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## Introduction

**OBJECTIVE:** To describe a novel case of congenital profound bilateral sensorineural hearing loss in a patient with bilateral nodular internal auditory canal and labyrinthine enhancement and temporal bone dysplasia.

**PATIENTS:** A 76-day-old female was referred to our clinic for congenital deafness. Behavioral observations and objective audiometric evaluation demonstrated bilateral profound sensorineural hearing loss and a comprehensive multidisciplinary evaluation identified compound heterozygous pathogenic variants in MYO7A, a gene associated with Usher Syndrome Type 1B or DFNB2. Computed tomography and contrast-enhanced magnetic resonance imaging studies demonstrated bilateral temporal bone anomalies with unique middle and inner ear malformations, as well as unique contrast enhancement in the membranous labyrinth and internal auditory canals that are not typically associated with MYO7A variants suggesting that a second unidentified process may be occurring.

**INTERVENTIONS:** Given the potential risk for progressive bilateral labyrinthitis ossificans, bilateral simultaneous cochlear implantation was performed at 4 months of age.

**MAIN OUTCOME MEASURES:** Subsequent audiologic follow up after implantation shows significantly improved access to auditory information and increased vocalizations. Speech/language skills were assessed using the Receptive-expressive Emergent Language Test (REEL-3) and both her receptive and expressive language abilities were found to be commensurate with her chronological age.

**CONCLUSION:** We report a novel presentation and imaging findings of congenital bilateral profound sensorineural hearing loss in a patient with nodular internal auditory canal and labyrinthine enhancement and co-existing inner ear dysplasia. Despite the radiologic findings of labyrinthine and internal auditory canal enhancement, the patient has demonstrated good benefit from cochlear implantation. Future study of rare variants of congenital deafness, such as this, is critical toward defining new disease processes and determining optimal treatment.

## Results

After 5 months of full time use and participation in intensive early intervention, the patient's auditory skills were measured using the LittleEARS Parent Auditory Questionnaire and were found to meet, or slightly exceed, performance expectations according to her chronological age. The patient is noted to consistently respond to her name, differentiate between different speakers, and recognize familiar environmental sounds. In addition, she is expanding her vocalizations. Speech/language skills were assessed using the Receptive-expressive Emergent Language Test (REEL-3) and both her receptive and expressive language abilities were found to be commensurate with her chronological age.

Genetic testing by targeted exome panel (Otoscope, University of Iowa, Molecular Otologic-Renal Laboratory (MORL) identified two missense variants in MYO7A, c.1456C>T (p.His486Tyr) and c.3931T>C (p.Ser1131Pro) based on transcript NM\_000260. Parental testing identified that each variant was inherited in trans. Neither variant has been previously reported as pathogenic, however, MORL laboratory interpretation based on software predictions of pathogenicity supported that the variants were causative. The first variant p.His485Tyr, has not been previously reported in ExAc (<http://exac.broadinstitute.org/> accessed Dec 29, 2016) nor ClinVar (accessed Dec 29, 2016). The second variant, p.Ser1131Pro, was identified in 5 out of 15068 individuals of South Asian origin for an allele frequency of 0.0003.

