

# Inner ear malformations in siblings presenting with vestibular symptoms in early childhood

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**Introduction.** Although the association between inner ear abnormalities and progressive sensorineural hearing loss is well known, vestibular signs or loss of vestibular function in these patients are often unrecognised by medical practitioners.

**Case report.** We report on two siblings with identical inner ear malformations, with special reference to the vestibular symptoms they displayed, as well as the rapid decline in hearing they both experienced in early childhood. We provide a brief overview of the latest classification of these inner ear defects as well as a review of the literature pertaining to children with inner ear malformations presenting with vestibular symptoms.

**Conclusion.** Gross anatomical defects of the inner ear are present in approximately 20% of cases of congenital hearing loss. These defects may result in a rapidly progressive hearing loss. Any child presenting with vestibular symptoms should be referred for an audiological assessment.

Although the association between inner ear abnormalities and progressive sensorineural hearing loss is well known,<sup>1-10</sup> vestibular signs or loss of vestibular function in these patients are often unrecognised by medical practitioners. We report on two siblings with inner ear malformations, with special reference to the vestibular symptoms they displayed, as well as the rapid decline in hearing they both experienced.

## Case reports

The elder sibling had no risk factors for congenital hearing loss and displayed normal developmental milestones during the first 18 months of his life. He suffered a minor fall shortly before his second birthday and developed an incapacitating loss of balance a few hours later. A paediatrician diagnosed concussion. A computed tomography (CT) scan performed at the time, aimed at detecting intracranial haemorrhage, was reported as normal, although a review of this scan does show enlarged vestibular aqueducts (Fig. 1).



Fig. 1. CT scan demonstrating enlarged vestibular aqueduct (white arrow).

One month later the patient was taken to an audiologist for a hearing test because his language development was slow. An auditory brainstem response (ABR) test indicated thresholds of approximately 50 dB in the left ear and 40 dB in the right ear (Fig. 2). One week later he fell backwards onto a tiled floor, this time without obvious balance disturbance. A second ABR test performed 8 weeks later indicated a dramatic worsening of the thresholds.

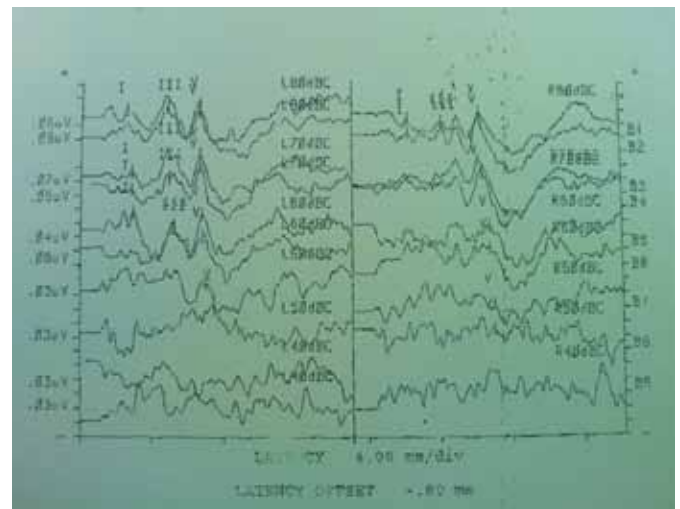


Fig. 2. First ABR test.

A paediatrician recommended that the patient's language and motor development be monitored over the next month. At this stage he had a vocabulary of approximately 30 words in two languages, but left out high-frequency 's' sounds. He demonstrated good mimicry of animal sounds, was aware of sounds such as his younger sibling crying and people knocking at the door, and enjoyed listening to and moving rhythmically to music.

