# AMSER Case of the Month September 2025

## A 3-month-old boy presents with hearing loss

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# Patient Presentation

- **HPI:** The patient is a 3-month-old male presenting with hearing loss. Auditory Brainstem Response (ABR) test demonstrates right profound to severe hearing loss and left profound rising to moderately severe hearing loss. Tympanograms are normal. The patient's mom is interested in future cochlear implants.
- **Birth History:** Born full term w/o complication but failed universal newborn hearing screening.
- **Family History:** Two maternal uncles are deaf.
- **Exam:**
  - **Ears:** Normal pinna bilaterally, mastoid is normal and non-tender bilaterally, external auditory canals are patent and tympanic membranes intact bilaterally. No abnormalities, effusions, or tenderness noted.

# What Imaging Should We Order?

# 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 |

These imaging modalities were ordered by the ENT physician along with genetic testing

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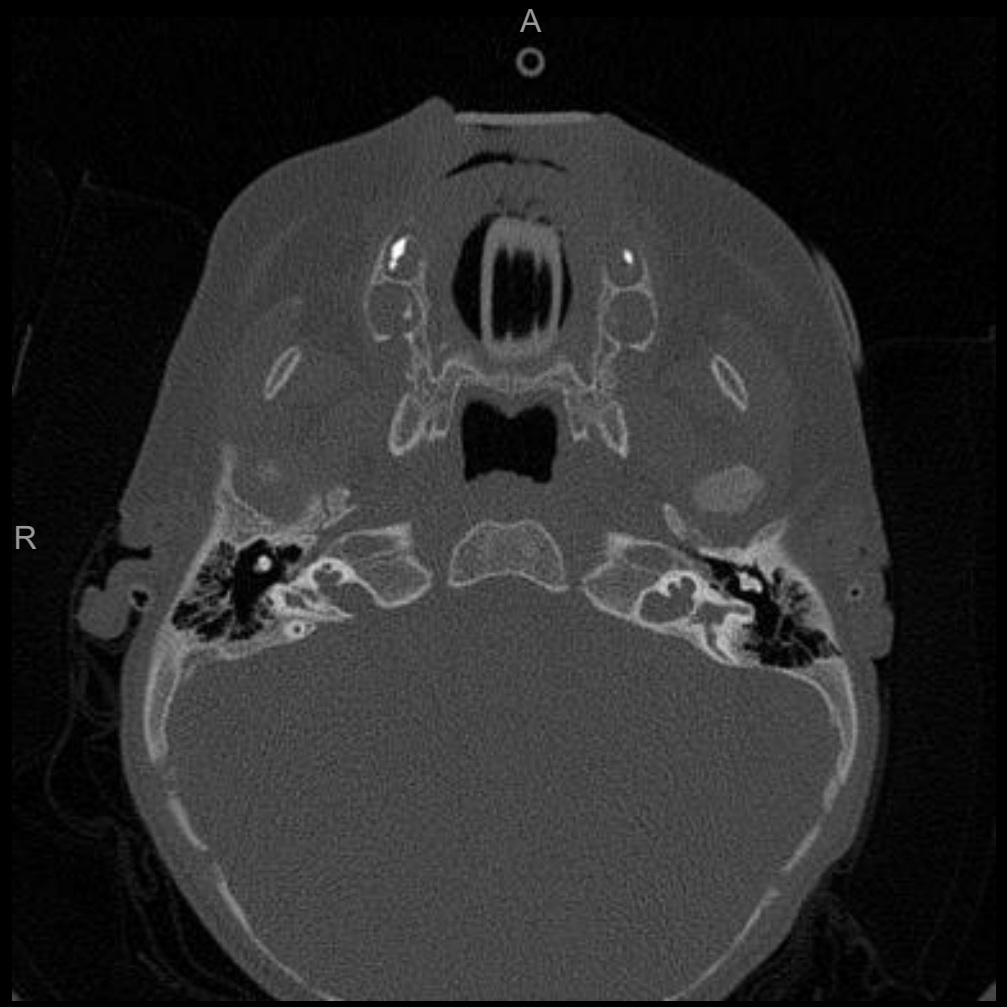
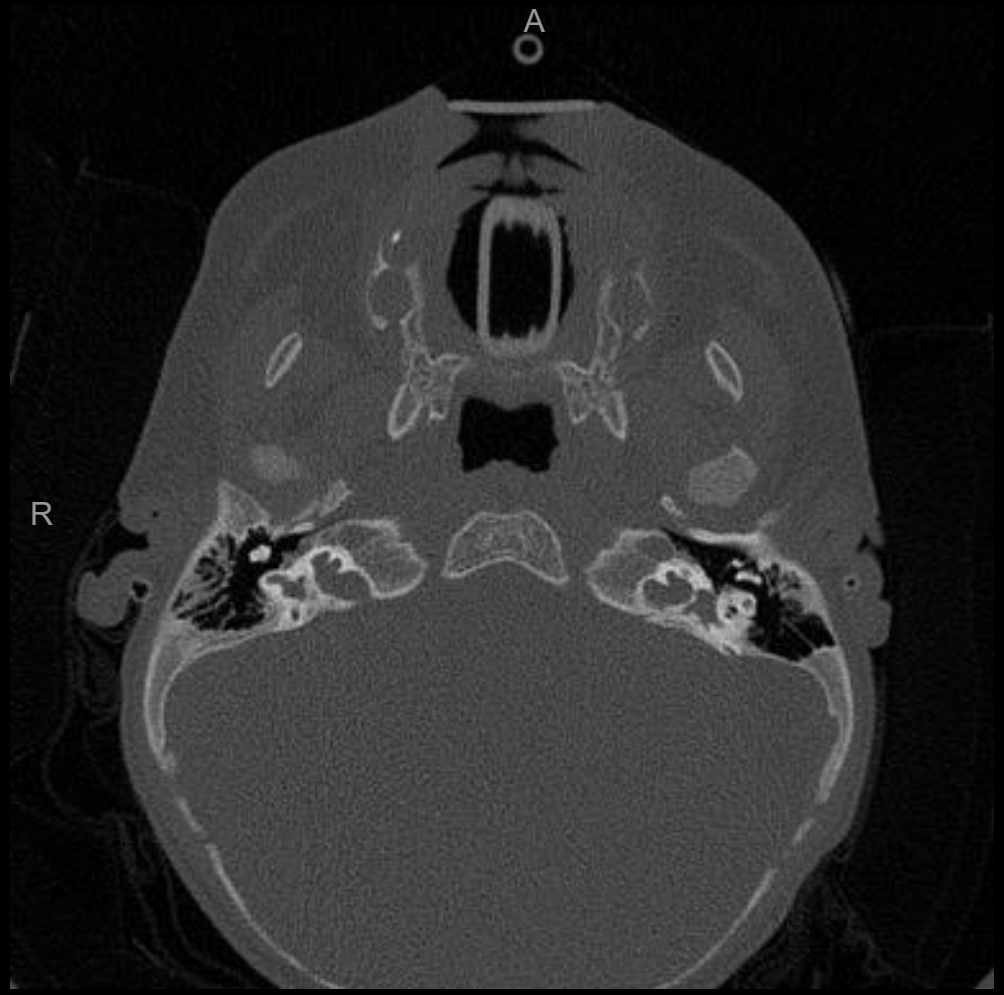
# CT Findings (unlabeled)



