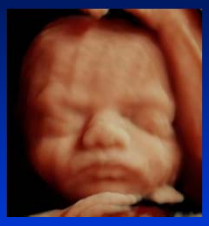
Evaluation of the Fetal Face

Judy A. Estroff, MD



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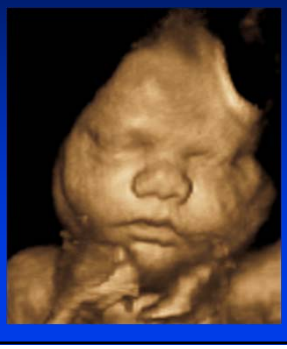
Approach and Goals

- Basic approach: speak the plastic surgeon's language
- Immediate goal: accurate diagnosis and classification of craniofacial anomalies
- Ultimate goal: improved parental counseling and patient outcome

Overview

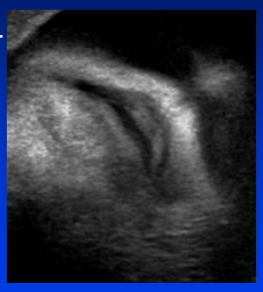
- The normal face
- Cleft lip and palate
- Abnormal profile
- Micrognathia
- Abnormal head shape
- Ear anomalies

The Normal Face



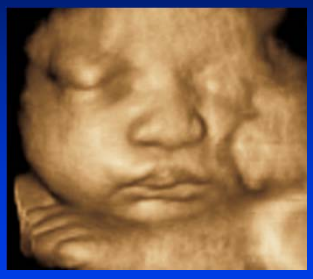
Anatomy of the Lip

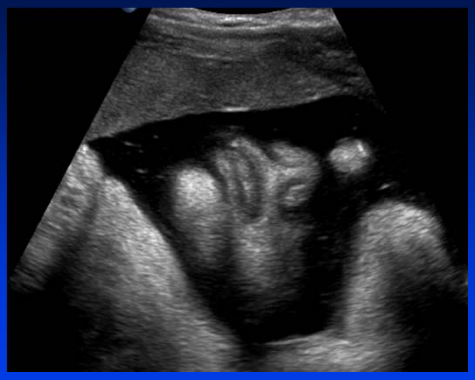
- Vermillion border
- White roll



Anatomy of the Nose

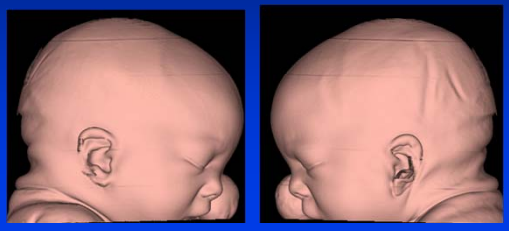
- Tip
- Nostril
- Alar base
- Philtrum





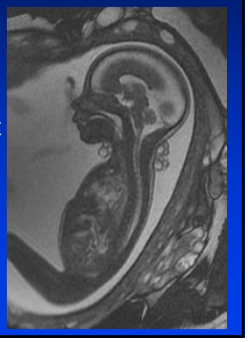
Anatomy of the Ears

- Top of helix should be at level of inner canthal line



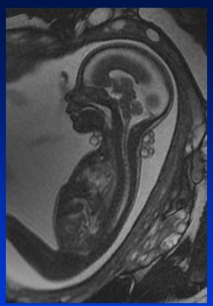
The Normal Profile

- Forehead and chin on same plane
- Nasal bone should be present
- Nose should project beyond plane of forehead and chin
- Top of ear at level of orbit

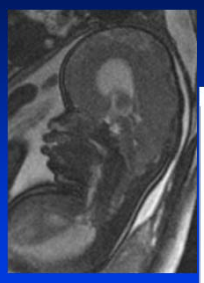


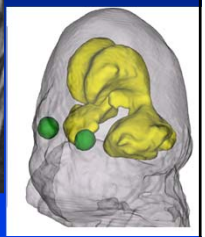
Abnormal head shape

- Brachycephaly
- Dolichocephaly
- Turribrachycephaly
- Microcephaly
- Macrocephaly
- Trigonocephaly



normal



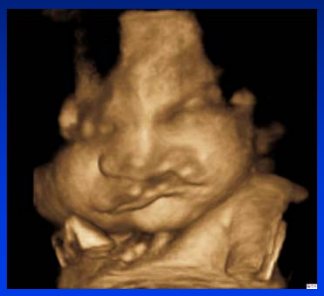


turibrachycephaly

Abnormal head shape: etiologies

- Craniosynostosis
- Microcephaly/Macrocephaly
- Open neural tube defect
- Hemifacial microsomy
- Local deformation (oligo, fibroids)
- Syndromes

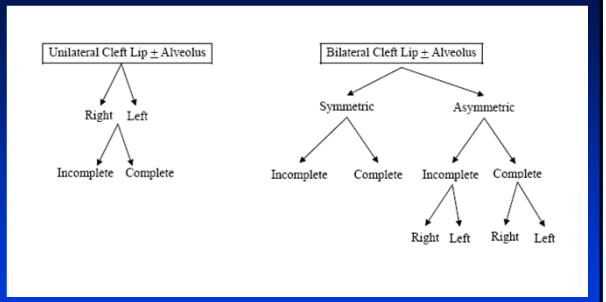
Cleft lip and palate



Cleft lip

Description of Cleft Lip

- Sidedness: unilateral, bilateral
- Symmetry: symmetric, asymmetric
- Extent: complete, incomplete
- Severity: mild, moderate, severe



Subtypes of Cleft Lip

- Mini-microform
- Microform
- Minor-form
- Incomplete
- Complete

Examples of minor form cleft lip

Problem

- Cleft lip and palate often go undetected on screening and targeted fetal sonography
- Detection rate low for cleft lip, lower for cleft palate
- It is important to know if the palate is cleft
  - accurate prenatal counseling of parents
  - to prepare parents for postnatal repair
  - if the palate is not cleft, the child will not have abnormal speech, recurrent ear infections or diminished midfacial growth

