Vestibular dysmorphia and cochlear hearing impairment in Down's syndrome

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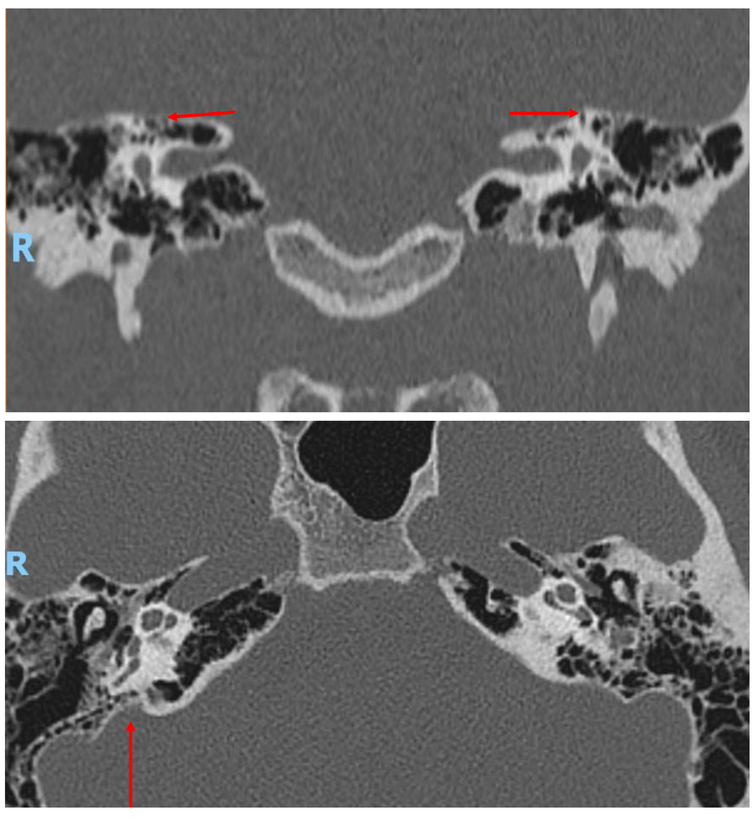
Introduction

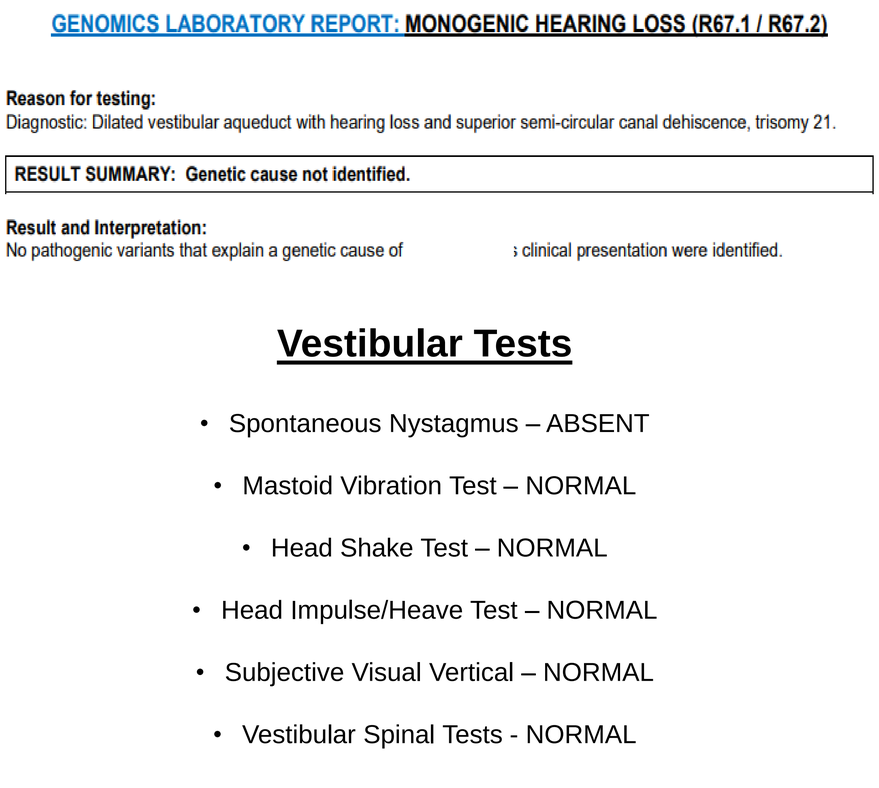
Patients with Down's Syndrome (DS) typically present with conductive hearing impairments in more than 75% of cases. Sensorineural hearing loss is rare and found in about 4.5%. Temporal bone abnormalities have been identified but the true prevalence is not known as children with this condition do not undergo routine temporal scans. In those who do, 75% have been reported with various abnormalities where less than 10% constitute third windows. This presentation highlights a mixed hearing loss in a child with DS and multiple third windows in the same subject that has not been reported before.