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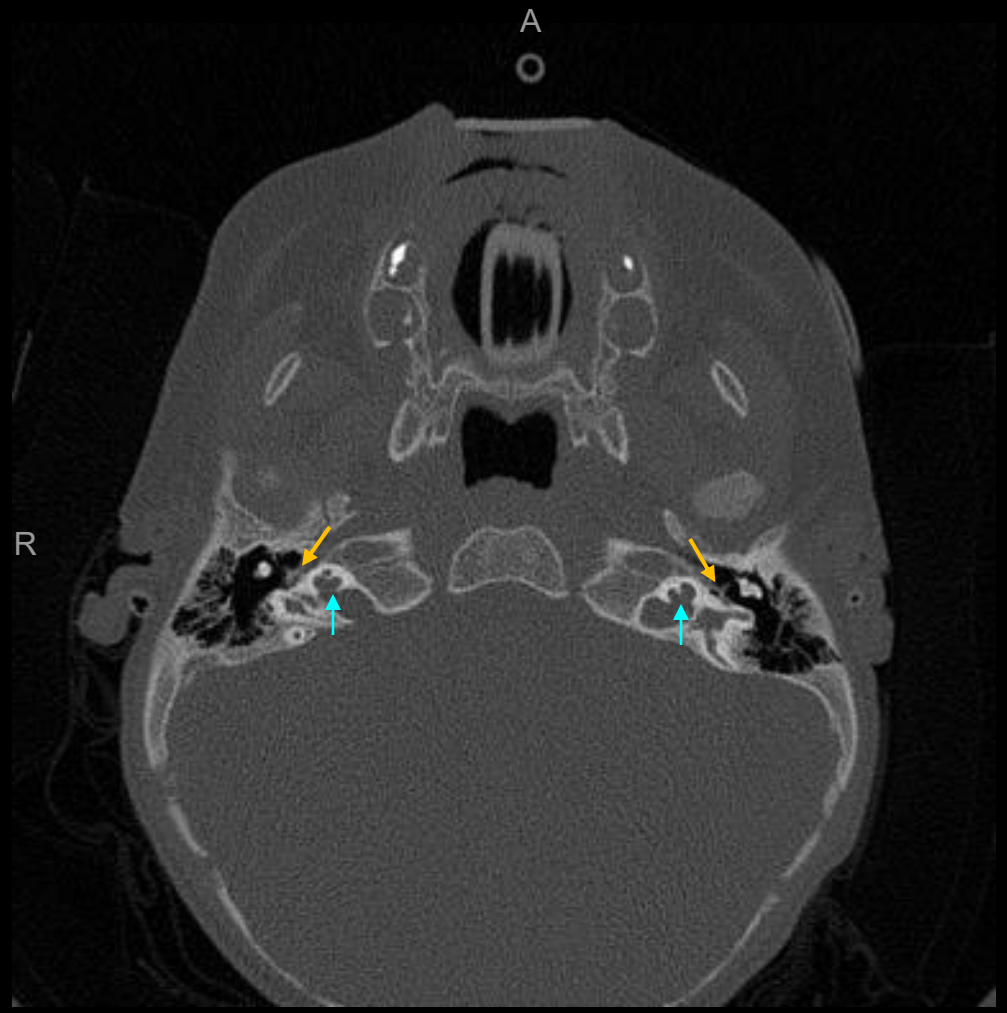