2D and 3D US of Cleft Palate

- Ultrasound accuracy in detecting cleft palate in the setting of cleft lip remains low, despite best 2D and 3D imaging
- Sensitivity 33-63%
- Specificity 84-95%

Pretorius, et al. J Ultrasound Med 2010; 29:357-364

Cleft Palate: Limitations of US

- Why is accuracy of US so low?
- Unfavorable fetal position
- Shadowing by adjacent bony structures

Pretorius, et al. J Ultrasound Med 2010; 29:357-364

Cleft Palate: Fetal MRI

- Preliminary work suggests sensitivity and specificity of fetal MR for detection of cleft palate is high (>90%)

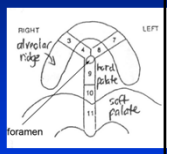
Manganaro L, Tomei A, Fierro F, et al. Fetal MRI as a complement to US in the evaluation of cleft lip and palate. Radiol Med 2011; 116:1134-48
Descamps MJ, Golding SJ, Sibley J, etal. MRI for definitive in utero diagnosis of cleft palate: a useful adjunct to antenatal care? Cleft Palate Craniofac J 2010;47:578-85
Estroff JA, et al. IFMSS 2011: Accuracy of Fetal MR in detection of cleft secondary palate in the setting of cleft lip

Cleft Lip and Palate: our approach

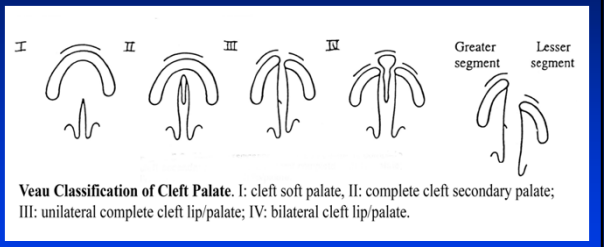
- Ultrasound for lip and alveolus=primary palate
- MRI for secondary palate, both hard and soft

Primary and Secondary Palate

- Primary palate= lip and alveolus; all structures anterior to incisive foramen
- Secondary palate= all structures posterior to incisive foramen. Includes part of the hard palate and all of the soft palate



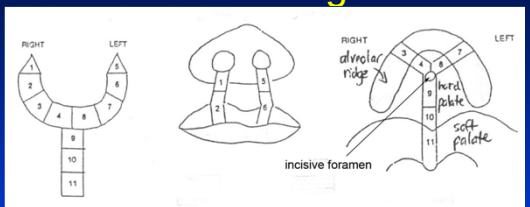
Types of Cleft Palate: Veau Classification



Veau Classification of Cleft Palate. I: cleft soft palate, II: complete cleft secondary palate; III: unilateral complete cleft lip/palate; IV: bilateral cleft lip/palate.

Modified by Mulliken, JB, data from Marrinan EM, LaBrie RA, Mulliken JB. Velopharyngeal function in nonsyndromic cleft palate: relevance of surgical technique, age at repair, and cleft type. Cleft Palate Craniofac J. 1998 Mar;35(2):95-100).

The "Y Diagram"

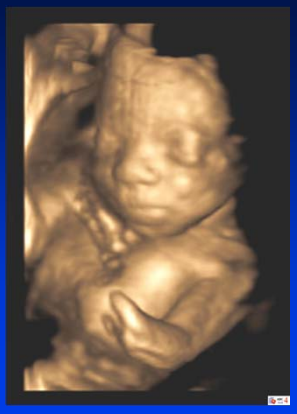


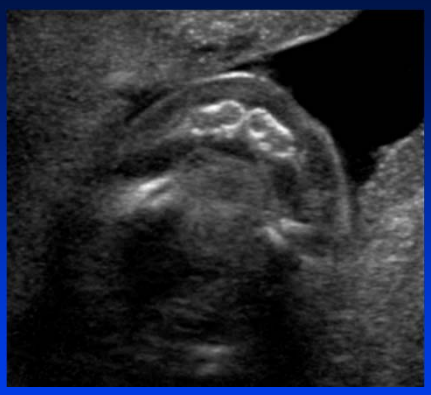
Key to Modified Kernahan Y-Diagram	
1 and 5	Nostril and nasal floor (R, L)
2 and 6	Lip (R, L)
3 and 7	Alveolus (R, L)
4 and 8	Hard Palate anterior to incisive foramen (still primary palate) (R, L)
9 and 10	Hard Palate posterior to incisive foramen (secondary palate) (midline)
11	Soft palate (midline)

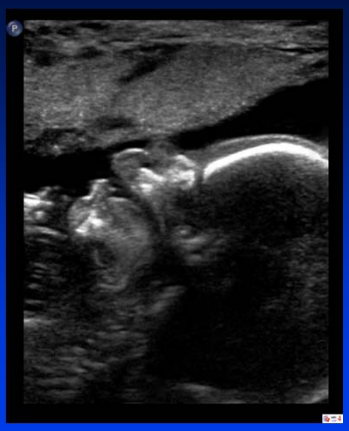
Adapted from: Kernahan, D.A. The striped Ya symbolic classification for cleft lip and palate. Plast. Reconstr. Surg. 47(5): 469, 1971.

