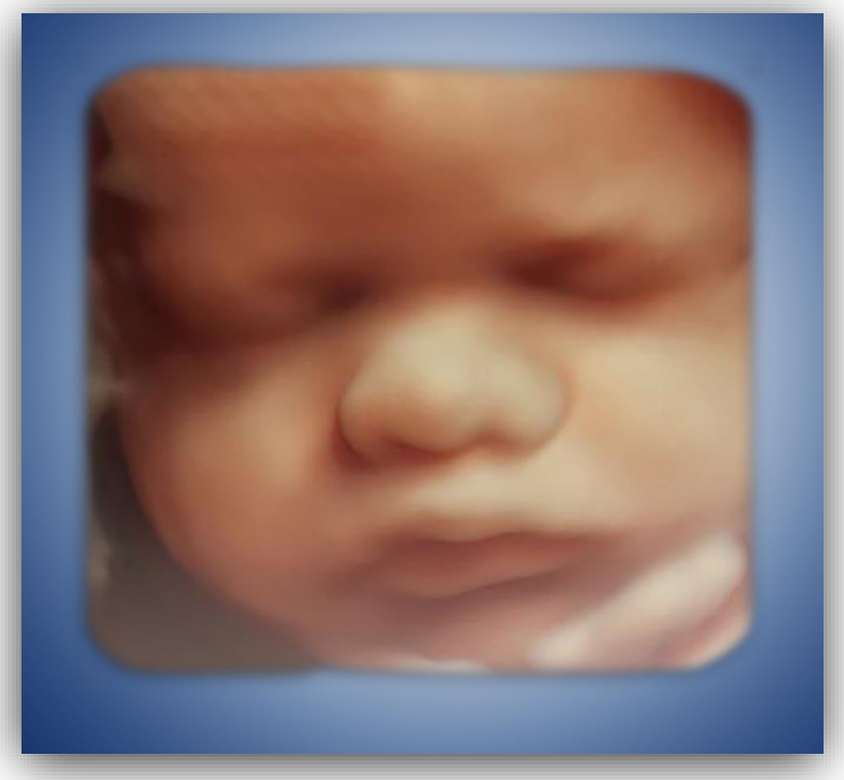
OB GYN SONOGRAPHY REVIEW

Fetal Face and Neck



Course Outline

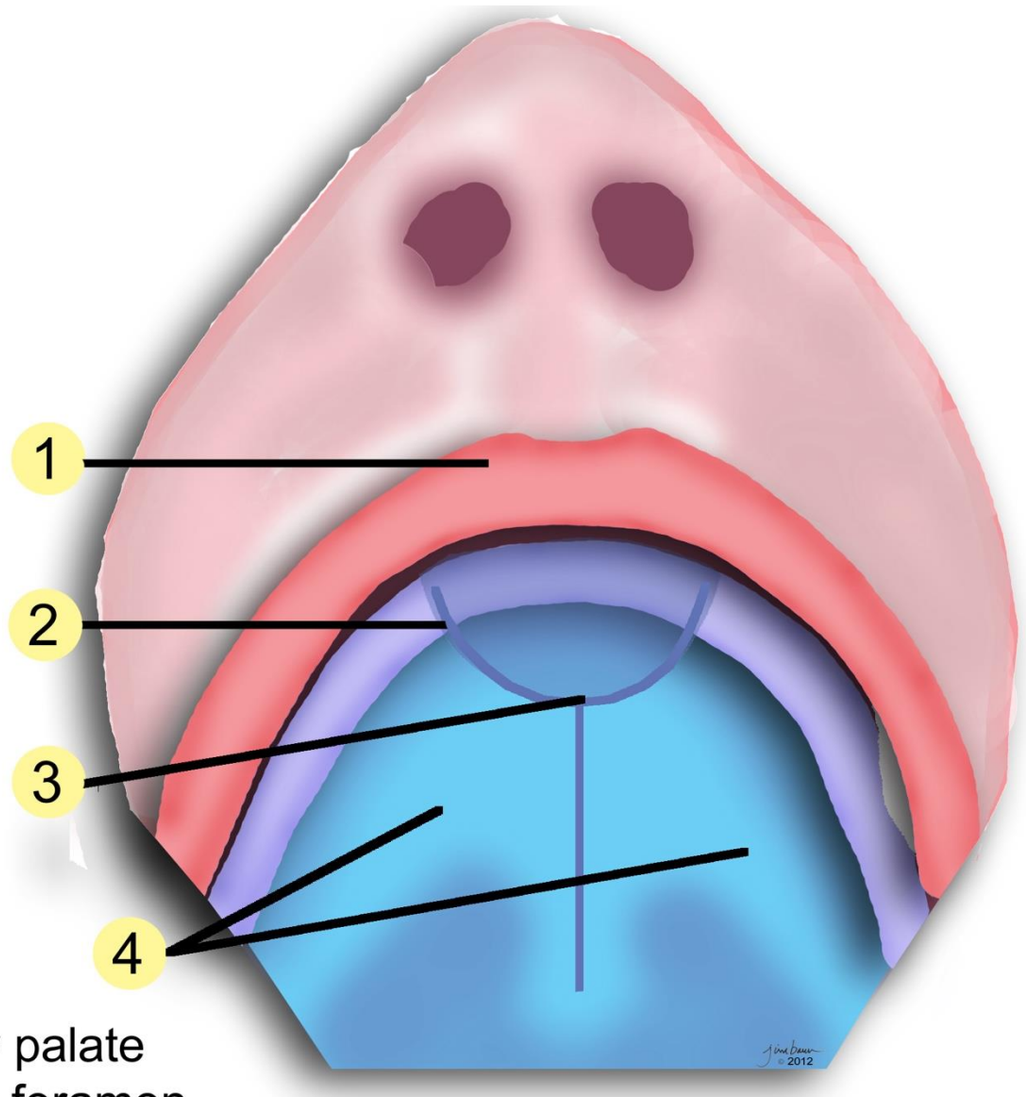
- Embryology
- Normal Sonographic Anatomy
  - Face
  - Neck
- Facial Abnormalities
- Chin & Neck Abnormalities

Embryology

Face

- Three primordial facial prominences present by 6 weeks
  - *Frontonasal prominence*: gives rise to forebrain, nose , and eyes
  - *Maxillary and mandibular prominences*: give rise to and create boundaries of primitive mouth
- Palate develops from bits of both *nasal* and *maxillary* prominences
- Complete formation by end of 11th week

EMBRYOLOGY

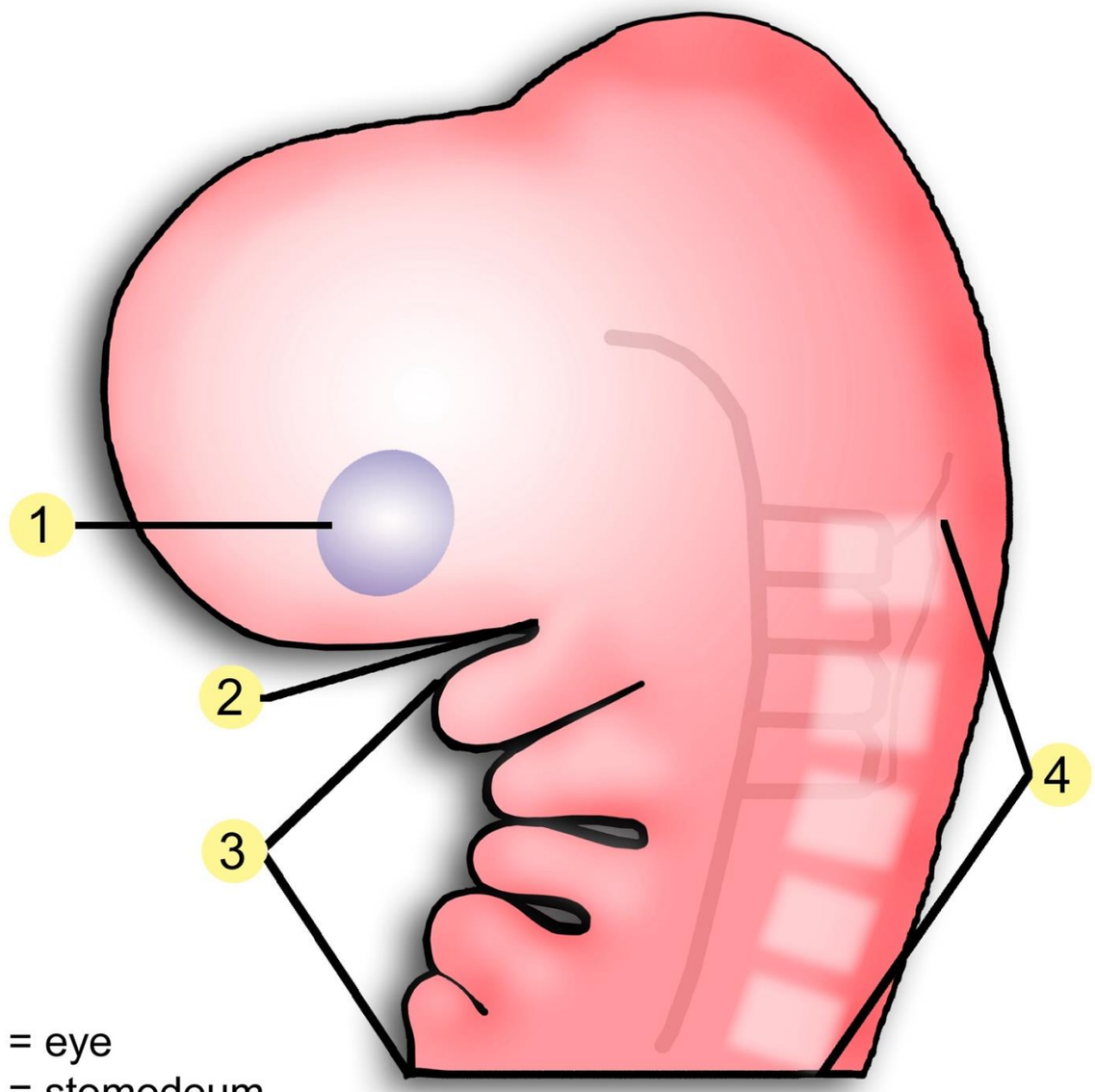


1 = lip
2 = primary palate
3 = incisive foramen
4 = secondary palate

Neck

- *Pharyngeal arches* and *grooves* metamorphose into:
  - *Face*
  - *Nasal cavities*
  - *Mouth*
  - *Tongue*
  - *Larynx, pharynx*
  - *Neck fascia*
- Formation completed by 12 weeks

EMBRYOLOGY



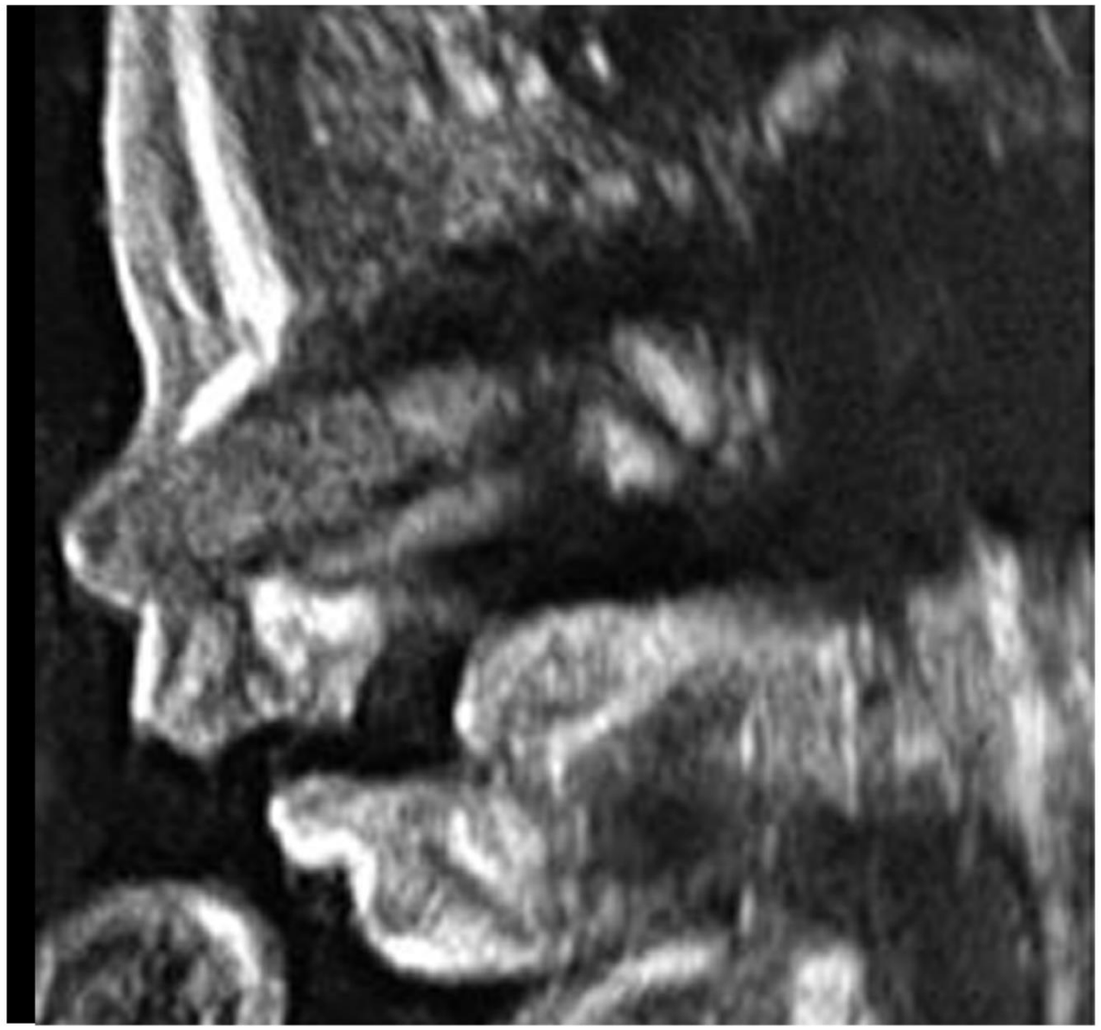
1 = eye
2 = stomodeum
3 = pharygeal arches
4 = somites

