Congenital Anomalies of the Temporal Bone

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Objectives

- Spectrum of congenital anomalies
- Imaging techniques
- Goals of imaging
- Imaging findings & clinical relevance
- Highlight important syndromes

Congenital anomalies of the temporal bone

Anatomy

- External auditory canal
- Middle ear space & contents
- Inner ear structures
- CEMEM: Congenital external & middle ear malformation

Causes

- Environmental causes
- Genetic mutations
- Chromosomal causes
- Uncertain cause

Clinical Presentation

Physical abnormality

- Craniofacial malformation
- Tags, pits, sinus tracts
- Abnormal ear & ear canal
- Ocular abnormalities
- Nasal obstruction
- White forelock
- Other organ system malformations

Hearing impairment

- SNHL, CHL, MHL

MDCT

- 0.625 mm axial images
- Reformats
  - Axial parallel to hard palate (HP)
  - Coronal parallel to HP
  - Oblique reformats
  - Decreased patient dose
- Cone beam units
- External & middle ear anomalies
  - CHL or MHL
  - Congenital cholesteatoma
  - SNHL
**MR (3T)**

Brain screen
Axial & oblique sagittal 3D T2
e.g. 3D T2 SPACE
advanced shimming
techniques to ↓ artifact
Non EPI DWI techniques
NO contrast!

SNHL (especially CI candidate)
HL & neurological abnormality

**Goals of imaging**

- Identify structural causes of HL
- Provide syndromic diagnosis
- Guide genetic testing
- Information for parental counseling
- Preoperative anatomy & surgical planning

Is the patient a surgical candidate?
Which is the optimal side for CI?
Are there anatomic risk factors?

**CEMEM**

External auditory canal
- EAC stenosis
- EAC atresia
- EAC duplication
- EAC dermoid
- EAC 1st BCC

Middle ear space & contents
- Congenital cholesteatoma
- MEC hypoplasia/anomaly
- Ossicular malformations
- Oval window stenosis/atresia
- Round window stenosis/atresia
- CN7 tympanic & mastoid segment anomalies

**External auditory canal malformations**

- Normal
- EAC stenosis
- EAC atresia
- EAC duplication
- Dermoids

**Paradoxical widening of stenotic EAC**

Bright on DWI trace & ↓ ADC

DDx cholesteatoma or keratosis obturans

**EAC anomalies**

Dermoid mimicking BCC
Evolve EAC opacities
Hole in tympanic plate
Decreased ADC on DWI
**Inner ear malformation**

- “Microscopic malformations”
- **Enlarged vestibular aqueduct (EVA)**
- Cochlear & cochlear aperture malformations
- Malformations of the vestibule
- Malformations of the SCC
- **IAC malformations & CN 7 & 8 anomalies**

**Ossicles in congenital external & middle ear malformation**

- Normal
- IC cone
  - 2 dots
  - 2 dashes
- Malformed
- Rotated
- Boomerang

**Ossicular anomaly with CEMEM**

- Malformed
- Rotated
- Fused to scutum
- Descending CN 7
  - Ventrally displaced
- Normal

**CEMEM: Windows & CN7**

- Normal
- Review the windows & tympanic CN 7
- Hypoplastic, opacified MEC; absent recesses
- Low tegmen mastoideum
- Atretic oval window; hypoplastic round window
- Anomalous tympanic CN7 along MEC floor
- Rudimentary or absent ossicles

**MR Interpreted as dermoid**

- Mural thickening on MR
- Hair on exam
- Cartilage on path
- Dx: Duplicated IAC
- Decreased ADC on DWI

**Unilateral micrognathia**

- e.g., BOR

**Symmetric micrognathia**

- e.g., TCS

**Asymmetric micrognathia**

- e.g., EOA

**Decreased ADC**

- on DWI

**Asymmetric micrognathia**

- e.g., EOA

**Unilateral micrognathia**

- e.g., HFM

**Symmetric micrognathia**

- e.g., TCS

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Conclusions: Measurements of the vestibular aqueduct can be performed reliably and accurately in the 45° oblique plane. The mean midpoint width was 0.5 mm, with a range of 0.3–0.9 mm. These may be considered normal measurement values for the vestibular aqueduct midpoint width when measured in the 45° oblique plane.

In contrast, the flared configuration of the normal vestibular aqueduct at the external aperture creates difficulty with defining the exact point at which a measurement there should be made, and it is virtually impossible to determine a width perpendicular to the bony margins without a large degree of uncertainty and very limited reproducibility.