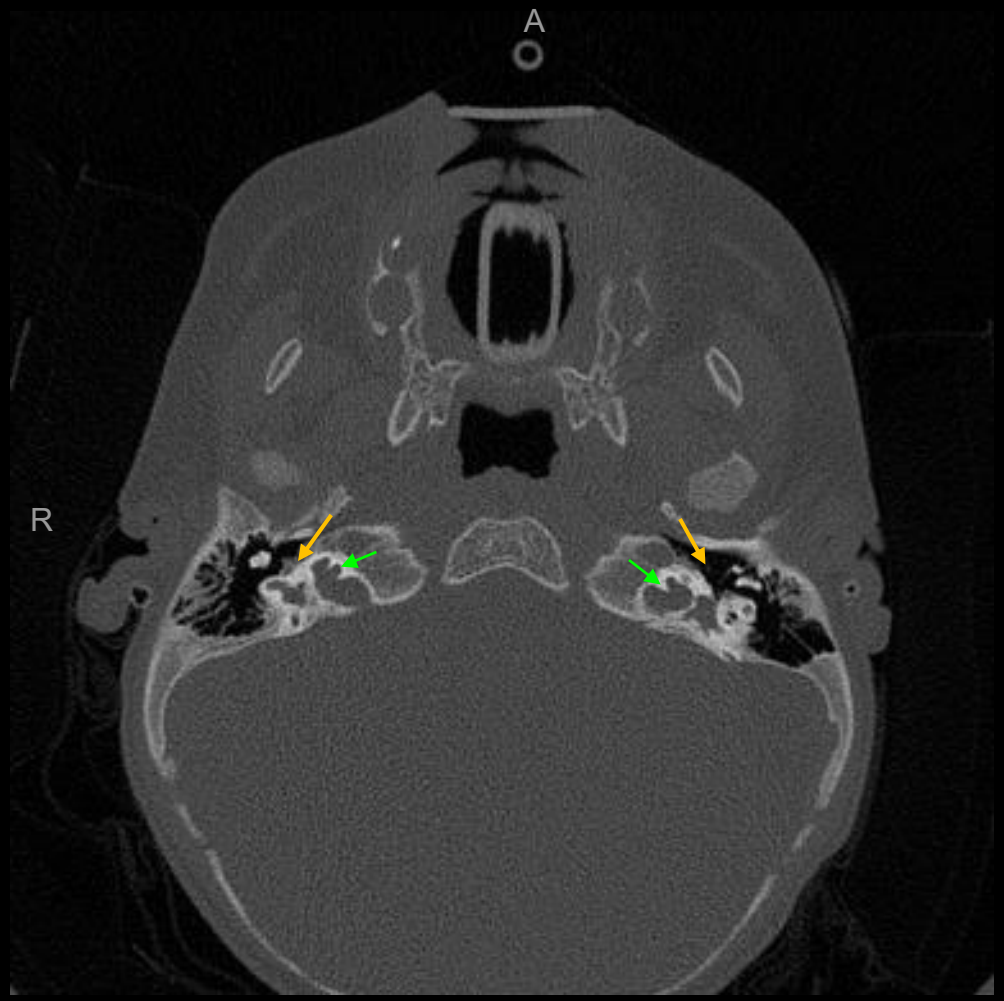
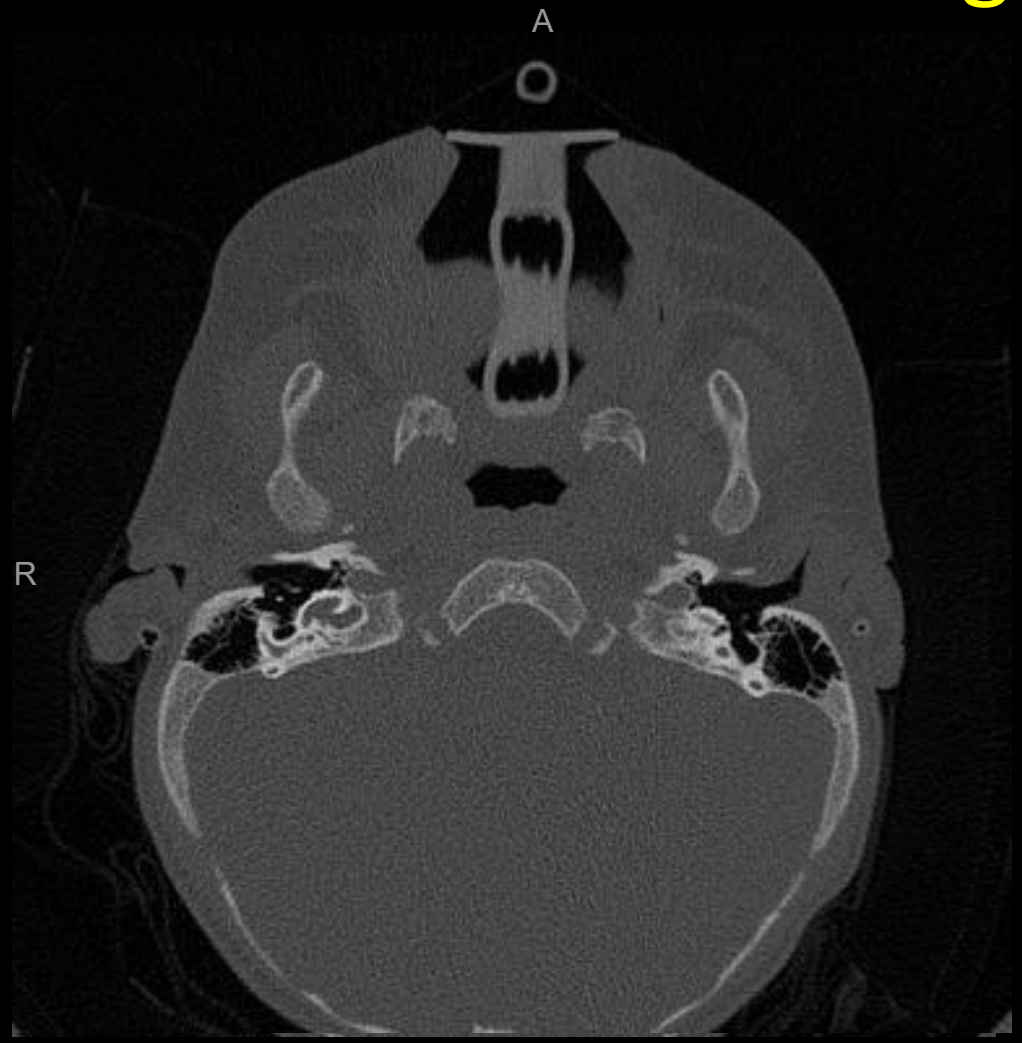
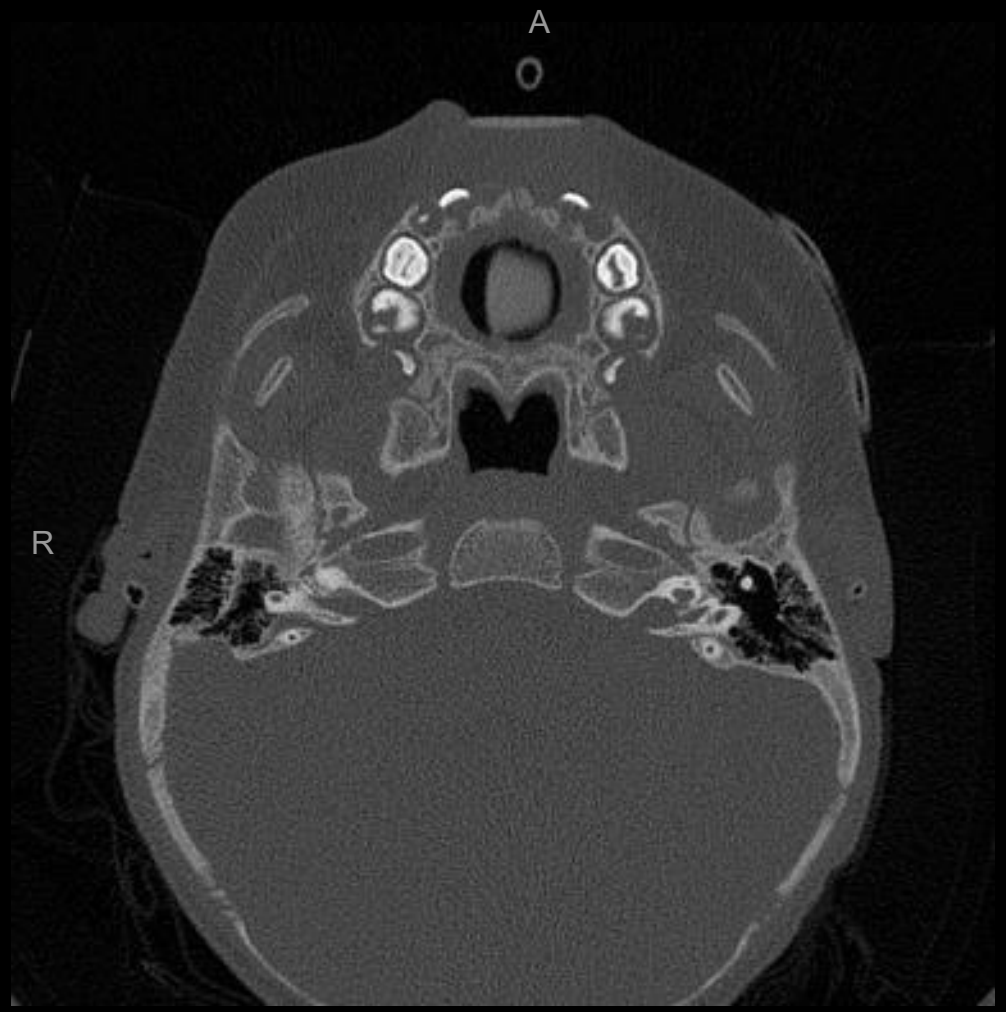