Normal Sonographic Anatomy

Imaging Protocol

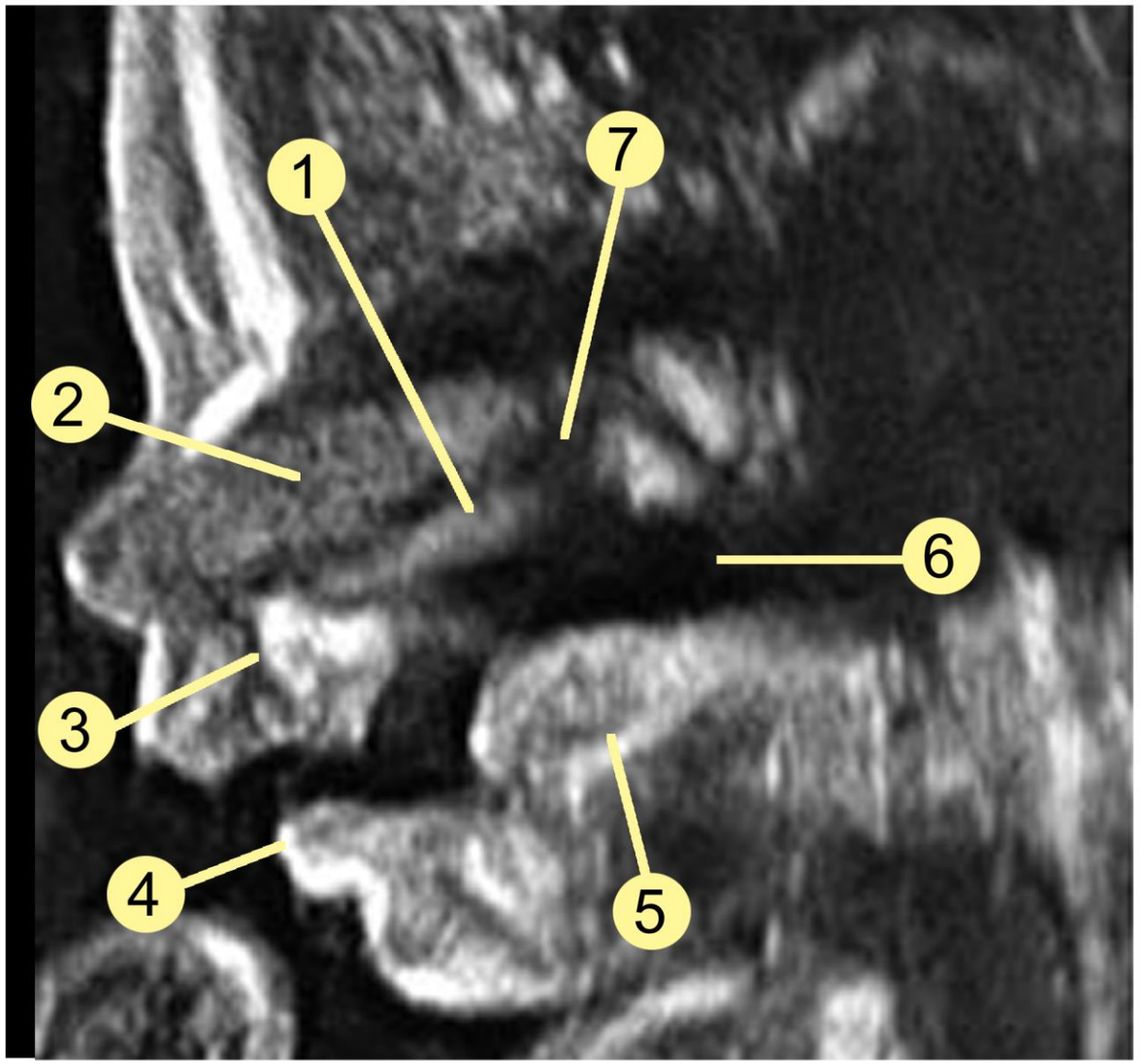
- Routine and targeted imaging of the fetal face and neck should include:
  - Sagittal view of orbits, nose, lips, maxilla and anterior mandible
  - Profile view of soft tissues of mouth and nasopharynx
  - Views of brow, cheeks, eyelids
  - View of lenses and nasal septum
  - Axial lips and nose

FETAL FACE



Profile view

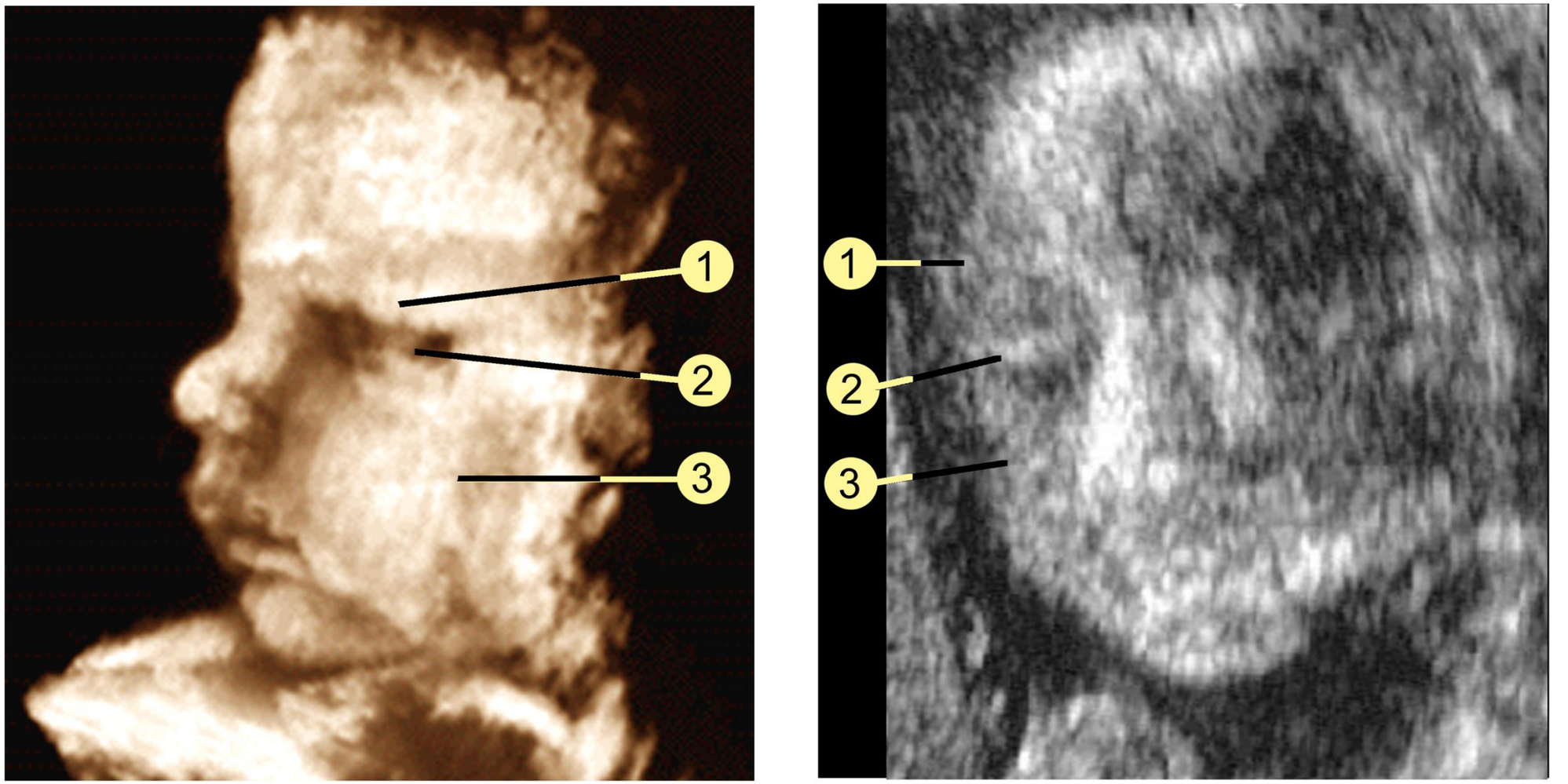
FETAL FACE



1 = soft palate
2 = nasal apparatus
3 = hard palate
4 = lower lip
5 = tongue
6 = oral pharynx
7 = nasal pharynx

FETAL FACE

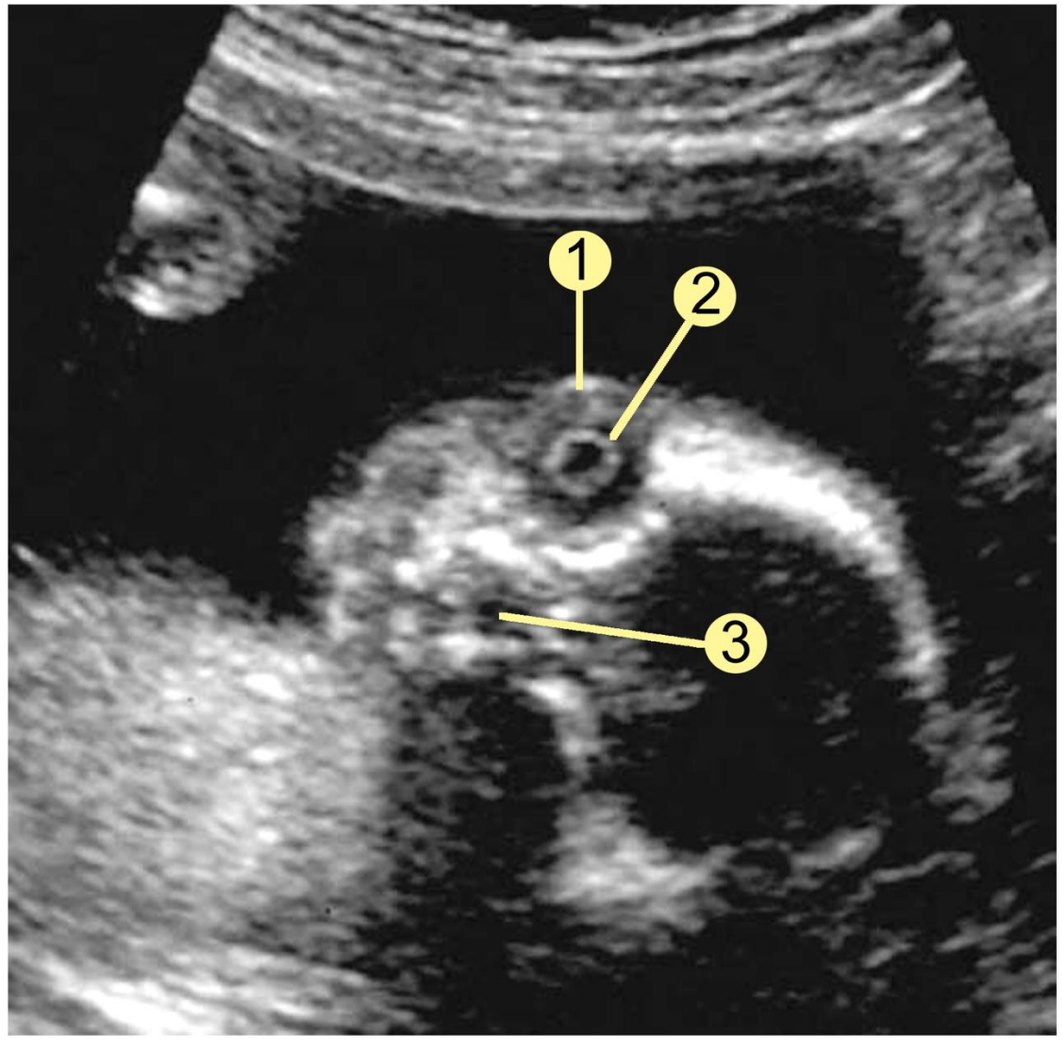
1 = brow
2 = eyelid
3 = cheek



Brow, cheeks. eyelid

FETAL FACE

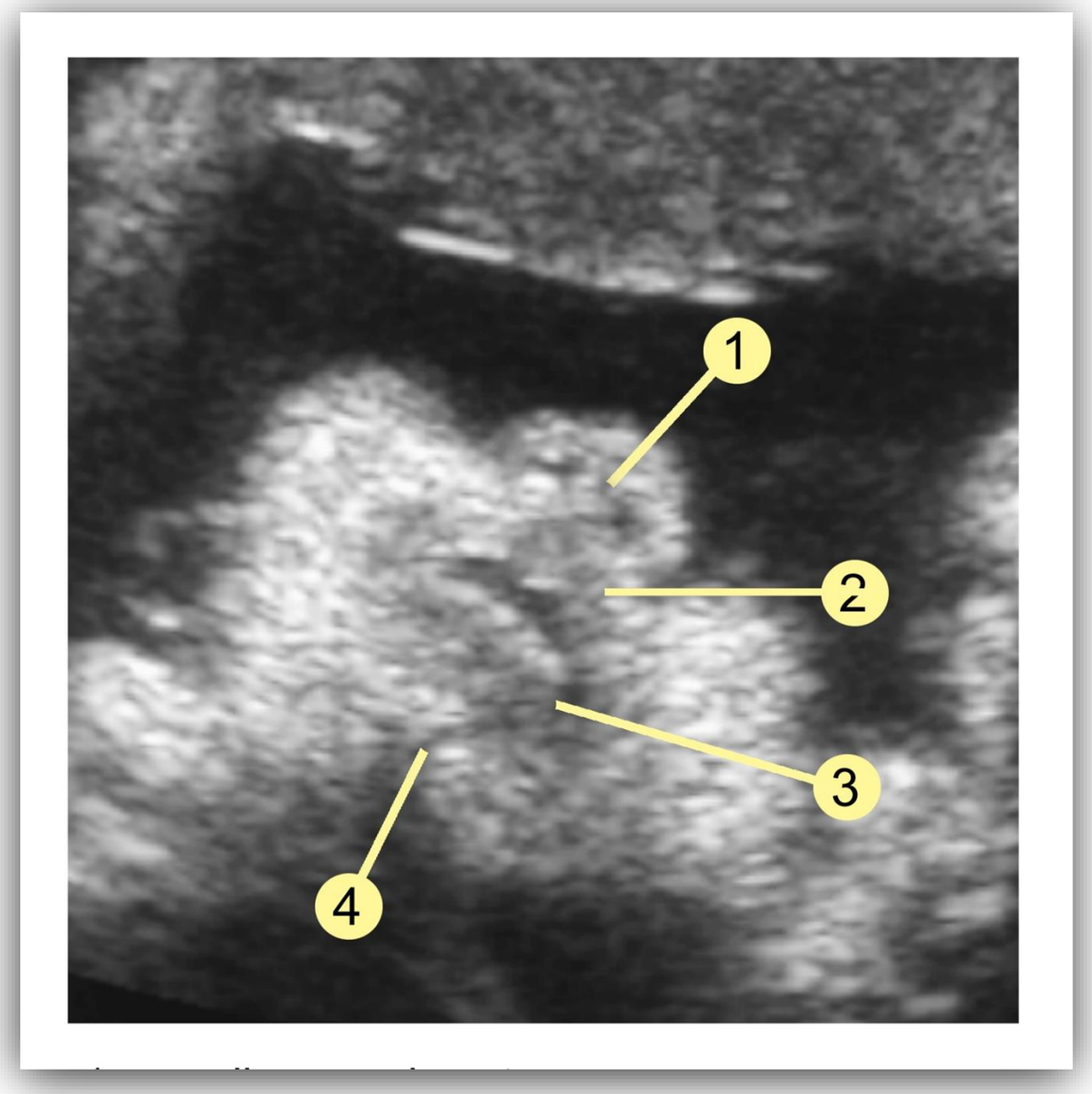
1 = orbital rim
2 = lens
3 = nasal septum



Lens & nasal septum

FETAL FACE

1 = nose
2 = upper lip
3 = lower lip
4 = chin

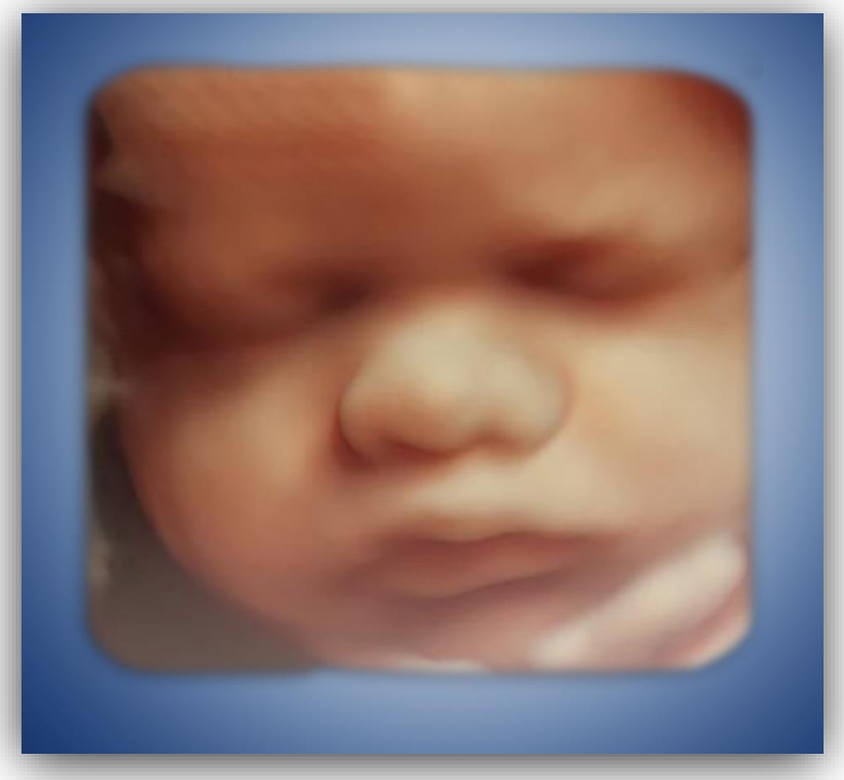


Axial lips & nose

Face Abnormalities

Categories of Abnormalities

- Face abnormalities
  - Clefting anomalies
  - Ocular and orbital abnormalities
    - Hypertelorism
    - Hypotelorism
    - Cyclopia
    - Microphthalmia
    - Anophthalmia



