

Fetal Face, Neck and Head

Dolores H. Pretorius, MD
Professor of Radiology
University of California, San Diego

Michael E. Hahn, MD, PhD
University of California, San Diego

Pretorius and Hahn



Disclosures

- None



Pretorius and Hahn

Learning Objectives

After completing this presentation, the learner will be able to:

1. Identify anomalies of the fetal face, head, and neck commonly seen on fetal ultrasound examinations
2. Learn how to evaluate fetuses at risk for cleft lip and palate
3. Understand how to utilize multiplanar and 3D views to avoid pitfalls in fetal facial diagnosis

Note: Videos will be bordered in yellow
Normal anatomy for comparison will be bordered in blue

Pretorius and Hahn

Lecture Outline

- **Face**
 - Profile
 - Orbits
 - Palate
 - Bonding
- **Head**
 - Sutures
- **Neck**
 - Nuchal Translucency
 - Neck Masses
 - Cystic hygroma
 - Jugular sacs

Pretorius and Hahn

Fetal Face Outline

- **Profile**
 - Technical factors/positioning
 - Nasal bone
 - Frontal bossing
 - Micrognathia
 - Masses
 - Proboscis
 - Encephalocele
- **Orbits**
 - Hypotelorism
 - Hypertelorism
 - Dacryocystoceles
- **Palate**
 - Normal anatomy
 - Retronasal triangle at 1st trimester
 - Palate at 2nd trimester
 - Overview of cleft types
 - Examples of clefts
 - MPRs and 3D views
- **Bonding**

Pretorius and Hahn

PROFILE

Pretorius and Hahn

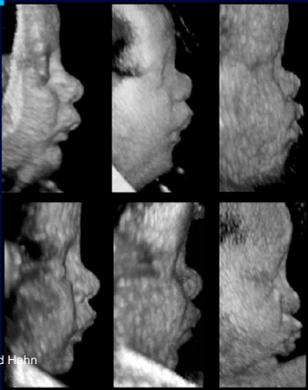
Profile

- Profile
 - Technical factors/positioning
 - Nasal bone
 - Micrognathia
 - Masses
 - Proboscis
 - Encephalocle
 - Tumors



Pretorius and Hahn

Various Profiles of Normal Fetuses



Courtesy of Dr. Platt

Pretorius and Hahn

Fetal Face: 125 Cases

- Orthogonal display
- Abdominal transducer
- True profile in 70%
- Off profile by up to 3-20 degrees in 30%



Merz et al, UOG 9:237, 1997

31 - WEEK PROFILE

Pretorius and Hahn

Normal Nasal Bone First Trimester



Look for the Equals = Sign + the Tip

Pretorius and Hahn

Likelihood Ratio for Increased Risk of Trisomy 21 with Absent Nasal Bone

- 1st Trimester
 - LR 4.4 – 85.6
 - Weingartner, et al. Fetal Diagn Ther 2006; 21:433
 - Recept Has, et al. Fetal Diagn Ther 2008; 24:61
 - LR 28
 - Cicero, et al. UOG 2004; 23: 218
- 2nd Trimester
 - LR 50
 - Moreno-Cid, et al. UOG 2014; 43:247
 - A meta analysis
 - Also, for nasal bone hypoplasia <2.5 mm
 - Cicero, et al. UOG 2003; 21:15

At UCSD:
30 X in 1st Tri
50 X in 2nd Tri

TODAY:
Most women
choose NIPT

Pretorius and Hahn

Absent or Short Nasal Bone

- Absence usually bilateral but may be unilateral
- Differential Diagnosis
 - Aneuploidy
 - Trisomies 21 (most common), 13, and 18
 - Delayed ossification
 - Ethnic variability
 - Technical
 - Idiopathic (up to 3%)

Pretorius and Hahn

Nuchal Screening

Is the nasal bone present?