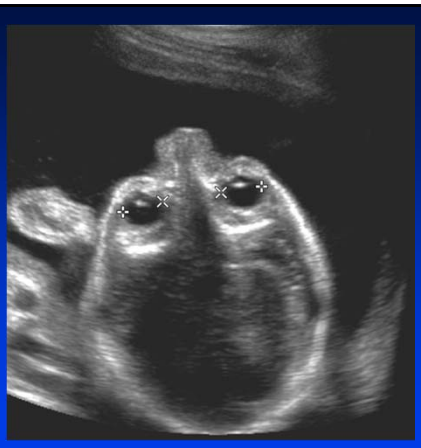
Sonography

- Lips
- Nostrils
- Vomer
- Maxilla and Mandible
- Orbits
- Profile
- Position of tongue









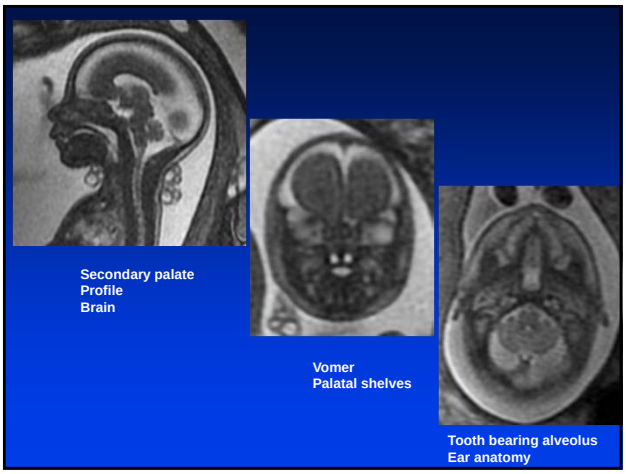
Sonographic Protocol for Evaluation of Fetal Face and Neck: 2D + 3D	
I. Skull and Brain Axial, Coronal and Sagittal planes	• Skull shape: (normal, dolichocephalic, brachycephalic, unusual) • Sutures: Open, no defects • Normal underlying brain • Measurements: Normal head size compared to long bone and abdominal biometry for given gestational age. (BPD/OFD/HC vs. GA ,AD, FL, HL) • Skin around head: normal, thickened, mass
II. Face Axial, Coronal and Sagittal planes	• Mouth: vermillion border intact • Nares: symmetric, not flattened or cleft • Vomer : straight, midline • OOD/IOD, OD, orbital shape, symmetry : 2 orbits with normal size globes and normal interocular distance • Maxilla : normal intact arc-shaped alveolus • Mandible: normal intact arc-shaped alveolus • Tongue: position, size • Ears – size, shape, position • Forehead: normal, sloped, bulging • Midface: normal, retruded • Chin: normal vs. small (micrognathia), retruded (retrognathia) or both
III. Neck Axial, Coronal and Sagittal planes	• Head position • Length of spine, curvature • Presence of nuchal cord, skin thickening • Integrity of vertebrae, lamina and spinous processes • Position of trachea, thyroid, strap muscles • Evaluate vallecula and pyriform sinuses, glottis, subglottic trachea, carina

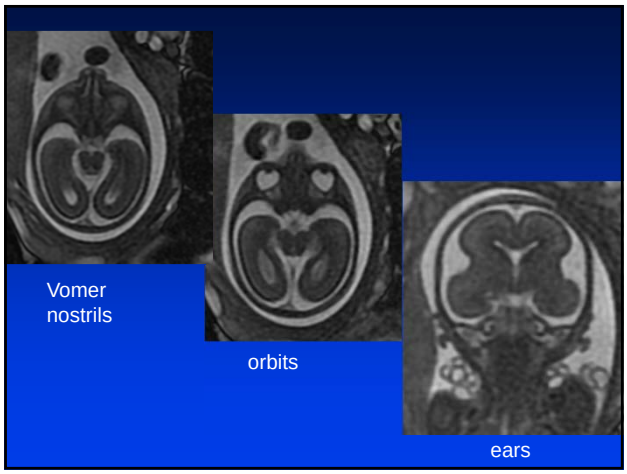
BPD= biparietal diameter
OFD= occipitofrontal diameter
HC= head circumference
GA= gestational age
AD= abdominal diameter
FL= femur length
HL= humeral length
OOD= outer orbital distance
IOD= inner orbital distance
OD= ocular diameter

Associated Signs of Cleft Palate in Presence of Cleft Lip	
Axial/Coronal views	• **Lips** – cleft • **Nares** –flattened or deformed • **Vomer** – deviated away from side of cleft; often midline if bilateral cleft lip and palate • **Maxilla** – interrupted alveolus, wide gap • **Orbits** – hypertelorism
Sagittal view	• **Profile** – midface retrusion • **Position of tongue** – high

MR

- Midline sagittal: primary bony palate and secondary soft palate; fetal profile
- Coronal: Vomer intersecting bony palate; inner and outer orbital distances
- Axial: Tooth-bearing alveolus





Always evaluate:

- Corpus callosum
- Cerebellar vermis
- Rest of fetal body for associated anomalies

Importance of accurate classification

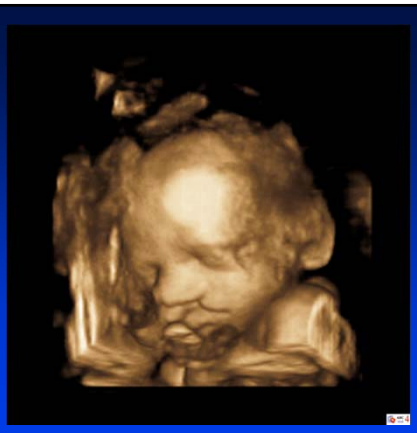
- If the secondary palate is NOT cleft, the child will NOT have hearing, speech and feeding difficulties
- If the secondary palate IS cleft, all of these problems are expected AND the surgery is more extensive

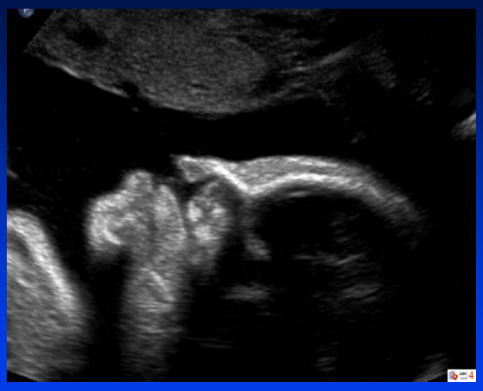
Abnormal fetal profile

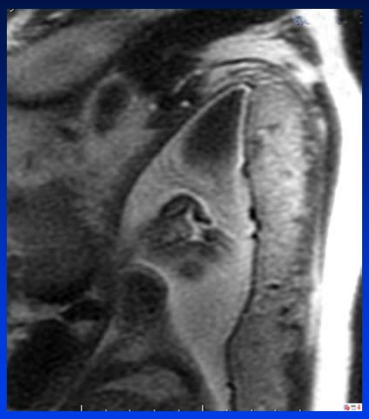
- Sloping forehead
- Bulging forehead
- Midface hypoplasia
- Micrognathia
- Retrognathia
- Arhinia
- Agnathia