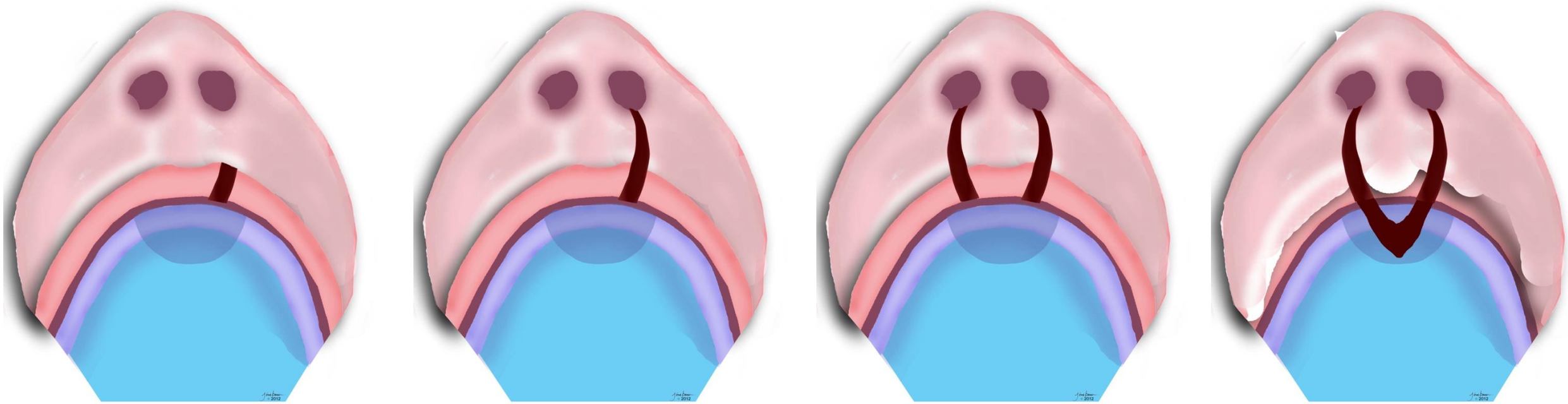
Clefting Abnormalities

- Relatively common congenital facial anomalies
- Cleft lip and cleft palate arise from two separate embryological events
- Disruption of *primary palate* (early 8th week) = *anterior cleft anomalies*
- Disruption of *secondary palate* (early 8th – 11th week) = *posterior cleft anomalies*

Clefting Abnormalities

- Anterior cleft palate anomalies
  - Incomplete unilateral cleft lip
  - Complete unilateral cleft lip
  - Complete bilateral cleft lip
  - Complete anterior cleft (uni- and bilateral) extending through lip and into maxilla

ANTERIOR CLEFTING VARIANTS



Incomplete unilateral cleft lip

Complete unilateral cleft lip

Complete bilateral cleft lip

Complete bilateral extending into maxilla

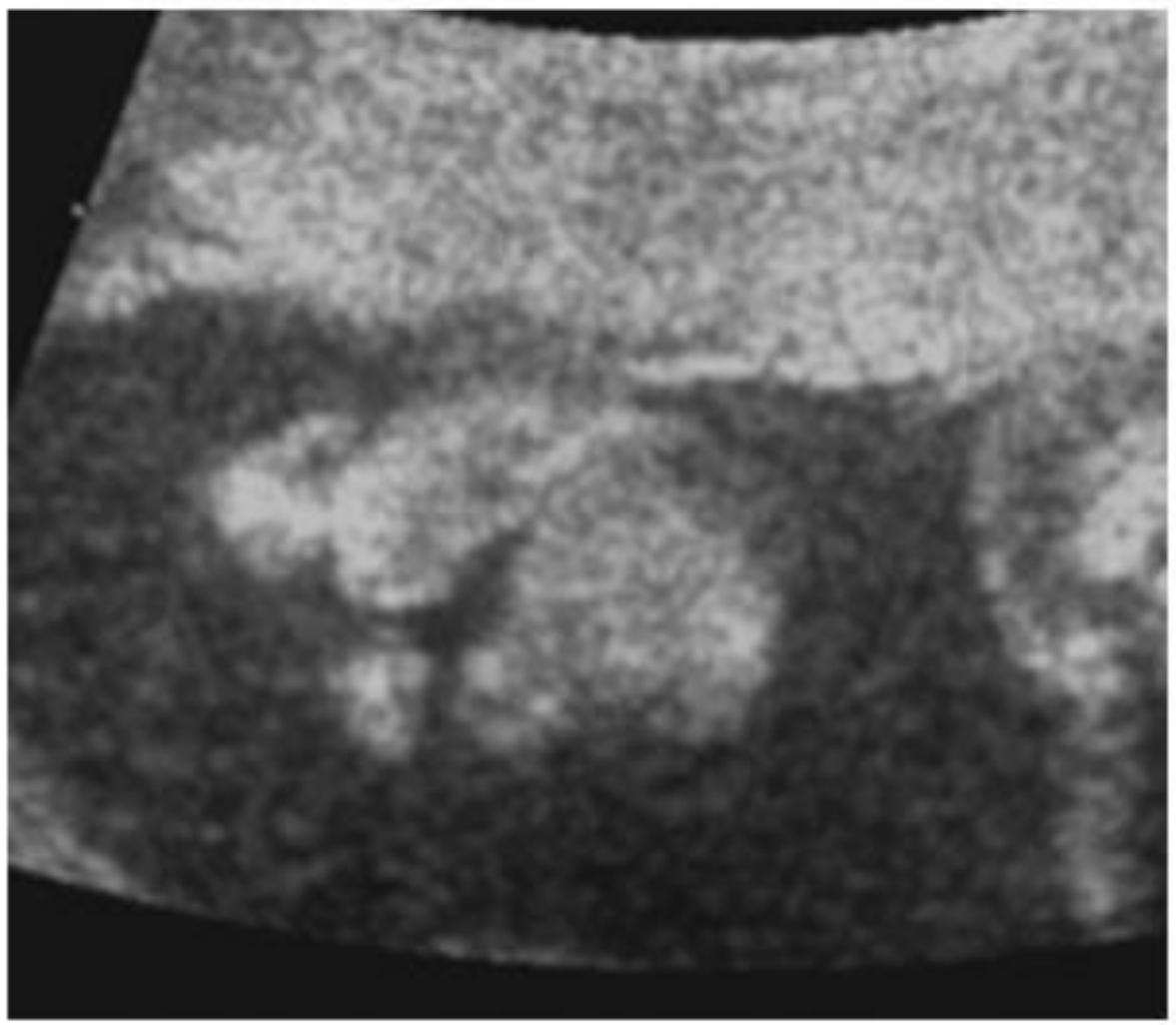
Clefting Abnormalities

- Imaging pearls & pitfalls
  - Cleft palate in absence of cleft lip is a diagnostic challenge
  - Cleft palate and associated cleft lip will demonstrate contiguity of lip defect with deeper structures
    - Amniotic fluid provides excellent US contrast
    - Cleft palate may be associated with an alveolar ridge

Clefting Abnormalities

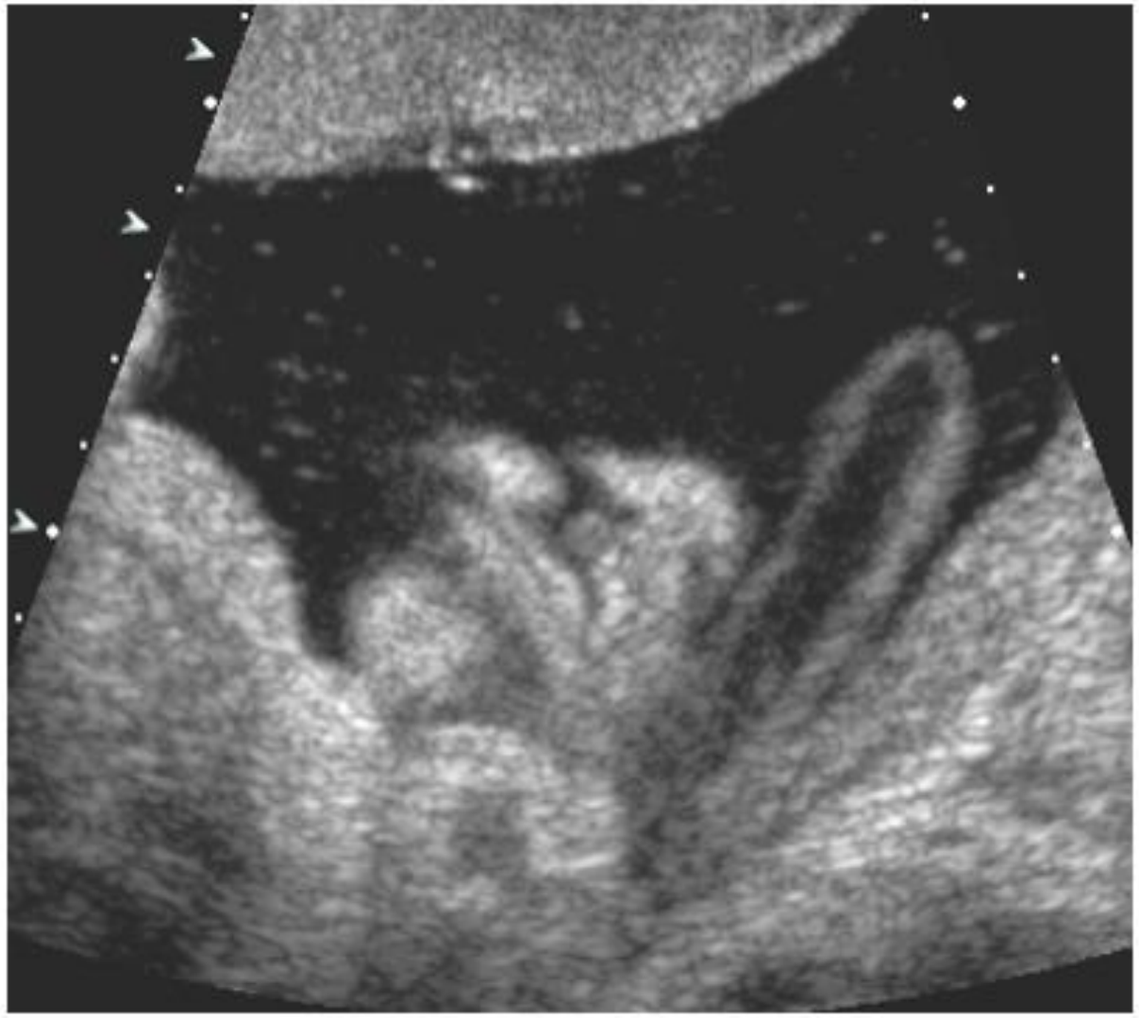
- Sonographic findings – anterior clefting anomalies
  - Defect may be demonstrated as a groove extending from one nostril through the lip
  - Usually best demonstrated coronally
  - Defect may be uni- or bilateral
  - Polyhydramnios may result from swallowing difficulties

