AMSER Case of the Month July 2024

8-month-old male presenting with progressive mixed bilateral hearing loss

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Patient Presentation

- 8 m.o. male born at 39w1d presenting with progressive mixed hearing loss
  - Unremarkable pregnancy with no perinatal complications
  - Failed newborn hearing screening x2
  - No family history of congenital hearing loss
  - No improvement after bilateral cerumen disimpaction or usage of bilateral hearing aids

Pertinent Labs

- **Maternal labs:** unremarkable
- **CMV PCR:** negative
- **Auditory testing:** profound severe mixed hearing loss in both ears (R>L)

What Imaging Should We Order?

Select the applicable ACR Appropriateness Criteria

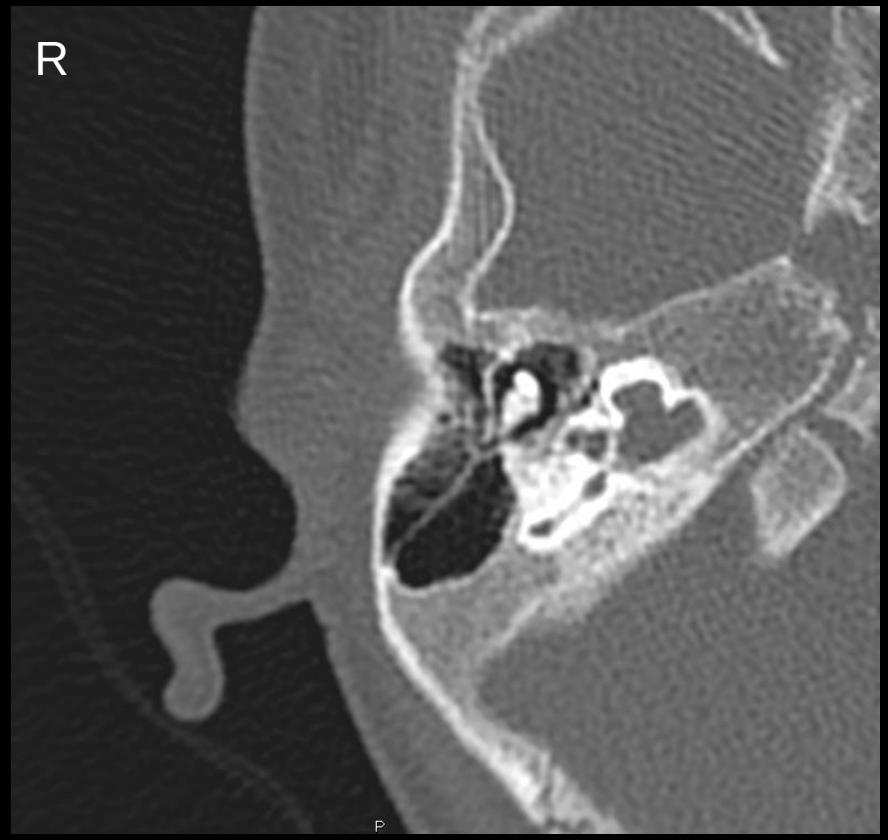
Variant 5: **Congenital hearing loss or total deafness or cochlear implant candidate. Surgical planning.**

