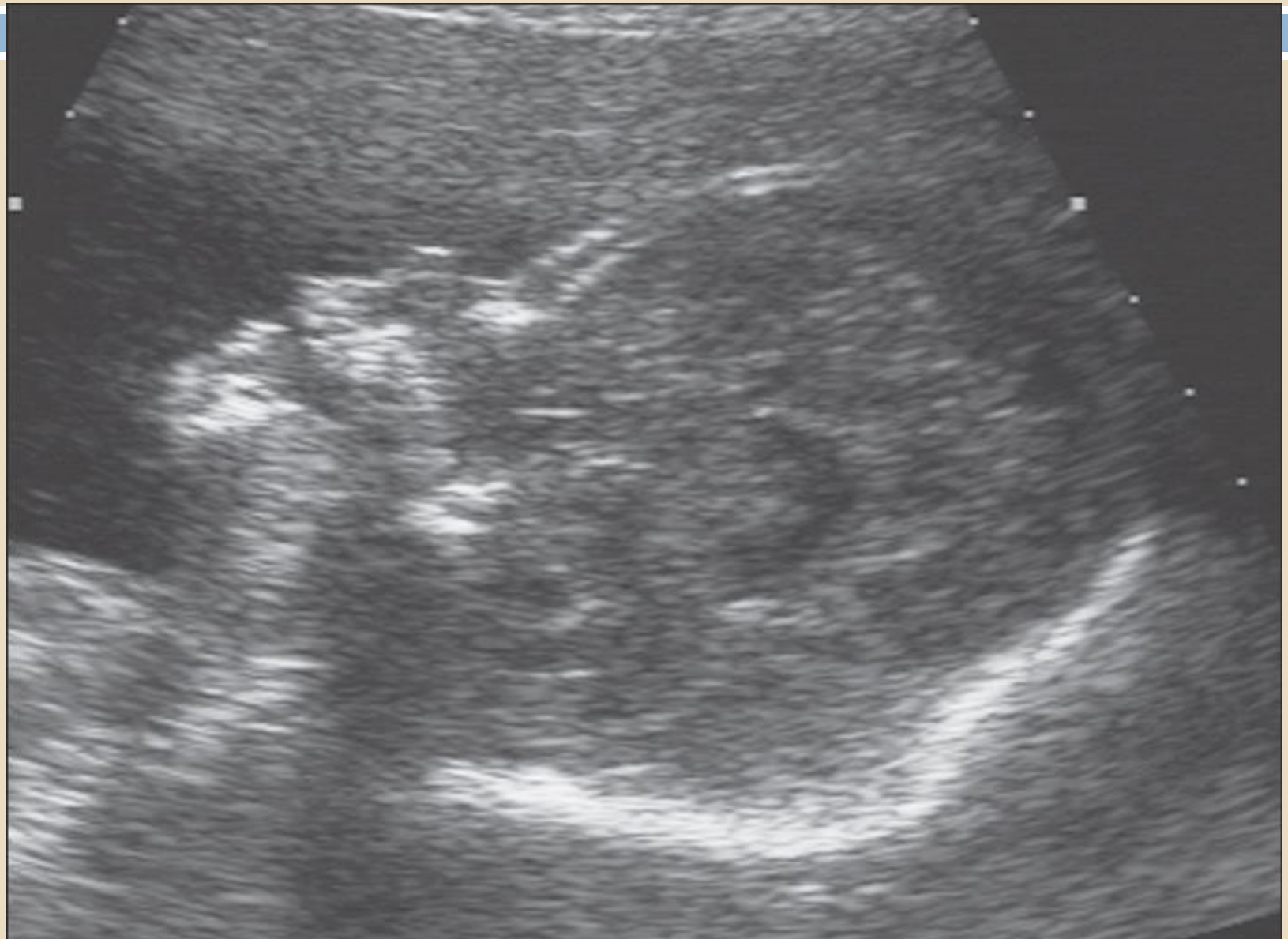
Forehead

- Appreciated by evaluation of the profile
- Series of mid-sagittal scans through the face
  - (frontal bone) appears as a curvilinear surface with differentiation of the forehead, nose, lips, and chin
- View allows diagnosis of anterior cephaloceles
  - Arise from the frontal bone
  - May cause widely spaced orbits
    - (Hypertelorism)

Skull

- Cloverleaf skull
  - Appears as an unusually misshapen skull with a cloverleaf appearance
  - Has been associated with
    - Skeletal dysplasias (dwarfism)
    - Ventriculomegaly
- Any irregularities in the contour of the forehead
  - Search for other malformations

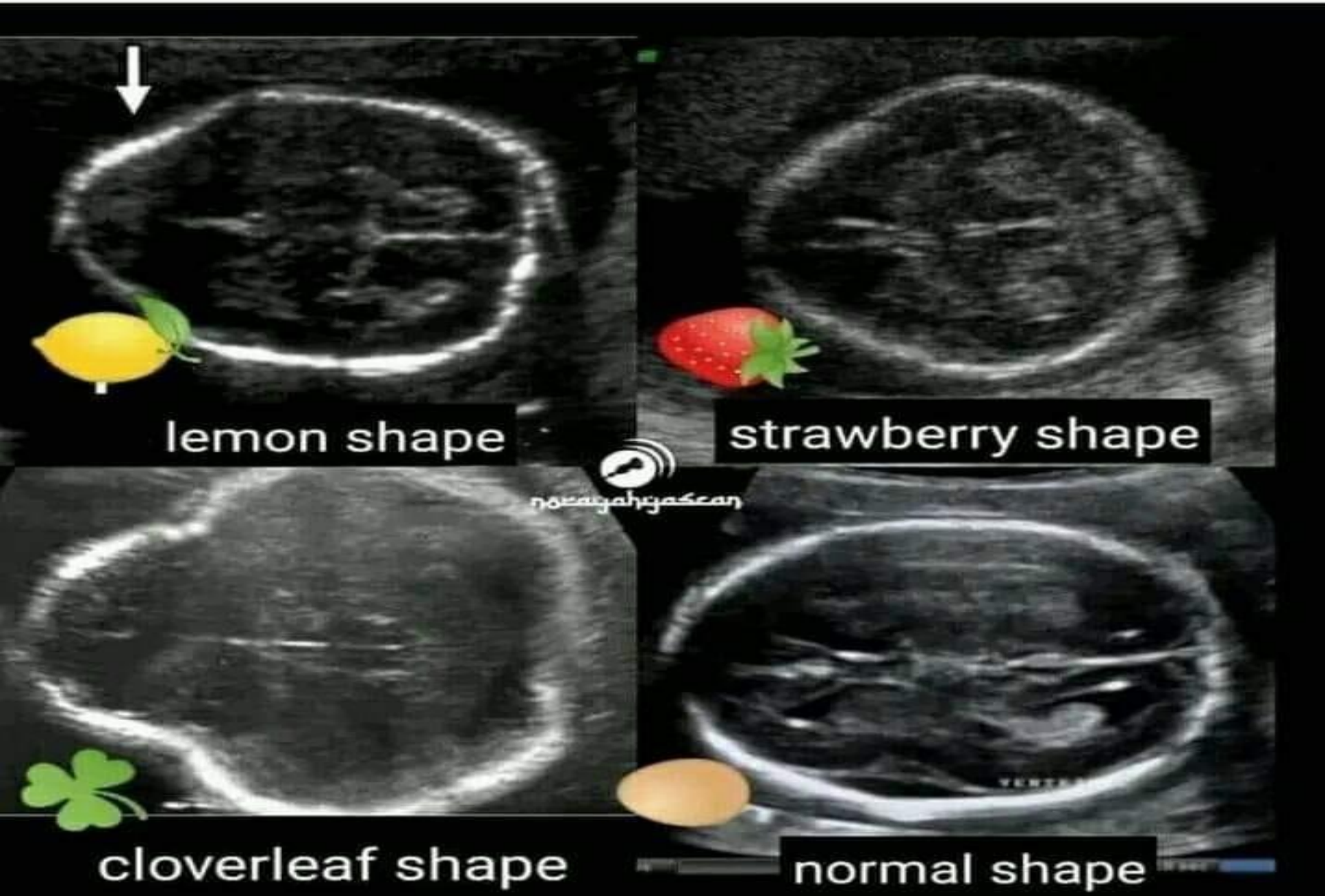
Cloverleaf skull



Skull

- Frontal bossing may be observed in a fetus
  - With a lemon-shaped skull
    - (from spina bifida)
  - When there is an absent or depressed nasal bridge
- Fetus with a strawberry-shaped cranium
  - May be bulging of the frontal bones

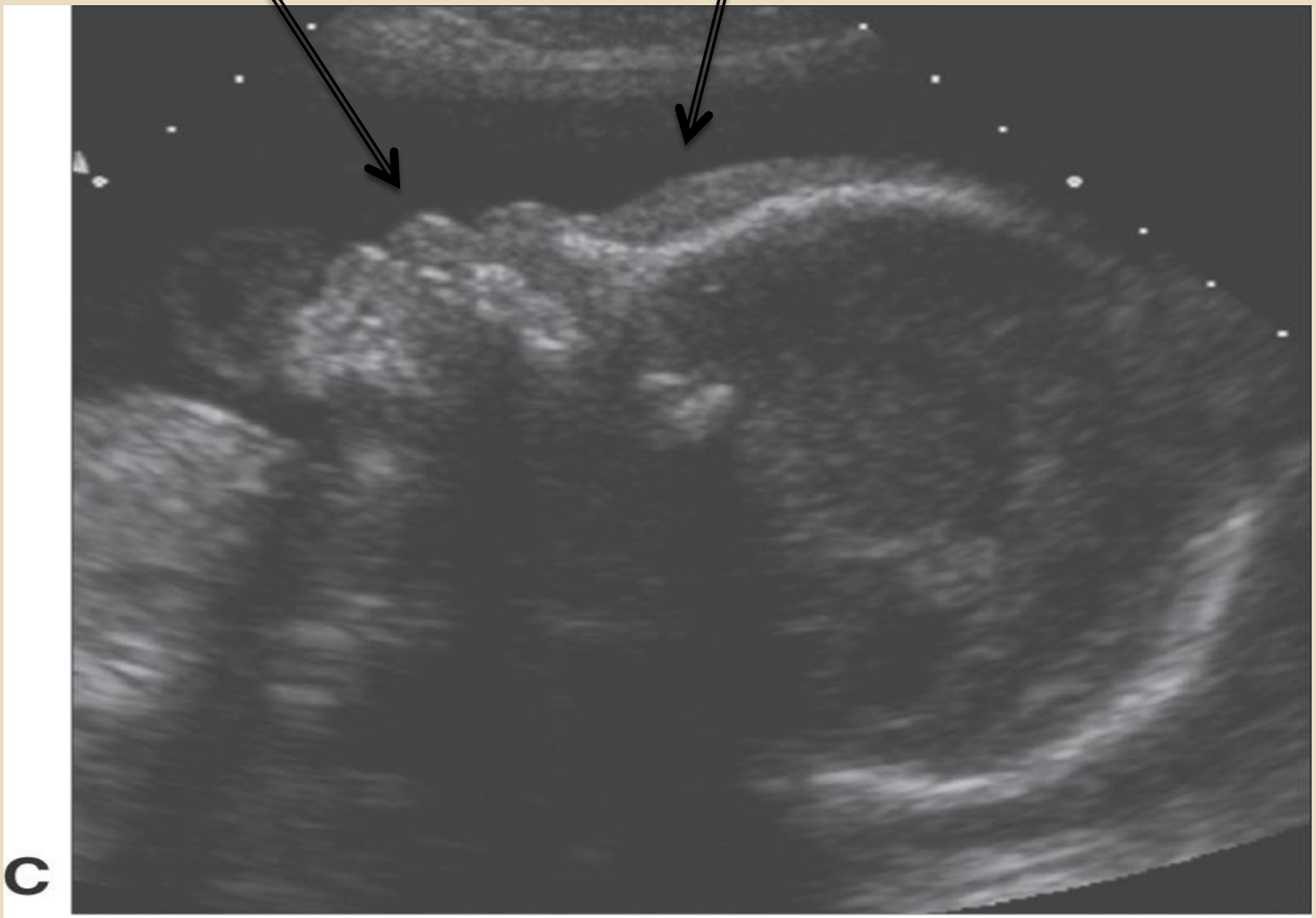
Fetal head anamolies with different shapes



Maxillary or Midface Hypoplasia

- Depressed or absent nasal bridge is an underdevelopment of the middle structures of the face
- Defect is easily noted with coexisting frontal bossing
- Nasal bone may be small or absent with some chromosome anomalies
  - Especially trisomy 21