CLEFTING ABNORMALITIES



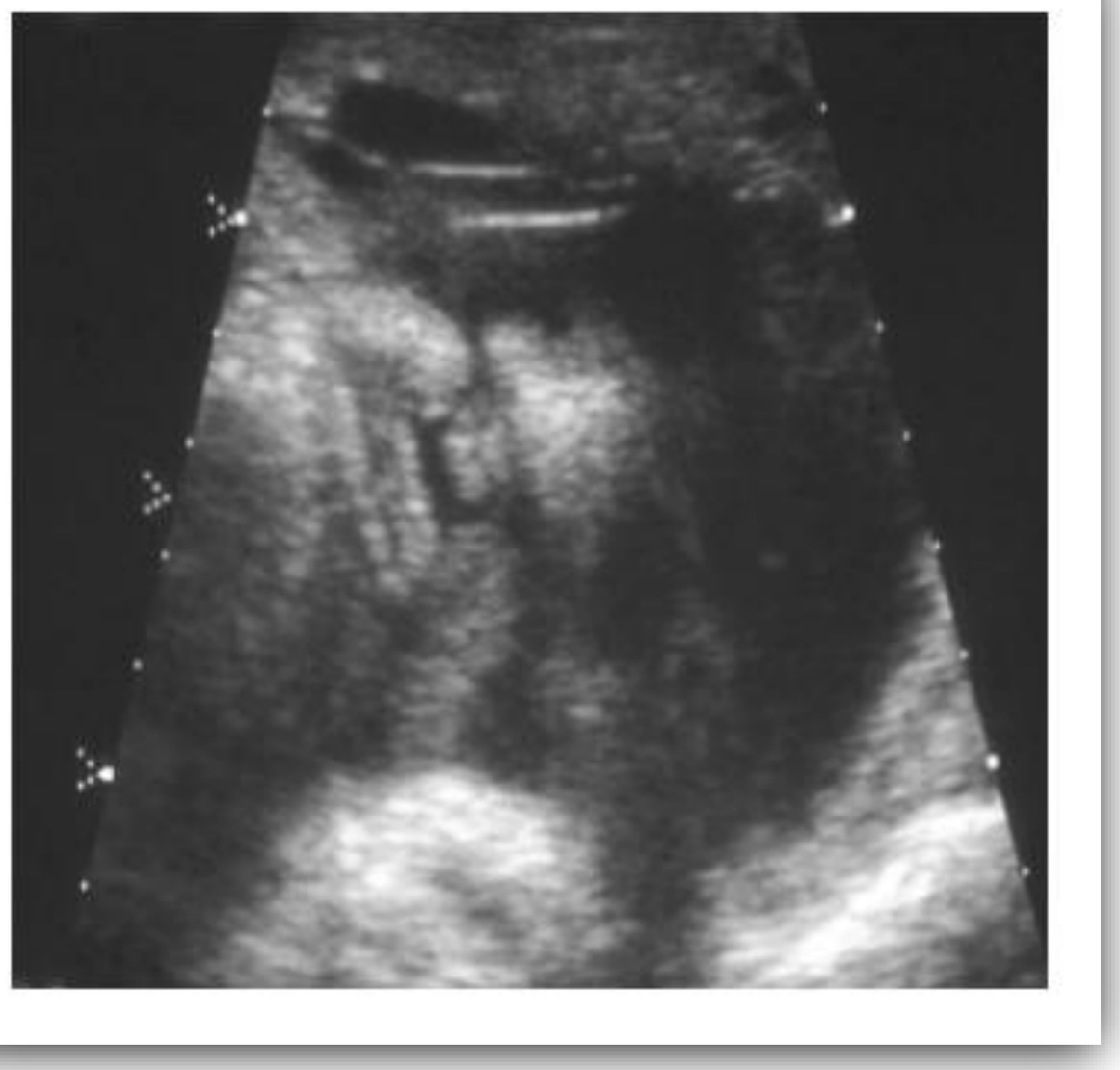
Cleft lip - unilateral

CLEFTING ABNORMALITIES



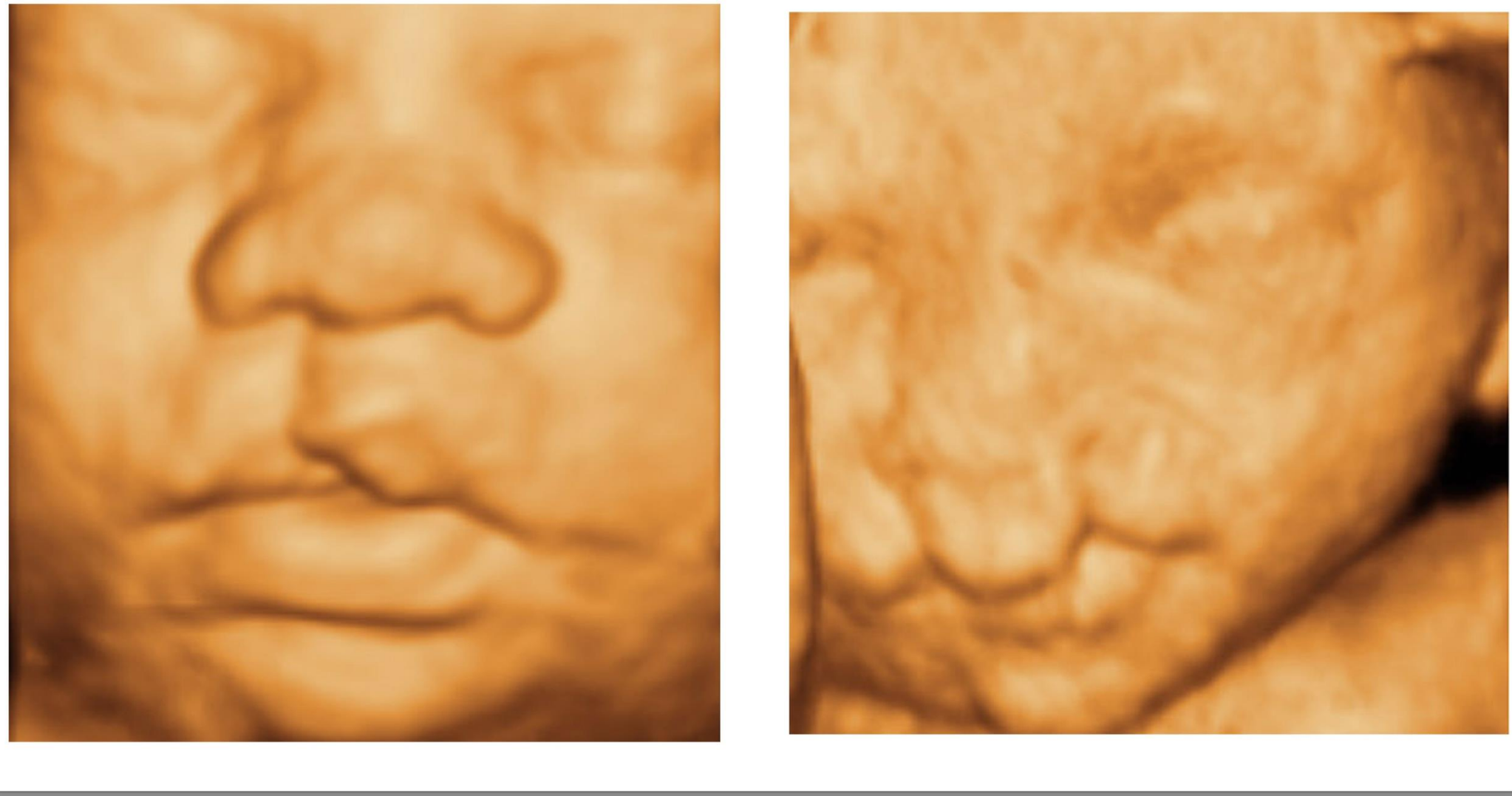
Cleft lip & palate - bilateral

CLEFTING ABNORMALITIES



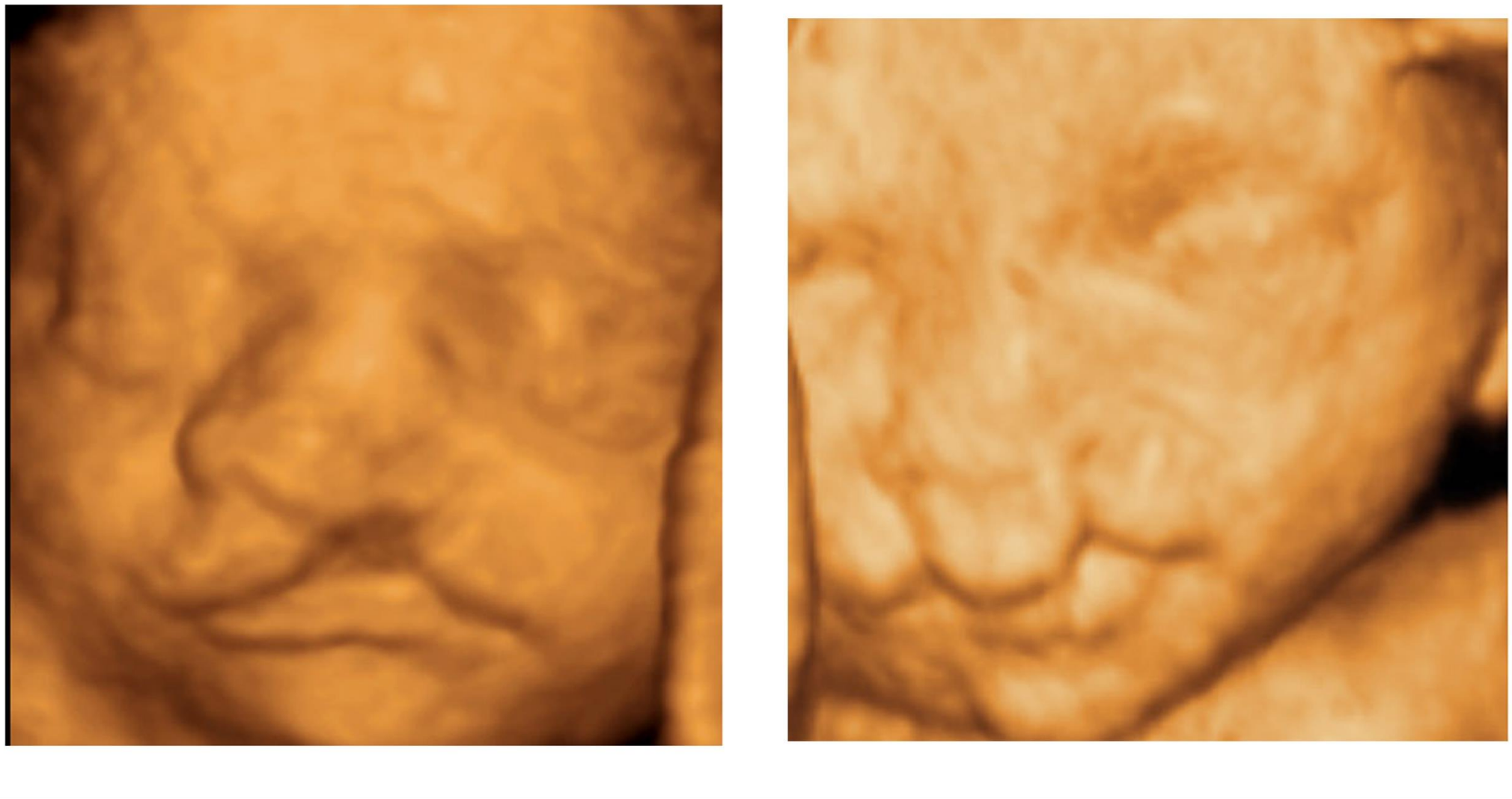
Cleft lip & palate - unilateral

CLEFTING ABNORMALITIES



Cleft lip

CLEFTING ABNORMALITIES

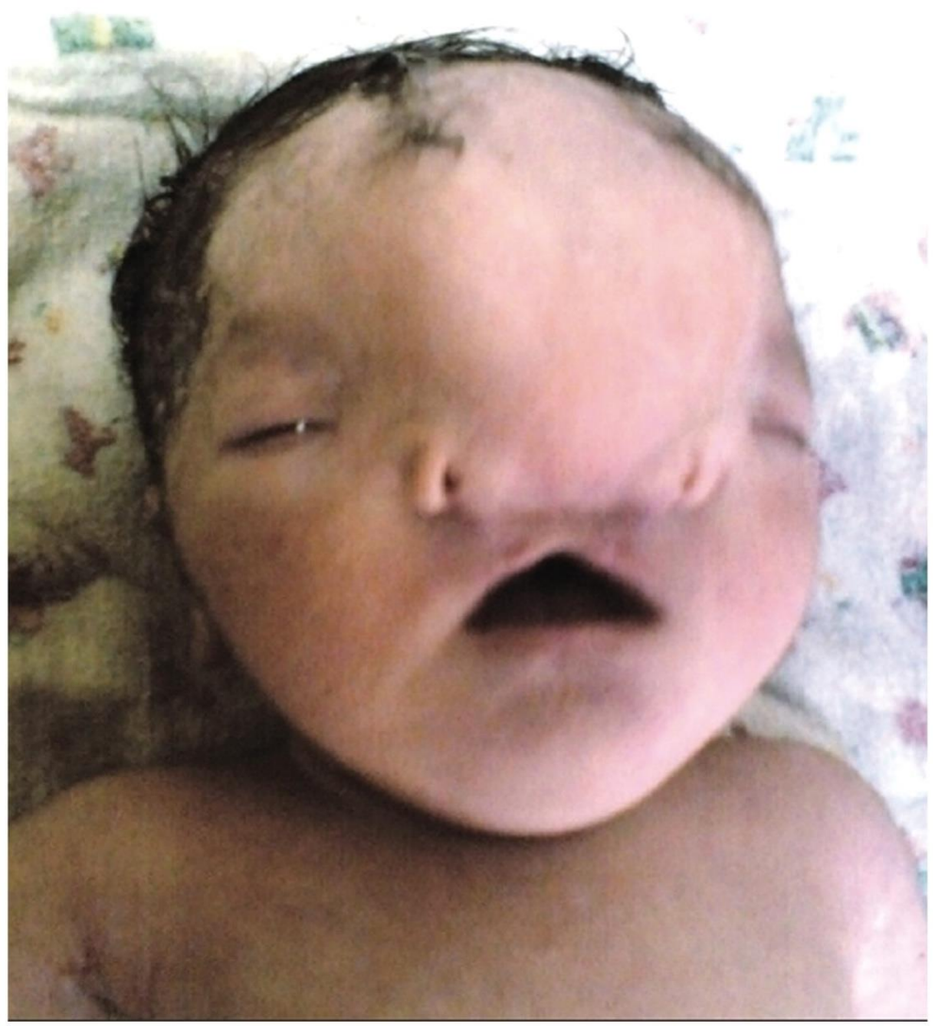


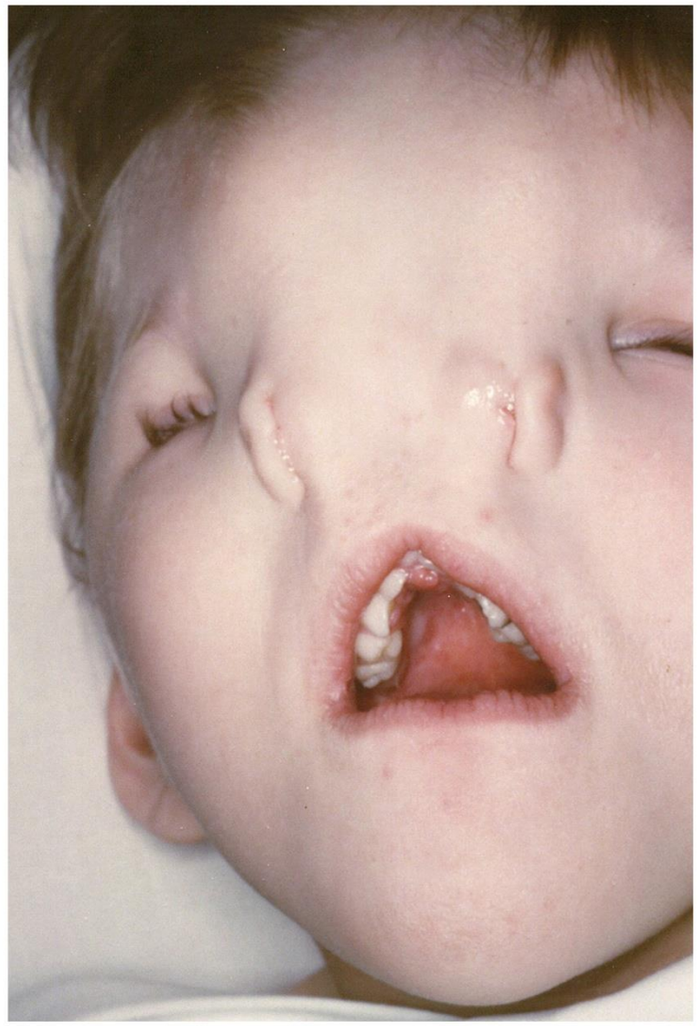
Cleft lip & palate

Median Facial Clefting

- Rare embryonic mesenchymal defect
- Characterized by severe derangement of internal anatomic relationship between nasal cavity and frontal bones
- Rarely associated with external lip or other facial clefting defects
- Also called *frontonasal dysplasia sequence*

MEDIAN FACIAL CLEFTING SYNDROME





Ocular & Orbital Abnormalities

- Are often principle signs of other significant fetal anomalies & syndromes
- Categories:
  - Hypertelorism
  - Hypotelorism
  - Cyclopia
  - Microphthalmia
  - Anophthalmia

Orbital Hypertelorism

- Orbits space too far apart
- Increase in intraorbital distance above 95th percentile
- Two conditions typically associated are:
  - Frontal encephalocele
  - Midline facial clefting syndrome

Orbital Hypertelorism

- Associated abnormalities include:
  - Trisomy 21 (Down syndrome)
  - Trisomy 13 (Patau syndrome)
  - Agenesis of the corpus callosum
  - Turner syndrome
  - Ehlers-Danlos syndrome
  - Apert syndrome

Orbital Hypertelorism

- Sonographic findings include:
  - Orbits placed far apart
  - Interocular distance and binocular distance at or above 95th percentile for dates