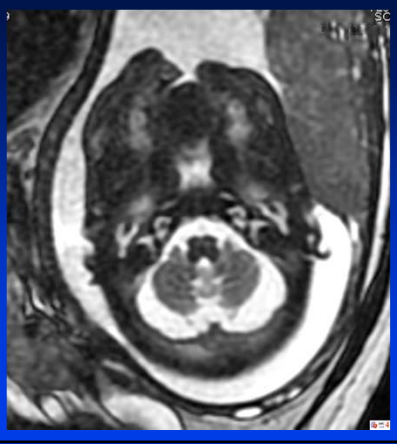
Cleft examples

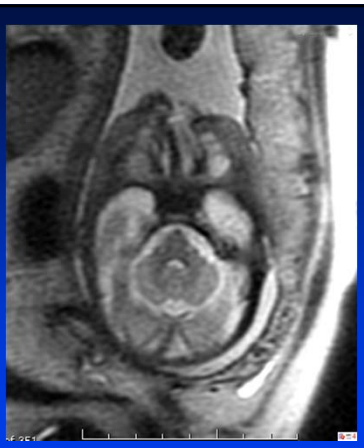
Unilateral complete cleft lip and palate

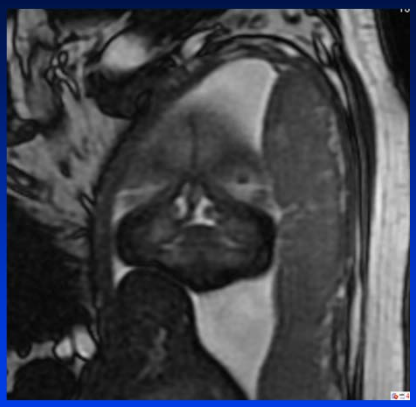


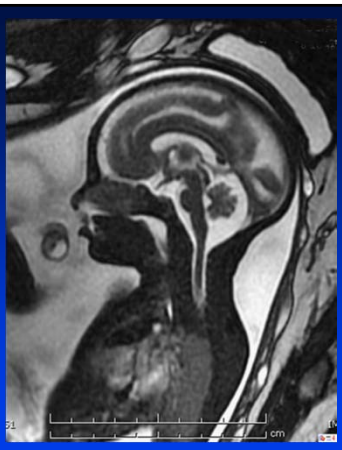






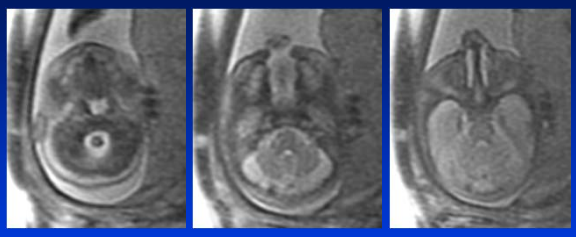




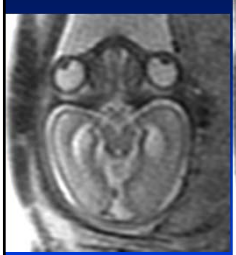


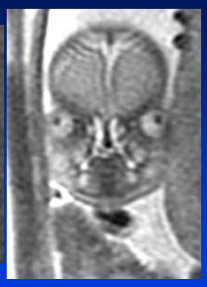
Bilateral cleft lip and palate

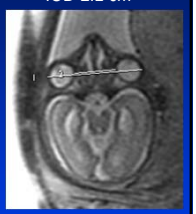
- Vomer usually midline
- 2 tooth buds in intermaxillary segment
- Hypertelorism



25w







OOD 4.8cm
IOD 2.1 cm

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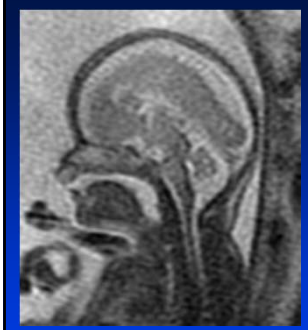

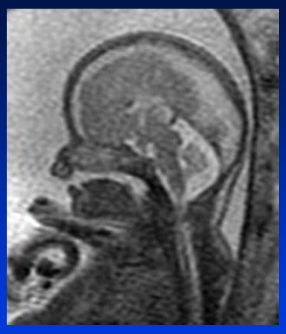

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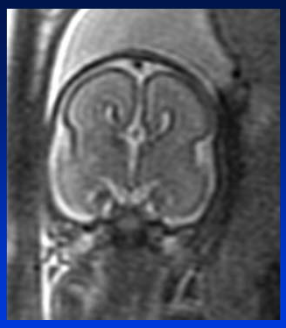
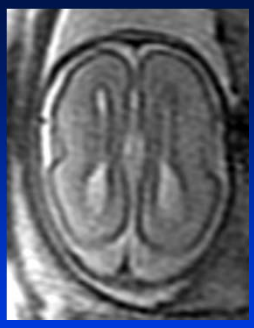

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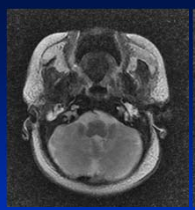
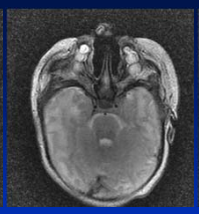
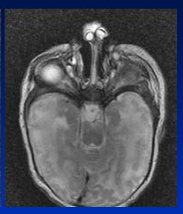
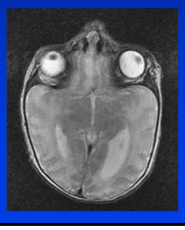
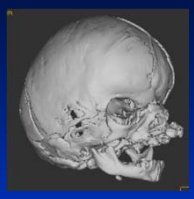
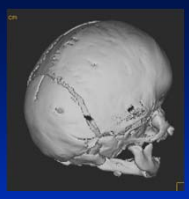
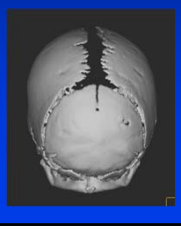
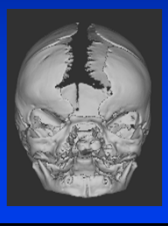