No, there's no = sign



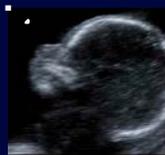
Non-ossified Nasal Bone



Normal

Pretorius and Hahn

Follow-up Scan at 18 Weeks



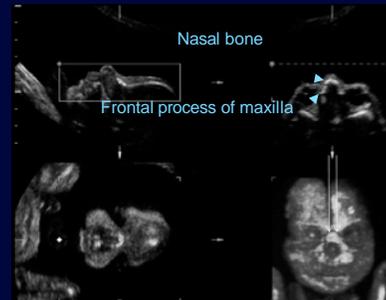
Non-ossified Nasal Bone



Normal for Comparison

Pretorius and Hahn

Normal Nasal Bone



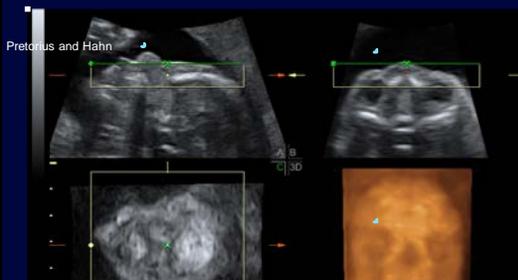
Nasal bone

Frontal process of maxilla

Benoit and Chaoui. UOG 2005; 25:19

Pretorius and Hahn

Absent Nasal Bones



Pretorius and Hahn

Solitary Absent Nasal Bone



LEFT



Right

Pretorius and Hahn

Solitary Absent Nasal Bone

Pretorius and Hahn

Micrognathia



425.1Kb
interstitial
deletion of
4q32.1



Treacher
Collins
Syndrome



13 weeks

Pretorius and Hahn

Micrognathia

- Underdeveloped mandible leads to small chin
- At 2nd trimester, use sagittal profile view
- At 1st trimester, use retronasal triangle (RNT) view
 - Normals have a mandibular “gap” (Sepulveda, et al. UOG 2012;39:152.)
- **Outcomes:** (Luedders, et al. Prenat Diagn 2011;31:146.)
 - 43% musculoskeletal disorders
 - 15% non-skeletal anomalies
 - 7% isolated
 - Overall, 15% alive post-natally
 - Rule out isolated cleft palate
- Associated with Pierre Robin sequence, Treacher-Collins Syndrome

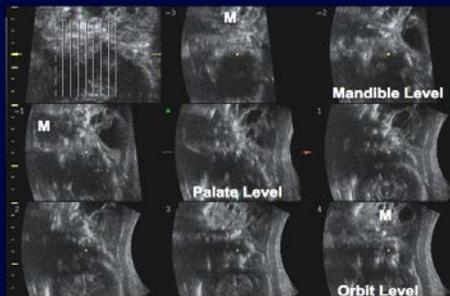
Pretorius and Hahn

Epignathus: Teratoma

Pretorius and Hahn



Epignathus: Multislice



Pretorius and Hahn

M = Mass

Encephalocele

- Referred for elevated MS-AFP
- 75% are occipital in Caucasians
- 75% are frontal in southeast Asian women
- Should take folate prior to conception for future pregnancies

Pretorius and Hahn

Pretorius and Hahn

Proboscis Holoprosencephaly

Pretorius and Hahn

Frontal Encephalocele

Pretorius and Hahn

Posterior Encephalocele

- Think Meckel Gruber Syndrome
 - Polydactyly
 - Large cystic kidneys

Pretorius and Hahn

Pretorius and Hahn

Pretorius and Hahn

Orbits

Normal

Hypotelorism
Holoprosencephaly

Hypertelorism
Apert Syn

Pretorius and Hahn

Pretorius and Hahn

Hypo- and Hypertelorism

- Hypotelorism
 - Interocular distance < 5th percentile
 - Suspect if space between orbits < size of one orbit
 - Cyclopia is most severe form
 - Associations:
 - Holoprosencephaly
 - T13 and T18
 - Microcephaly
 - Trigonocephaly
 - Numerous rare syndromes
 - *Very rarely isolated*
- Hypertelorism DDx
 - Interocular distance > 95th percentile
 - Associations:
 - Typical facial clefts
 - Craniosynostoses
 - Agnesis of Corpus Collosum
 - T13 and Turner's Syn
 - Median facial cleft syndrome
 - Frontonasal dysplasia
 - Maternal antiepileptics
 - Numerous rare syndromes
 - *Normal variant*