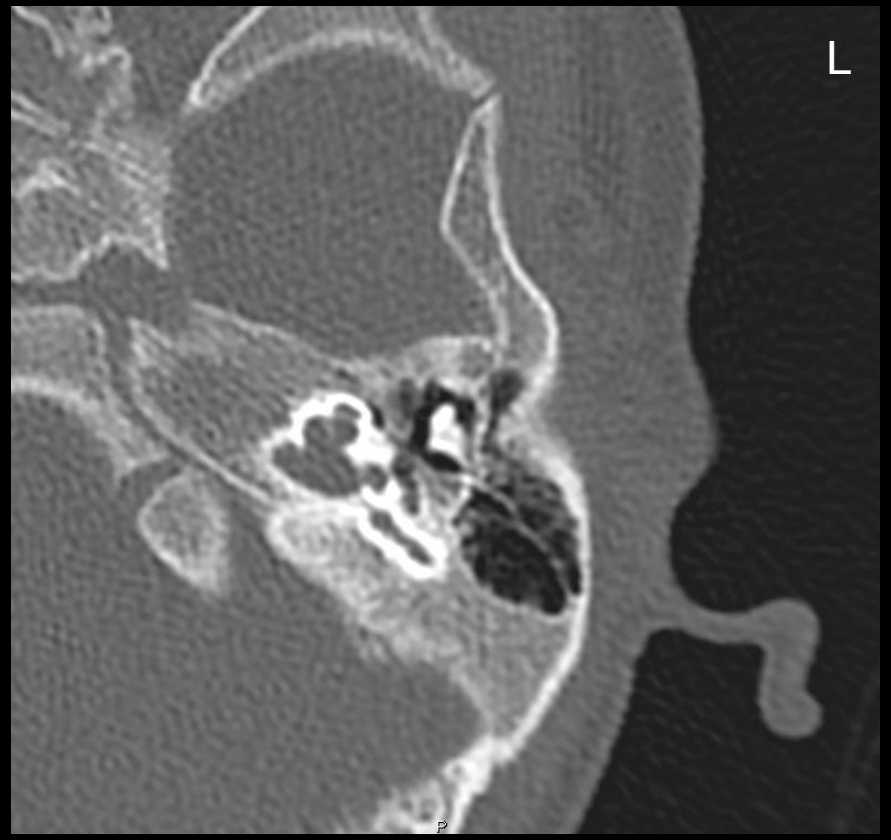
Procedure	Appropriateness Category	Relative Radiation Level
CT temporal bone without IV contrast	Usually Appropriate	☢☢☢
MRI head and internal auditory canal without and with IV contrast	Usually Appropriate	O
MRI head and internal auditory canal without IV contrast	Usually Appropriate	O
CT head with IV contrast	Usually Not Appropriate	☢☢☢
CT head without and with IV contrast	Usually Not Appropriate	☢☢☢
CT head without IV contrast	Usually Not Appropriate	☢☢☢
CT temporal bone with IV contrast	Usually Not Appropriate	☢☢☢
CT temporal bone without and with IV contrast	Usually Not Appropriate	☢☢☢
CTA head with IV contrast	Usually Not Appropriate	☢☢☢
MR venography head with IV contrast	Usually Not Appropriate	O
MR venography head without IV contrast	Usually Not Appropriate	O
MRA head without and with IV contrast	Usually Not Appropriate	O
MRA head without IV contrast	Usually Not Appropriate	O

This imaging modality was ordered by the ENT surgeon

AMSER

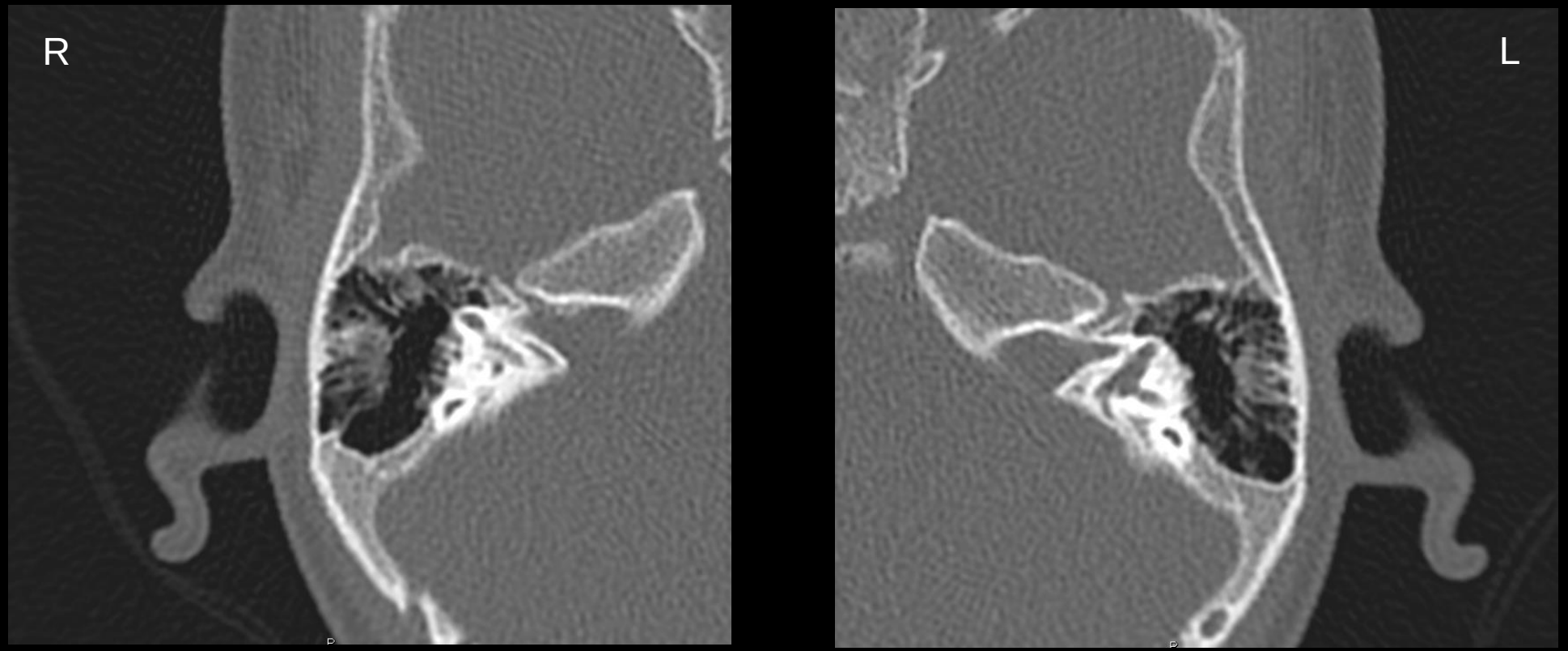
Findings (unlabeled)