DOL 2

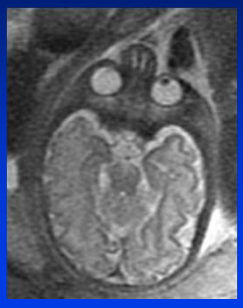






DOL 2: bilateral coronal craniosynostosis

Final diagnosis: Apert Syndrome


Atypical clefts

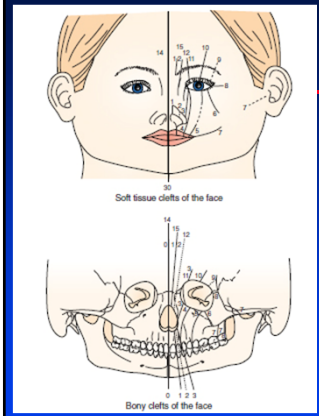
Twins: Referred at 35 weeks for a facial abnormality in one twin.



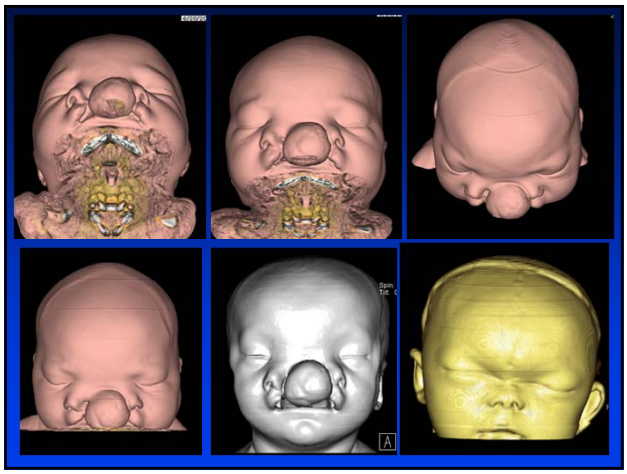
35w

Day 1 of life

Tessier Cleft



Lorenco AP, Estroff JA. The Fetal Face and Neck. In: Rumack CM, Wilson SR, Charboneau JW, Levine D, ed. Diagnostic Ultrasound, 4th ed, Mosby, 2010. *(modified from Tessier, P MD, Anatomical classification facial, cranio-facial and latero-facial clefts. Journal of Maxillofacial Surgery, 1976;4:69-92)*



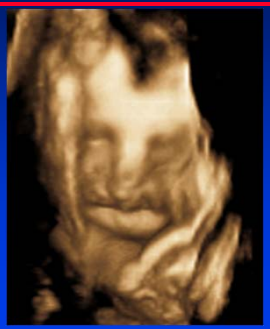
Tessier I4 facial cleft

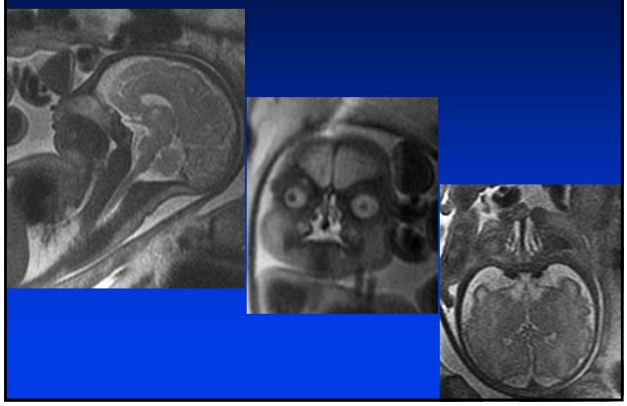
- Frontal encephalocele
- Cleft palate
- Hypertelorism
- Airway obstruction

Abnormal Fetal Profile

- Sloping forehead
- Midface hypoplasia
- Absent nose
- Absent mouth
- Micrognathia
- Protuberant tongue
- Masses; hydrops

Sloping forehead



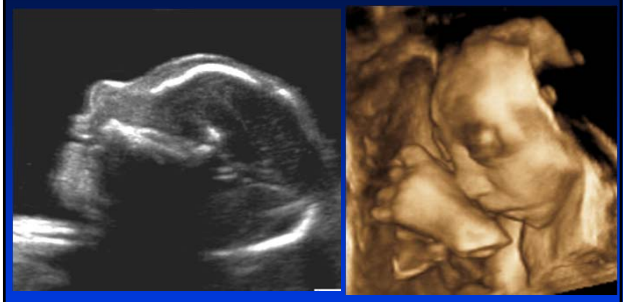


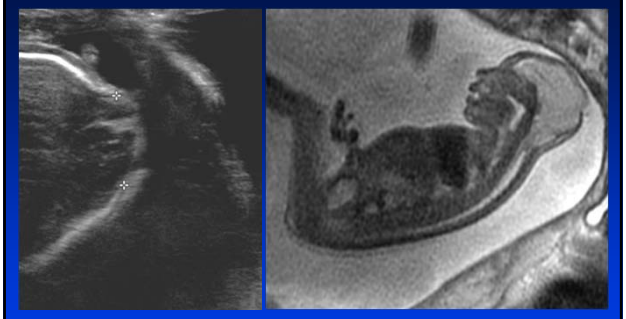
Wolf-Hirschhorn Syndrome (4p-)

- Broad bridge of the nose continuing to forehead
- Microcephaly
- High forehead with prominent glabella
- Ocular hypertelorism, epicanthus, highly arched eyebrows, short philtrum, downturned mouth
- Micrognathia
- Poorly formed ears with pits/tags
- IUGR, hypotonia