Large endolymphatic sac on MR

Cochlear malformations

- Complete labyrinthine aplasia
- Cochlear aplasia
- Common cavity malformation
- Cochlear hypoplasia
- Cystic cochleovestibular anomaly
- Incomplete partition type I (IP-I)
- Incomplete partition type II (IP-II)
- Modiolar deficiency
- Cochlear aperture stenosis or atresia

Enlarged vestibular aqueduct & IP-II anomaly

Potential risk of CSF leak at cochleotomy

Enlarged vestibular aqueduct

- Congenital lesion
- Delayed onset SNHL
- Fluctuating SNHL
- Progressive SNHL
- Post traumatic SNHL
- Dysequilibrium
- Familial cases
  - SLC26A4 mutation
  - ~27% to 40%
  - Pendred syndrome
  - Non syndromic EVA (DFNB4)

Cochlear malformations

- CLA (Michel)
- C aplasia
- C hypoplasia
- Single turn
- IP-I

Normal

Minimal CN hypoplasia
Common cavity malformation & CCVM

**Cochlear hypoplasia**
- Small, underdeveloped cochlea usually < 2 turns
- CH type I (small cochlear bud)
  - No modiolus or ISS
- CH type II (small cystic cochlea)
  - No modiolus or ISS
- CH type III (small cochlea, < 2 turns)
  - Shortened modiolus & ISS

Cochlear nerve likely hypoplastic or absent

In complete partition type I (IP-I)

Potential risk of CSF leak at cochleotomy

Incomplete partition type II (IP-II), EVA

Incomplete partition type II (IP-II), EVA

Potential risk of CSF leak at cochleotomy

Cochlear malformations in syndromes with HL

- "Baseball cap" IP-II & EVA Pendred
- "Corkscrew" IP-III XLMHL
- "Umwound" cochlea Branchio-oto-renal
- C, V, SCC hypo CHARGE

Potential risk of CSF leak at cochleotomy

IAC & cochlear nerve aperture

- Wide IAC
- Stenotic IAC (width < 2mm)
  - Hypoplasia/aplasia CN VIII components
- Wide cochlear aperture
  - Deficient or absent modiolus
- Stenotic/atretic cochlear aperture
  - Hypoplasia/aplasia CN VIIIC

Potential risk of CSF leak at cochleotomy

Oval window atresia
- Anomalous course CN7 Large emissary veins
Wide IAC
- NF1 dural ectasia
- NF2 vestibular schwannoma
- X-linked MHL (boys)
- PHACES (unilateral)

Wide cochlear aperture
- Deficient or absent modiolus

IAC stenosis

Wide cochlear aperture
- Deficient or absent modiolus

Vestibule & SCC
- Hypoplastic V
- Absent SCC
- CHARGE

Vestibule & Semicircular Canals
- Complete labyrinthine aplasia
- Hypoplasia of the vestibule
- Malformed or absent SCC
- Globular vestibule & SCC

"Baseball cap" cochlea & EVA/LES
- Pendred syndrome
- Non syndromic EVA (DFNB4)
- Autosomal recessive
- SLC26A4 gene mutations, chr 7q22.3
- FOX11, KCNJ10 mutations < frequent
"Corkscrew" cochlea + widened IAC
X linked stapes gusher; DFNX2
Transcription factor *POU3F4* mutation, chr Xq21.1

"Unwound cochlea" hypoplastic middle & apical turns
Branchio-oto-renal (BOR) syndrome
Autosomal dominant, *EYA1* gene, chr 8q13

Hypoplastic vestibule & SCC
Charge syndrome
Autosomal dominant
*CHD7* chromodomain mutations, chr 7q21
Coloboma *
Heart defects
Atresia choanae *
Retardation
Genital
Ear *
* Major criteria

Small LSCC bone island & CNC
Trisomy 21

Posterior SCC anomaly
Waardenburg syndrome (IV)
Alagille syndrome
BOR
Conclusion

- Reviewed CEMEM & inner ear malformations
- Pathognomonic syndromes
  - Large vestibular aqueduct
  - Flared EVA
  - PDS & DFNB4
  - Funnel shaped EVA
  - BOR
  - CHARGE

Deficiency of the cochlear modiolus
- Risk of CSF leak cochleotomy
- IP-II cochlear anomaly + EVA/LES
- X-linked mixed hearing loss

Stenosis of CN canal
- Hypoplasia/absence of CN
- Failure of cochlear implantation
- CHARGE syndrome, trisomy 21