Evaluation of Fetal Orbits and Ears

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Disclosure

• I have no disclosures
Goals & Objectives

• Review basic US anatomic views for the evaluation of the orbits and ears

• Describe some of the major malformations involving the orbits and ears
Background on Facial Abnormalities

• Important themselves

• May also indicate an underlying problem
  – Chromosome abnormality/ Syndromic conditions
Background on Facial Abnormalities

• Assessment of the face is included in all standard fetal anatomic surveys

• Recheck the face if you found other anomalies

• And conversely, if you see facial anomalies look for other systemic defects
Background on Facial Abnormalities

• Fetal chromosomal analysis is often indicated

• Fetal MRI frequently requested in search for additional malformations

• US / Fetal MRI, as complementary techniques: information for planning delivery / neonatal treatment
• Anatomic evaluation

• Malformations (orbits, ears)
Orbits Axial View

- **Bony orbits:**
  IOD
Orbits Axial View

- **Bony orbits:** IOD and BOD, which correlates with GA, will allow detection of hypo-/hypertelorism
Orbits Axial View

- Axial
  - Bony orbits
  - Intraorbital anatomy:
    - Globe
    - Lens
Orbits Axial View

• Axial
  – Bony orbits
  – Intraorbital anatomy:
    • Globe
    • Lens
Orbits Axial View

- Hyaloid artery is seen as an echogenic line bisecting the vitreous

- By the 8th month the hyaloid system involutes
  - If this fails: **persistent hyperplastic primary vitreous**
Malformations of the orbits

- Hypotelorism
- Hypertelorism
- Anophthalmia / microphthalmia
- Persistent hyperplastic primary vitreous
- Masses
Alobar Holoprosencephaly
hypotelorism
Orbital Hypertelorism

- Excessive separation between the eyes
  - Skeletal: Craniosynostosis, Camptomelic dysplasia, arthrogryposis, Roberts syndrome
  - CNS (Median cleft face syndrome, Anterior encephalocele, oral teratomas)
  - Chromosomal
  - Noonan syndrome
  - CHARGE association

Trisomy 13: Hypertelorism and persistent hyperplastic primary vitreous
Associations with micro / anophthalmia (uni / bilat)

- Isolated
- **CNS anomalies** (Meckel-Gruber syndrome, Walker-Warburg Syndrome, AICARDI syndrome, Median cleft face syndrome)
- **Trisomies** (T13, T18)
- Hemifacial microsomia/OAVS
- TORCH
- **CHARGE association**
- **Other syndromes:**
  - Goltz (Focal Dermal Hypoplasia)
  - Fraser
Persistent Hyperplastic Primary Vitreous

- Frequently seen with trisomy syndromes and other forms of abnormal development of the brain.

- US: There is a band of tissue attached to the posterior surface of the lens and a small globe.

- MRI: dark band attached to the posterior surface of the lens, thick irregular lens and small globe.

33 w GA fetus with Trisomy 13
Orbital masses
Mucocele of the Nasolacrimal Duct/Dacrocystocele
Mucocele of the Nasolacrimal Duct

- Uni/bilateral

- Dilatation of the lacrimal drainage system due to obstruction

- **Typical cystic appearance** different from other possible masses:
  - Hemangioma
  - Glioma
  - Dermoid cyst
  - Frontonasal encephalocele
Evaluation of the ears

- The auricles are not frequently targeted
- They can be seen (axial, coronal or sagittal) + 3D - US
  - Usually evaluated in the setting of multiple anomalies
Evaluation of the ear on fetal MRI

Fetal MRI will help to better assess the ears and potential associated malformations.

Fast gradient imaging (SSFP) is best, but Single shot fast spin echo T2w imaging can also be useful.

Sagittal image of the auricle on SSFP sequence.

Coronal SSFP image of the auricles, external auditory canals, fluid filled middle ear and inner ear structures.

Auricle, inner ear structures.
Basic Embryology

- 1\textsuperscript{st} and 2\textsuperscript{nd} branchial arches are involved in the development of the ear (1\textsuperscript{st} arch also in the face)

- Neural crest cell migration induce differentiation and development

- Deficient neural crest cell migration: Hypoplasias and abnormal development
  - Otomandibular dysplasias (AKA branchial arch syndromes)
Basic Embryology

- The auricles start in the lower neck.
- With the development of the mandible, the ears ascend to the level of the eyes.
- Impaired ascent is associated with mandibular hypoplasia:
  - Micrognathia with low set ears
  - Otocephaly
Pathologic conditions of the auricles

• Microtia / anotia (Spectrum of hypoplastic to absent ear)

• Malposition – Low-set ears
Microtia / anotia

- Sporadic and isolated
- Syndromic (including branchial arch syndromes)
- Chromosome defects
- Environmental insults (alcohol, thalidomide, isotretinoin, diabetic embryopathy)
Major Branchial Arch Syndromes

- Typically unilateral defects in Hemifacial microsomia/Oculo-Auriculo-Vertebral Spectrum

- Bilateral defects in Treacher Collins Syndrome (AKA Mandibulofacial dysostosis)
Hemifacial microsomia/Oculo-Auriculo-Vertebral Spectrum

- The second most common type of facial anomaly, after cleft lip and cleft palate

- Asymmetry is present in 65% of the cases
Hemifacial microsomia/
Oculo-Auriculo-Vertebral Spectrum

• Usually involve the maxillary, temporal, and zygomatic bones, which are small and flat

• Eye (dermoids, upper eyelid coloboma, microphthalmia / anophthalmia)

• Ear (anotia, microtia)

• Vertebral (fused and hemivertebrae, spina bifida) defects are common

• About 15% cleft lip or palate
Hemifacial microsomia/ Oculo-Auriculo-Vertebral Spectrum

• Other malformations include

  • Cardiac abnormalities, such as tetralogy of Fallot and ventricular septal defects

  • GI defects (Esophageal Atresia)

  • Renal anomalies

  • Lung hypoplasia/agenesis
Hemifacial microsomia/OAVS
Hemifacial microsomia/OAVS

CASE TO COMPARE:
MICROGNATHIA BUT
NORMAL HARD
PALATE
Treacher Collins Syndrome

- Bilateral facial defect
- Malar hypoplasia due to underdevelopment of the zygomatic bones
- Mandibular hypoplasia
- Down-slanting palpebral fissures, lower eyelid colobomas
- Malformed external ears
Treacher Collins Syndrome

Micrognathia, cleft palate

Absent bilateral auricles+ EAC
Treacher Collins Syndrome

Absent bilateral auricles + EAC

Normal case to compare:
- bilateral ears and EAC
Treacher Collins Syndrome
Reduced dose CT with post-processing (maximum intensity three dimensional image)

3yo now post mandibular distraction and other facial reconstruction surgeries. Tracheostomy dependent
Pathologic conditions of the auricles

- Microtia / anotia (Spectrum of hypoplastic to absent ear)

- Malposition – Low-set ears
The most extreme form of low-set ears: Otocephaaly

**Most extreme form of auricular malposition**

- Marked underdevelopment or almost total absence of the mandible
- Ears: anteriorly and inferiorly, approaching fusion near the midline under the jaw
- It is considered a lethal condition (airway)
- Frequent h/o “polyhydramnios and unable to visualize the mandible” in isolation or with holoprosencephaly, situs inversus or visceral anomalies
Summary

• Reviewed basic US anatomic views for the evaluation of the orbits and ears

• Described some of the major malformations involving the orbits and ears
Thank you!!!!

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