## Figures

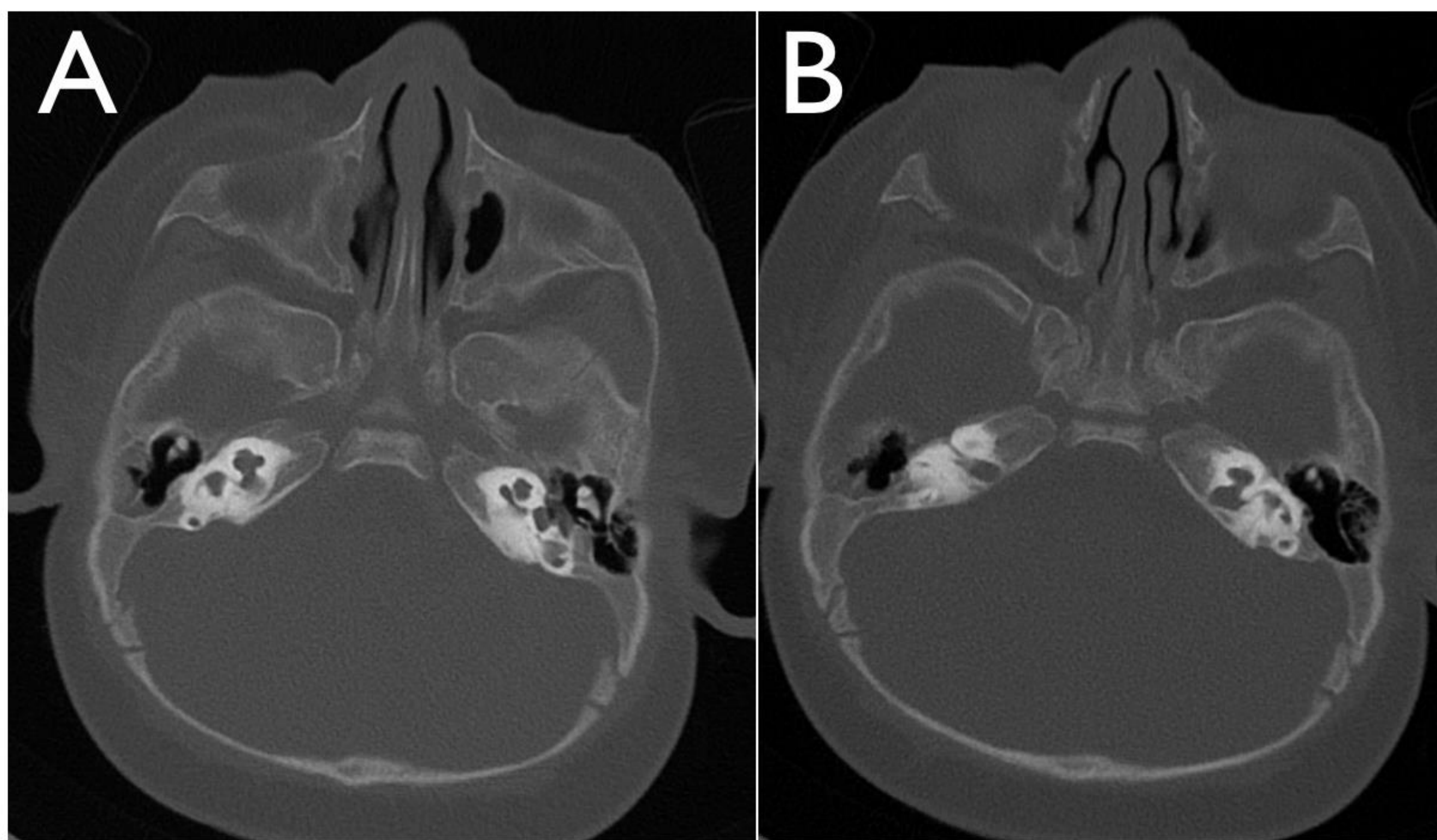


Figure 1: CT Axial gadolinium enhanced imaging at the level of the bony labyrinth shows dysmorphic cochleae (A) unwound/corkscrew morphology, as evidenced by bulbous middle turns, flattened apical turns, and asymmetric modiolus and Patulous cochlear apertures. There is also evidence of a duplicate external auditory canal (B)