Hypoplasia with Frontal Bossing

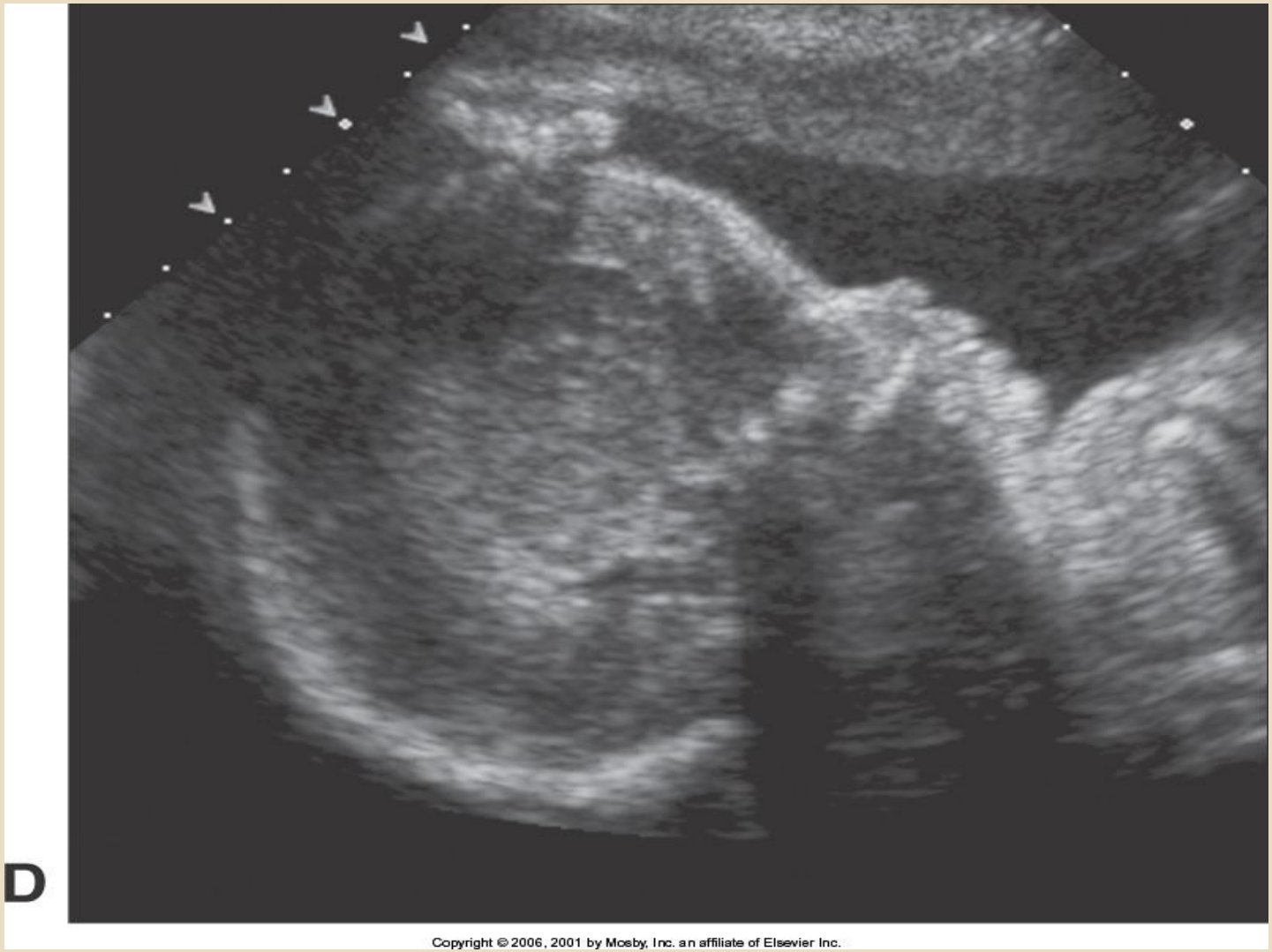


Midface Hypoplasia

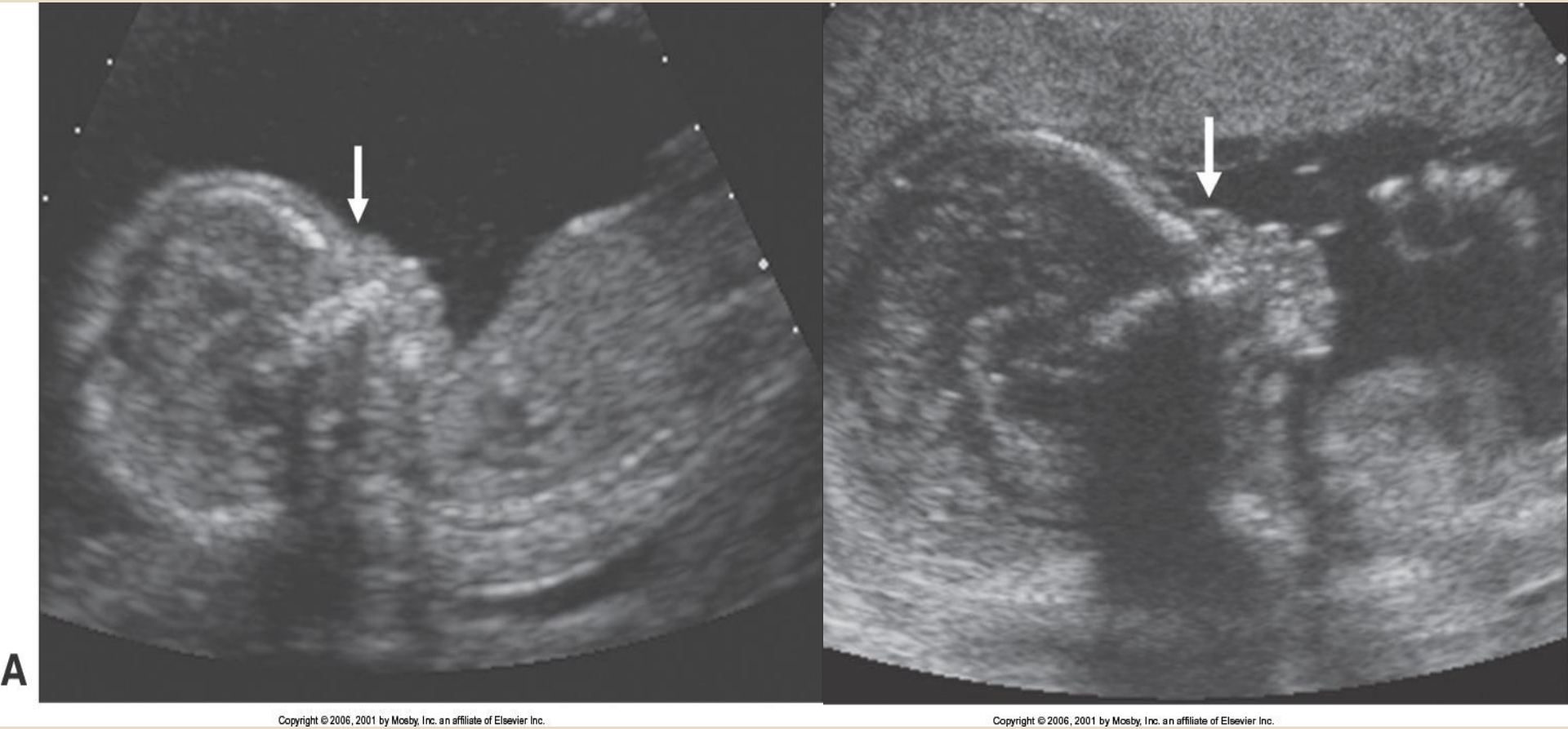
Trisomy 21
Down's
Syndrome

Progeroid Syndrome

Small Facial Profile Compared With Head



Trisomy 21



Absent Nasal Bone

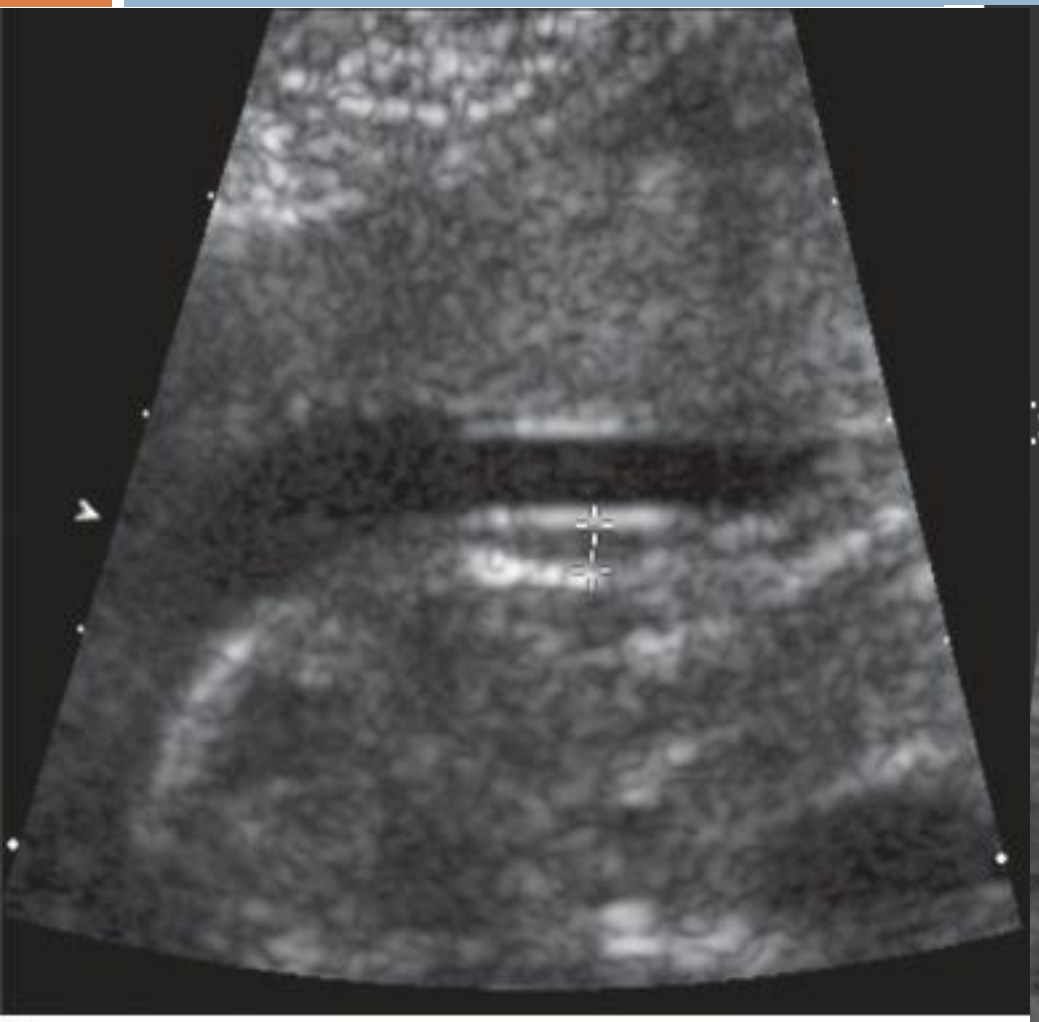
Frontonasal Dysplasia

- Median cleft face syndrome consisting of a range of midline facial defects involving
  - Eyes
  - Forehead
  - Nose
- Abnormalities include
  - Ocular hypertelorism
  - Broad nasal bridge
  - Midline defect of the frontal bone
- By ultrasound - primary finding is hypertelorism
- If one cranial abnormality is found
  - Look for additional ones

Nuchal Area

- Association of first trimester fetal nuchal lucency with aneuploidy is well established
- Is dependent on the size and extent of the nuchal abnormality
- Fetuses with diffuse fetal nuchal fluid and hydrops involving the fetal torso
  - Have a high prevalence of chromosomal anomalies and poor outcome

Greater than 3 mm is abnormal



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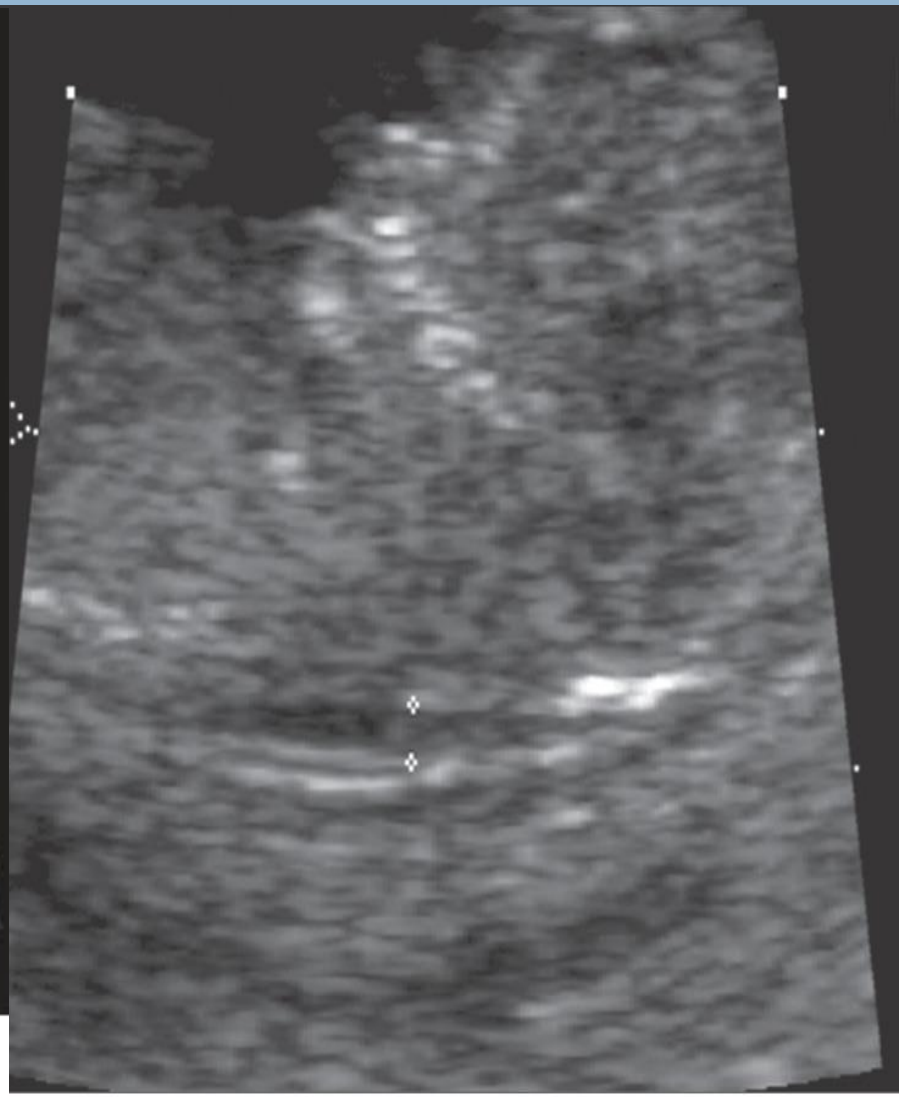


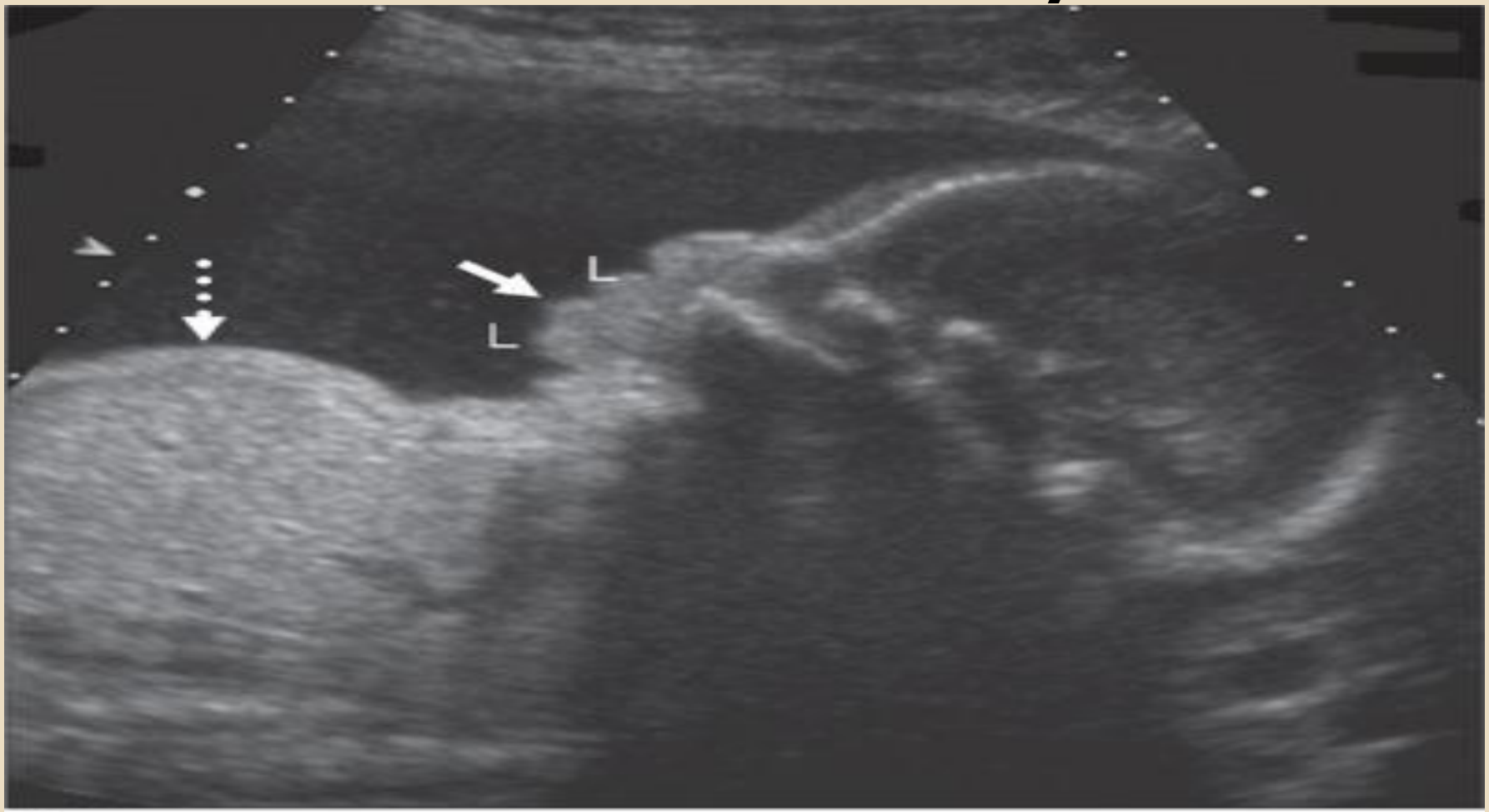
Nose and Upper Lip

- Masses of the nose and upper lip may distort the facial profile and indicate a cleft lip
- To exclude a cleft lip and palate look for
  - Nostril symmetry
  - Nasal septum integrity
  - Continuity of upper lip
- Tumors may disrupt facial contours
  - Epignathus
  - Teratoma