ORBITAL ABNORMALITIES

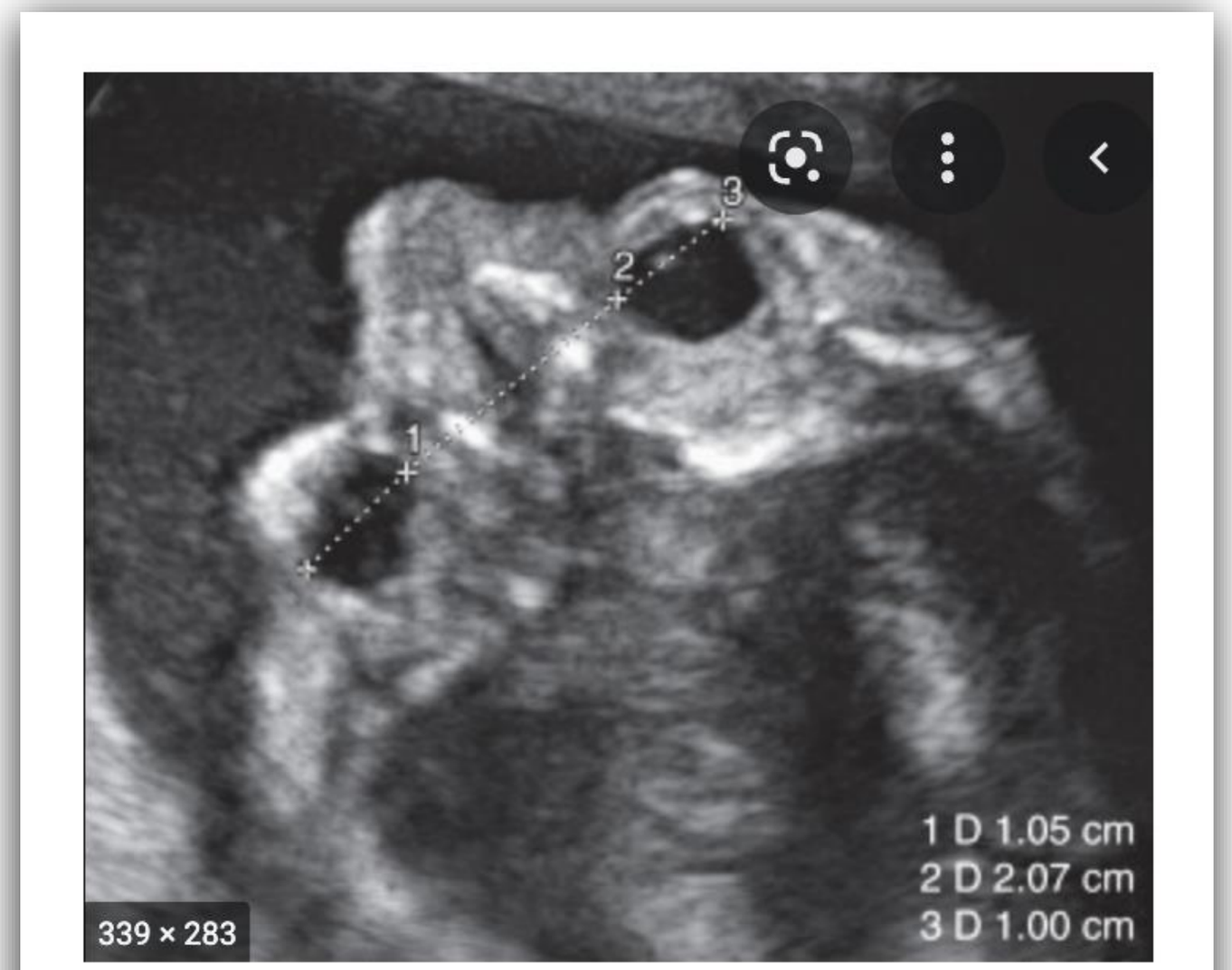
Table 1. The fetal interocular distance (mm): GA, gestational age (n = 595)

GA (weeks)	n	Mean ± SD	Percentile		
			10th	50th	90th
15	22	8.78 ± 0.92	7.69	8.70	10.15
16	22	9.88 ± 1.10	8.40	10.00	11.41
17	23	10.66 ± 0.96	9.24	10.90	11.56
18	31	11.40 ± 1.10	10.12	11.10	13.16
19	22	11.72 ± 0.97	10.26	11.80	13.21
20	26	13.05 ± 0.97	11.71	13.25	14.25
21	24	13.65 ± 1.40	11.45	13.60	15.45
22	22	14.02 ± 1.29	12.20	14.20	15.37
23	23	14.43 ± 1.22	12.70	14.30	16.40
24	26	14.90 ± 1.08	13.28	15.05	16.30
25	23	15.59 + 1.40	13.90	15.30	17.70
26	22	15.91 ± 0.98	14.73	15.85	17.42
27	22	16.17 ± 0.93	15.09	16.05	17.83
28	22	16.97 ± 1.58	15.13	16.85	19.55
29	22	17.33 ± 1.44	15.62	17.15	19.66
30	23	17.57 ± 1.50	16.00	17.30	19.24
31	22	17.90 ± 1.46	16.00	17.50	19.98
32	22	18.21 ± 1.27	16.21	18.05	19.99
33	22	18.70 ± 1.42	17.10	18.75	20.17
34	22	19.82 ± 1.38	17.92	19.55	21.91
35	22	20.25 ± 1.46	18.01	20.05	22.40
36	22	20.37 ± 1.41	18.22	20.40	22.50
37	22	20.75 ± 1.53	18.55	20.75	22.82
38	22	21.93 ± 1.94	19.22	21.70	24.72
39	22	22.18 ± 1.95	19.79	22.00	25.00
40	22	22.41 ± 1.29	21.13	22.10	25.16

Table 2. The fetal binocular distance (mm): GA, gestational age (n = 595)

GA (weeks)	n	Mean ± SD	Percentile		
			10th	50th	90th
15	22	20.20 ± 2.10	17.22	20.00	22.96
16	22	23.19 ± 1.34	21.80	22.90	25.04
17	23	25.59 ± 1.30	24.08	25.40	27.70
18	31	27.30 ± 1.75	24.88	27.10	30.28
19	22	29.52 ± 1.74	27.49	29.20	31.71
20	26	31.14 ± 1.56	29.17	31.10	33.19
21	24	32.88 ± 1.12	31.40	33.00	34.25
22	22	35.06 ± 1.03	33.69	35.10	35.94
23	23	36.17 ± 1.85	34.12	36.00	39.18
24	26	37.52 ± 1.54	34.87	37.85	39.42
25	23	39.45 + 1.85	36.74	39.40	42.00
26	22	41.05 ± 1.52	39.23	40.95	43.79
27	22	42.63 ± 1.87	39.62	43.00	44.72
28	22	43.87 ± 2.02	41.12	44.10	46.07
29	22	45.43 ± 1.95	43.00	45.15	48.88
30	23	46.31 ± 1.88	43.70	46.00	48.98
31	22	47.89 ± 1.93	44.46	48.00	50.04
32	22	48.24 ± 2.02	45.37	48.25	51.13
33	22	49.31 ± 2.07	46.23	49.60	51.70
34	22	51.98 ± 2.64	48.66	51.95	55.77
35	22	52.99 ± 2.35	49.32	53.00	56.99
36	22	53.32 ± 2.36	50.18	53.15	57.08
37	22	54.04 ± 1.89	51.65	53.80	57.44
38	22	55.49 ± 2.12	52.58	55.60	58.27
39	22	55.79 ± 2.26	52.66	56.05	58.69
40	22	55.97 ± 1.89	53.66	56.15	58.77

ORBITAL HYPERTELORISM



Increased interocular and binocular distance

Orbital Hypotelorism

- Orbits space too close together
- Reduction in intraorbital distance below 5th percentile
- Condition typically associated :
  - Holoprosencephaly

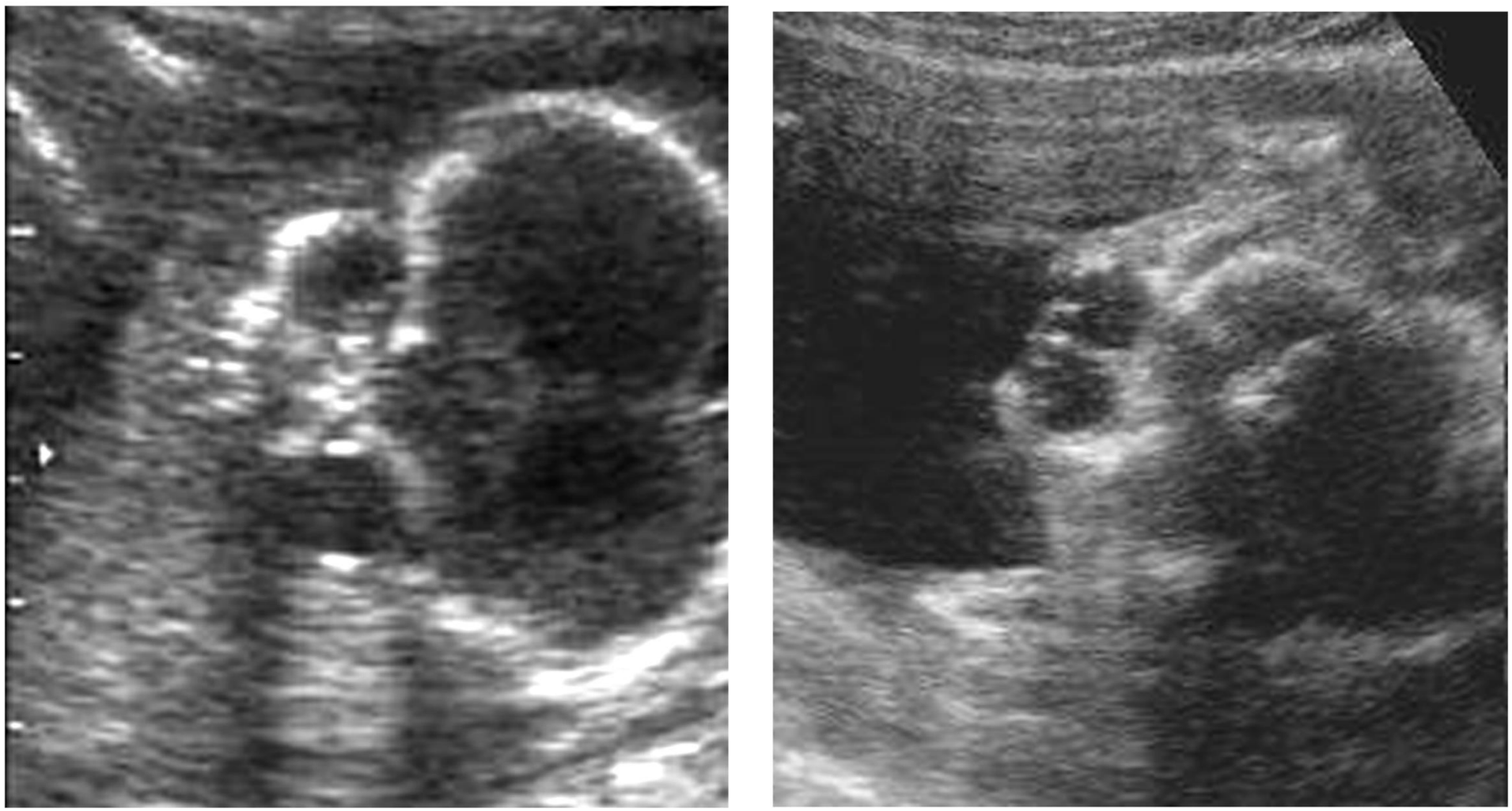
Orbital Hypotelorism

- Associated abnormalities include:
  - Trisomy 13 (Patau syndrome)
  - Encephalocele
  - Cleft palate
  - Cardiac anomalies
  - Imperforate anus
  - Diaphragmatic hernia
  - Meckel-Gruber syndrome

Orbital Hypotelorism

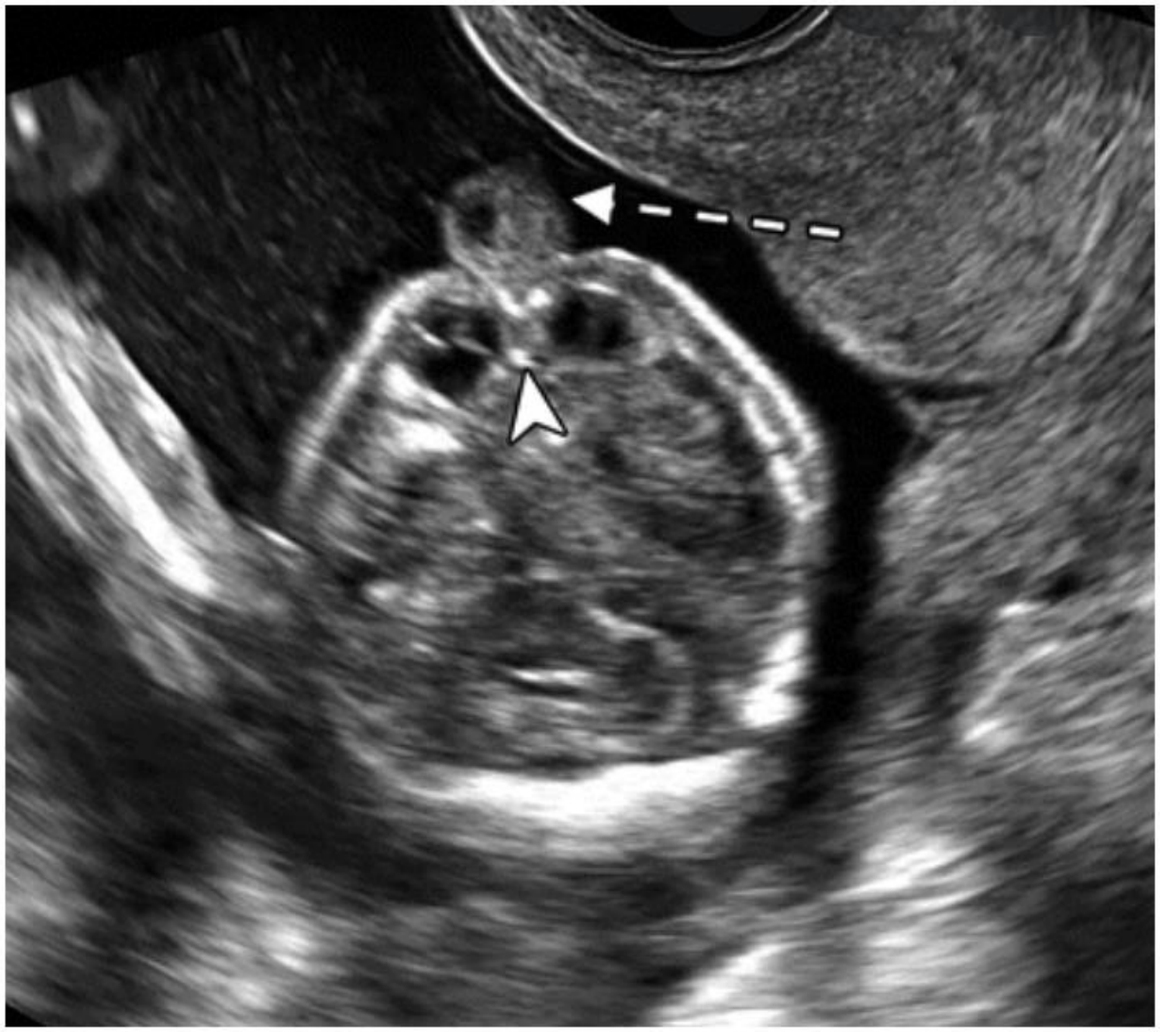
- Sonographic findings include:
  - Close appearing orbits
  - Interocular distance and binocular distance below 5th percentile for dates

ORBITAL HYPOTELORISM



Moderate **Severe**

ORBITAL HYPOTELORISM



Holoprosencephaly

Cyclopia

- Presence of single midline orbit
- May be single, fused or no eyes present
- Can be induced by exposure to certain teratogenic toxins
- Condition typically associated :
  - Alobar holoprosencephaly with proboscis