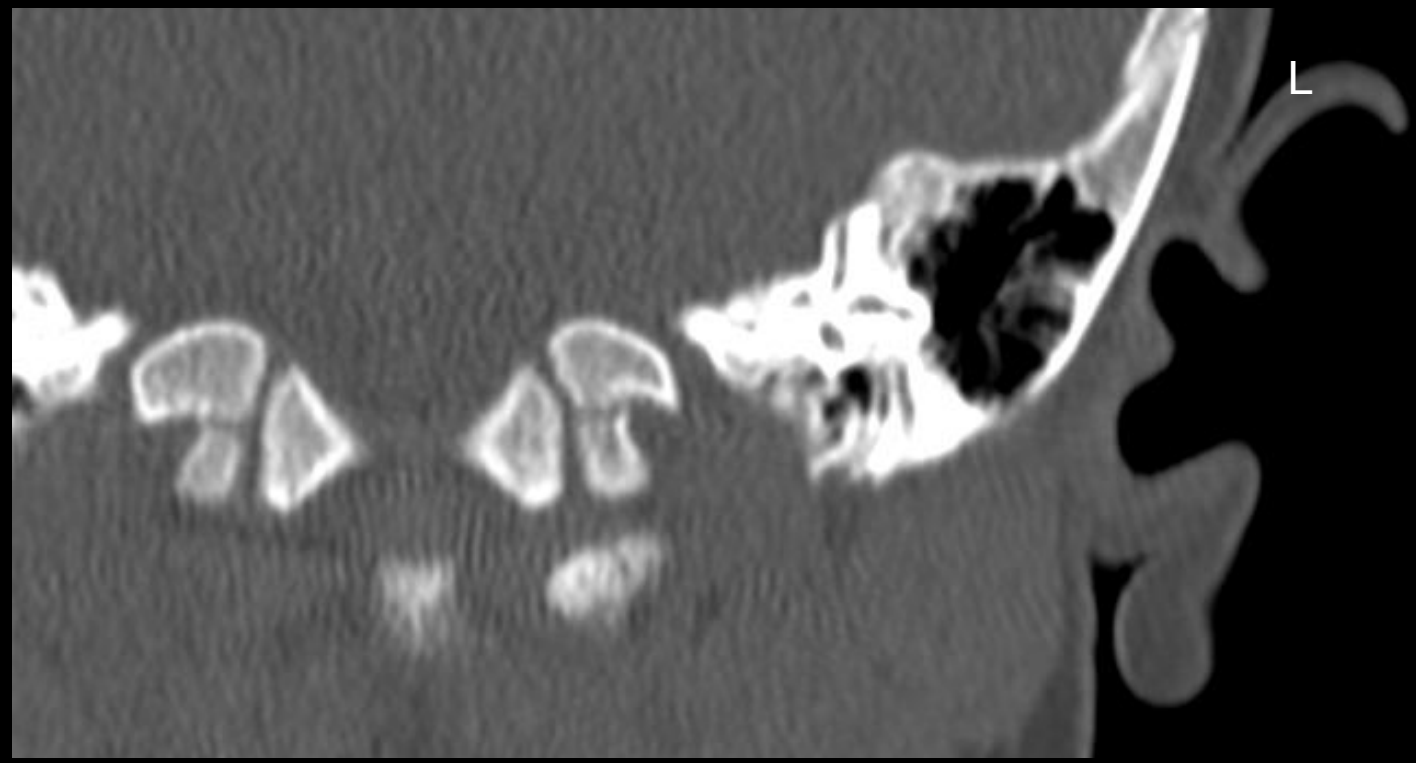
CT Temporal Bone (Axial)