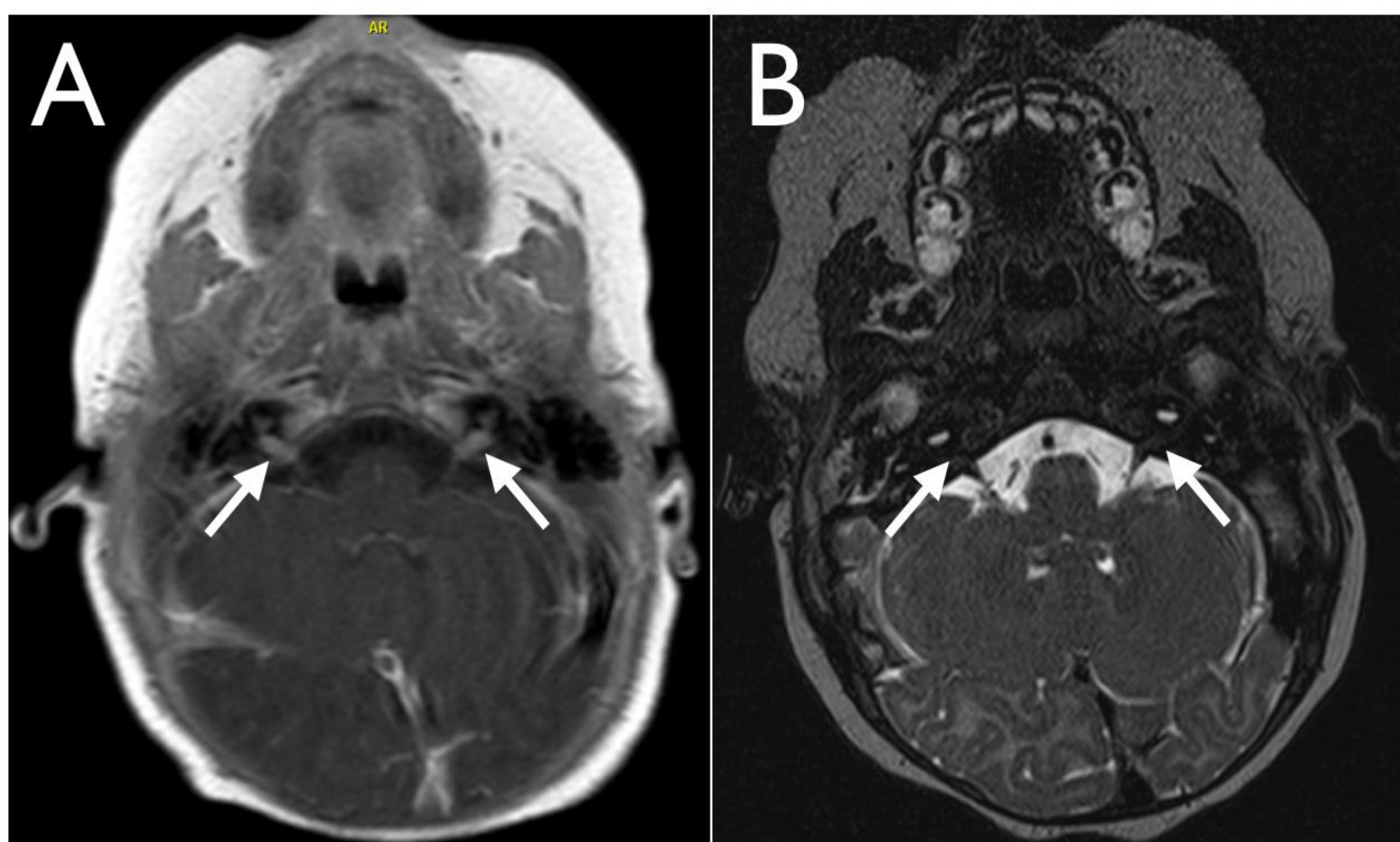


Figure 2: T1 Axial (A) gadolinium enhanced imaging through the level of the internal auditory canal shows bilateral circumferential enhancing tissue which displaces the normal CSF signal. Within the IAC the nerve configuration is obscured by the circumferential enhancing tissue.

## Discussion / Conclusion

Early diagnosis of SNHL in children is paramount as it is well established that a delay in identification and treatment of hearing impairment can adversely affect speech and language development. We report a novel presentation of congenital bilateral profound SNHL in a patient with nodular IAC and labyrinthine enhancement and inner ear dysplasia. A comprehensive evaluation under the direction of a multidisciplinary team was performed and identified biallelic pathogenic variants in MYO7A, however, the MRI findings are not consistent with this diagnosis suggesting second etiology may be occurring in this child. The patient has demonstrated good benefit from cochlear implantation. Future study of rare variants of congenital deafness, such as this, is critical toward defining new disease processes and determining optimal treatment.

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