Stapes, oval and round windows are slightly dysplastic bilaterally. There is corkscrew appearance of the cochlea with absence of modiolus, interscalar septa and cribriform plate between the basal turn of the cochlea and internal auditory canal bilaterally.

# CT Findings (unlabeled)



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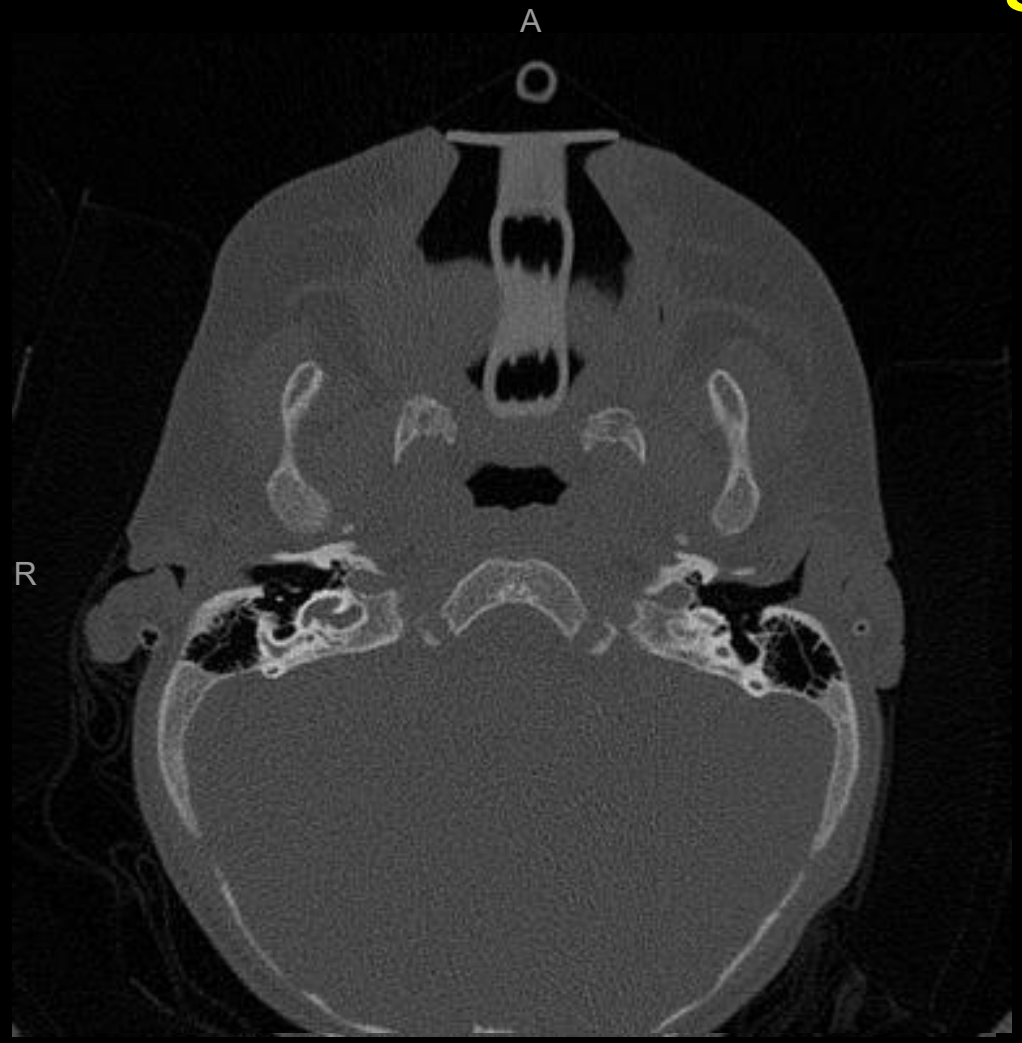