Pretorius and Hahn

Abnormal Orbits



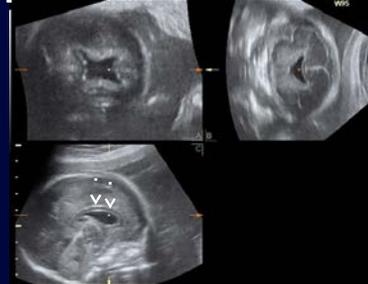
Anophthalmia

Pretorius and Hahn



Cataracts

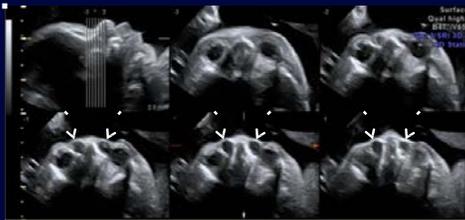
Abnormal Orbits



Septo-Optic Dysplasia

Pretorius and Hahn

Dacrocystocele

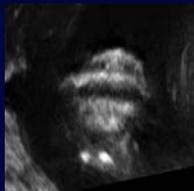


Pretorius and Hahn

LIP/PALATE

Pretorius and Hahn

Normal Lip/Nostrils



2D: Lips



3D: Lips

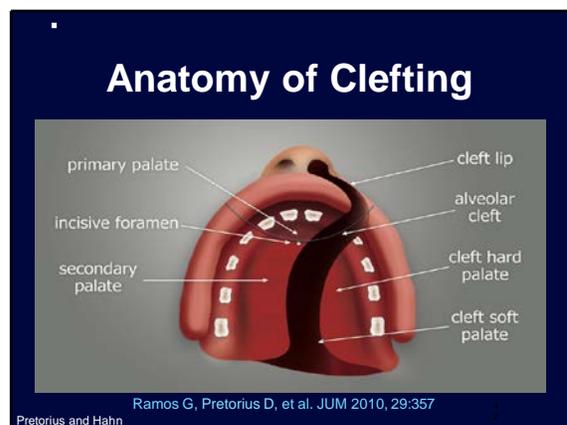
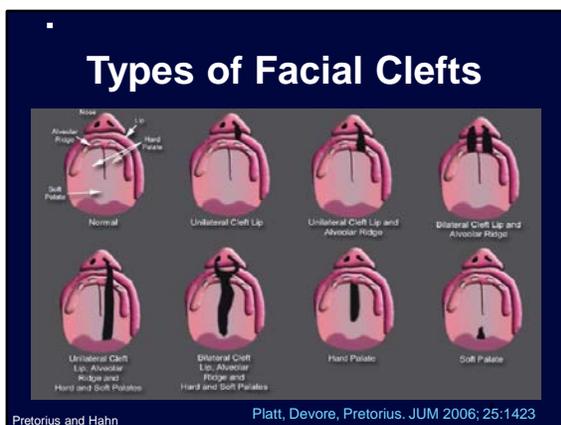
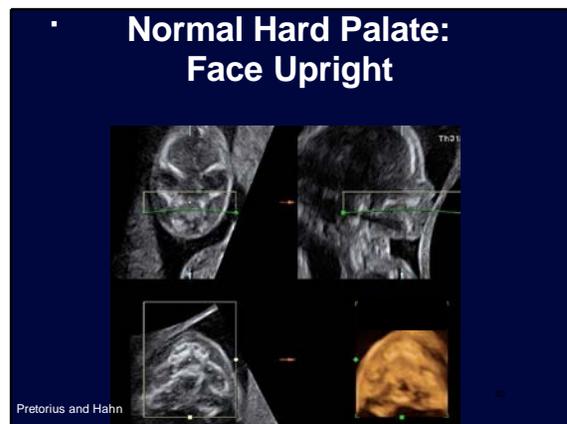
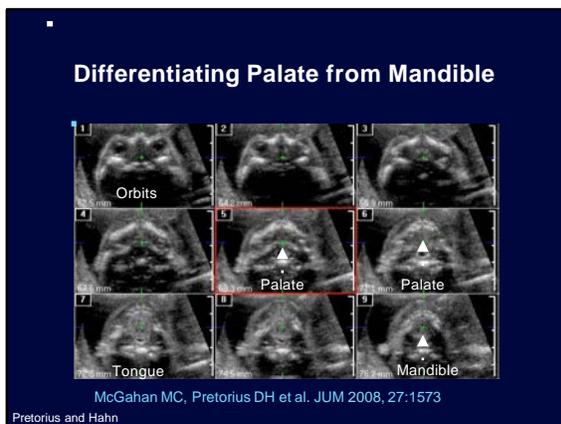
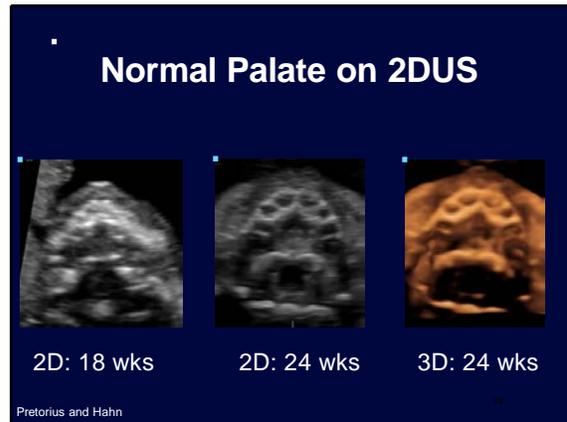
Pretorius and Hahn

Nostrils too Close



Holoprosencephaly

Pretorius and Hahn



Secondary Palate

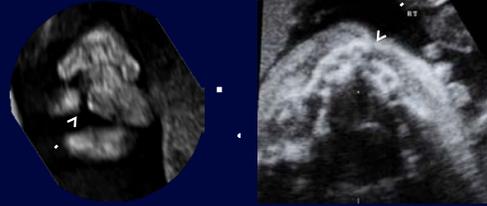
- Hard palate (between blue arrows)
- Soft palate (between white arrows)