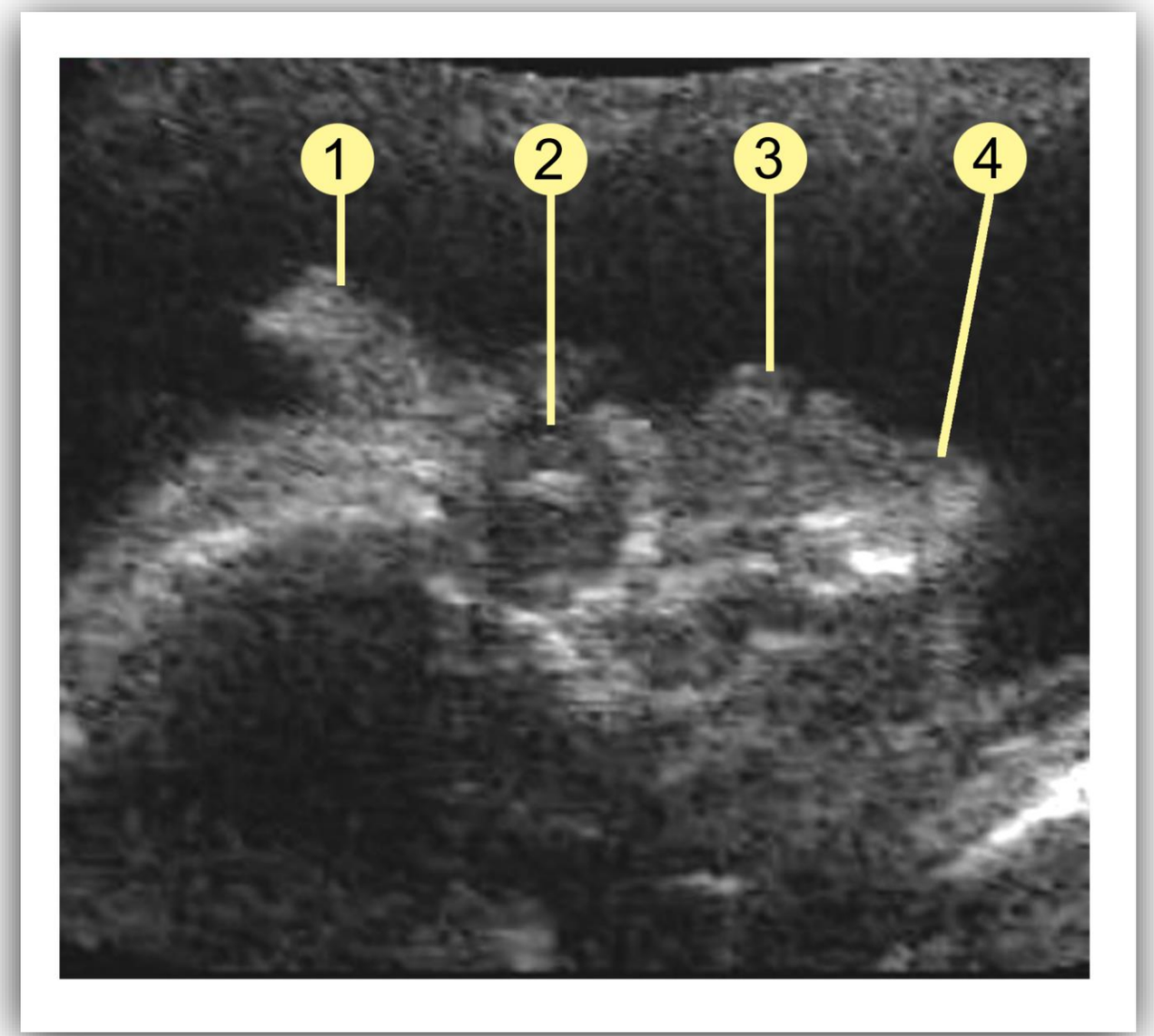
Cyclopia

- Associated abnormalities include:
  - Alobar holoprosencephaly
  - Median facial clefting
  - Trisomy 13 (Patau syndrome)
  - CHARGE association

Cyclopia

- Sonographic findings include:
  - Single midline orbit
  - Microcephaly (64% of cases)
  - Other findings associated with holoprosencephaly

CYCLOPIA



1 = proboscis
2 = single midline orbit
3 = upper lip
4 = chin

Microphthalmia

- Reduction in orbital size
- Objectively when affected orbit is disproportionately small relative to other facial structures
- May be uni- or bilateral
- May occur in isolation or as part of a syndrome

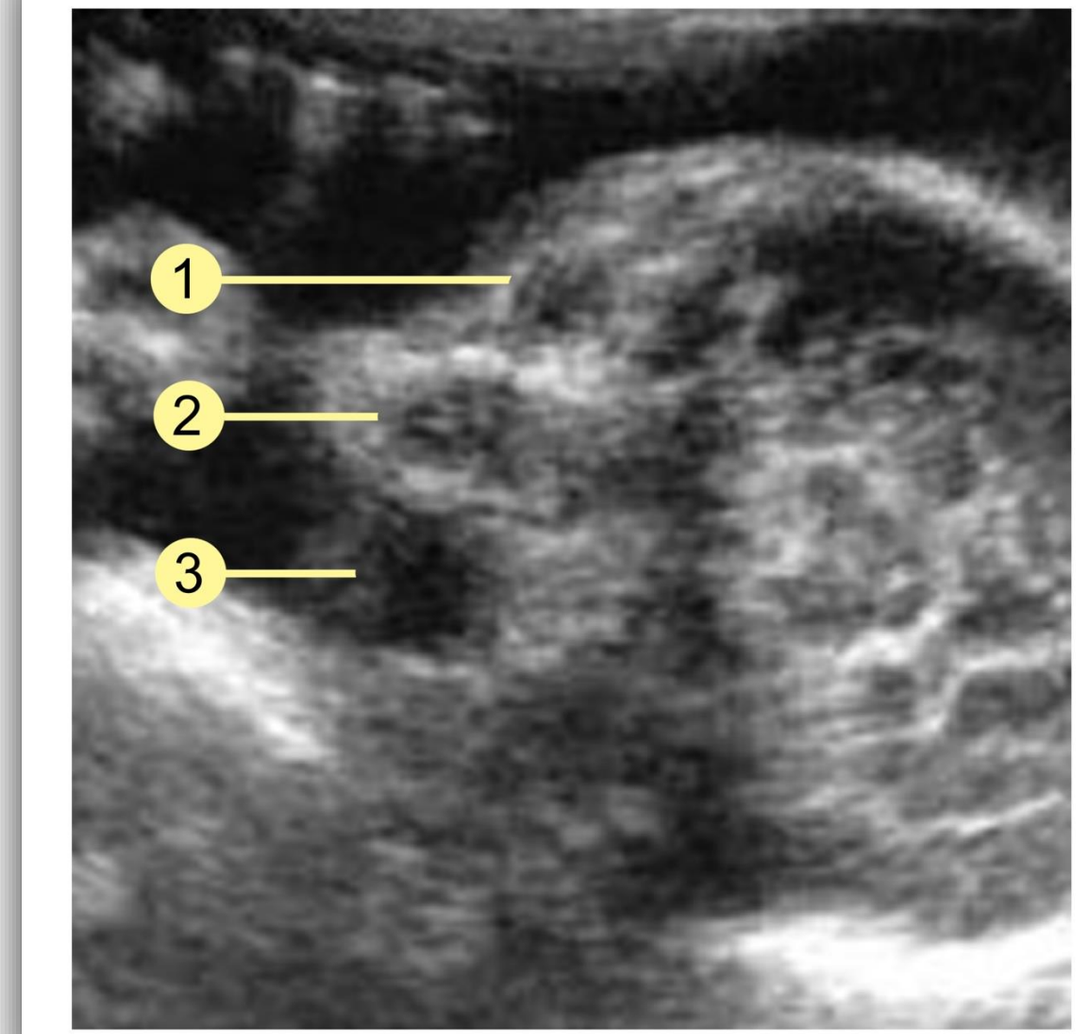
Microphthalmia

- Associated abnormalities include:
  - Holoprosencephaly
  - *In utero* infection
  - Fetal alcohol syndrome
  - Triploidy
  - Trisomy 13 (Patau syndrome)
  - Treacher Collins syndrome

Microphthalmia

- Sonographic findings include:
  - Reduced orbital size
  - Characteristic findings associated with other concomitant congenital conditions

MICROPHTHALMIA



1 = microphthalmic right eye
2 = nasal apparatus
3 = normal left eye

Anophthalmia

- Congenital absence of an eyeball within an orbit
- May be uni- or bilateral; isolated occurrence or syndromic in nature
- Causes include :
  - Failure of eye tissue to grow
  - Optic vascular maldevelopment
  - Secondary tissue degeneration due to toxic insult

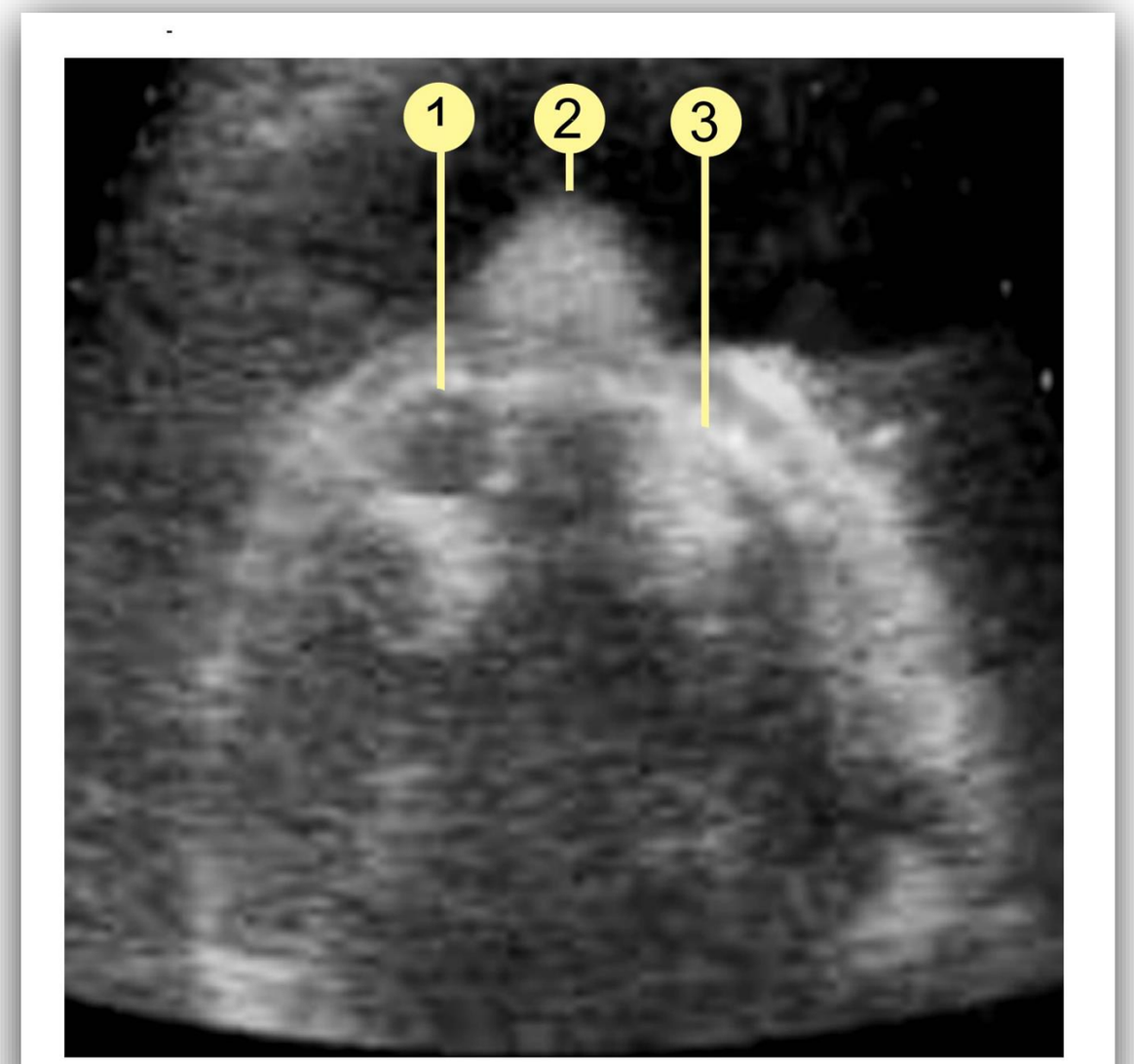
Anophthalmia

- Associated abnormalities include:
  - Trisomy 13 (Patau syndrome)
  - CHARGE association
  - Heterotaxy
  - Lenz microphthalmia syndrome (rare, inherited disorder)

Anophthalmia

- Sonographic findings include:
  - Absent anechoic eye chamber
  - Echogenic filling of orbital cavity

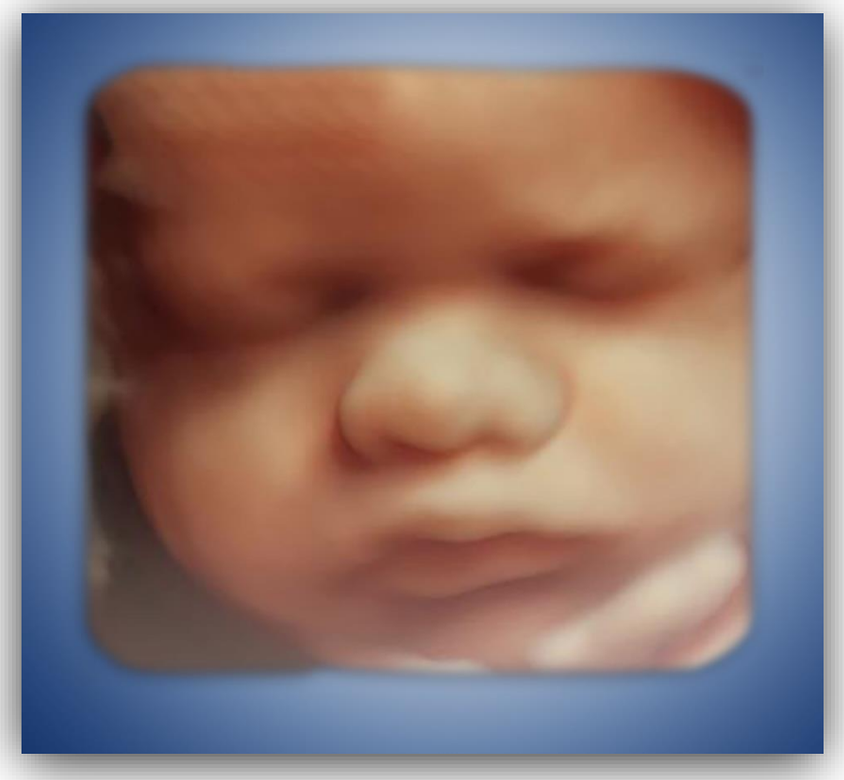
ANOPHTHALMIA



1 = normal left eye
2 = nose
3 = absent right eye

Categories of Abnormalities

- Chin & neck abnormalities
  - Micrognathia
  - Macroglossia
  - Neck cysts
  - Neck teratomas
  - Epignathus
  - Cystic hygromas



Chin & Neck Abnormalities

Micrognathia

- Abnormally small mandible
- Occurs as part of many syndromes
- If severe, may compromise postnatal respiration
- Diagnosis is usually a subjective one