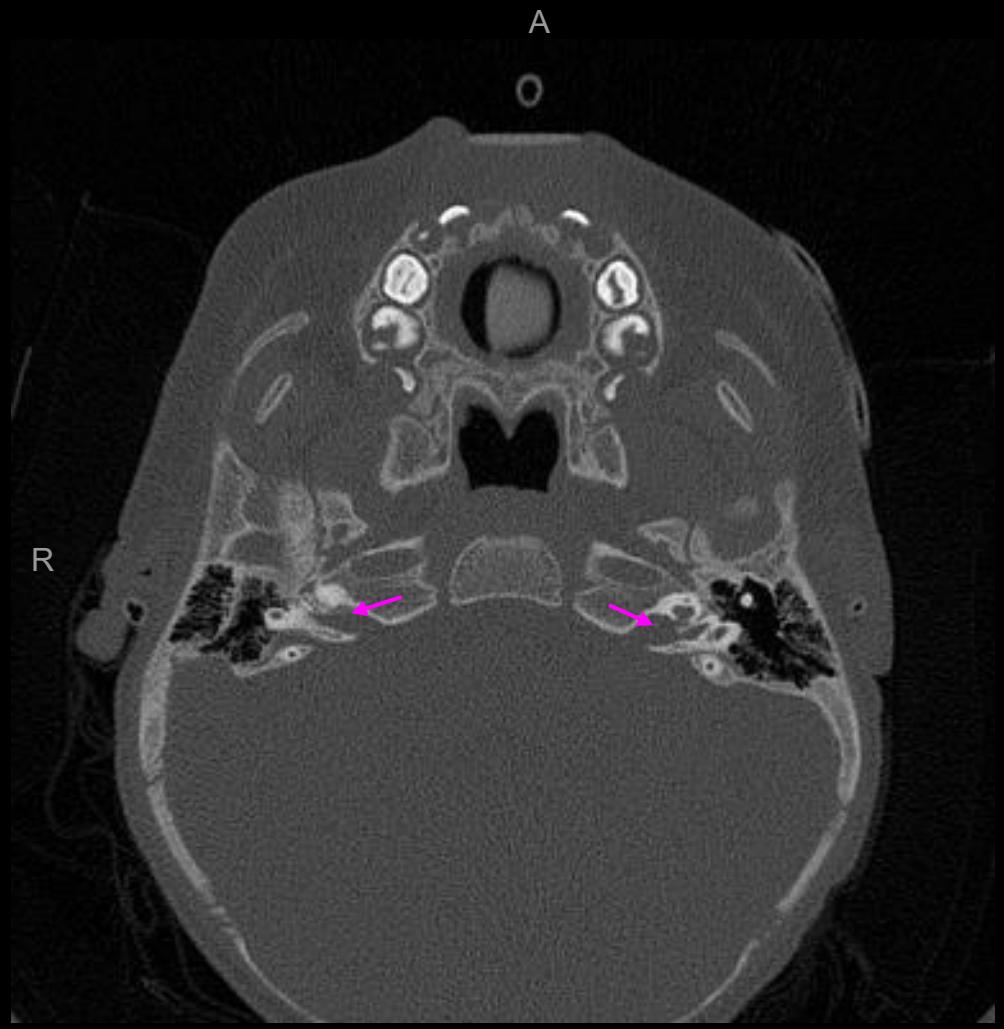


Stapes, oval and round windows are slightly dysplastic bilaterally. There is corkscrew appearance of the cochlea with absence of modiolus, interscalar septa and cribriform plate between the basal turn of the cochlea and internal auditory canal bilaterally. There is widening of the fundus of the internal auditory canal bilaterally.

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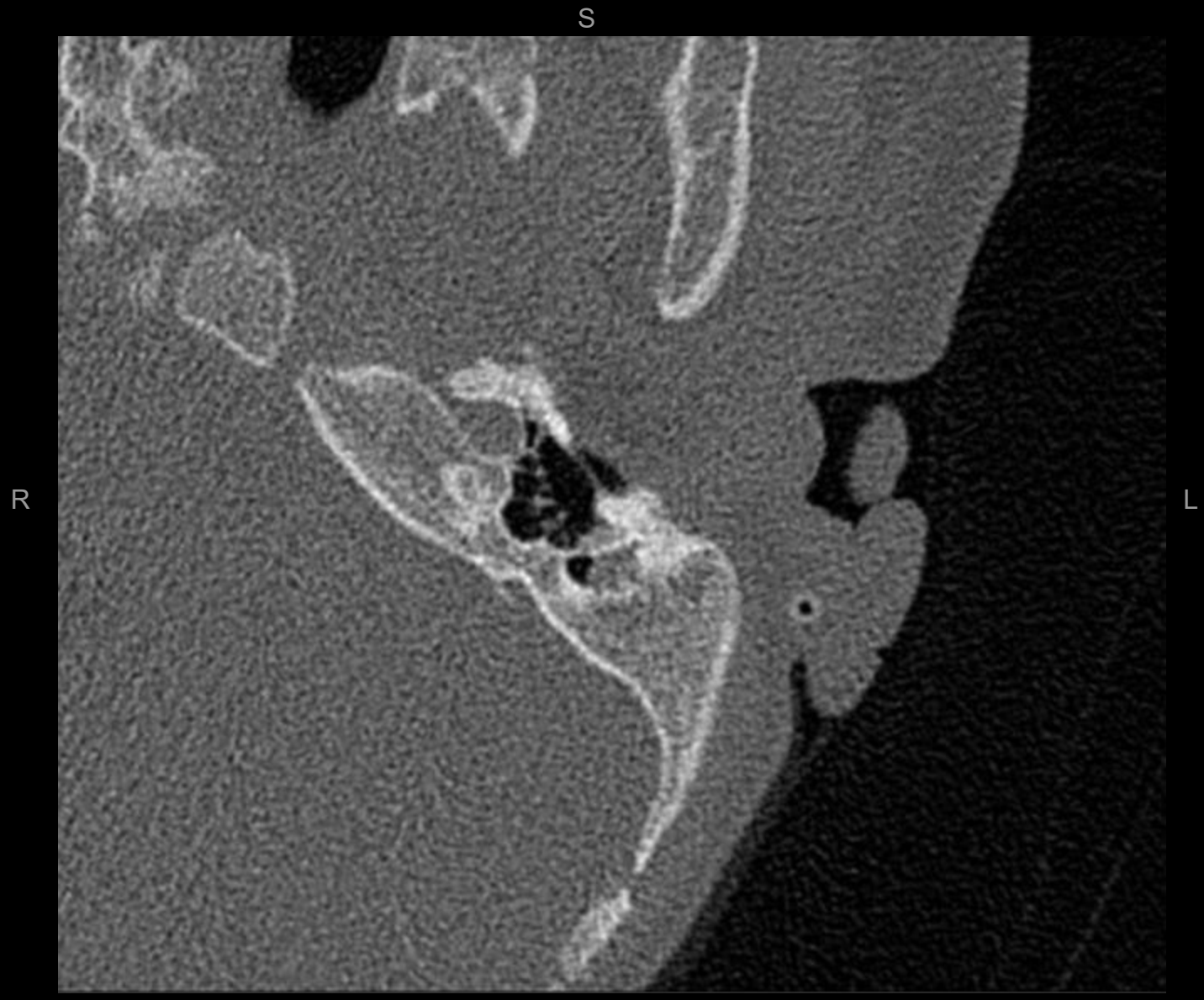