Campbell. UOG 2007, 29:124

Pretorius and Hahn

Cleft Lip and Palate: 2D



- 27 weeks

Pretorius and Hahn

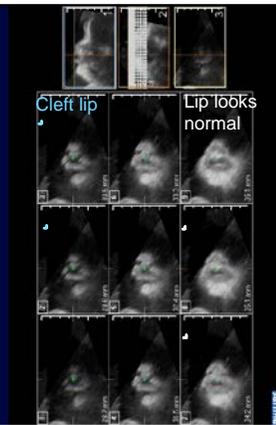
Unilateral Cleft Lip and Palate



26 wk

Pretorius and Hahn

Cleft Lip and Palate: Frontal plane thru Cleft Lip



Pretorius and Hahn

Bilateral Cleft Lip and Palate 35 weeks



Pretorius and Hahn

Cleft Lip and Palate: Pre and Post- Repair



With permission from the family

Pretorius and Hahn

Cleft Lip and Palate: 3D Multiplanar

Pretorius and Hahn

CLEFT LIP

3D Cleft Lip and Palate

Pretorius and Hahn

CLEFT HARD PALATE

Normal hard palate

**First Trimester Diagnosis
Normal Retronasal Triangle: 2D**

Nasal Bones

Maxilla

12w 1d

Pretorius and Hahn

1st Trimester Cleft Palate: Trisomy 13

13 Weeks

Bilateral Cleft L/P at 18 wks

Normal for Comparison

Pretorius and Hahn

**Cleft Lip and Palate:
It can be subtle...**

13 wk 2 d

19 wk 2 d

Pretorius and Hahn

BONDING

Pretorius and Hahn

42 weeks and 31 weeks



Pretorius and Hahn With permission from the family

Reassurance for Patients: Very Beneficial

- Prior anomalies or fetal demise
- Surrogate parents
- Hospice patients carrying fatal anomalies
- Infertility
- Family history of anomalies
- Surgery during pregnancy - added
- Anomaly in current pregnancy - added

Pretorius. UOG 2000, 16(S)1:94

Pretorius and Hahn

Face Important to Mom



19 wks: Adnexal Mass

Ovarian Luteoma



28 wks: Post Op

Pretorius and Hahn

HEAD

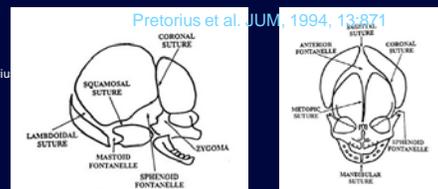
Pretorius and Hahn

Fetal Head Outline

- Skull views
 - Normal sutures
 - Craniosynostosis
 - Cloverleaf skull (kleeblattschädel)
 - Frontal bossing

Pretorius and Hahn

Normal Sutures



Pretorius et al. JUM, 1994, 13:871

Pretorius

Craniosynostosis

- Overall prevalence 1:2,500 births
- Sagittal synostosis most common, followed by coronal synostosis
- Apert syndrome accounts for 4.5% of all cases - next case
- Seen in more than 150 genetic disorders
- Airway obstruction is observed in 40% of severe cases
- Mental delay may occur in 50% of case