Tongue

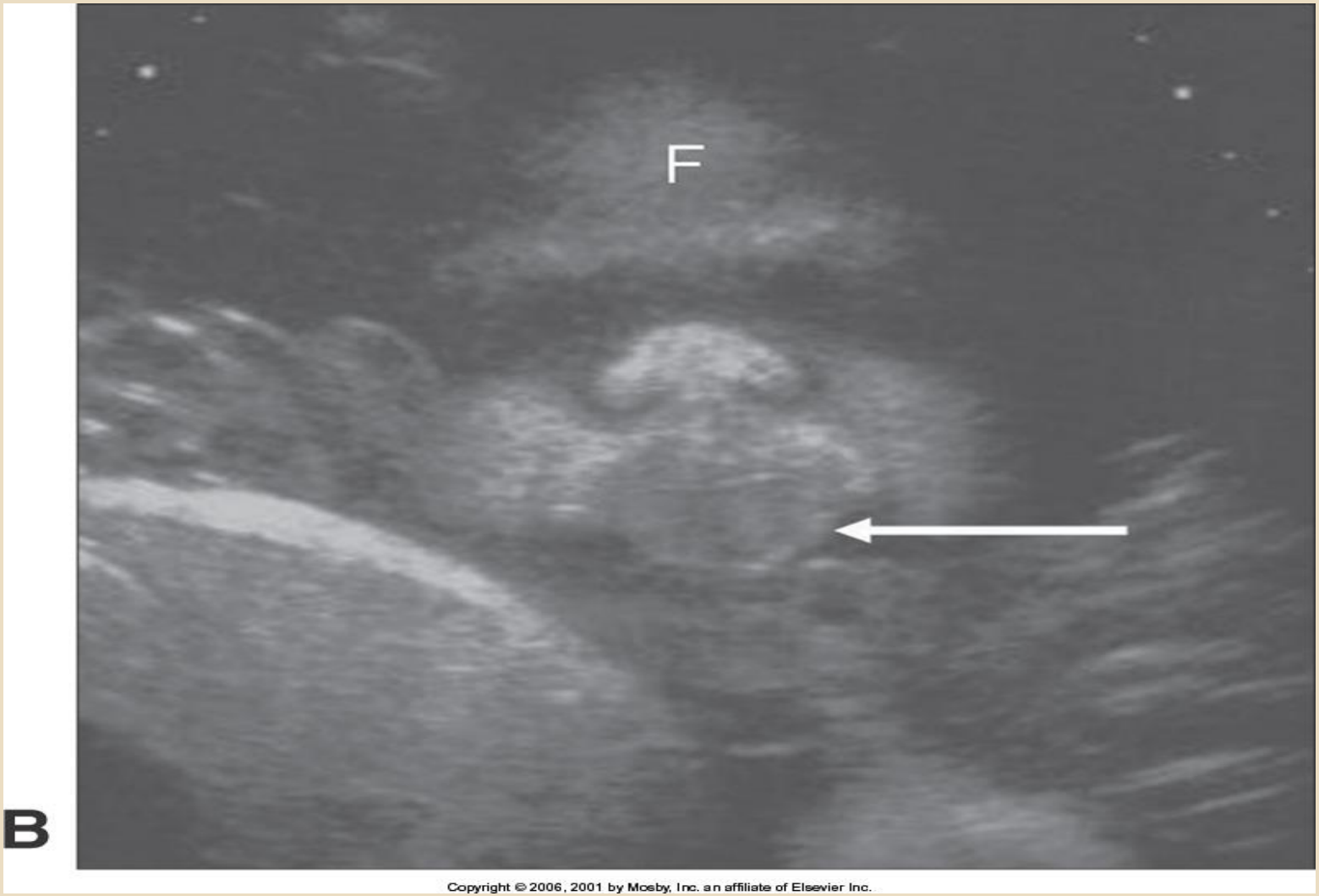
- Tongue protrusion may suggest *macroglossia* (enlarged tongue)
  - Condition found in *Beckwith-Wiedemann syndrome*
    - Congenital overgrowth of tissues

Beckwith-Wiedemann Syndrome



Tongue protruding from mouth *(arrow)* between lips (*L*) and enlarged fetal liver *(dotted arrow)*

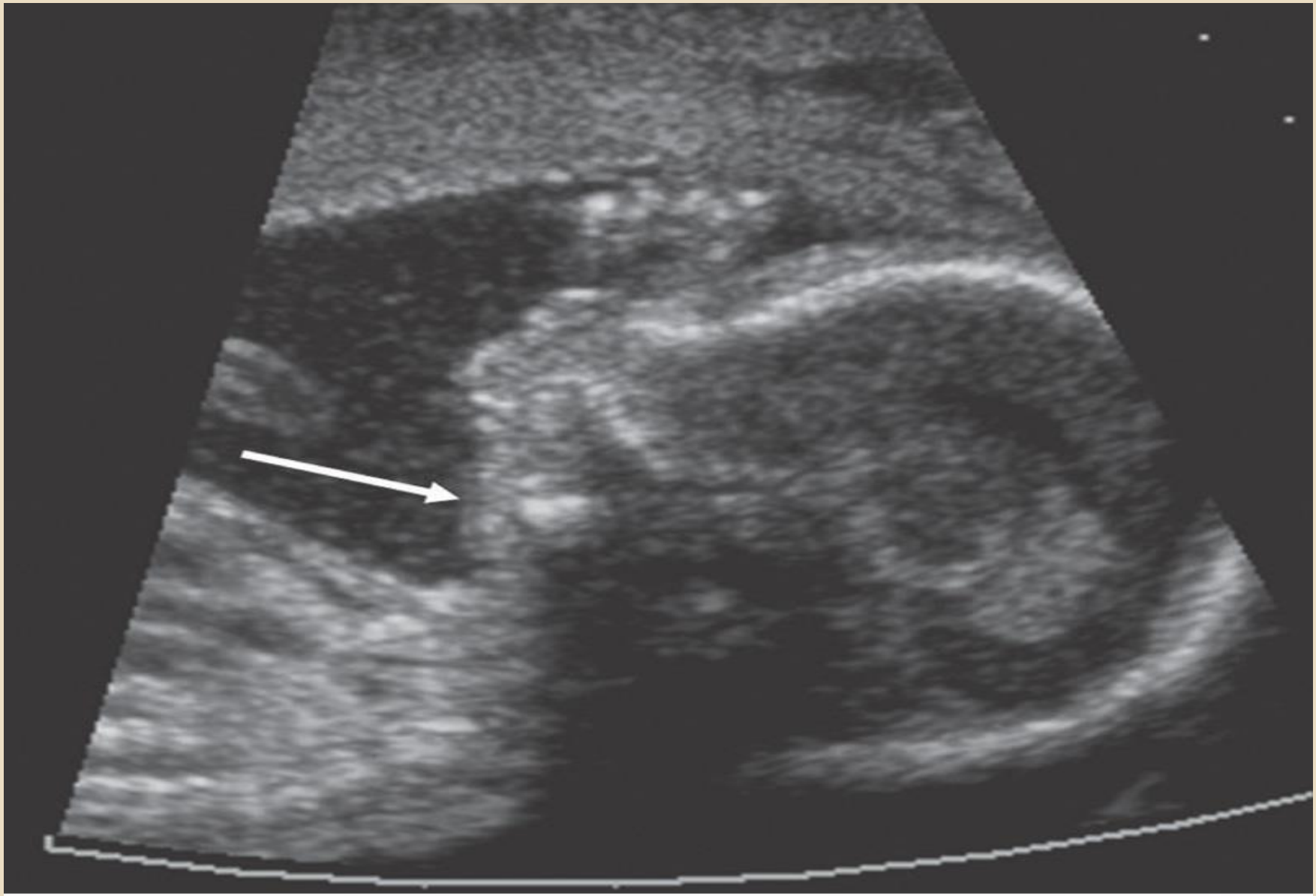
Beckwith-Wiedemann Syndrome



Coronal view of the fetal face demonstrates a protruding tongue suggesting macroglossia *(arrow)*

Mandible

- Congenital micrognathia may be suspected when a small chin is observed
  - Most cases of micrognathia are detected from the subjective appearance of a small chin when imaging the fetal profile
- Abnormally small chin may be so severe that polyhydramnios occurs
  - Because of the inability of the fetus to swallow



Micrognathia

Ear

- Ear malformations
  - Rarely predicted prenatally
    - Low-set ears may be appreciated in a longitudinal view when the placement of the ear appears lower than usual
  - May be observed in Goldenhar's syndrome with anophthalmia (absent eye) and hemifacial microsomia
- May be observed prenatally
  - Small ears (Robert syndrome)
  - Inadequate development of the ear (Nager acrofacial dystocics syndrome)

Coronal plane

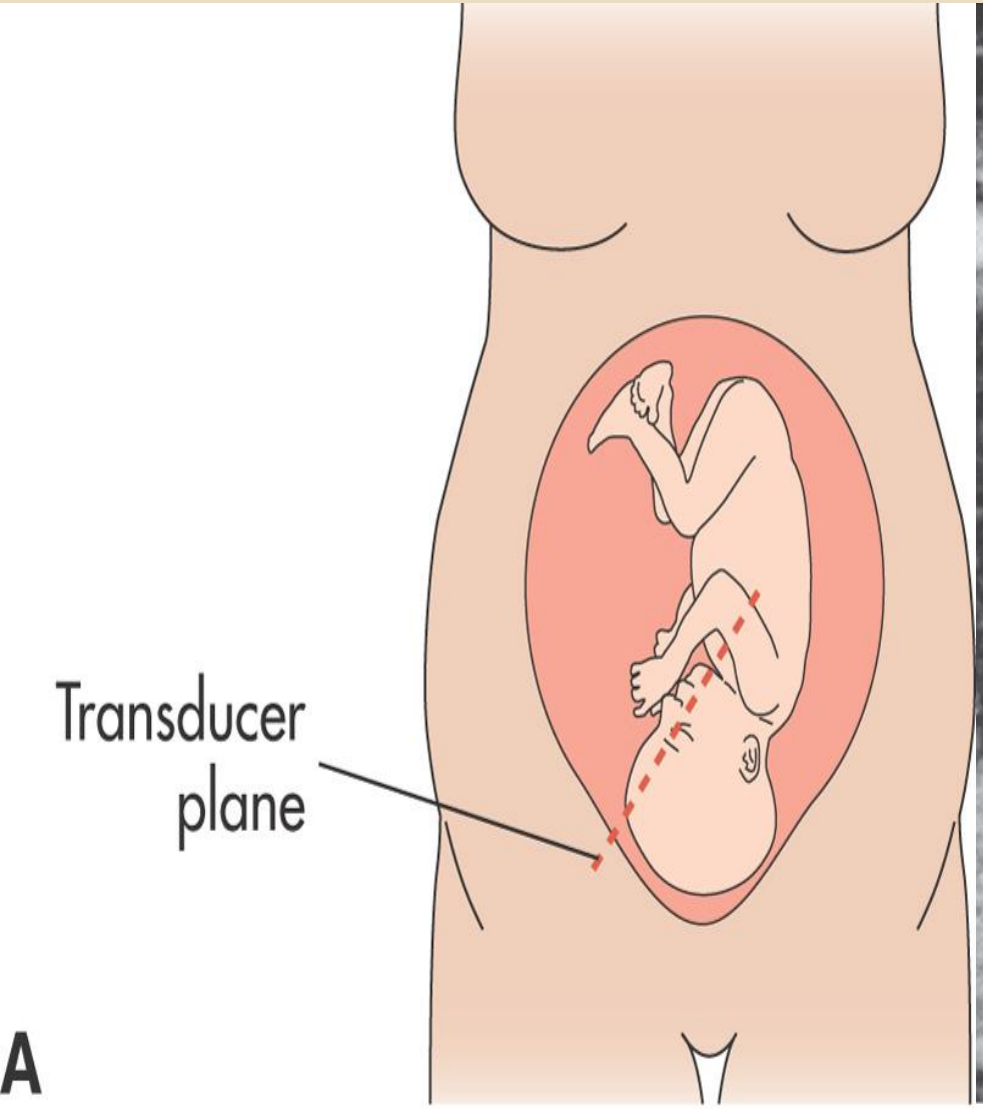
Unusual appearing ear lobe of a posteriorly rotated ear

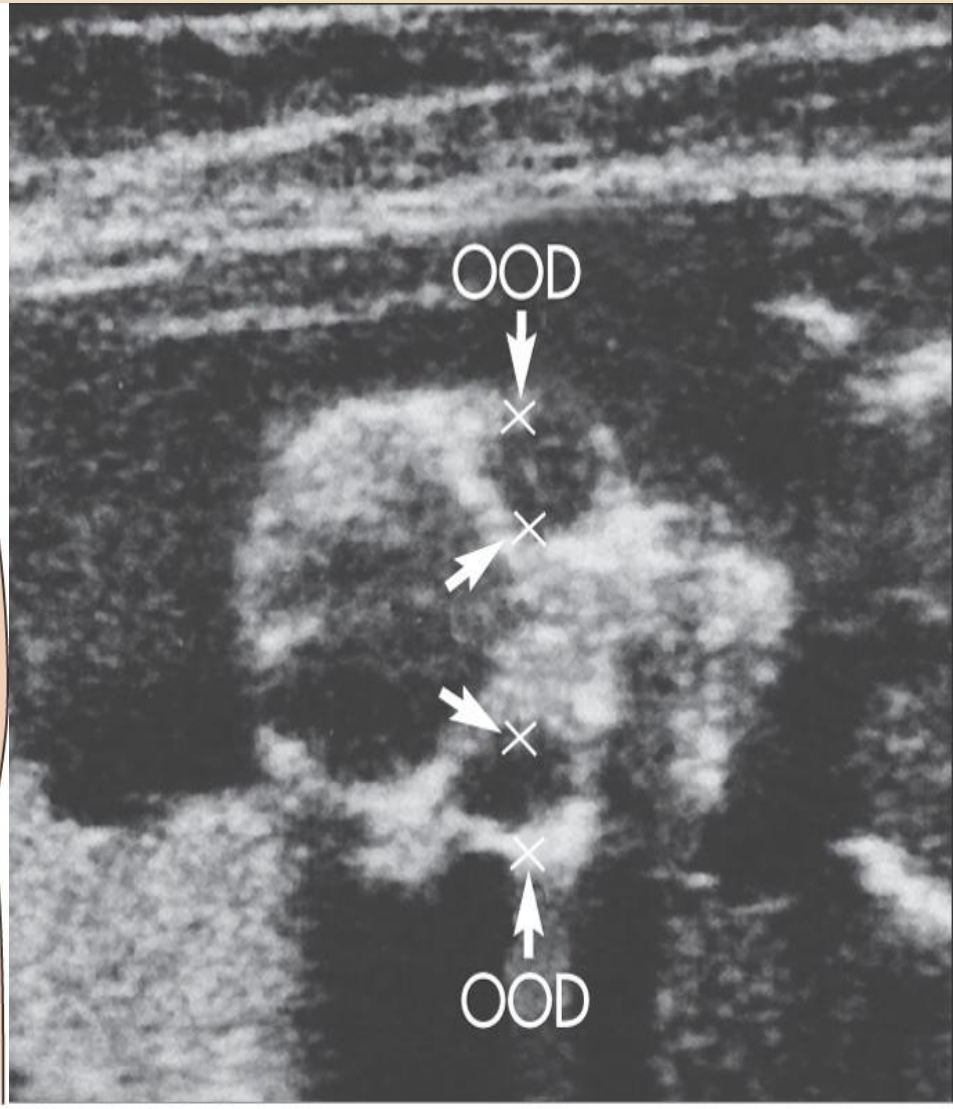
Abnormalities of the Orbits

- Orbital architecture has become increasingly important in the evaluation of craniofacial anomalies
- Must document the presence of both eyes and assess the overall size of the eyes to exclude
  - *Microphthalmia* (small eyes)
  - *Anophthalmia* (absent eyes)

Abnormalities of the Orbits

- Orbital distance measurements are helpful in the diagnosis of
  - Hypotelorism (eyes too close together)
  - Hypertelorism (eyes too far apart)
- Both of these conditions are associated with other anomalies