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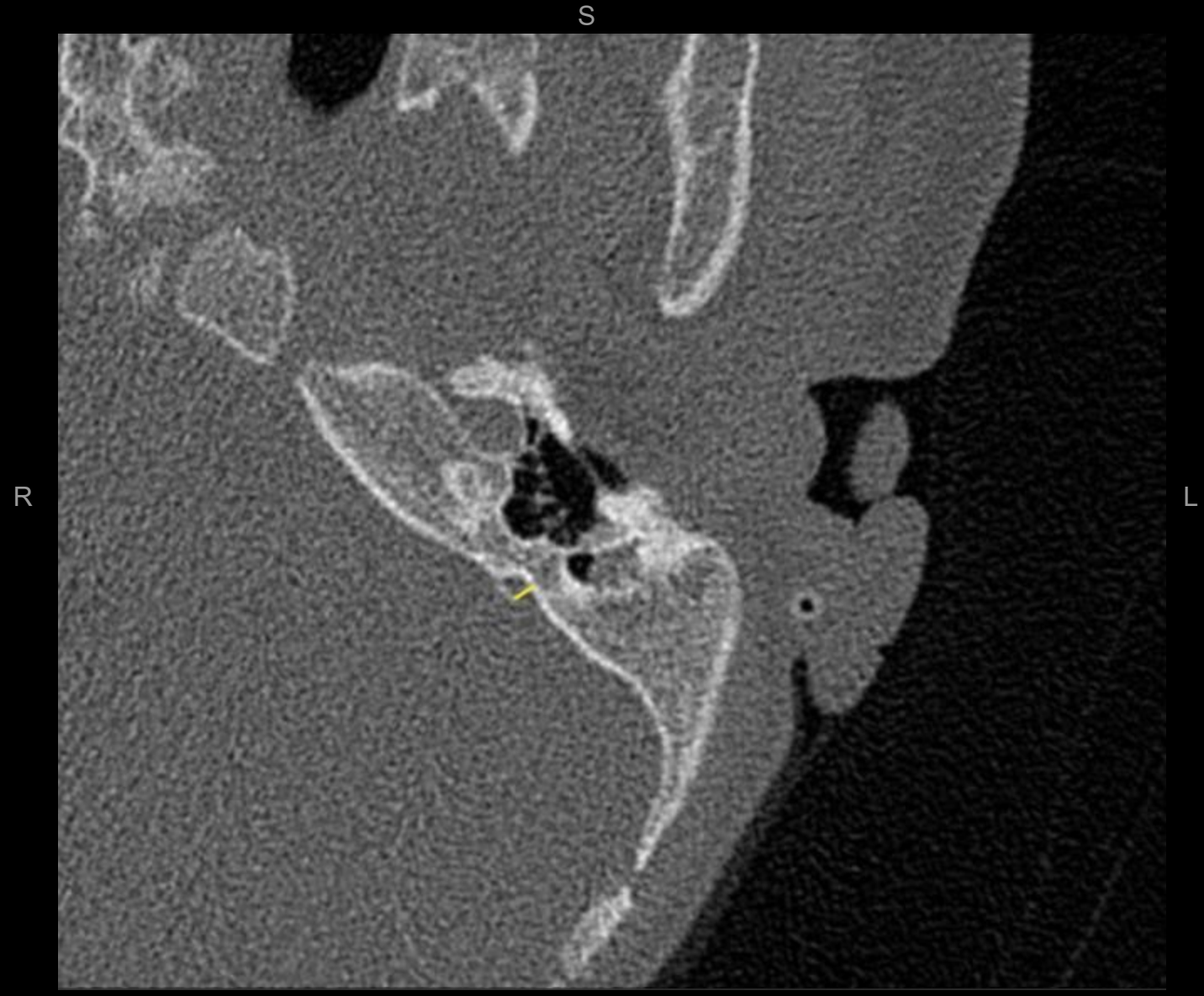
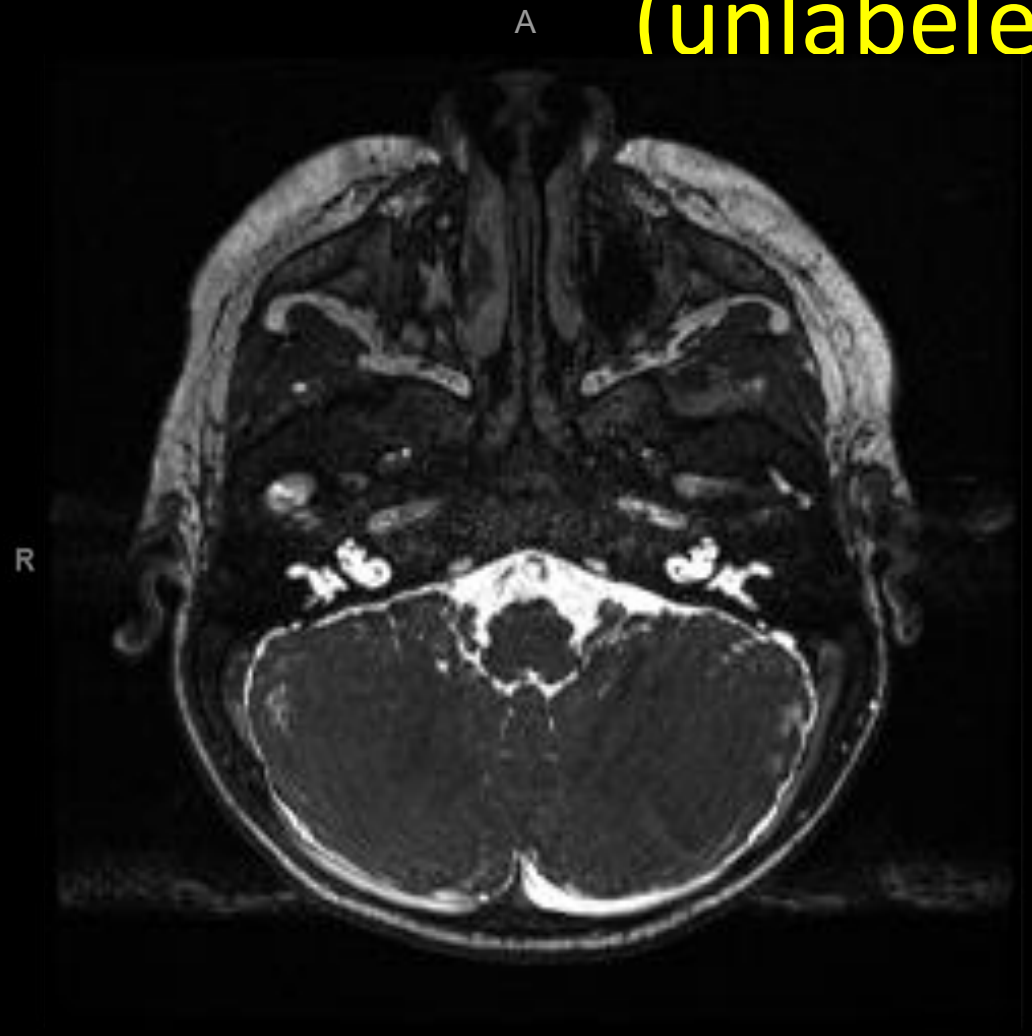
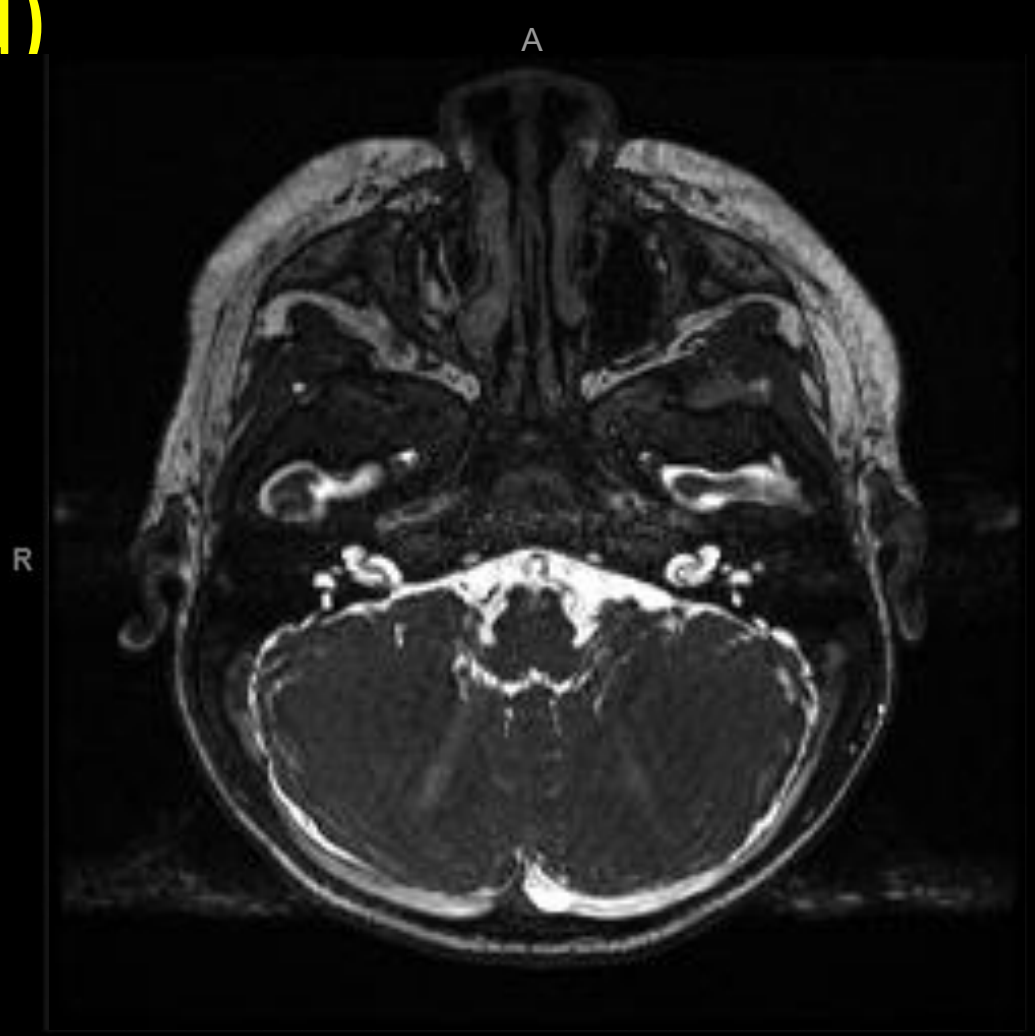