Findings (unlabeled)



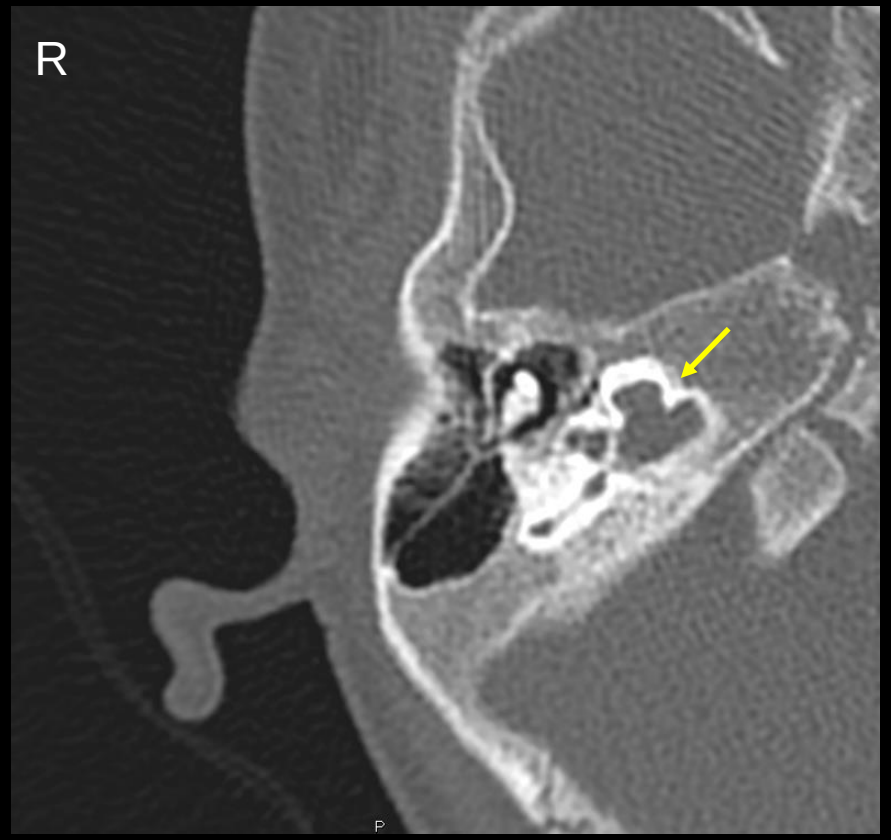
CT Temporal Bone (Axial)