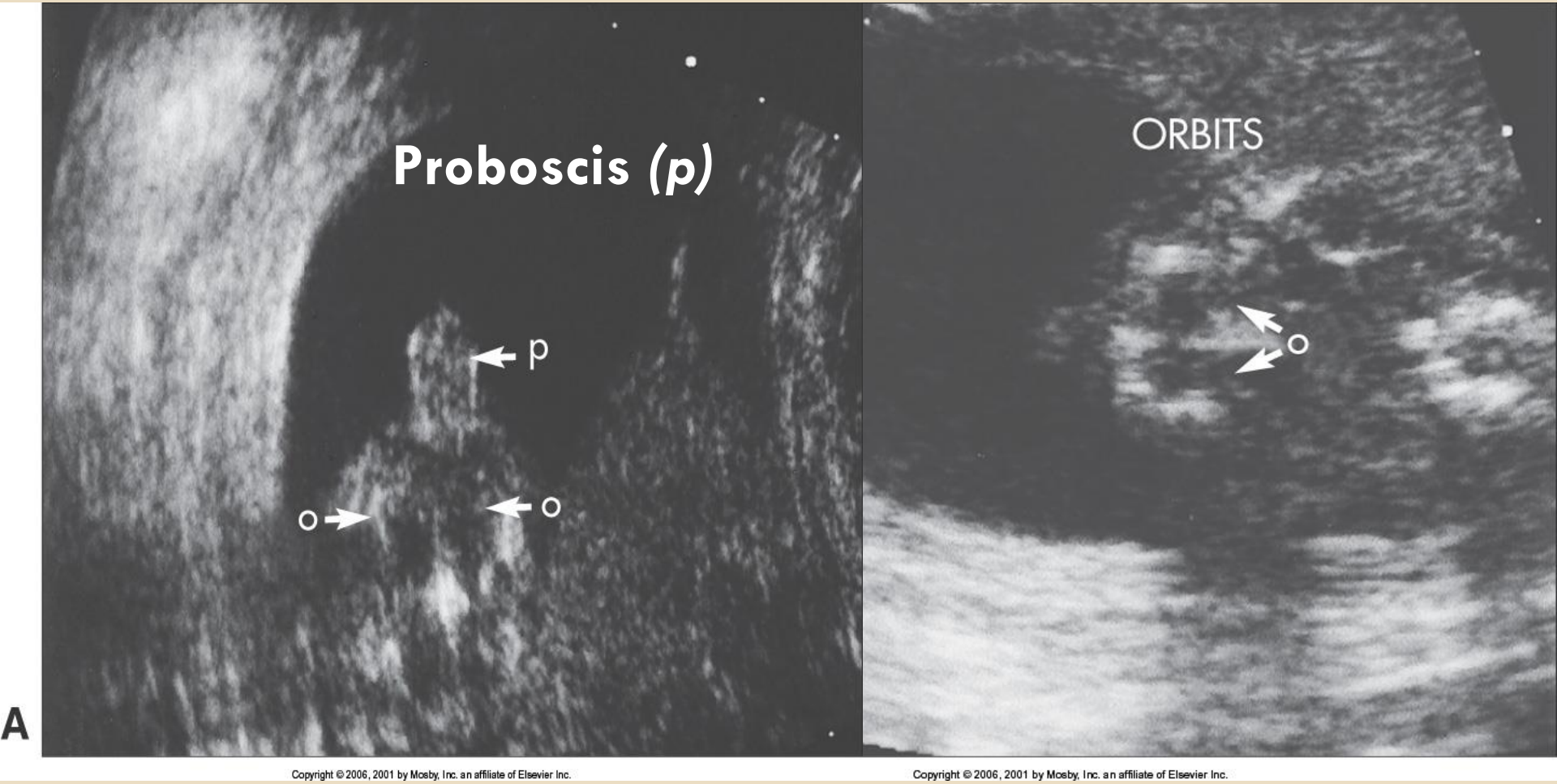




Hypotelorism

- Characterized by a decreased distance between the orbits
- Associated with several syndromes and other anomalies
- Conditions include
  - Holoprosencephaly
  - Microcephaly
  - Craniosynostoses
  - Phenylketonuria
- Orbital width measurement may identify these fetuses

Hypotelorism



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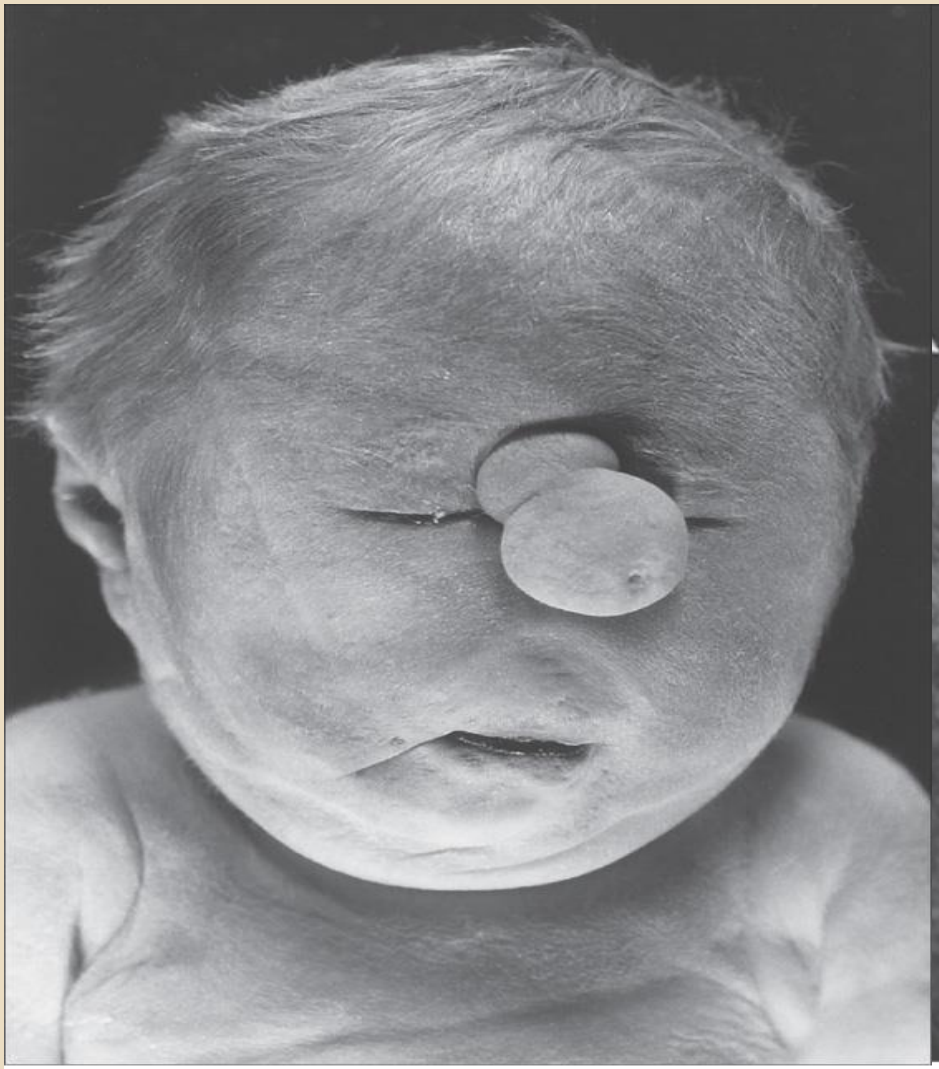
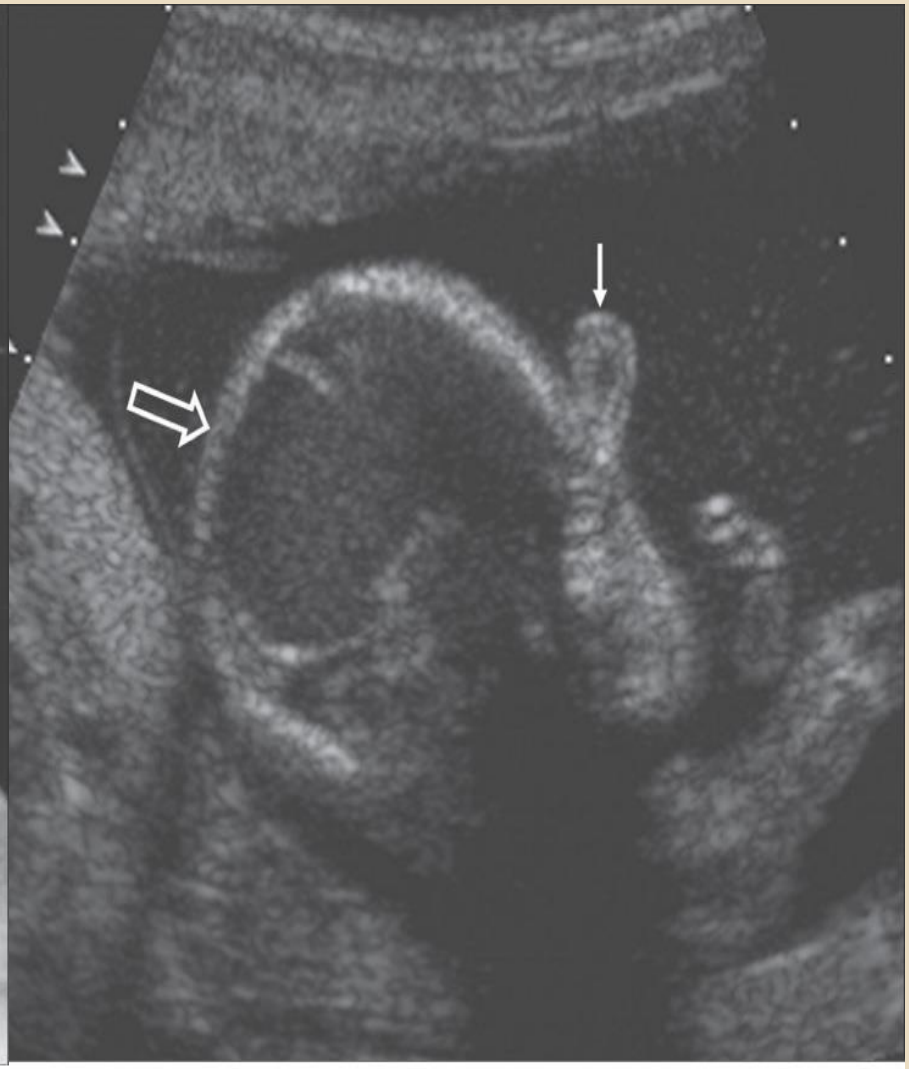
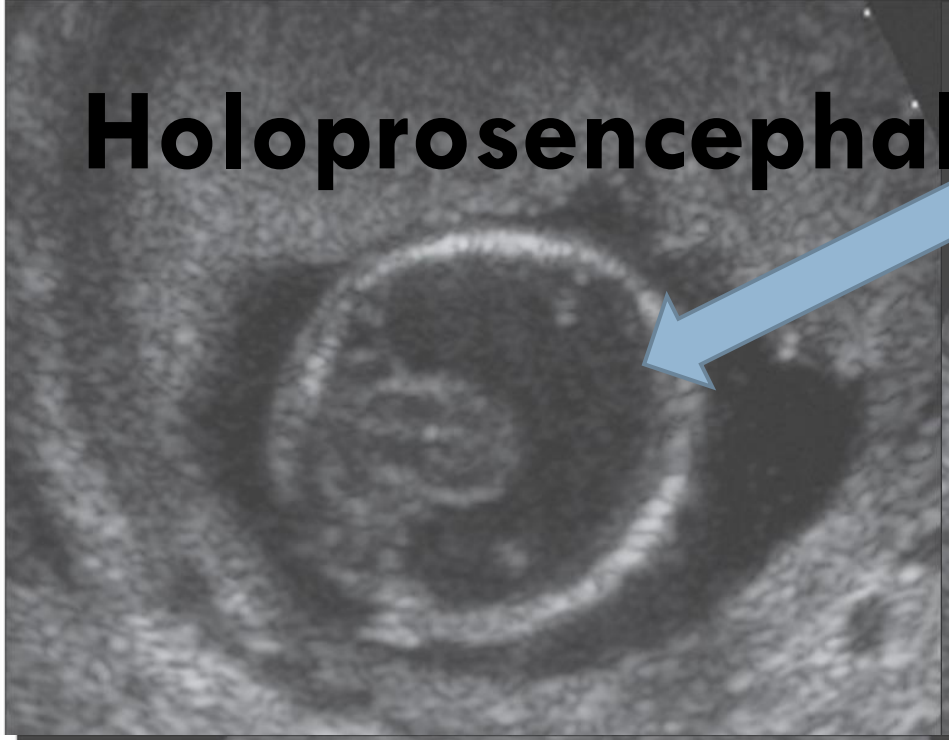
Trisomy 13 was found after delivery

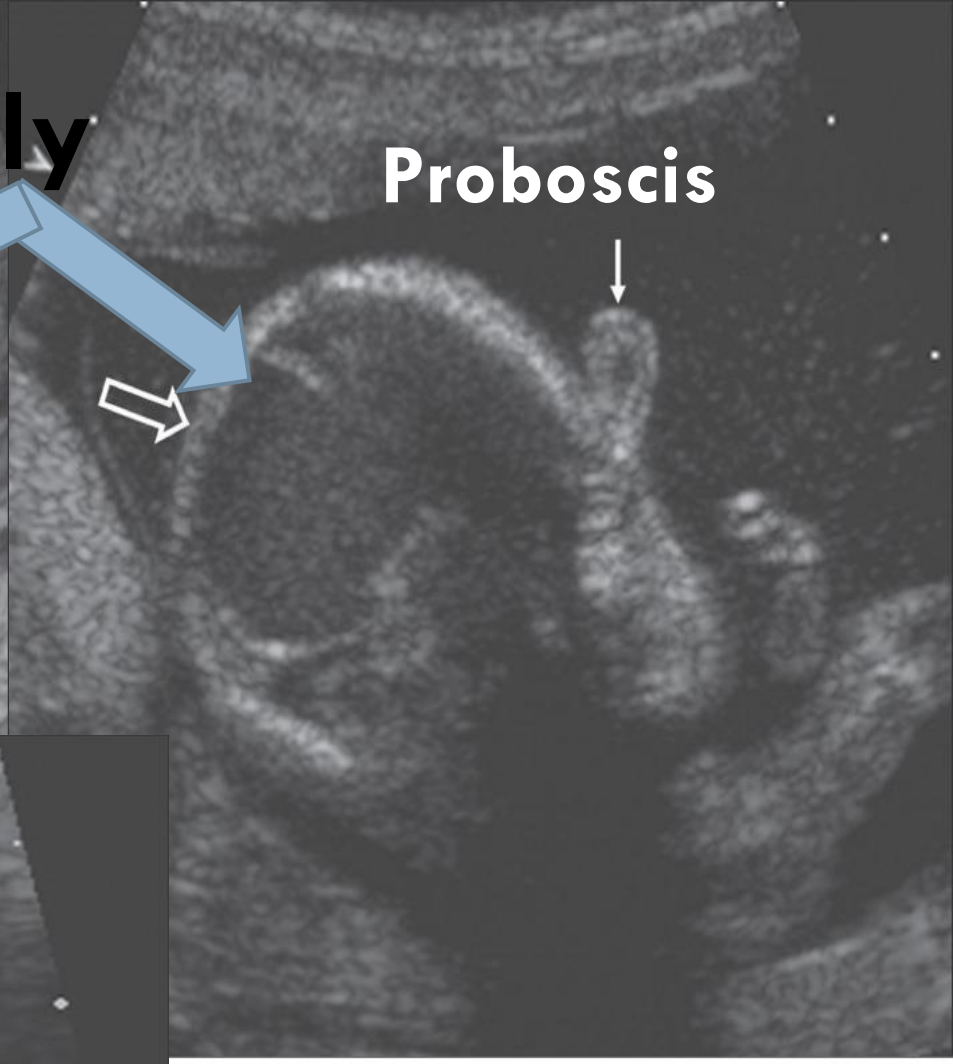
Proboscis

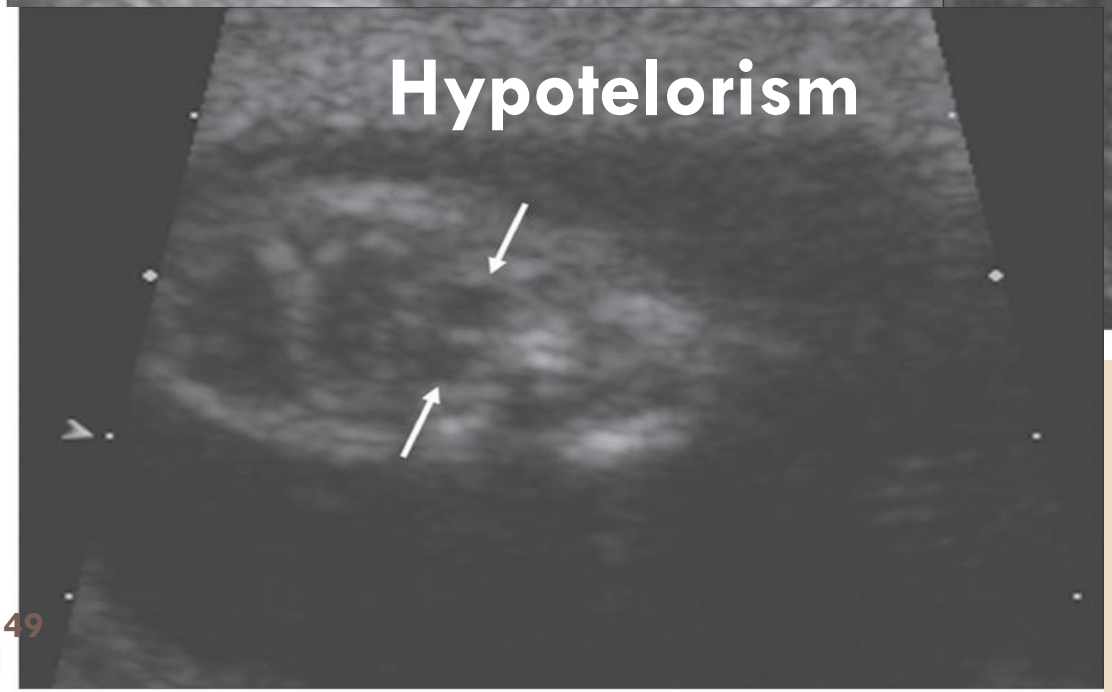




Holoprosencephaly







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Hypertelorism

- Characterized by abnormally widely spaced orbits
- Found in a number of:
  - Abnormal fetal conditions
  - Genetic syndromes
    - Ventriculomegaly may be present
  - Chromosomal anomalies