Battaglia A, Carey JC, South ST, et al. Wolf-Hirschhorn Syndrome. 2002 Apr 29 [Updated 2010 Jun 17]. In: Pagon RA, Bird TD, Dolan CR, et al., editors. GeneReviews™ [Internet]. Seattle (WA): University of Washington, Seattle; 1993-. Available from: http://www.ncbi.nlm.nih.gov/books/NBK1183/

History:
26 week fetus with sloping forehead





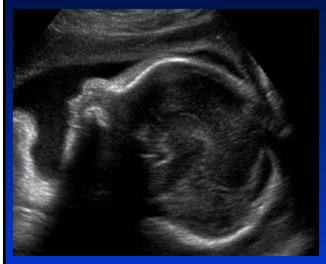
Occipital encephalocele

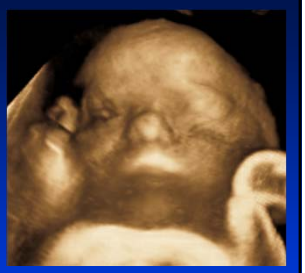
Mouth and jaw anomalies

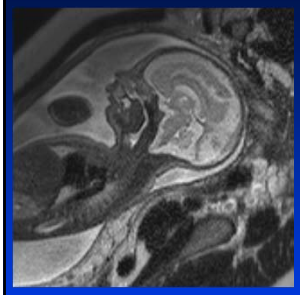
- Micrognathia: small mandible
- Agnathia: absence of mandible
- Otocephaly: hypoplasia or absence of mandible
- Microstomia or aglossia: small or absent mouth
- Macrostomia: large or wide mouth
- Macroglossia: enlarged tongue

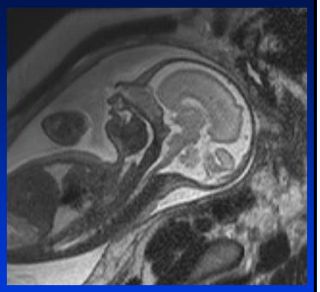
Micrognathia

- Micrognathia: small mandible
- Retrognathia: posteriorly displaced receding chin
- Glossoptosis: abnormal posterior position of tongue









Robin Sequence: Triad

- Cleft palate
- Micrognathia
- Glossoptosis

Robin Sequence: Etiology

- Primary abnormality: ? small mouth
- Tongue falls back= glossoptosis
- Inhibits fusion of palate leading to cleft

Robin Sequence: Prenatal Diagnosis

- Polyhydramnios
- Micrognathia
- High arched cleft palate

Robin Sequence: Prognosis and Management

- Concern for upper airway obstruction
- Neonatal respiratory distress
- Feeding problems
- If survive infancy, jaw variably grows, and child often does well
- Autosomal recessive recurrence risk
- Differential dx includes: T13, T18

Syndromes Associated with Micrognathia

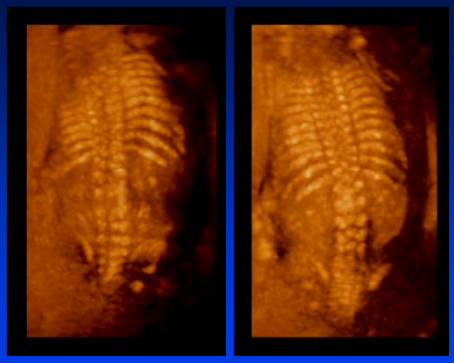
- Etiology: monogenic, chromosomal, teratogenic, disruptive
- Nager syndrome
- Stickler Syndrome
- CHARGE syndrome
- Miller-Dieker syndrome
- Goldenhar syndrome
- Treacher-Collins syndrome
- Etc
  - Cohen, MM. The Child With Multiple Birth Defects. 1997 2nd edition

Craniosynostosis

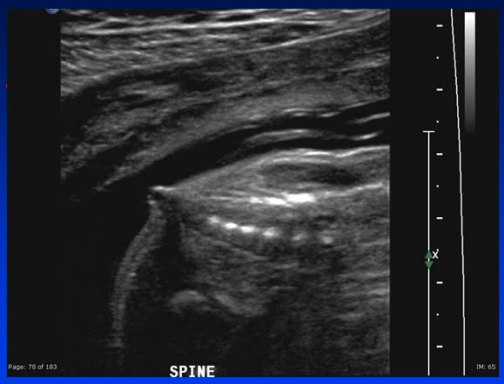
- Premature fusion of one or more cranial sutures
- Abnormal calvarial shape
- Exorbitism/exophthalmos
- FGFR (fibroblast growth factor related)
- Often syndromic (Apert, Crouzon, Pfeiffer)

Referral History

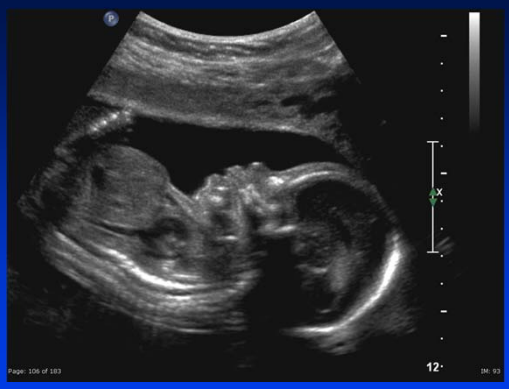
- 21 weeks: scoliosis and possible meningocele