The patient was brought to our institution on the advice of a mother whose son had received a cochlear implant at our centre. The findings on clinical examination were unremarkable, with behavioural responses to loud sounds. His speech suggested a lack of access to high-frequency sounds. A CT scan and magnetic resonance imaging (MRI) showed bilateral enlarged vestibular aqueducts, enlarged endolymphatic ducts and sacs, and a type II incomplete partition (classic Mondini) defect (Figs 3 and 4). ABR/auditory steady-state

response (ASSR) and behavioural testing demonstrated thresholds greater than 100 dB in the left ear and in the 60 - 80 dB range in the right ear. He was fitted with bilateral digital hearing aids and was enrolled in an intensive speech therapy programme at age 2 years 7 months. Receptive and expressive language development was monitored.

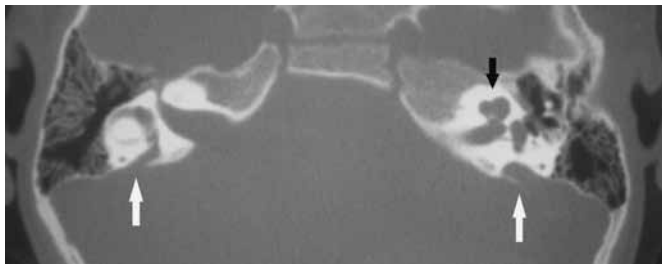


Fig. 3. High-resolution CT scan demonstrating enlarged vestibular aqueducts (white arrows) and cystic second and apical turns (black arrow).

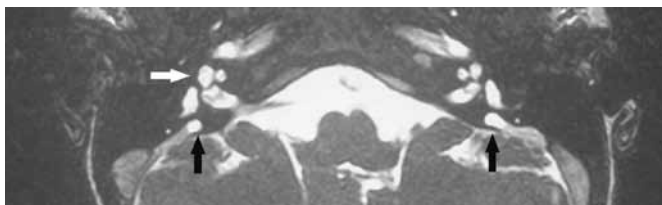


Fig. 4. T<sub>2</sub>-weighted MRI demonstrating enlarged endolymphatic sacs (black arrows) and cystic second and apical turns (white arrow).

Fourteen months later he suffered another minor fall, which resulted in significant hearing loss (approximately 15 dB deterioration) in the better (right) ear when tested 2 weeks after the incident (Fig. 5), without any clinical acute vestibular loss. A decision was taken to implant a Nucleus cochlear implant in his left ear (at age 3 years 11 months) and continue with the hearing aid in his right ear. Surgery was uncomplicated, with full insertion of a curved array. There was no vestibular fallout postoperatively and he was walking on the day of surgery. Five months later minor head trauma resulted in the loss of the remainder of his hearing in the right ear and he received a second Nucleus implant. Postoperatively he demonstrated acute vestibular fallout which compensated over a few weeks. No gushers were encountered with either of the surgeries, but the cochleostomies did result in an initial excess leakage of fluid.

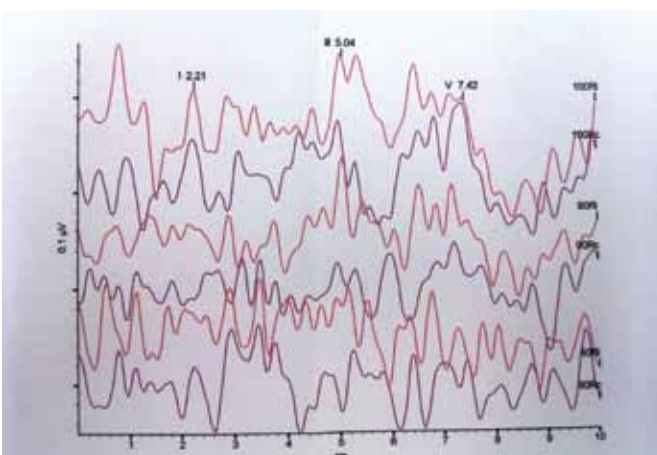


Fig. 5. Final ABR indicating profound hearing loss in the right ear.