ABNORMALITIES OF THE NOSE, MAXILLA, LIPS, AND PALATE

- Facial structures may be viewed by placing the transducer in:

  - Lateral coronal plane
    - Nasal structures in relationship to the orbital rings and maxillae is studied
  - Sagittal profile plane
    - This is an important view in assessing the presence or absence of the nose, lips, and chin
  - Modified tangential maxillary view
    - (inferior–superior projection) demonstrates nostrils and nasal septum

- Evaluation of the nasal triad should assess:
  - 1. Nostril symmetry
  - 2. Nasal septum integrity
  - 3. Continuity of the upper lip to exclude cleft lip and palate
- Do not mistake the normal nostrils or mucous membrane for a cleft

Lip and Palate

- **Medial cleft lip**
  - Caused by incomplete merging of the two medial nasal prominences in the midline
- **Oblique facial cleft**
  - Failure of maxillary prominence to merge with the lateral nasal swelling, with exposure of the nasolacrimal duct
- **Complete bilateral cleft lip and palate**
  - Large gap in upper lip on modified coronal view; nose is flattened and widened

Lip and Palate

- **Unilateral complete cleft lip and palate**
  - Incomplete fusion of maxillary prominence to the medial prominence on one side; modified coronal view
- **Incomplete cleft**
  - Nose is intact; modified coronal view of lip

Lip and Palate

- Cleft lip with or without cleft palate
  - Most common congenital anomaly of the face
- Cleft lip occurs because of failure of fusion of
  - Primary palate
  - Secondary palate
    - Results in a clefting defect coursing anteriorly through the upper lip
- Cleft palate occurs when the lateral palatine processes fail to fuse in the midline
- Cleft lip and palate occur together when both fusions are absent

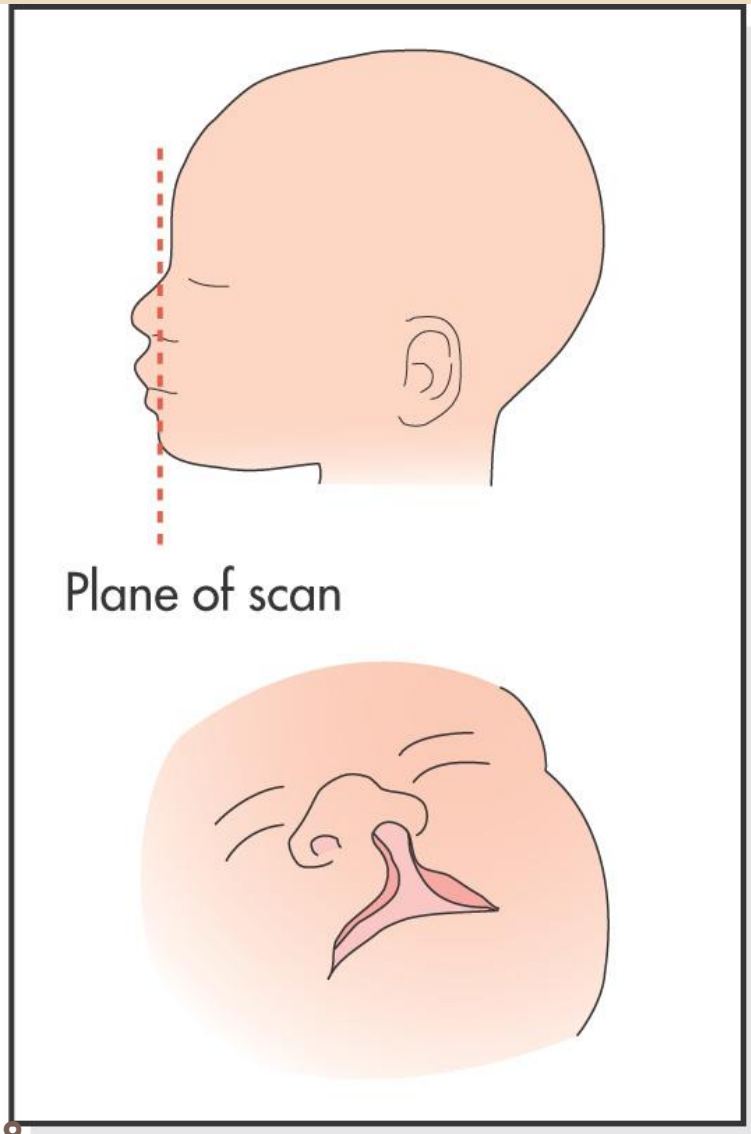
Lip and Palate

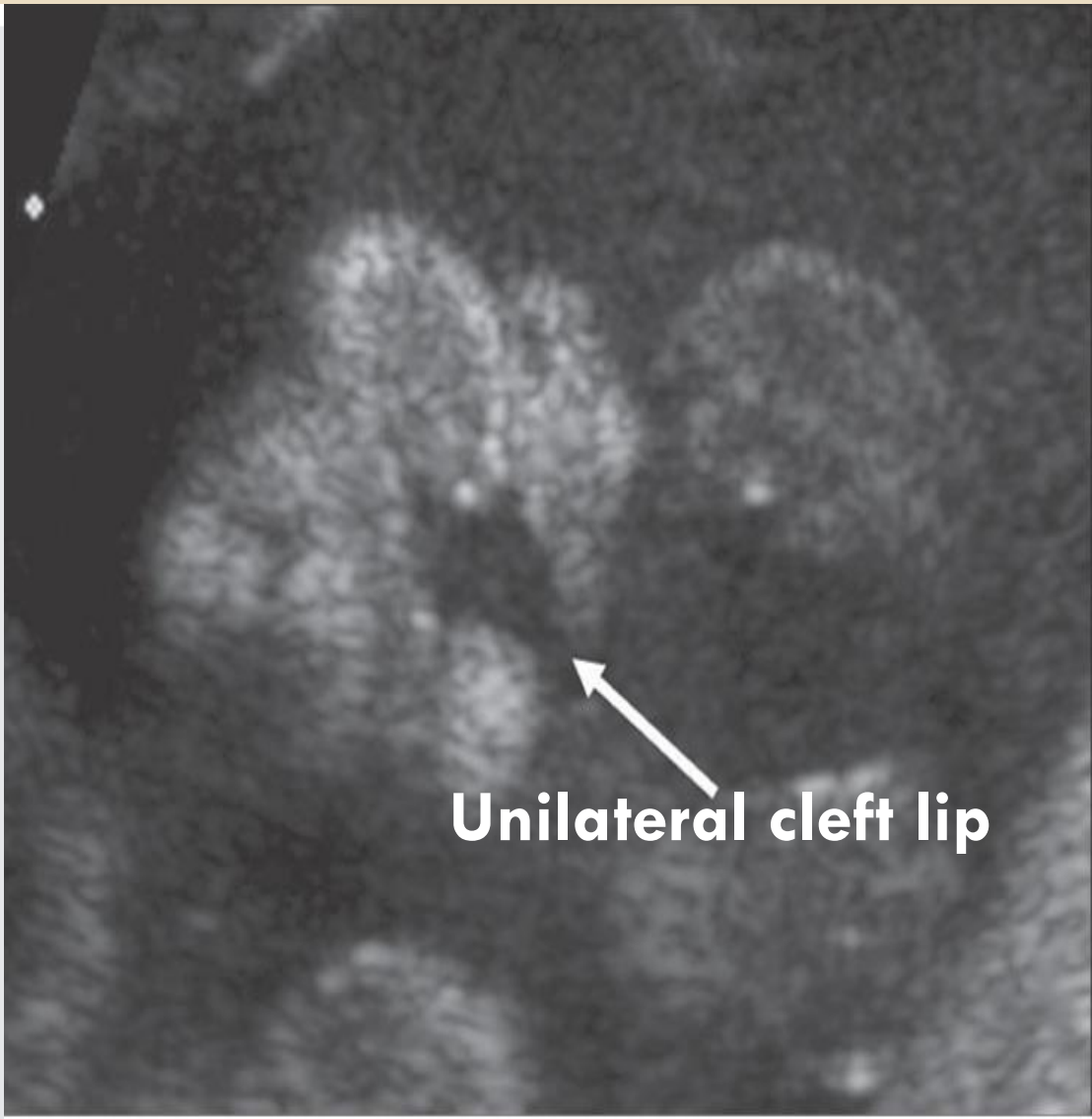
- Clefts may be unilateral or bilateral
  - May occur
    - Singular
    - In association with other anatomic abnormalities
- Frequency of cleft lip with or without a cleft palate shows ethnic variation
  - 1 in 3000 births in African Americans
  - 1 in 600 births in Caucasians
  - 1 in 350 births in Asians
  - 1 in 150 to 1 in 250 births in Native Americans

Lip and Palate

- Sonographically
  - Visualization of the hard and soft palates is a challenge because of considerable bony shadowing
- Use a systematic approach to examine the fetal face for clefts in the coronal and axial planes
- 3-D ultrasound may help to image the palate and lip

Modified coronal plane





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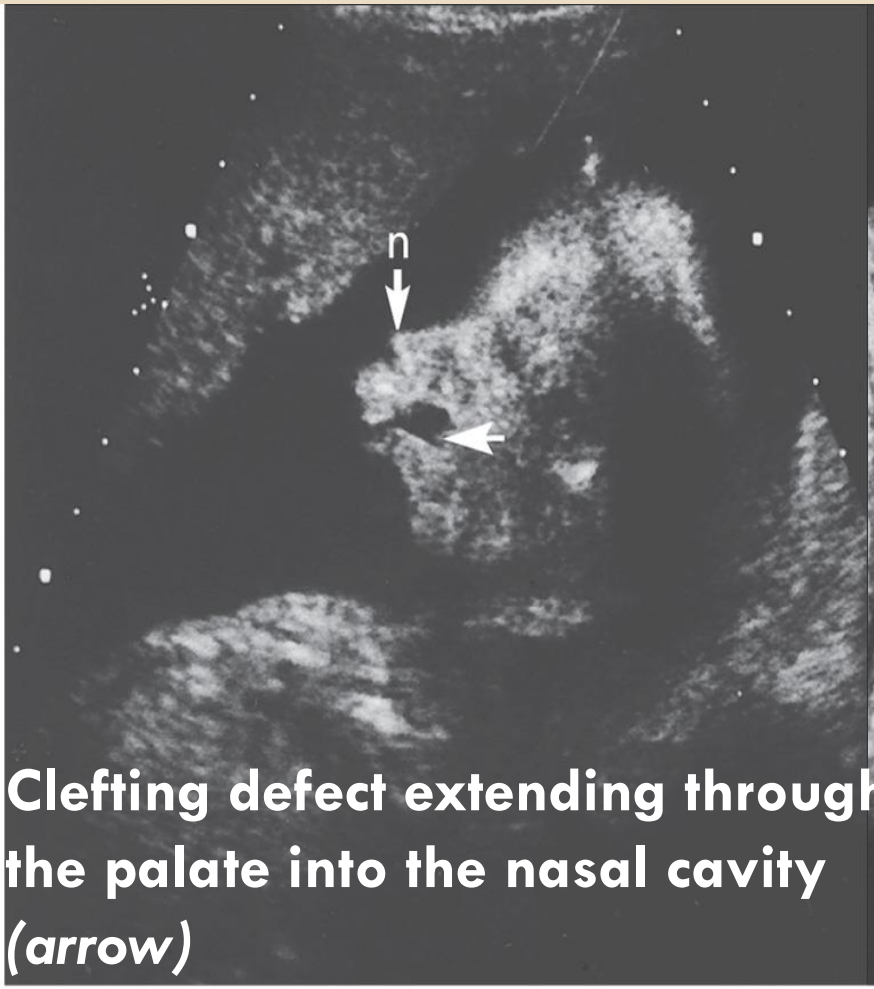
Unilateral cleft lip