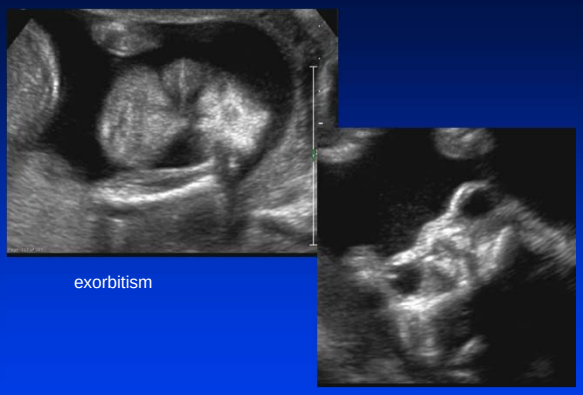
21w6d



21w6d



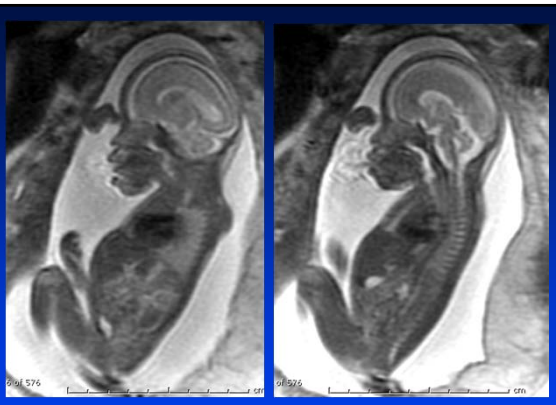
21w6d



exorbitism

hypertelorism

21w6d

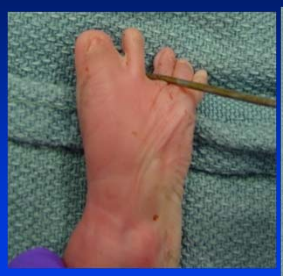


21w6d

Summary of imaging findings at 21 weeks, 6 days

- Brachycephaly
- Hypertelorism
- Exorbitism
- Midface hypoplasia
- Broad short mandible and maxilla
- Macroglossia
- Scoliosis
- “Tail”

Outcome: demise at 27 weeks Presumed Pfeiffer syndrome



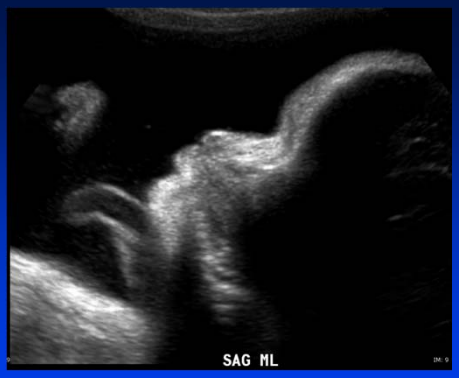
Broad great toe

Flat midface, lowset ears

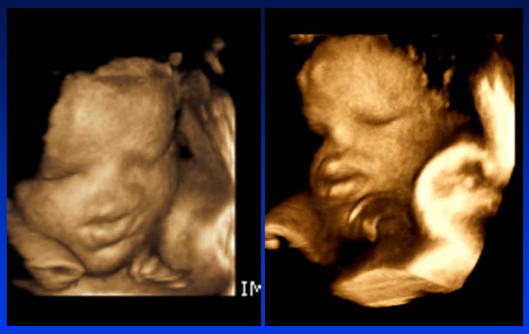
Nose anomalies

- Arhinia: absent nose
- Midface hypoplasia
- Nasal glioma
- Encephalocele
- Accessory nostril

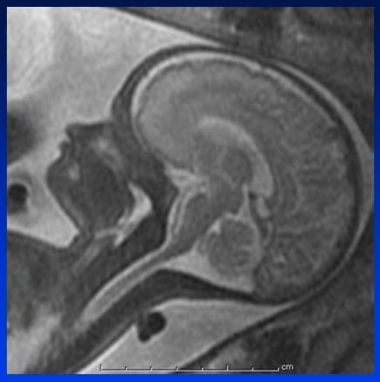
Arhinia: absent nose



34w2d



34w2d



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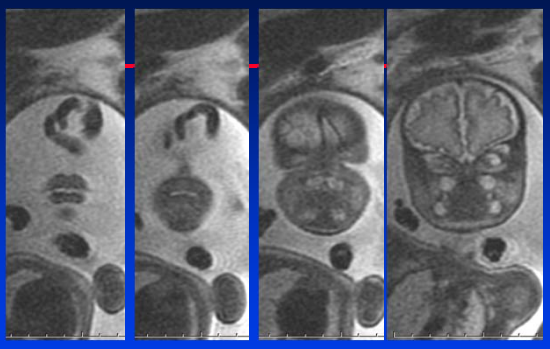
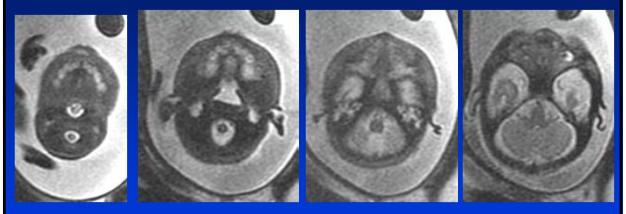


34w2d



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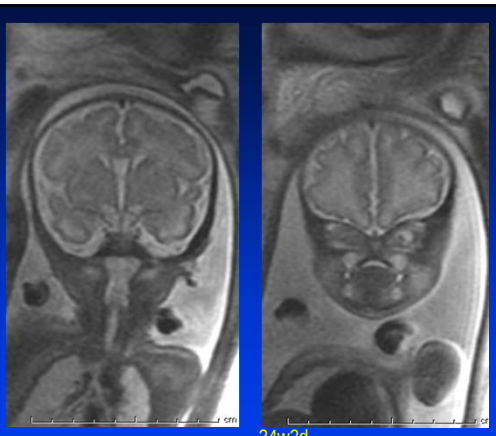
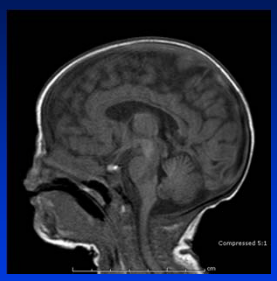
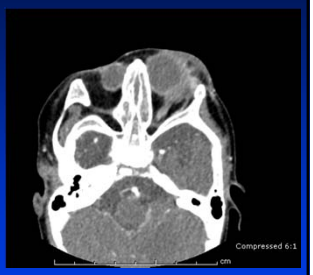