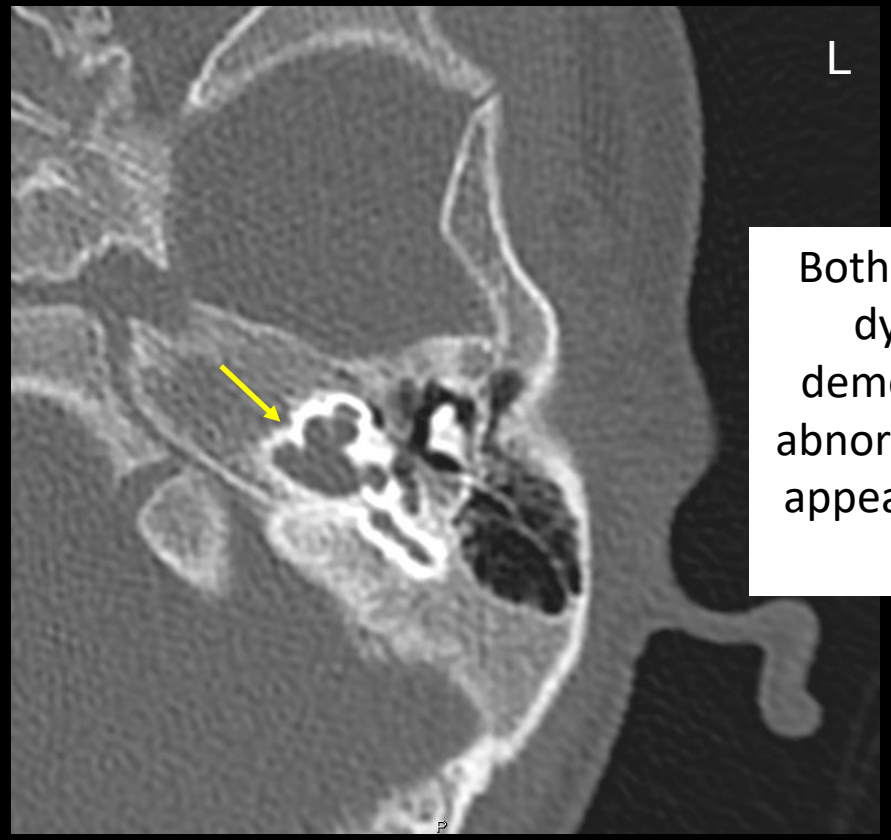
Findings (unlabeled)



CT Temporal Bone (Coronal)

Findings (labeled)