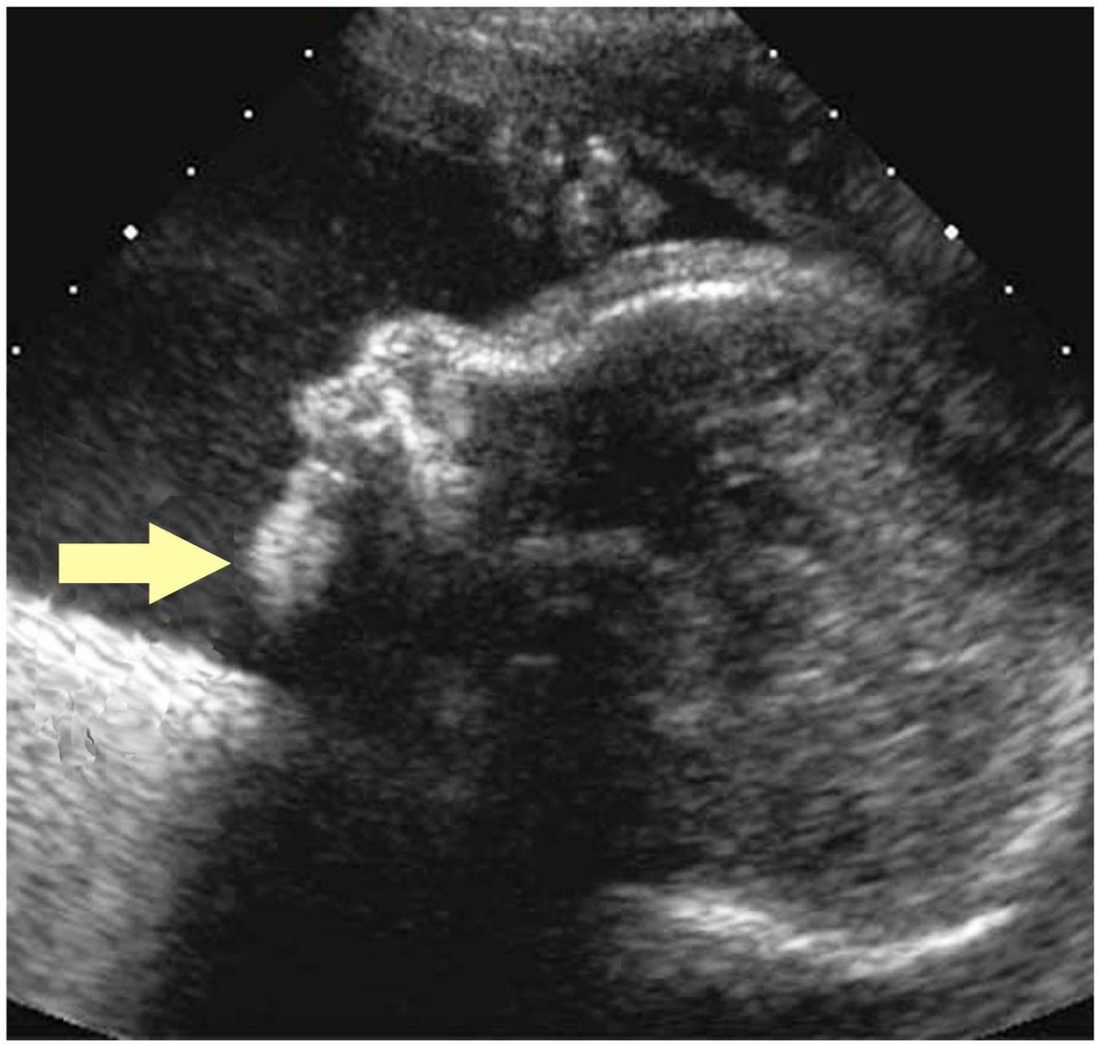
Micrognathia

- Associated conditions include :
  - True sagittal facial image demonstrated receding chin
  - Jaw index measurements consistent with observations
  - Polyhydramnios suggests impairment of fetal swallowing

Micrognathia

- Sonographic findings include:
  - Sagittal section demonstrates receding chin
  - Jaw index (AP mandible diameter ÷ BPOD) < 21
  - Polyhydramnios suggests impairment of fetal swallowing mechanism

MICROGNATHIA



Receding chin (arrow)

Macroglossia

- Abnormally large tongue
- Most common syndromic association is Beckwith-Wiedemann syndrome
- May also be caused by posterior solid mass displacing tongue forward

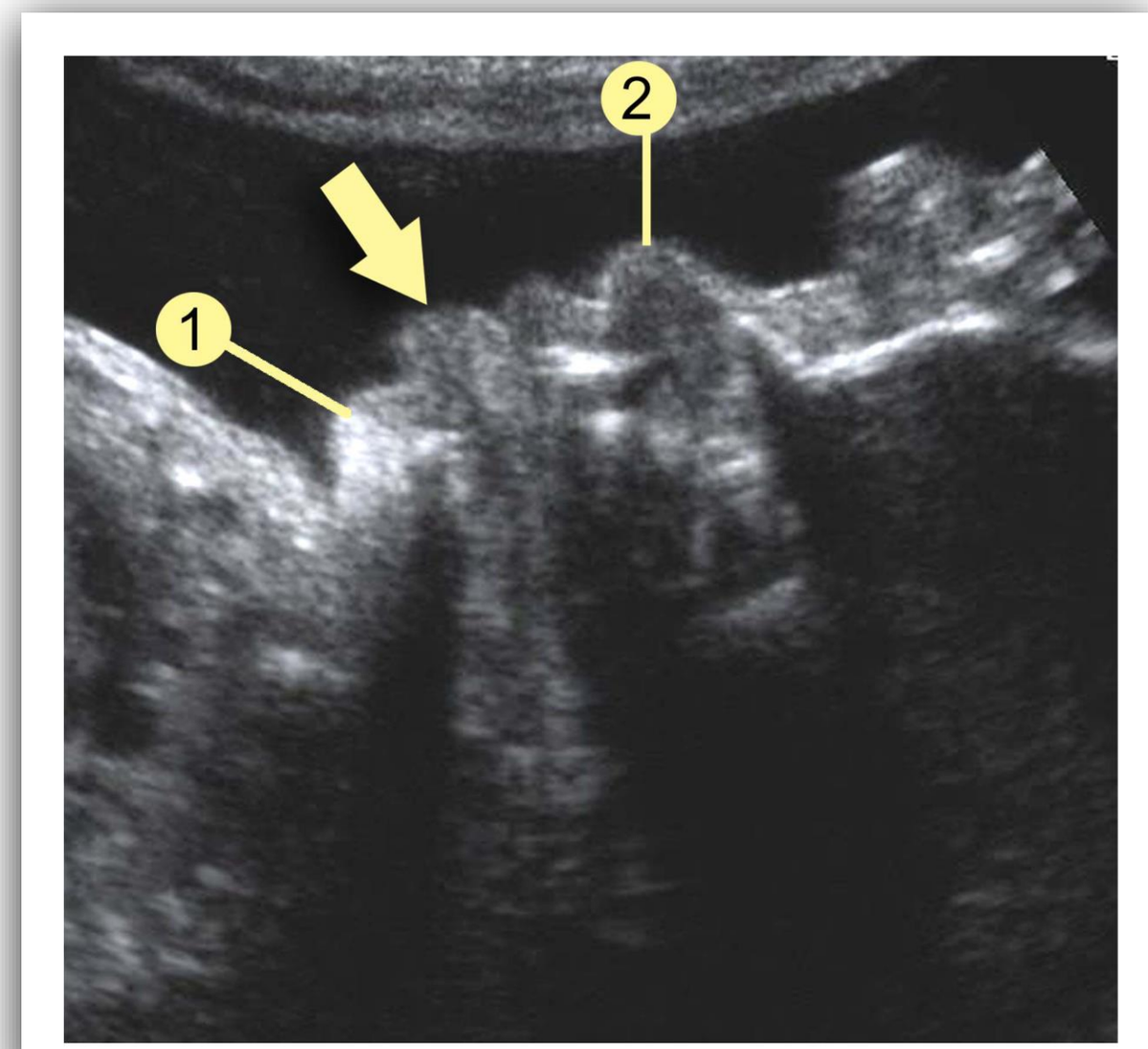
Macroglossia

- Associated conditions include :
  - Beckwith-Wiedemann syndrome
  - Congenital hyperthyroid syndrome
  - Trisomy 21 (Down syndrome)
  - Triploidy
  - Hunter syndrome

Macroglossia

- Sonographic findings include:
  - True midline sagittal image shows tongue protruding between upper and lower lips
  - Subjective appearance of a big tongue
  - Tongue remains outside of mouth regardless of swallowing movement of lips
  - Polyhydramnios suggests swallowing impairment

MACROGLOSSIA



1 = lower lip
2 = nose

Protruding tongue (arrow)

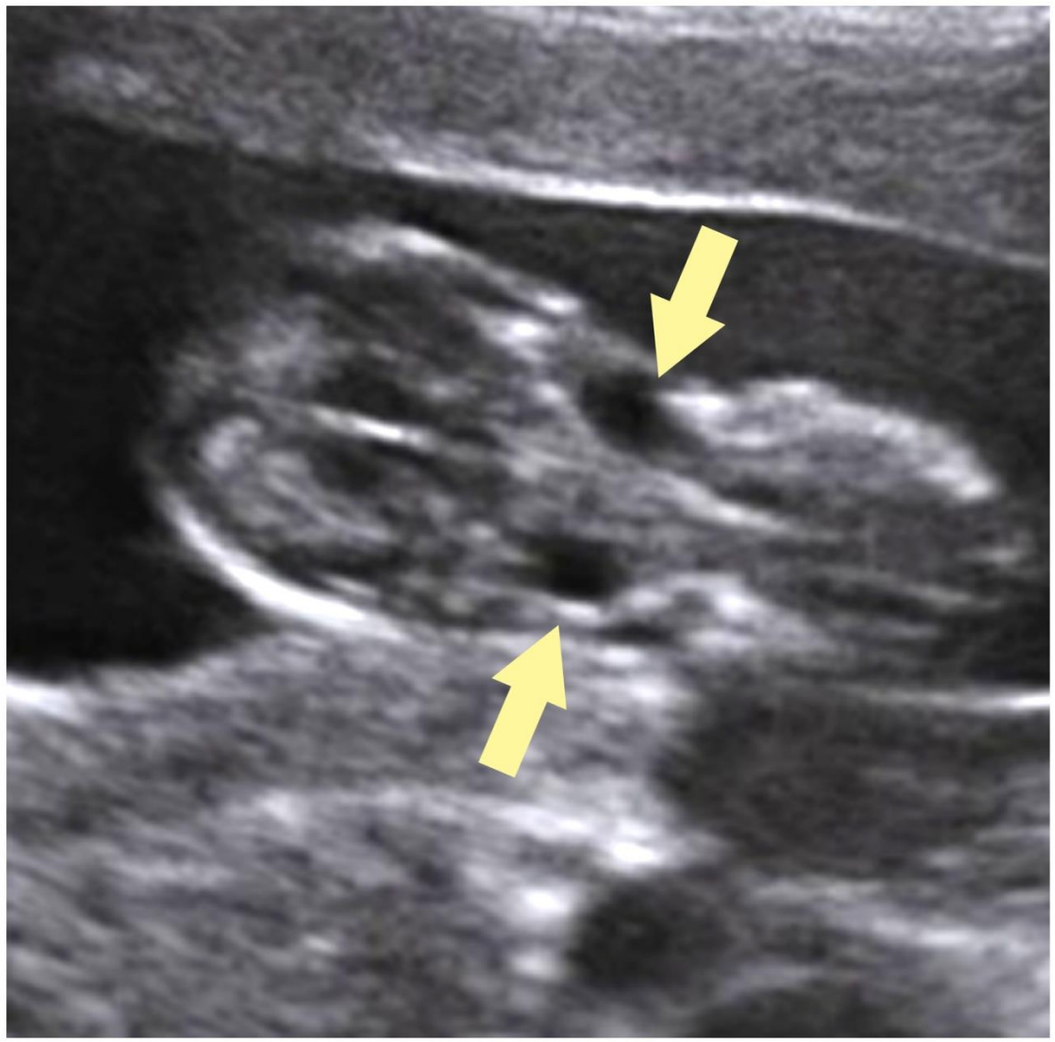
Neck Cysts

- Rarely encountered in prenatal imaging appearing more frequently as subdermal masses in children and young adults
- Two types:
  - Branchial cleft cysts: failure of branchial cleft to undergo embryonic obliteration. ***located laterally***
  - Thyroglossal duct cysts: p[persistence of thyroglossal tract. ***located midline***

Neck Cysts

- Sonographic findings include:
  - Smooth-bordered, anechoic mass in neck
  - Posterior acoustic enhancement
  - Displacement of normal midline structures

NECK CYSTS



Branchia cleft cysts (arrows)

Neck Teratomas

- Encapsulated tumors containing tissue from all three embryonic germ cell layers
- Gross pathological appearance varied widely
- Range in size and location
- Most immature teratomas are benign, however, mature teratomas carry malignant potential

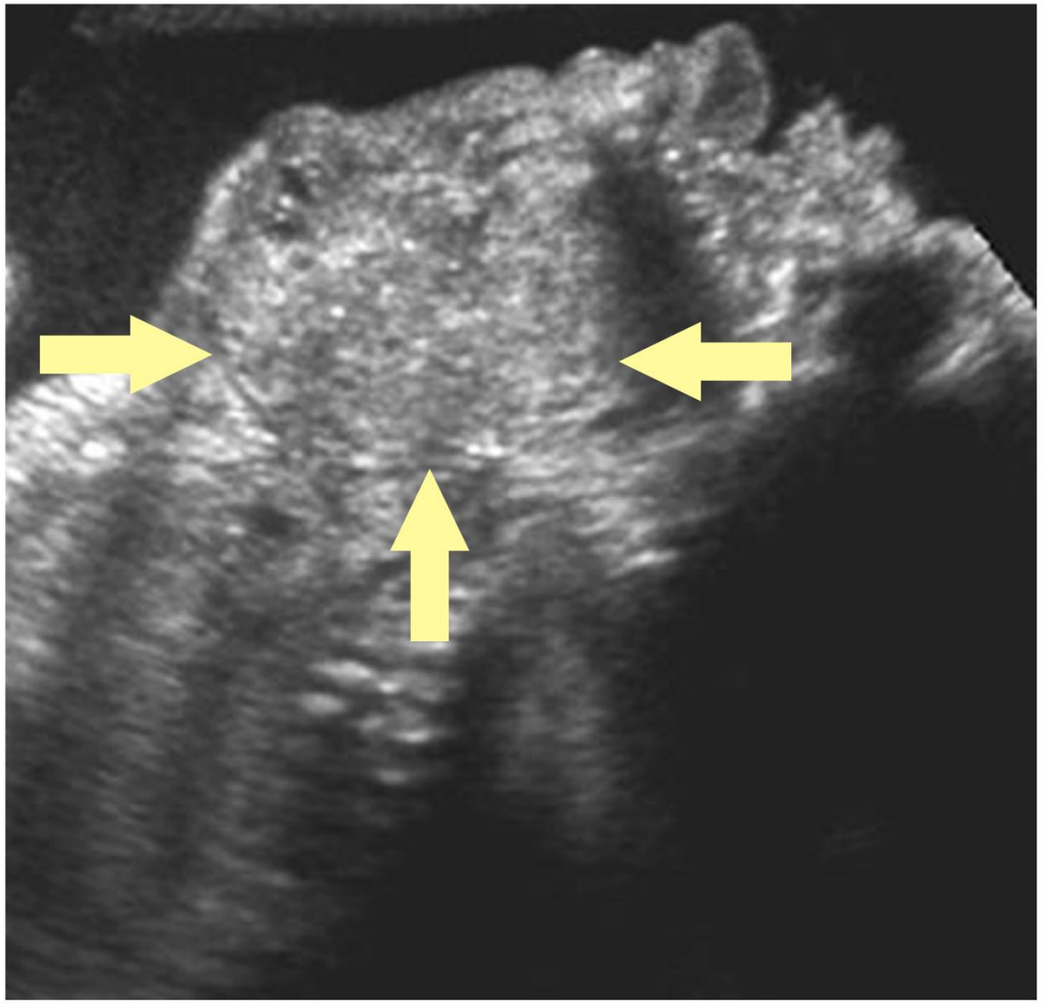
Neck Teratomas

- Associated conditions include :
  - Hydrops fetalis
  - Neonatal airway obstruction