Clefting defect extending through the palate into the nasal cavity *(arrow)*

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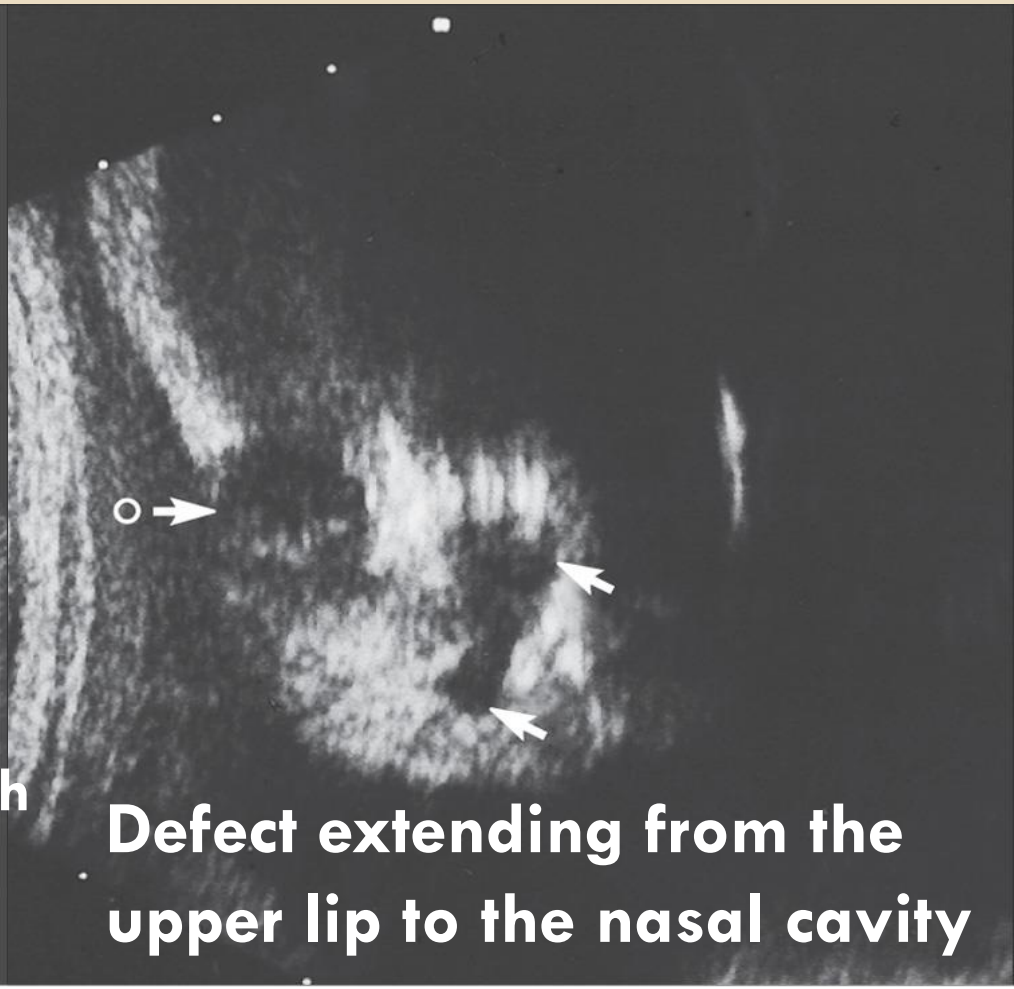
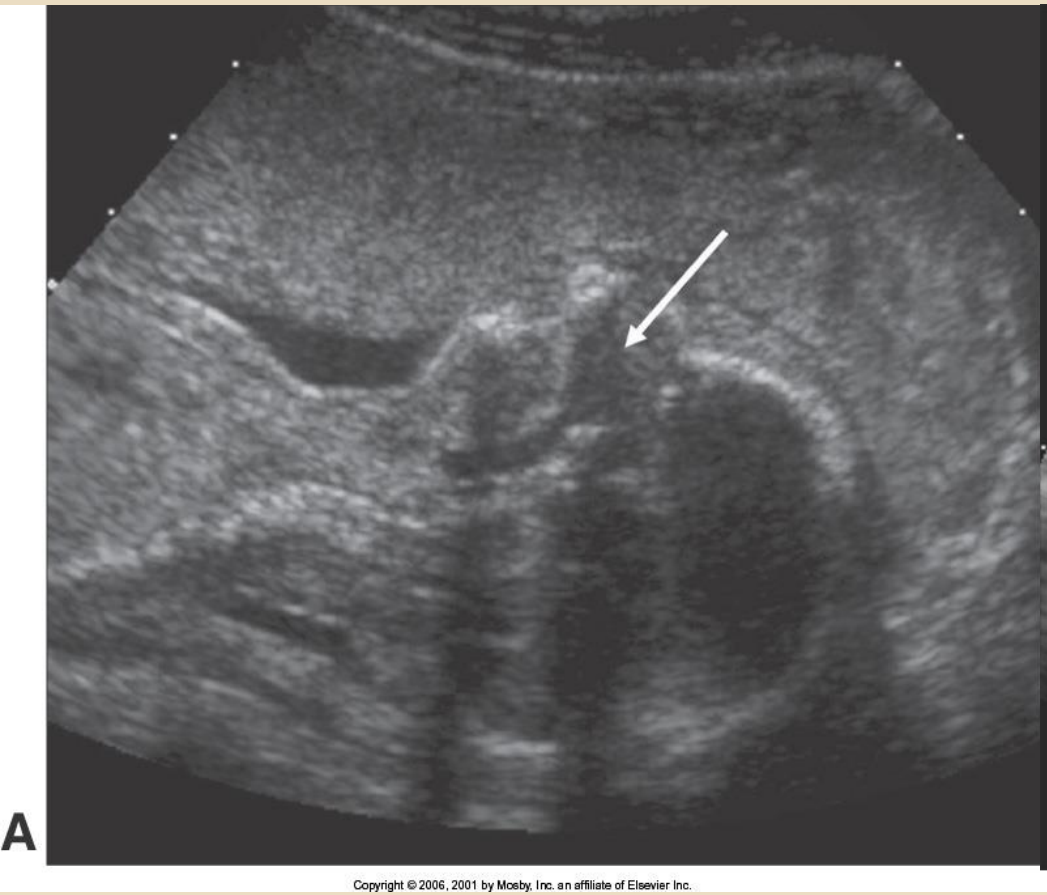
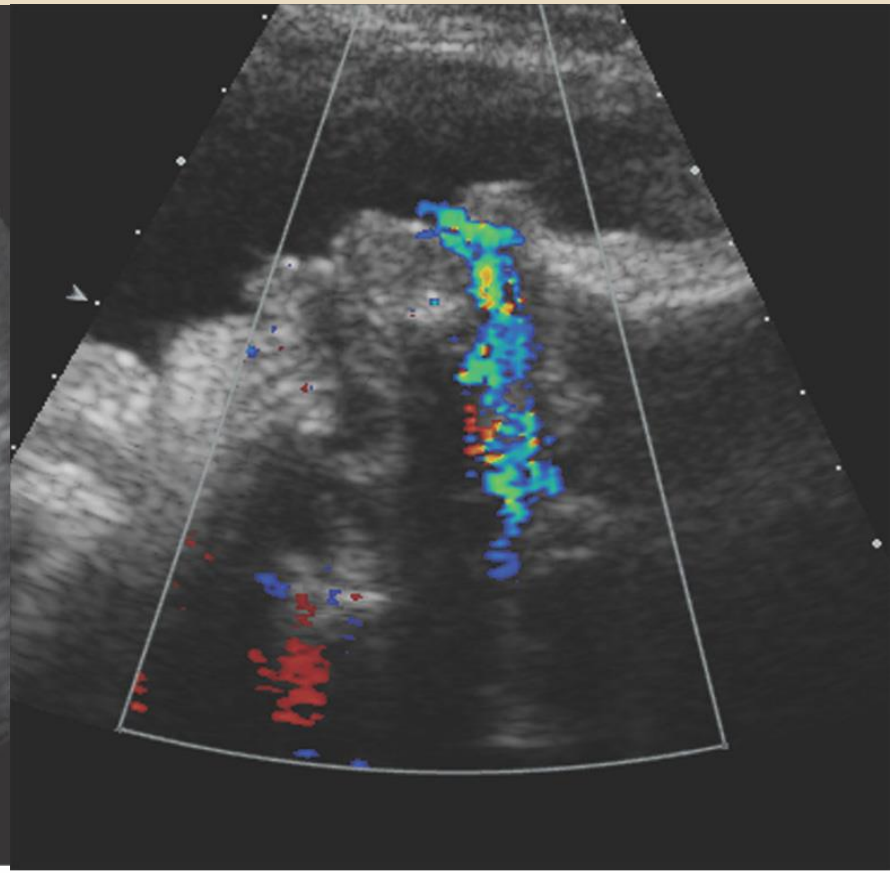


Defect extending from the upper lip to the nasal cavity

Median facial cleft seen with holoprosencephaly





Fluid seen in common oropharynx-nasopharynx area suggesting a cleft palate *(arrow)*

Normal palate demonstrating color only seen in the nasopharynx

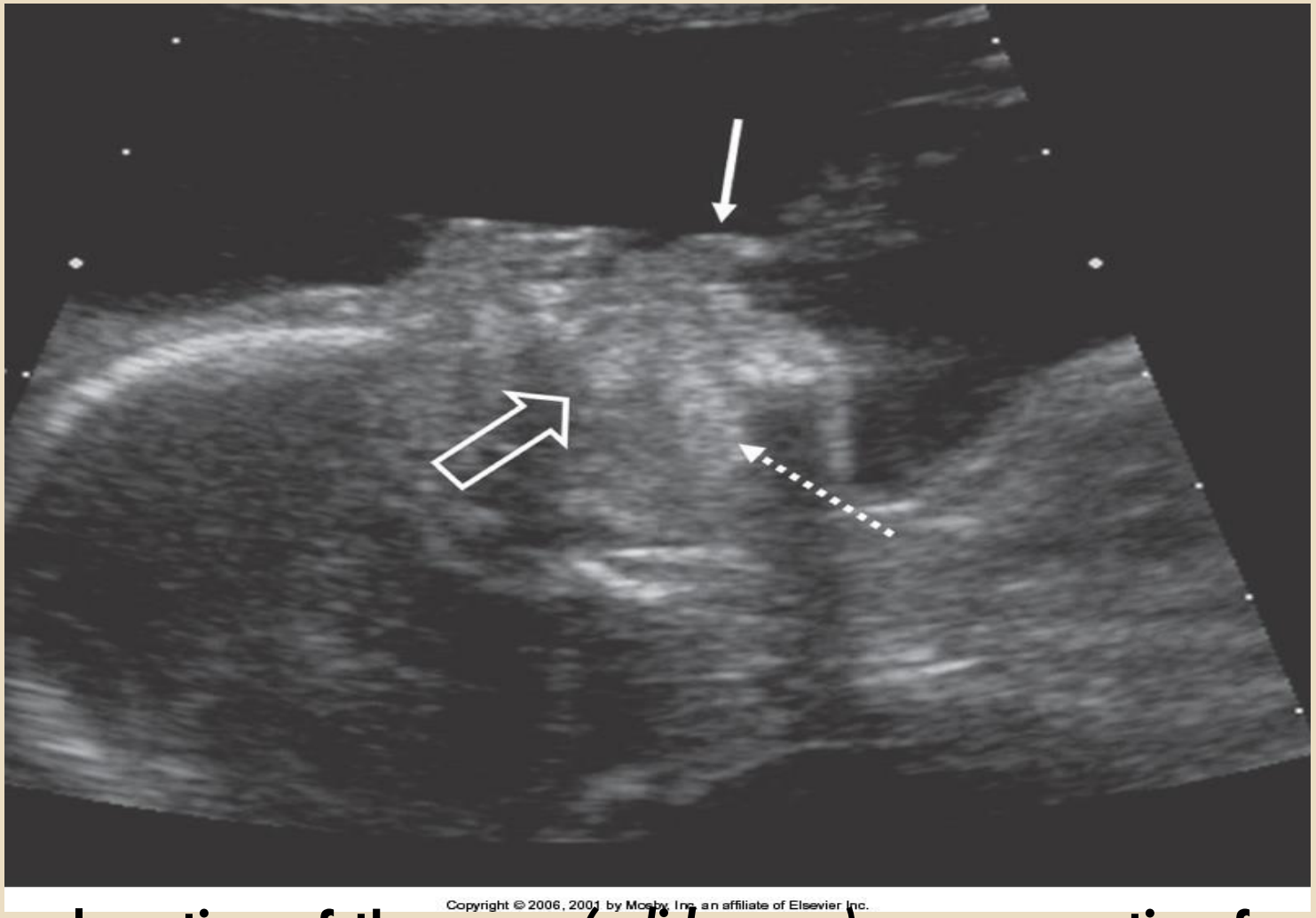
Oral Cavity Abnormalities

- Few congenital malformations of the oral cavity
- Normal fetus may exhibit
  - Swallowing
  - Protrusion and retrusion of the tongue
  - Hiccoughing
- Abnormal positioning of the tongue may be indicative of
  - Mass
  - Obstructive process
  - Macroglossia (large tongue in Beckwith-Wiedemann syndrome)

Epignathus

- A teratoma located in the oropharynx
- Masses may be highly complex and contain
  - Solid
  - Cystic
  - Calcified components
- Swallowing may be impaired
  - Resulting in polyhydramnios
  - In these cases a small stomach may be present

Sagittal view of a fetus with a small epignathus



External portion of the mass *(solid arrow)*, mass erupting from the maxilla *(open arrow)*, and tongue compressed against the lower jaw *(dotted arrow)*

Neck Abnormalities

- Congenital anomalies of the neck are rare
  - When present may represent life-threatening disorders
- Masses are usually large and obvious because their presence causes distortion of the neck contour and adjacent structures
- Most common mass
  - Cystic hygroma colli
    - (lymphatic obstruction)

Neck Abnormalities

- Rarer lesions include
  - Cervical meningomyelocele
  - Hemangiomas
  - Teratomas
  - Goiter
  - Sarcoma
  - Metastatic adenopathy

Neck Abnormalities

- Clinically, a fetal neck mass is cause for concern
- When a large tumor exists
  - Delivery is complicated
    - Tumor may cause
      - Delivery dystocia
        - (inability to deliver the trunk once the head has been delivered)
      - Obstruction of the airway
        - (requires immediate intubation)

Neck Abnormalities

- Goiter
  - Suggests that the mother may have hypothyroidism
- Cystic hygroma
  - High risk for Turner syndrome (45 X)
  - Other chromosome defects are also associated
  - May also result from heart failure
    - (because of a cardiac malformation)

Cystic Hygroma

- Result from a malformation of the lymphatic system
  - Leads to single or multi-loculated lymph-filled cavities around the neck
- Failure of the lymphatic system to properly connect with the venous system results in distention of the jugular lymph sacs and the accumulation of lymph in fetal tissues
  - Abnormal collection of lymph causes distention of the lymph cavities
    - Causes fetal hydrops and even fetal death

Cystic Hygroma

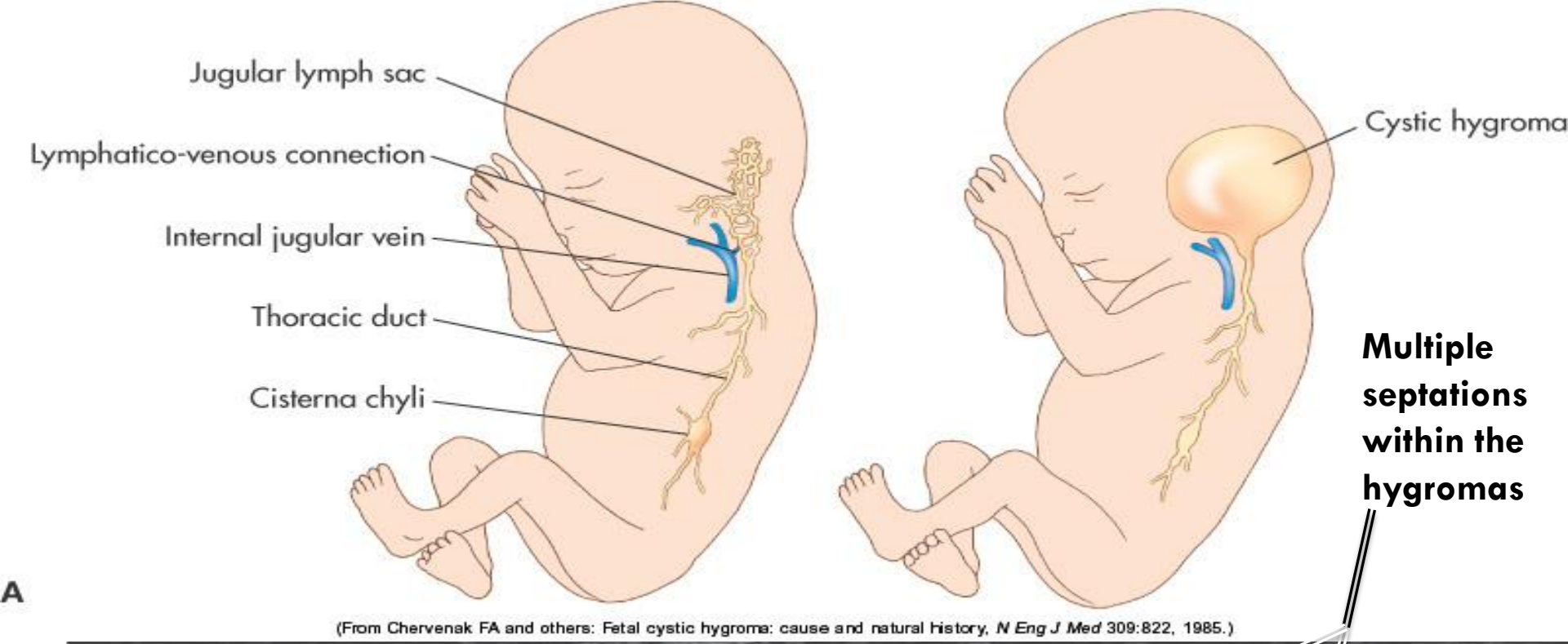
- Developmental defects of the lymphatic vessels
- Cardiac and renal diseases are common
- May present as isolated small cystic cavities with or without septations may arise from
  - Posterior – most common
  - Anterior - rare
  - Lateral - rare

Cystic Hygroma

- Large hygromas have a typical sonographic appearance
  - Bilateral large cystic masses at the posterolateral borders of the neck may surround the neck and head in severe cases
- Typically, a dense midline septum divides the hygroma, with septations noted within the dilated lymph sacs
- Heart failure commonly results in intrauterine death