Pretorius and Hahn

2D Can Tip you Off by Shape of Skull



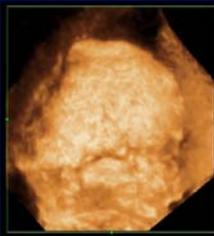
Normal



Craniosynostosis

Pretorius and Hahn

Apert Syndrome



Coronal Suture



Metopic Suture

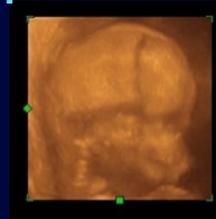
21 weeks

Pretorius and Hahn

Craniosynostosis of Coronal Suture



Craniosynostosis
21 wks



Normal
18 weeks

Pretorius and Hahn

3D Rendering of Metopic Suture



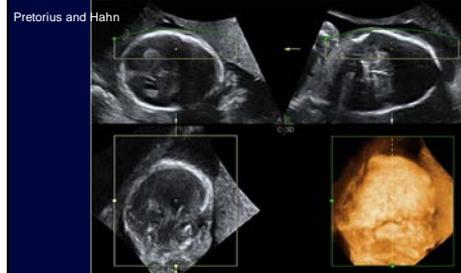
Abnormal



Normal

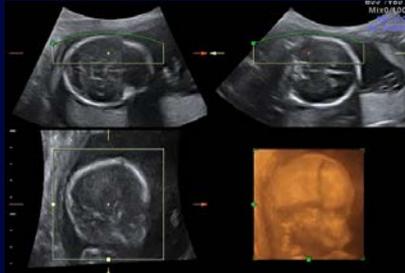
Pretorius and Hahn

Craniosynostosis on 3DUS How to Get the Image



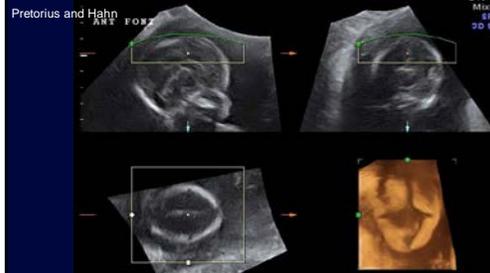
Pretorius and Hahn

How to Render the Volume Acquire in Axial Plane



Pretorius and Hahn

Sagittal Suture Acquire in Sagittal Plane



Pretorius and Hahn

Metopic Sutures: How to Do It



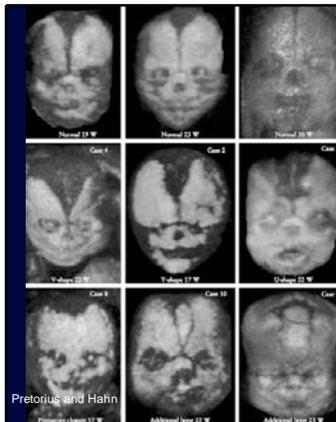
Pretorius and Hahn

Rendering in Maximum mode

Metopic Sutures:

1st Row: Normal

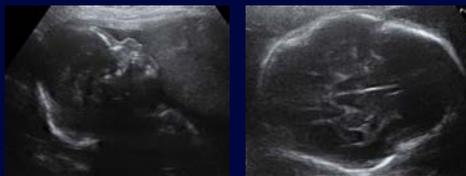
2nd & 3rd Rows: Abnormal



Pretorius and Hahn

Chaoui et al. UOG 2005, 26:761

Kleeblattschädel (Cloverleaf Skull)

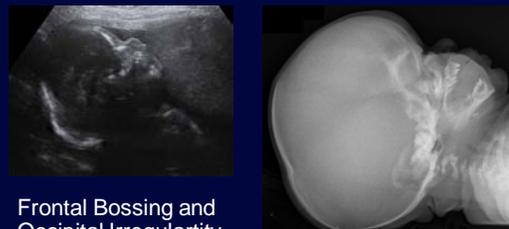


Frontal Bossing and Occipital Irregularity

Cloverleaf Skull Shape

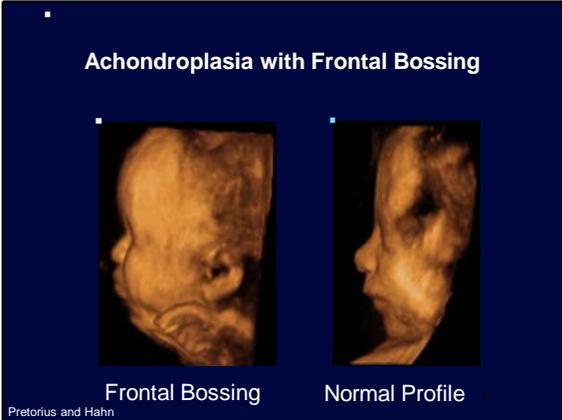
Pretorius and Hahn

Kleeblattschädel (Cloverleaf Skull)