Neck Teratomas

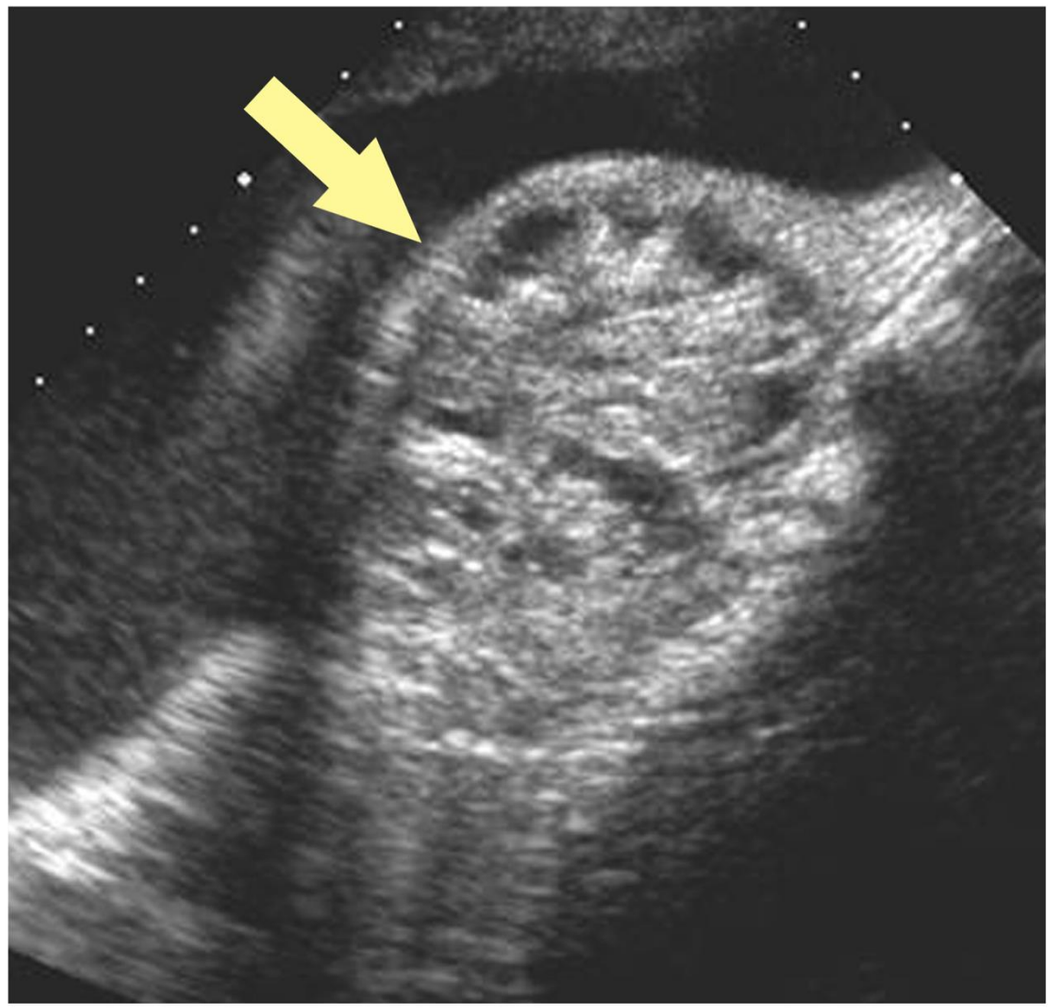
- Sonographic findings include:
  - Complex, cystic/solid tumor seen near fetal neck
  - Calcifications may be present
  - Polyhydramnios (30% of cases)

NECK TERTATOMAS



Sagittal view with hyperextension of neck

NECK TERATOMAS



Exophytic complex neck mass (arrow)

Epignathus

- Rare teratoma arising from oral cavity or pharynx
- Most commonly arise from sphenoid bone, however other sites of origin include: *soft palate, tongue, pharynx, jaw*
- Typical of teratoma, they include tissue from all three germ cell layers

Epignathus

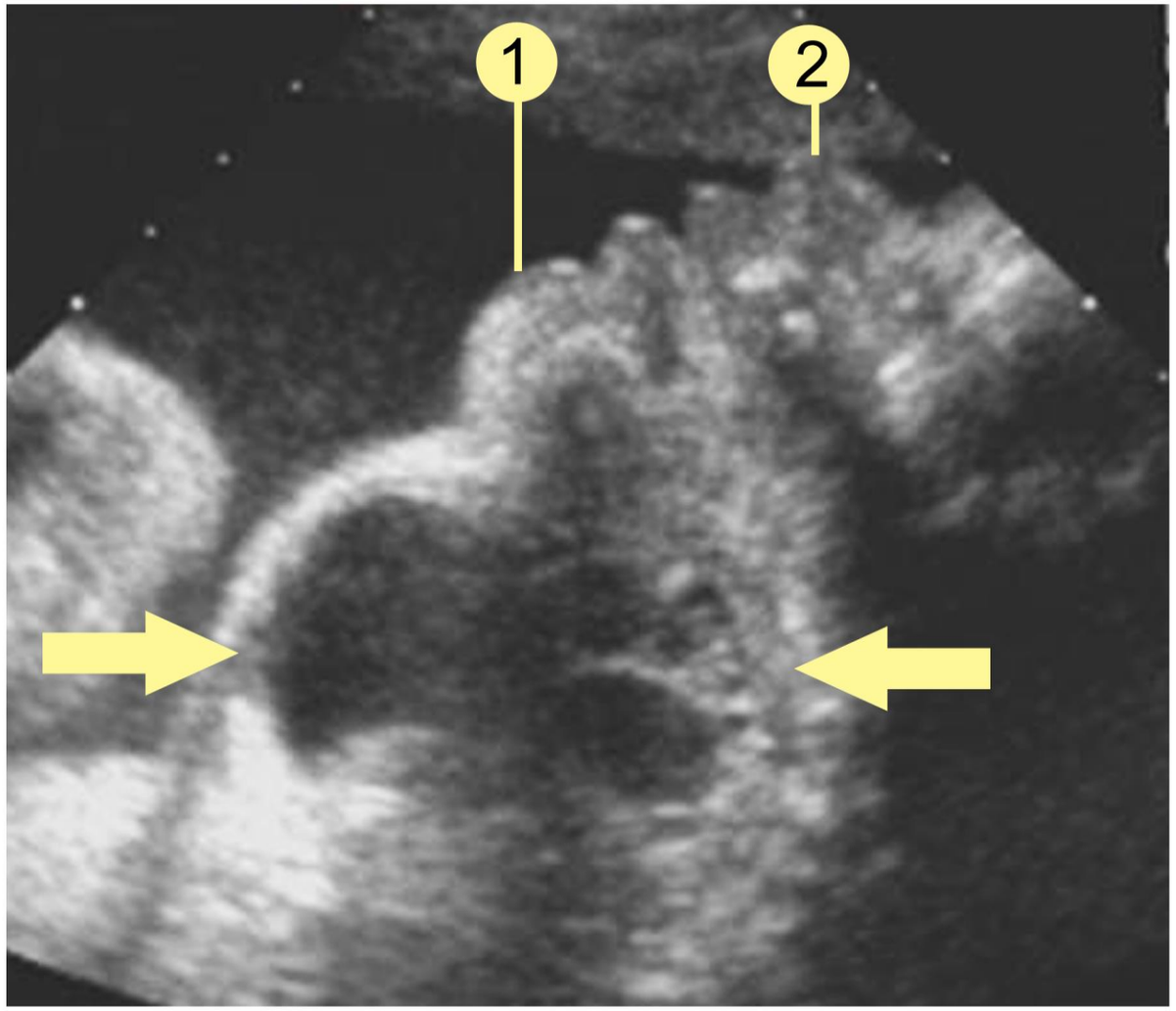
- Associated conditions include :
  - Cleft palate
  - Facial hemangiomas
  - Branchial cysts
  - Hypotelorism
  - Umbilical hernia
  - Congenital heart defects

Epignathus

- Sonographic findings include:
  - Complex, cystic/solid tumor protruding from mouth and/or neck
  - Polyhydramnios suggests impairment of swallowing mechanism

EPIGNATHUS

1 = chin
2 = nose



Complex mass arising from neck (arrows)

Cystic Hygromas

- Benign, developmental anomaly of lymphatic origin
- Characterized by single or multiple cystic fluid collections within soft tissue surrounding fetal neck
- Sizes of cystic lesions vary greatly
- May be simple or septated

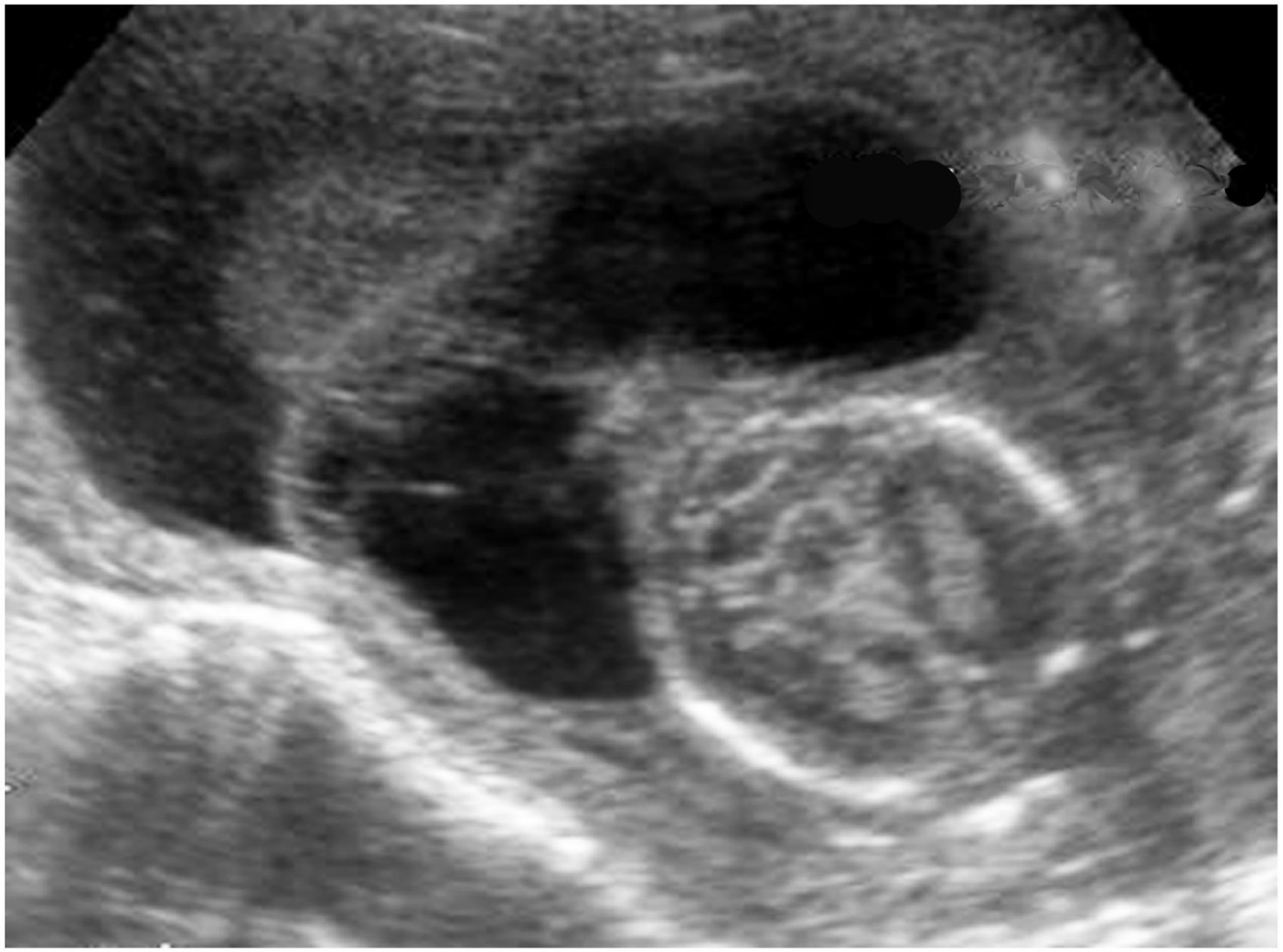
Cystic Hygromas

- Associated conditions include :
  - Turner syndrome
  - Trisomies 13, 18, 21
  - Noonan syndrome

Cystic Hygromas

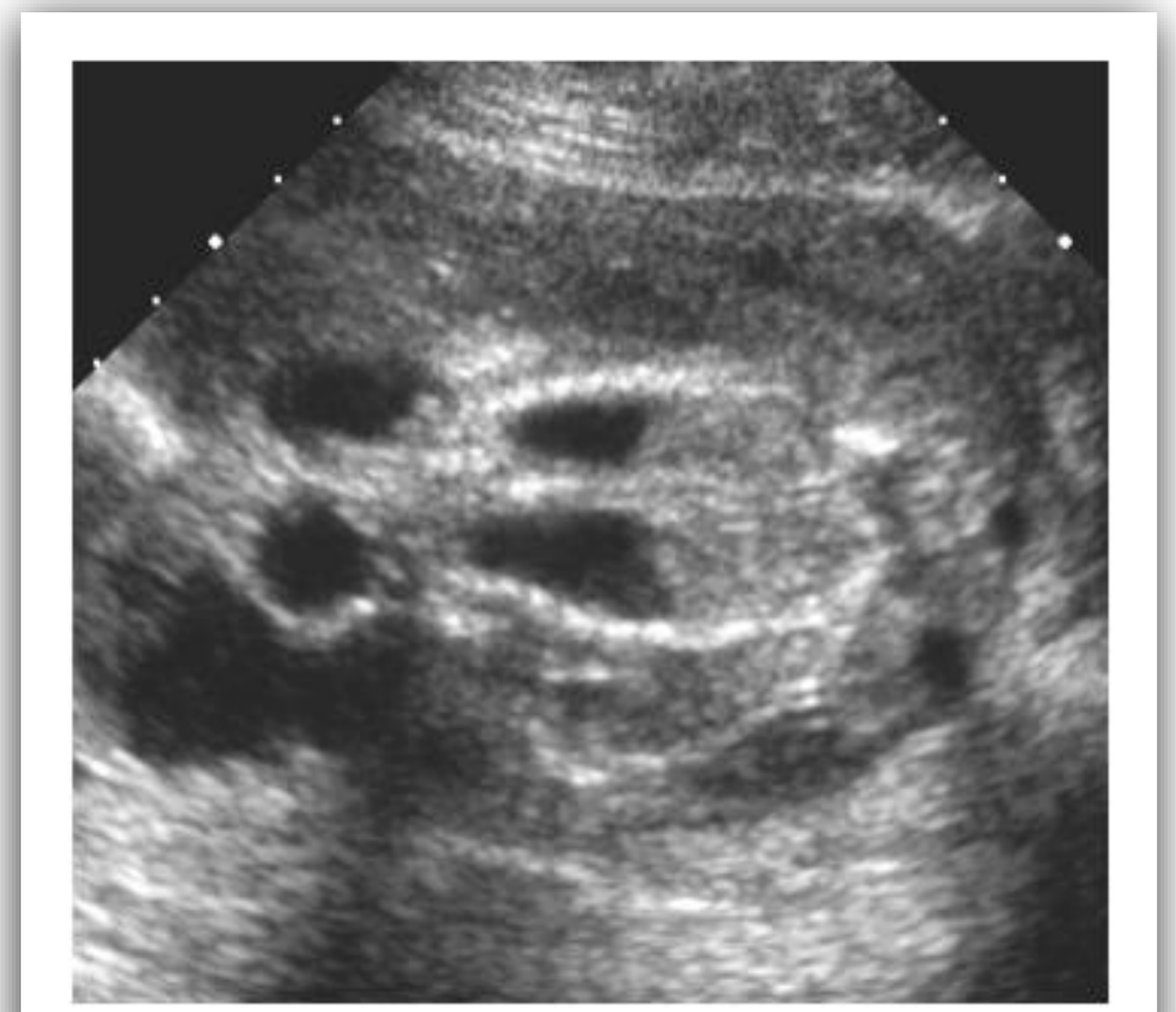
- Sonographic findings include:
  - Fluid-filled structure(s) presenting as cystic masses contiguous with chest wall
  - Thin-walled, multiseptated cysts usually located posterior to fetal head/neck
  - Associated with fetal ascites, fetal edema (anasarca), enlarged edematous placenta, intradermal fluid collections (cystic cutaneous lymphangiectasis)
  - May mimic a cervical teratoma or NTD

CYSTIC HYGROMAS



Cystic mass arising from neck

CYSTIC HYGROMAS



Cystic neck mass, pleural effusion

OB GYN SONOGRAPHY REVIEW

Fetal Face and Neck