There is mild left vestibular aqueduct dilatation measuring 1.5 mm.

# MRI Findings (unlabeled)



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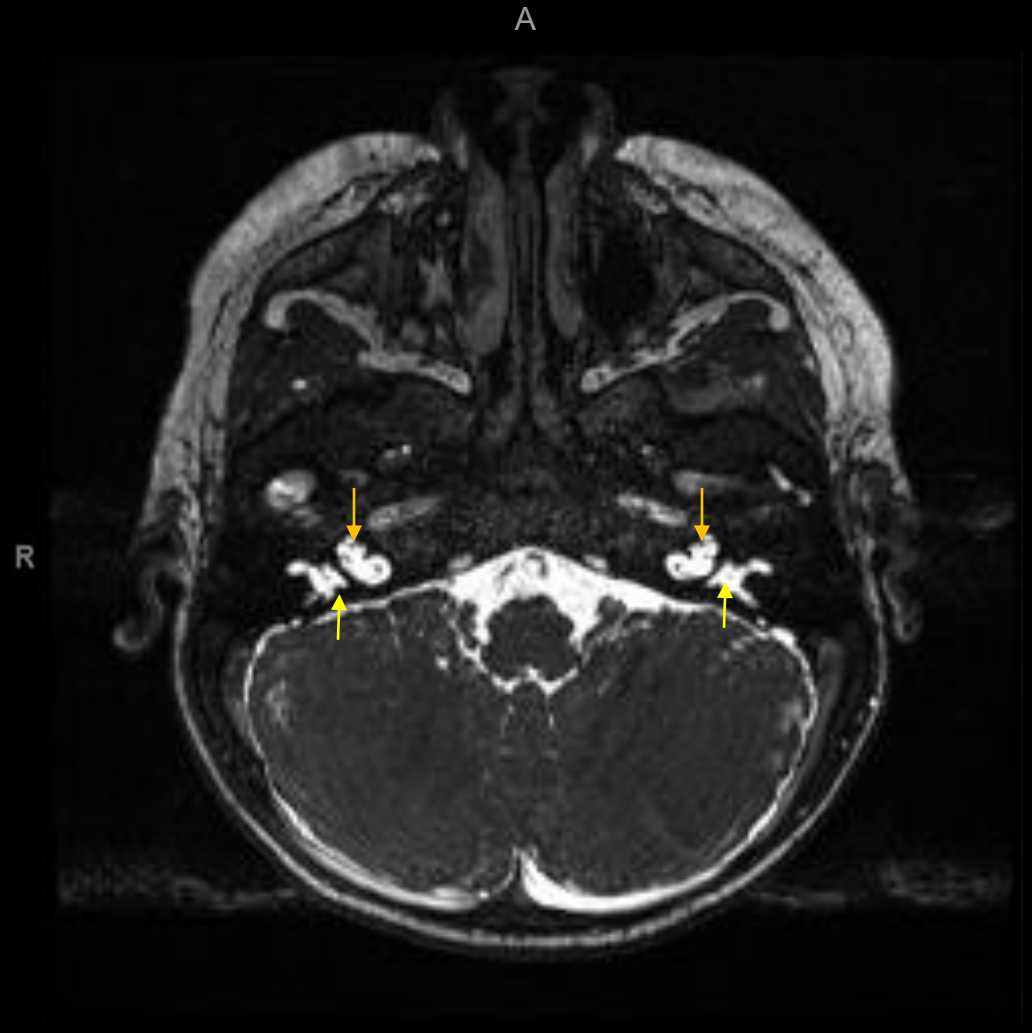