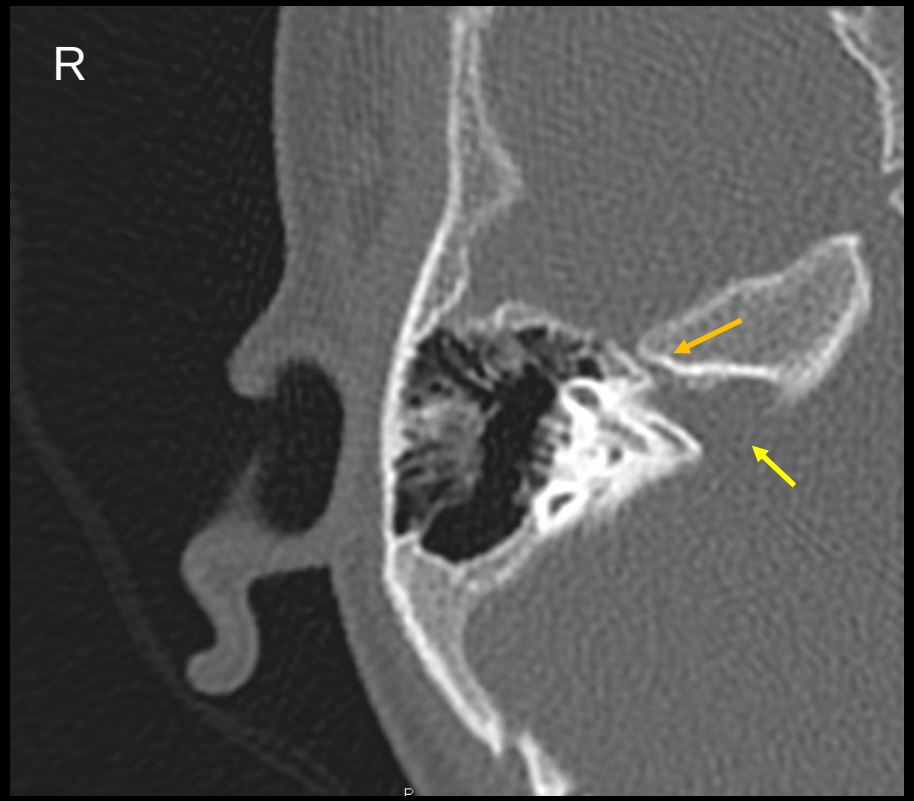


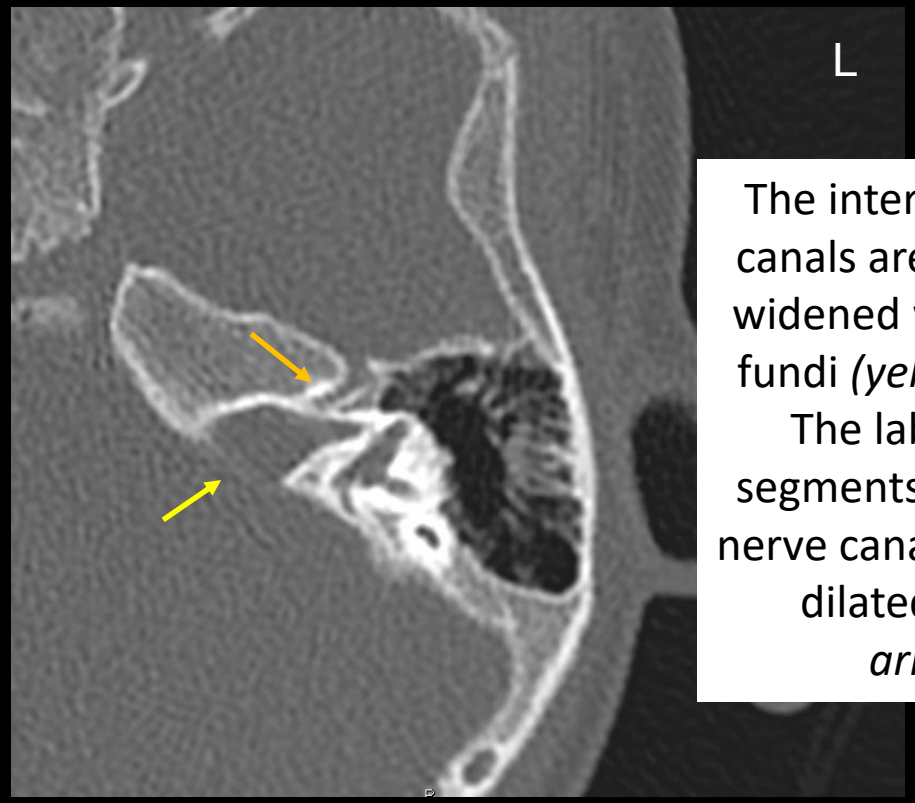


Both cochleae are dysmorphic, demonstrating an abnormal corkscrew appearance *(yellow arrows).*

CT Temporal Bone (Axial)

Findings (labeled)