Frontal Bossing and Occipital Irregularity

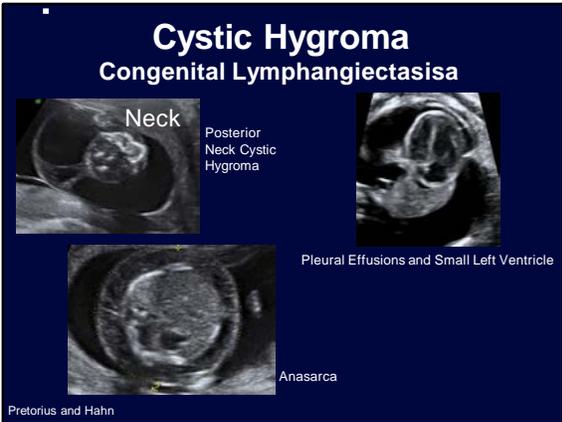
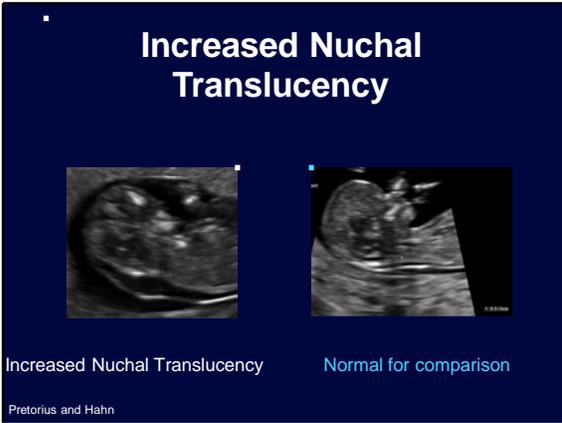
Pretorius and Hahn



NECK

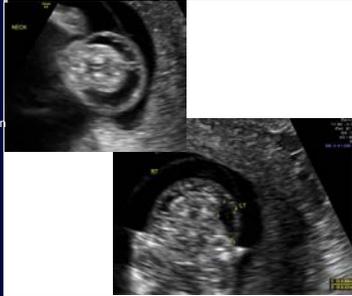
Pretorius and Hahn

- ### Fetal Neck Outline
- First trimester nuchal translucency
 - Normal
 - Abnormal
 - Neck masses
 - Nuchal cystic hygroma
 - Jugular sacs
- Pretorius and Hahn



- ### Nuchal Cystic Hygroma
- Edema and fluid-filled spaces within posterior neck and back
 - Thought to be due to lymphatic obstruction
 - First trimester frequency is 1/285 (0.35%)
 - Associations: (Scholl, Benson, et al. Obstet Gynecol 2012, 120:551)
 - 55% karyotype abnormality
 - 29% major anomaly
 - 39% perinatal loss
 - Overall abnormal outcome in 87%
- Pretorius and Hahn

Jugular Sacs



Pretorius and Hahn

Jugular Sacs

- Significant association with increased NT and presence of JLS
- 22/26 had JLS identified
- 10/26 with aneuploidy – terminated/demised
- 16/26 with normal karyotype
 - 3 with other anomalies:
 - Holoprosencephaly, tricuspid atresia, and Pierre-Robin sequence

Bekker et al. UOG 2005, 25:239

Pretorius and Hahn

Thyroid Goiter



Pretorius and Hahn

Conclusions

Think of Fetal Face:

- Profile
- Cleft lip/palate
- Orbits
- Neck
- Cranium

Pretorius and Hahn

Key References

Pettit KR, Tran N, Pretorius DH. Ultrasound evaluation of the fetal face and neck: In Callen's *Ultrasonography in Obstetrics and Gynecology*, 5th edition, edited by Norton M, Saunders WB, Saunders Company, Philadelphia, PA, 2016.

Scholl J, Durfee SM, Russell MA, Heard AJ, Benson CB, et al. First-trimester cystic hygroma: relationship of nuchal translucency thickness and outcomes. *Obstet Gynecol* 2012; 120: 551-9.

Kagan KO, Cicero S, Staboulidou I, Wright D, Nicolaides KH. Fetal nasal bone in screening for trisomies 21, 18, and 13 and Turner syndrome at 11 – 13 weeks of gestation. *Ultrasound Obstet Gynecol* 2009; 33: 259-64.

Martinez-Ten P, Adiego B, Illescas T, Bermejo C, Wong AE, Sepulveda W. First-trimester diagnosis of cleft lip and palate using three-dimensional ultrasound. *Ultrasound Obstet Gynecol* 2012; 40: 40-6.

Pretorius and Hahn



Where discoveries are delivered™

UC San Diego
HEALTH SCIENCES