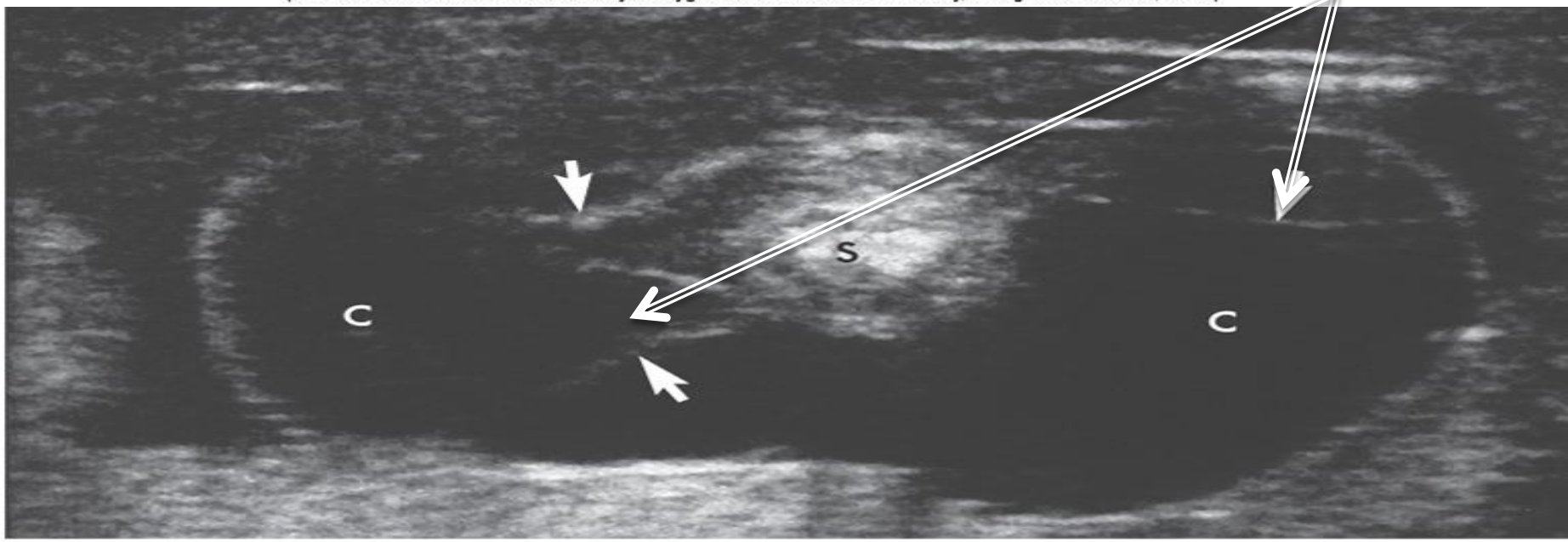
Cystic Hygroma

- Associated with other chromosomal anomalies
- Prognosis for fetuses with hydrops is dismal
- Differential diagnosis includes:
  - Meningomyelocele
  - Encephalocele
  - Nuchal edema
  - Branchial cleft cyst
  - Cystic teratoma
  - Hemangiomas
  - Thyroglossal duct cysts

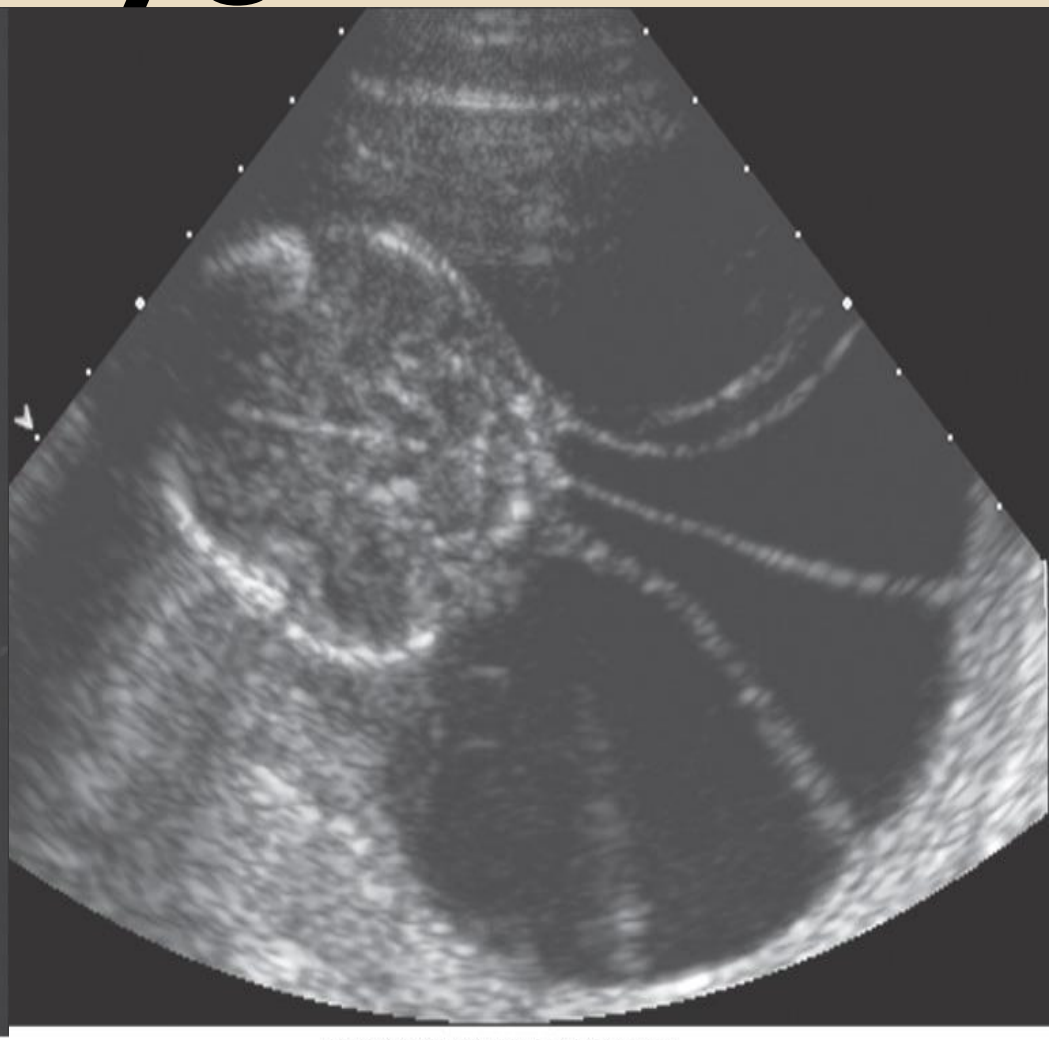


(From Chervenak FA and others: Fetal cystic hygroma: cause and natural history, *N Eng J Med* 309:822, 1985.)