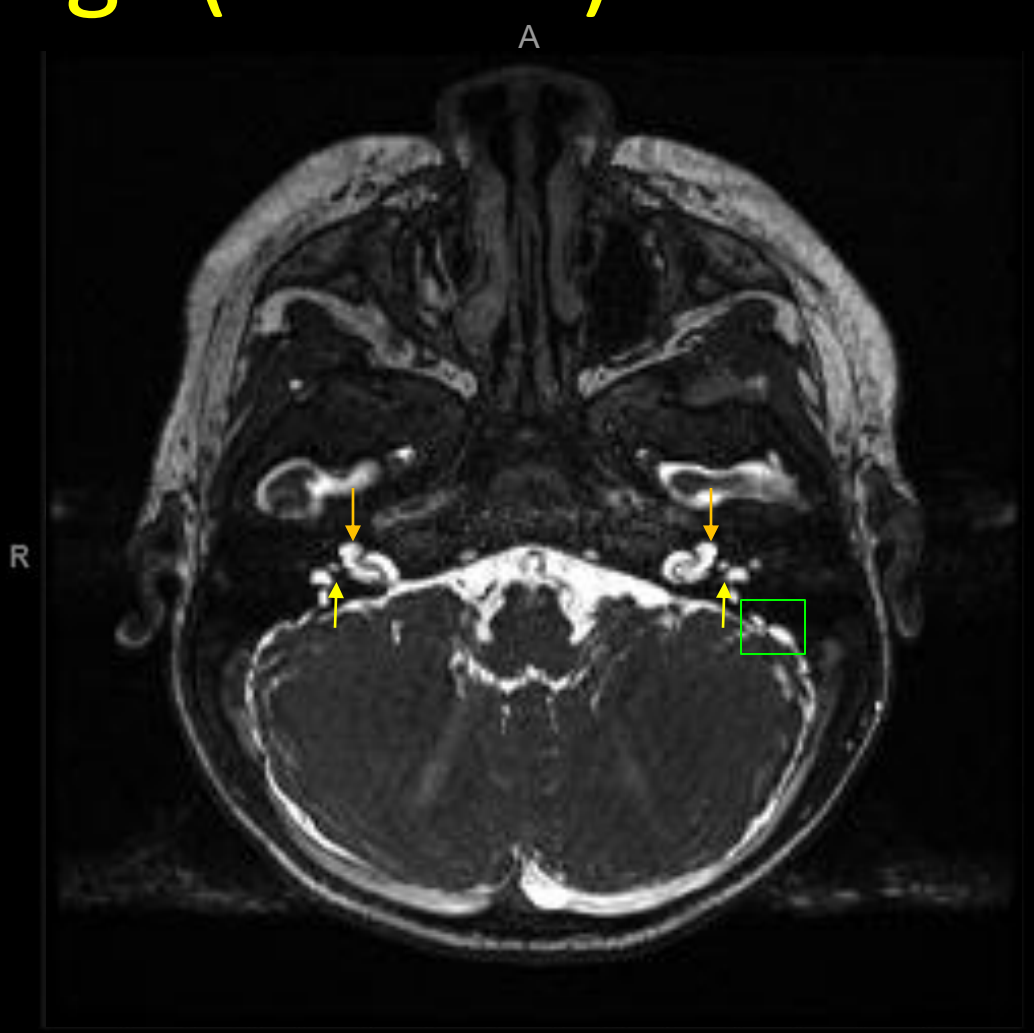


There is corkscrew appearance of the cochlea bilaterally with symmetrical widening of the fundus of bilateral internal auditory canals. Irregular appearance of the vestibule bilaterally. There is mild dilation of the left vestibular aqueduct with diameter of approximately 1.5 mm.

# Final Dx:

X-Linked Gusher Syndrome

(Bilateral cochlear incomplete partition type III)

# Case Discussion

## Etiology:

- The *POU3F4* gene is identified as the location where the mutation occurs. *POU3F4* is located at Xq13-q21.1 and the mutation of the gene that causes the condition is DFNX2. DFNX2 leads to a loss in the conductive element of hearing.[1-3]

## Epidemiology:

- As an X-linked syndrome, a vast majority of cases are males with hearing loss with some evidence of milder phenotypes for females with the mutation. The true prevalence is unknown due to the rare nature of the condition and the unclear inheritance pattern, an estimate of the prevalence is 0.0018 in 1100 live births.[1,2]

## Clinical Features:

- Moderate to severe conductive to sensory cochlear hearing loss, with the mixed variety being the most common.[1,2]

# Case Discussion

## Imaging Findings:

- The CT phenotype features are bulbous internal auditory meatus, incomplete separation of the coils of the cochlea from the internal auditory meatus, and wide first and second parts of the intratemporal facial nerve along with an absent bony modiolus.[1,2]

## Management:

- Bilateral cochlear implant along with digital amplification is the first step in management. Vestibular rehabilitation is recommended as well.[2]

# References:


1. Anger GJ, Crocker S, McKenzie K, Brown KK, Morton CC, Harrison K, MacKenzie JJ. X-linked deafness-2 (DFNX2) phenotype associated with a paracentric inversion upstream of POU3F4. Am J Audiol. 2014 Mar;23(1):1-6. doi: 10.1044/1059-0889(2013/13-0018). PMID: 24096866; PMCID: PMC4644427.
2. 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. Published 2023 Aug 4. doi:10.3390/audiolres13040052
3. Vore AP, Chang EH, Hoppe JE, Butler MG, Forrester S, Schneider MC, Smith LL, Burke DW, Campbell CA, Smith RJ. Deletion of and novel missense mutation in POU3F4 in 2 families segregating X-linked nonsyndromic deafness. Arch Otolaryngol Head Neck Surg. 2005 Dec;131(12):1057-63. doi: 10.1001/archotol.131.12.1057. PMID: 16365218; PMCID: PMC6775642.