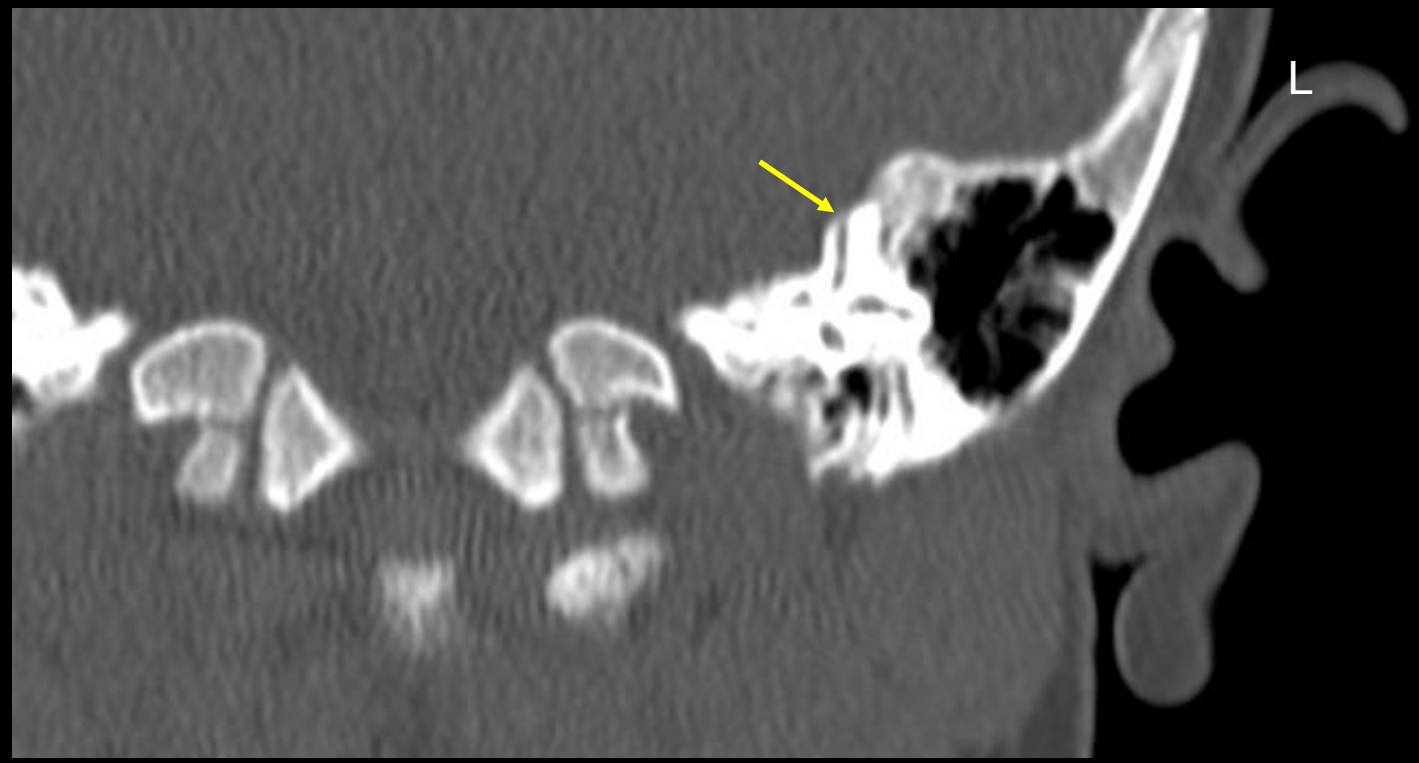




The internal auditory canals are abnormally widened with bulbous fundi *(yellow arrows).* The labyrinthine segments of the facial nerve canals are slightly dilated *(orange arrows).*

CT Temporal Bone (Axial)

Findings (labeled)



There is bony dehiscence of the roof of the left superior semicircular canal *(yellow arrow)*.

CT Temporal Bone (Coronal)

Final Dx:

X-Linked Stapes Gusher

Case Discussion (1)

- *POU3F4* gene at Xq13-q21.1 involved in otic capsule remodeling[1-3]
  - Loss of function causes incomplete separation between the base of the cochlea and internal auditory canal, creating abnormal CSF communication
- Presents as profound progressive mixed hearing loss
  - Conductive hearing loss due to stapes footplate fixation
  - Sensorineural hearing loss due to cochlear dysplasia
- Other possible symptoms: disequilibrium, dizziness/vertigo, tinnitus, autophony, conductive dysacusis, perilymph “gushing” during stapedectomy[3]

Case Discussion (2)

- Classic radiographic findings in X-linked stapes gusher:
  - Dilated internal auditory canal
  - Incomplete cochlear partition type III
- Other commonly seen/associated imaging findings[1,2]:
  - Dilated labyrinthine segment of the facial nerve canal
  - Abnormalities of the vestibular aqueduct
  - Semicircular canal dehiscence

Case Discussion (3)

- The patient was confirmed via genetic testing to have a pathogenic variant of *POU3F4.*
- Bilateral cochlear implants were placed.
- The patient is developing well with Speech and Audiology support!

References

1. Dasgupta S, Hong J, Morris R, Iqbal J, Lennox-Bowley A, Saniasiaya J. X-linked gusher disease DFNX2 in children, a rare inner ear dysplasia with mixed hearing and vestibular loss. Audiol Res 2023;13(4):600-614.

2. Eluvathingal Muttikkal TJ, Nicolasjilwan M. Congenital X-linked stapes gusher syndrome in an infant. A case report. Neuroradiol J 2012;25(1):76-80.

3. Kumar G, Castillo M, Buchman CA. X-linked stapes gusher: CT findings in one patient. AJNR Am J Neuroradiol 2003;24(6):1130-1132.