Multiple septations within the hygromas

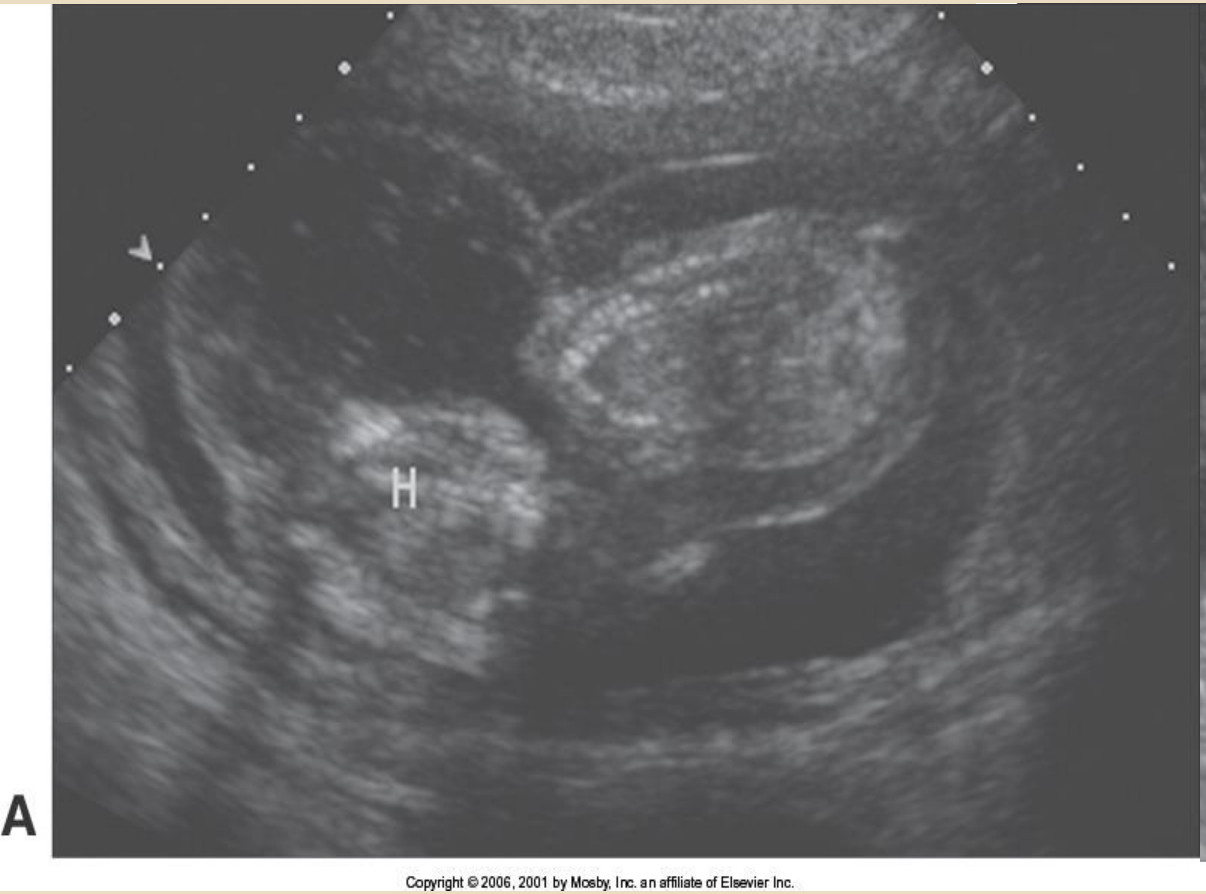


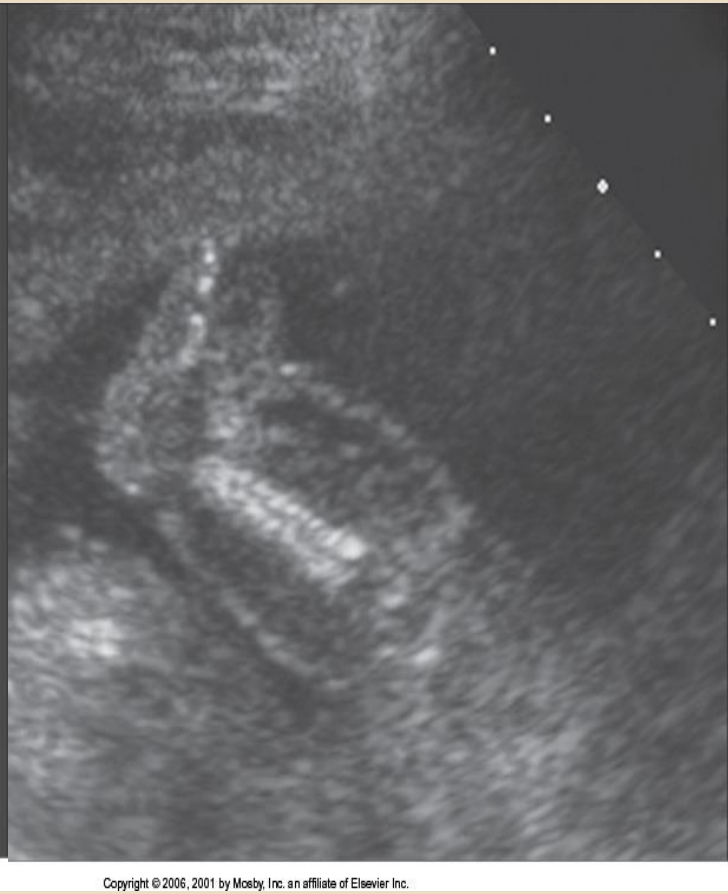
Cystic Hygroma



SMALL **LARGE**

Cystic Hygroma

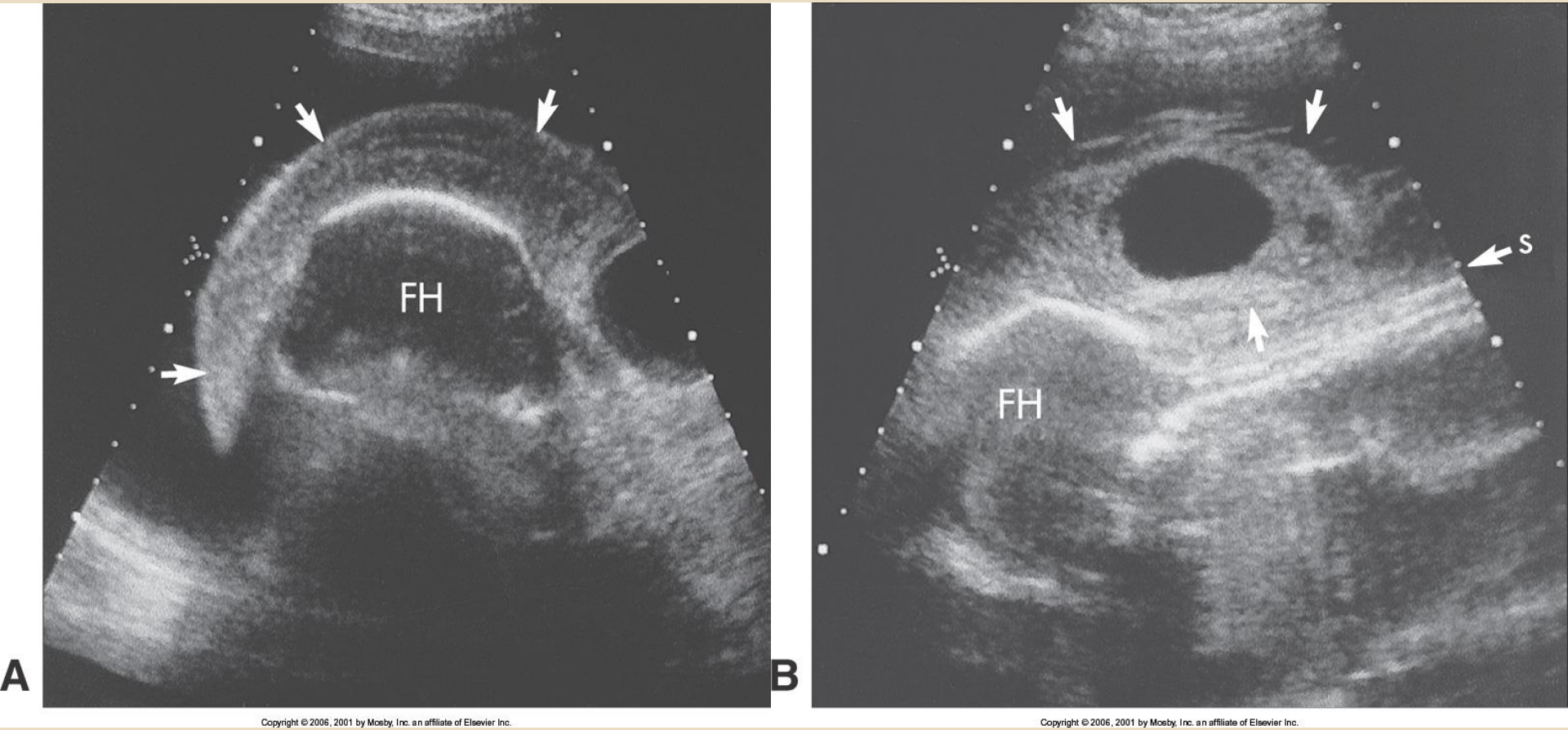




Extends to include most of the fetal trunk

Limb edema

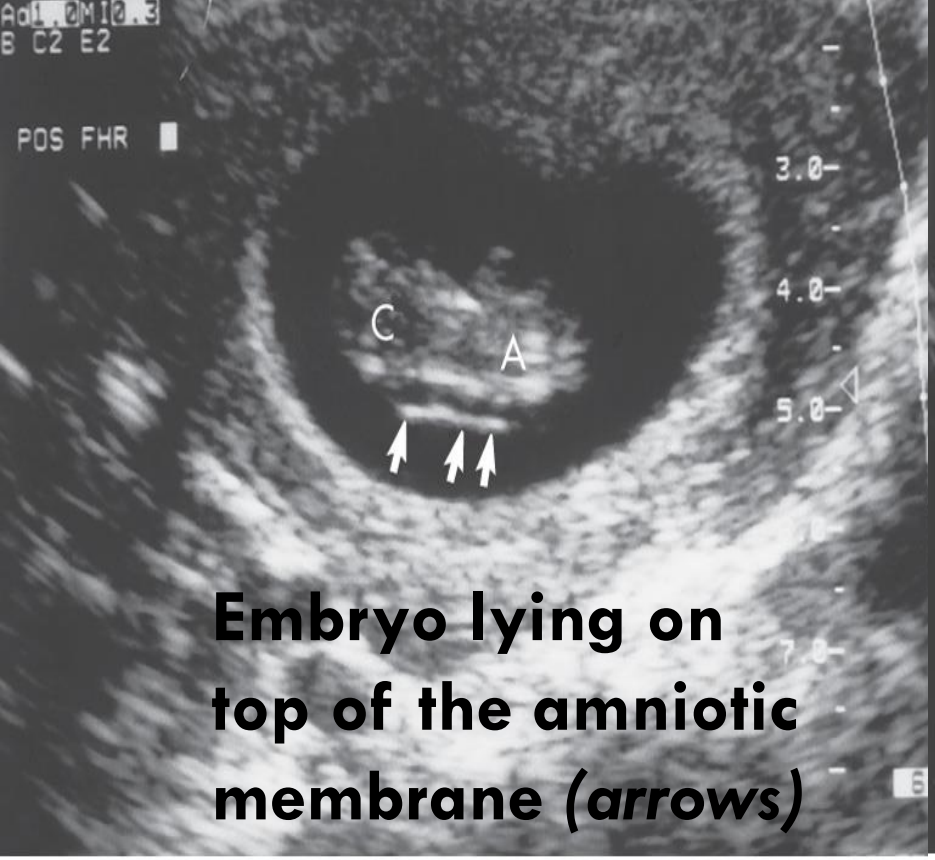
Cystic Hygroma



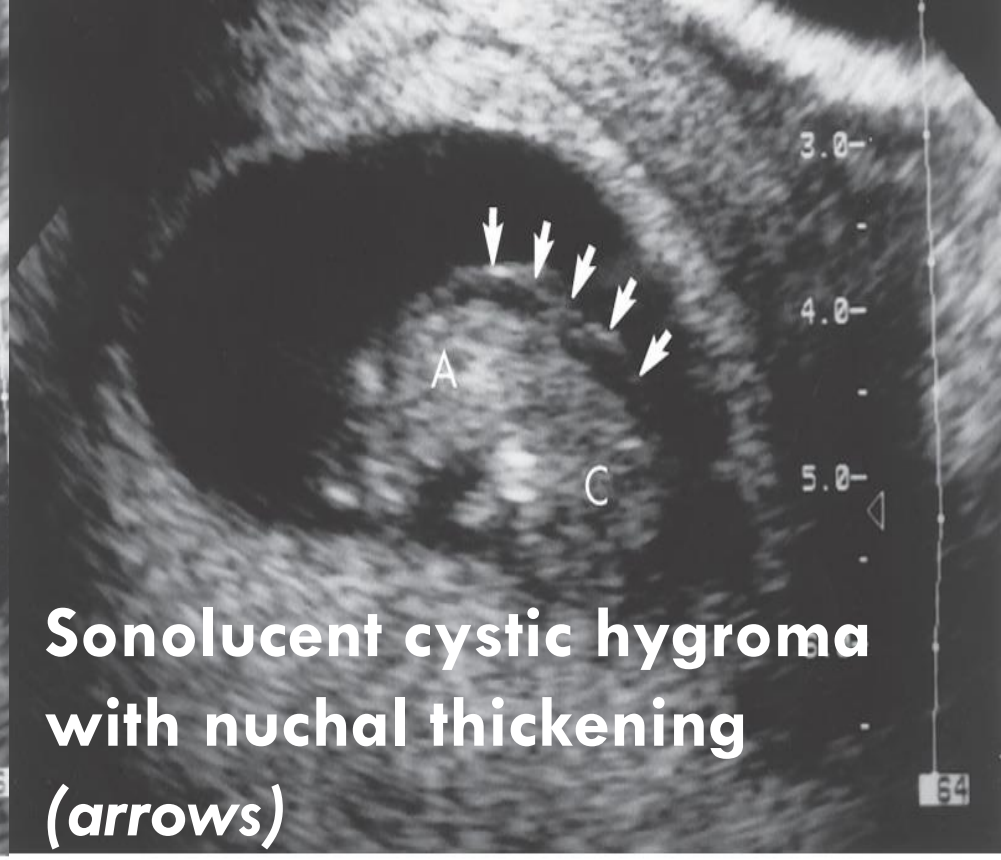
Helmet-like appearance of the scalp edema *(arrows)*

Normal Finding

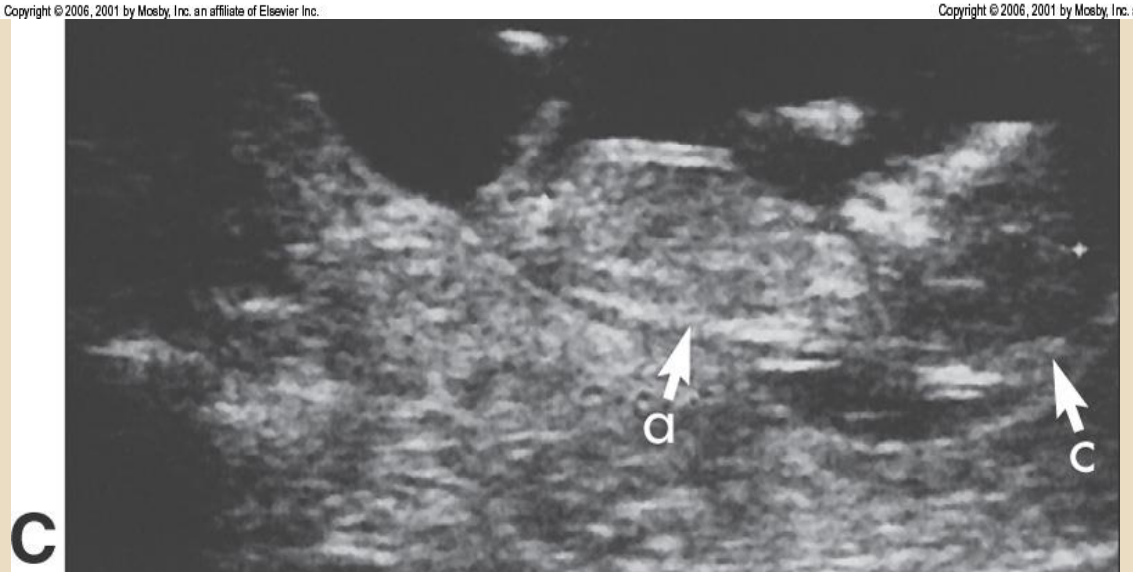
- Linear specular reflection routinely imaged along the back of the fetal neck in the late first trimester
  - Should not be mistaken for a cystic hygroma in the first trimester
- Remember
  - Typically, cystic hygromas appear as multiseptated, thin-walled cystic masses near the fetal head or neck



Embryo lying on top of the amniotic membrane *(arrows)*



Sonolucent cystic hygroma with nuchal thickening *(arrows)*

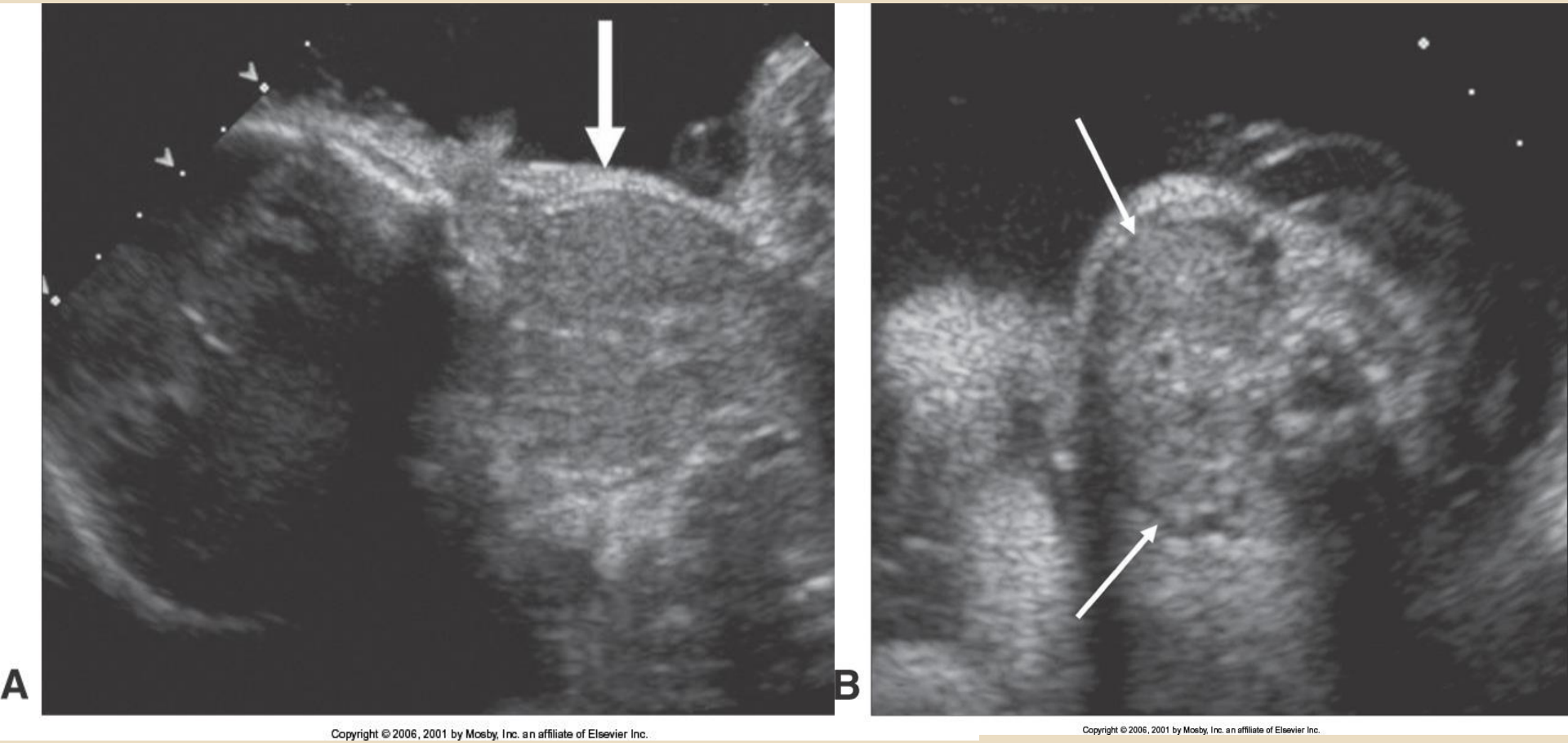


Normal Amnion

Fetal Goiter

- Appears as a solid mass arising from the anterior fetal neck
- Bi-lobed appearance may be visualized
- Esophagus may be obstructed by a goiter
  - Results in polyhydramnios and a small or absent stomach
- Follow-up studies of fetuses with goiter should include
  - Size estimations
  - Presence and size of fetal stomach
  - Amniotic fluid

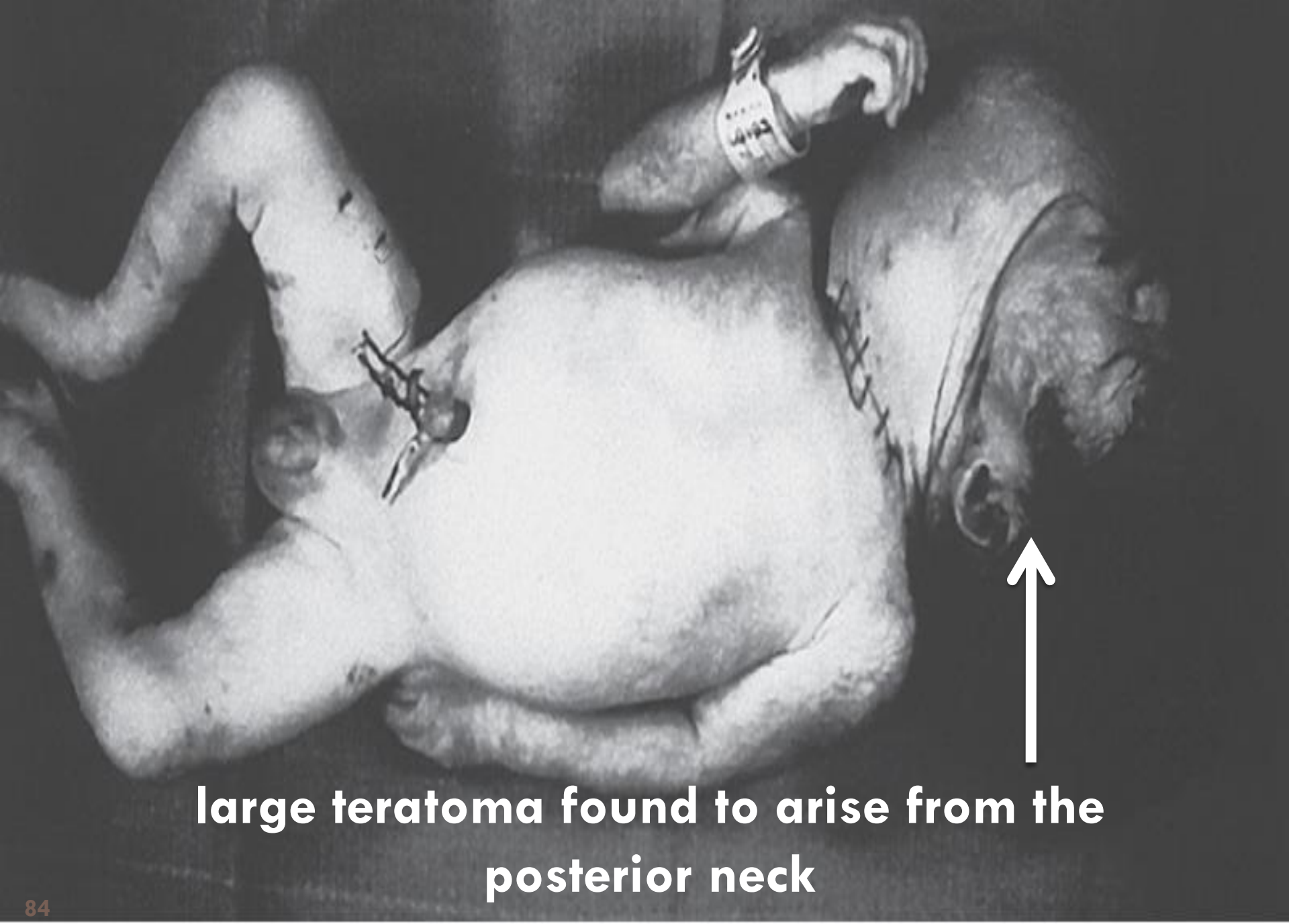
Goiter



Bi-lobed enlarged fetal thyroid gland *(arrow)*

Teratoma

- Solid tumor
  - Usually
    - Unilateral
    - Anterior
    - May have complex sonographic patterns
      - (cystic, solid, echogenic)



large teratoma found to arise from the posterior neck

Neck Masses

- Evaluate neck mass for the following characteristics:
  - Position of mass (anterior, posterior, lateral, or midline)
  - Unilateral or bilateral lesion
  - Polyhydramnios (common finding)
  - Heart failure and hydrops
  - Coexisting anomalies
  - Hyperextension may suggest neck mass or iniencephaly (fusion of occiput to spine)