34w2d



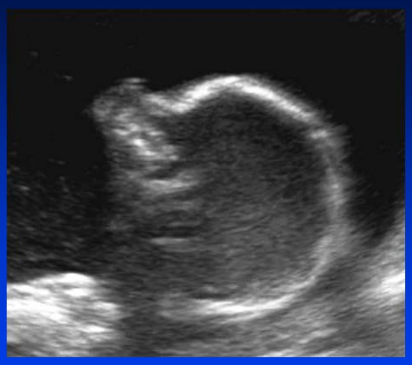
Absent nasal cavity

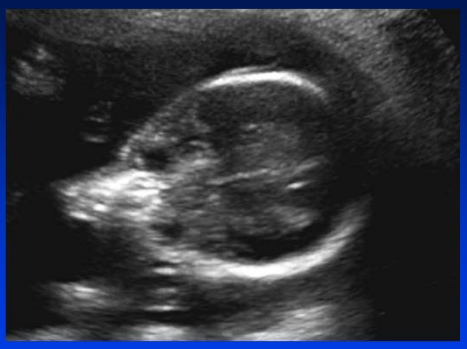


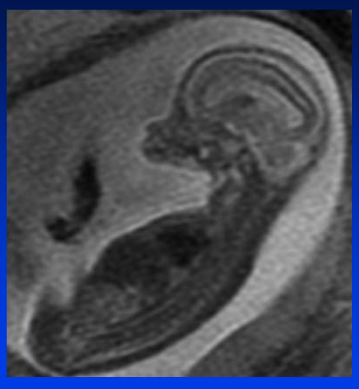
micropthalmia

1 wk

Agnathia: congenital absence of the lower jaw



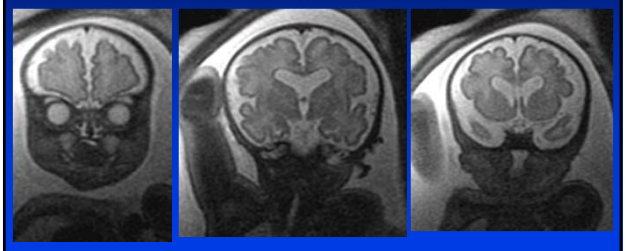




Ear anomalies

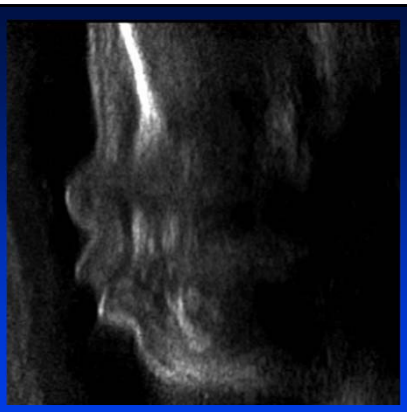
- Anotia
- Microtia
- Low set ears
- Protruding ears
- Auricular tags

Goldenhar Syndrome:
hemifacial microsomia



Trisomy 18

Midface hypoplasia, hypotelorism, lowset ears, ACC, clenched hands:

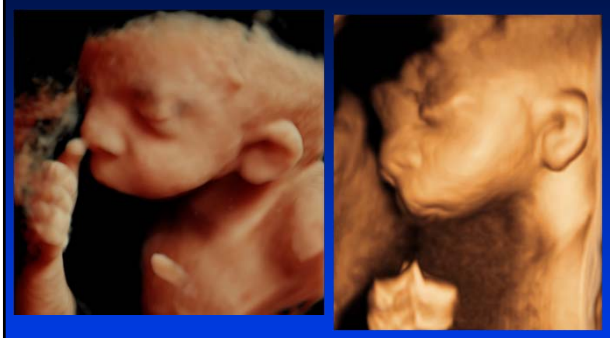


Profile looks normal, but no nasal bone: T21

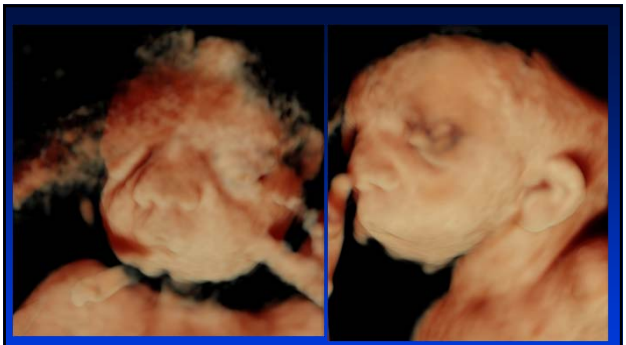
History

- 25 w: Multiple anomalies:
  - agnathia, microstomia, pelvic kidney and polyhydramnios
- 29 2/7w: admitted for PPROM
- 29 3/7w: STAT C-section- found fully dilated and fetus in breech position

BWH



26w5d poly, micrognathia, microstomia, low large ears

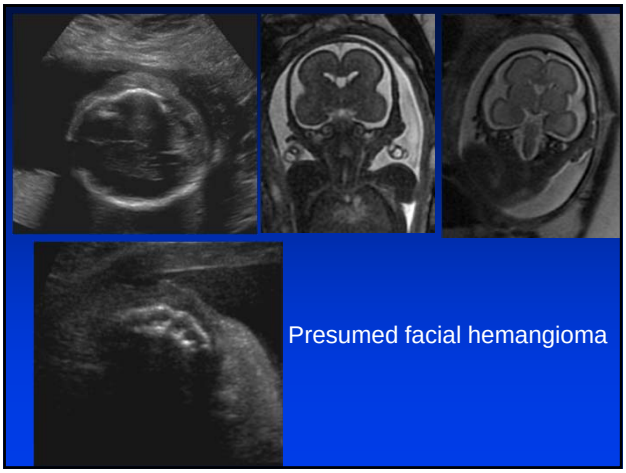


Outcome

- Delivery of a 2 lb 10 oz infant
- Cyanotic, floppy, no heart rate
- Chest compressions
- Emergency tracheostomy
- During procedure infant became cyanotic and lost HR. CPR was initiated.
- Transillumination: right pneumothorax
- Unresponsive; died.

BWH

Face Masses



Presumed facial hemangioma

Thank you for your attention.