Patient history

A child diagnosed with DS was identified and regularly monitored in the Audiology department. The child presented with mild aural dysplasia and persistent otitis media with effusion. Pure tone audiometry and tympanometry testing were performed in accordance with BSA guidelines and a conductive hearing loss was identified. In 2017 the conductive air-bone gap increased and this was reflected in tympanometry testing which showed a flat response. Two years later a mixed loss appeared in the left ear, with the right side still presenting with a mild conductive impairment. The patient was fitted with a contact-mini bone conductor hearing aid at this time. Post pandemic, the child then presented with a more pronounced mixed hearing loss in the left ear, with middle ear recovery proven by normal, peaked tympanometry suggesting a 3rd window pathology. The patient received some benefit from the hearing aid use and continued to use it even when the conductive element of the hearing impairment had resolved.
This phenotype led to aetiological investigations to determine the underlying cause, including the full vestibular test battery. A high resolution CT scan of the temporal bones showed bilateral superior semicircular canal dehiscence and a right sided enlarged vestibular aqueduct. R65 and R67 gene panel testing and karyotype testing to investigate sensorineural hearing loss were normal. Vestibular function tests assessing static, low, mid, and high frequency semicircular canal function and gravitational sensor function were all normal. The child did not exhibit any 3rd window symptoms.



(Top) Bilateral Superior Semi-Circular Canal Dehiscence **(Bottom)** Right Enlarged Vestibular Aqueduct



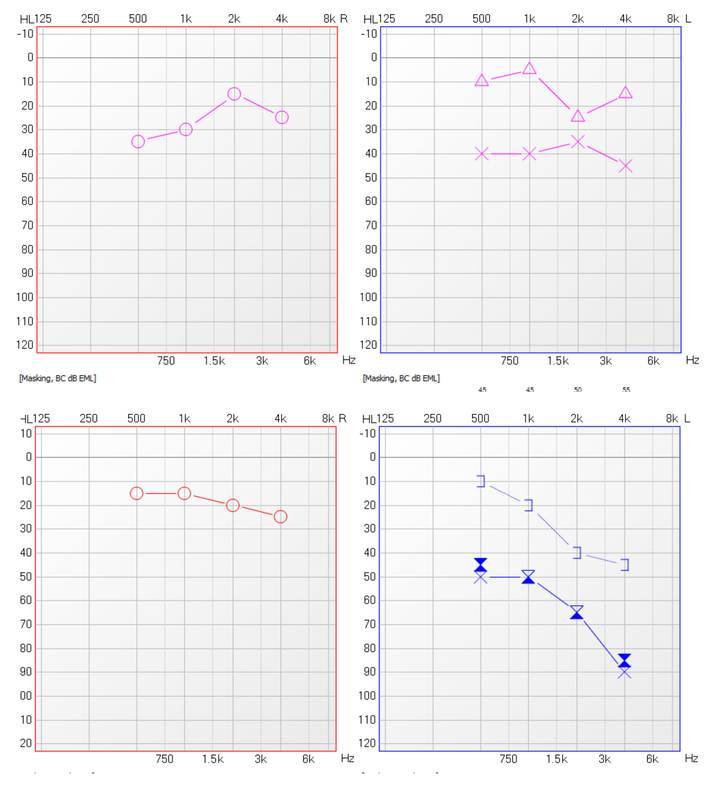
GENOMICS LABORATORY REPORT: **MONOGENIC HEARING LOSS (R67.1 / R67.2)**

Reason for testing:
Diagnostic: Dilated vestibular aqueduct with hearing loss and superior semi-circular canal dehiscence, trisomy 21.

RESULT SUMMARY: Genetic cause not identified.

Result and Interpretation:
No pathogenic variants that explain a genetic cause of ; clinical presentation were identified.

(Top, middle) Genetics Report



(Top) Audiogram June 2015 **(Bottom)** Audiogram Sept 2021

Vestibular Tests

- Spontaneous Nystagmus – ABSENT
- Mastoid Vibration Test – NORMAL
- Head Shake Test – NORMAL
- Head Impulse/Heave Test – NORMAL
- Subjective Visual Vertical – NORMAL
- Vestibular Spinal Tests - NORMAL

Discussion

The commonest cause of hearing loss in Down's syndrome is persistent conductive hearing loss due to otitis media with effusion. Sensorineural hearing loss is very rare and usually occurs in approximately 5% of patients, manifesting in the mid-teens. Temporal bone abnormalities are sporadically reported and this cohort of paediatric patients do not routinely undergo temporal CT scans. When such abnormalities are found in temporal bone structures, less than 10% present with a 3rd window pathology.
Our child initially presented with persistent otitis media with effusion, followed by recovery of middle ear function but the conductive hearing loss continued and we began to see the development of a mixed hearing impairment on the left side. The patient was extensively investigated and it was proven through imaging investigations that the bilateral 3rd window was the underlying cause of the hearing impairment. Therefore, we cannot discount the possibility of a mixed hearing loss developing in the right ear in the future.

Conclusions

It is crucial that children with DS, with persistent conductive/mixed hearing losses are fully investigated for future management implications. Our case highlights the sensorineural aspect in Down's, therefore such children must undergo regular monitoring and situational counselling as to how to manage these audio vestibular phenotypes and mitigate risk factors for optimal function. Vestibular quantification is also important as vestibular structural abnormalities may accompany such a hearing loss as was in our case, with the mixed loss being caused by the third windows.
Alongside the hearing impairment and canal dehiscence, our child was identified as having a second vestibular abnormality in the right ear, therefore we should not rule out the possibility that multiple vestibular dysmorphias may be present in the same patient and look closely at the vestibular system holistically through imagining. Multiple third windows have not been reported as yet as shown in our case. Such findings have important connotations for future vestibular symptom manifestation and management. Parents and patients need to be educated on the distinct possibility of hearing loss progression and vestibular deterioration as part of the individualised management plan.

References

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