The younger sibling was born approximately 2 weeks before his older brother fell for the first time. He was accidentally hit on the head with a mobile phone by his brother at 6 weeks of age and bumped his head again at 12 weeks of age. Both these events resulted in nystagmus, emesis and head tilt to the right. He was diagnosed with concussion on both occasions. He failed oto-acoustic emission (OAE) screening twice, and was first seen at our institution at 5 months of age because of grave parental concern about his lack of consistent

auditory responses. His motor milestones were appropriate for age and he was sitting unaided. ABR/ASSR testing confirmed bilateral sensorineural hearing loss, profound in the left ear and severe in the right ear, which was confirmed one month later (Fig. 6). CT and MRI confirmed bilateral enlarged vestibular aqueducts and type II incomplete partition defects. He was fitted with bilateral digital hearing aids and enrolled in a speech therapy programme.

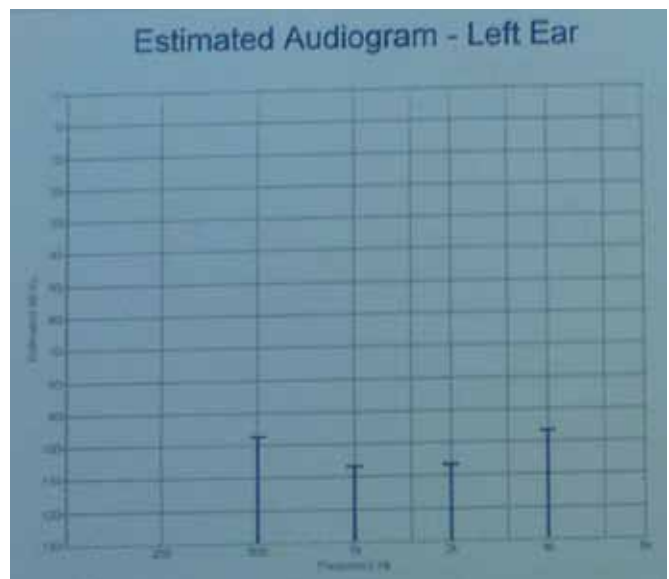
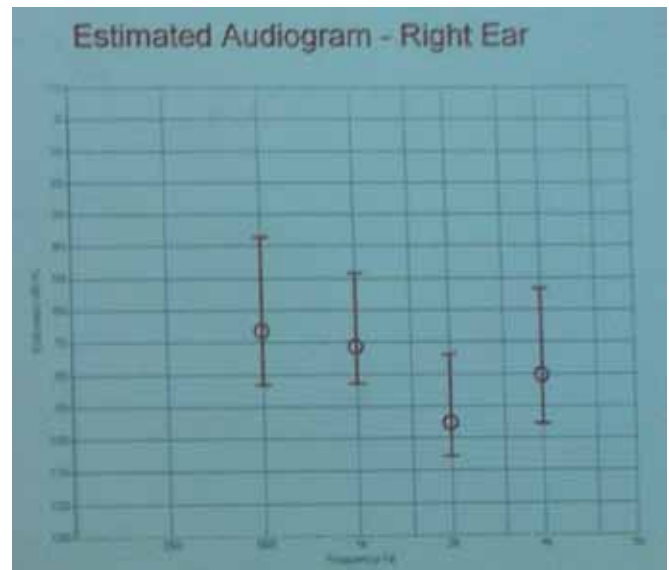


Fig. 6. ASSR test indicating profound loss in the left ear and moderate-severe loss in the right ear.

Over the next 7 months, he developed obvious vestibular fallout on at least four occasions while attempting to stand and walk. Each episode resulted in a regression of these key motor milestones, and he displayed left-beating nystagmus. With compensation, he was able to sit, stand and then walk again.

Although aided audiograms showed thresholds of 45 - 50 dB in the right ear, it was decided to implant the left ear because of poorer than expected performance. The procedure was undertaken at age 1 year 4 months. This was the first operation undergone by the two siblings, and a straight electrode array was used. Unfortunately, only 16 of the 22 electrodes could be inserted. Postoperatively he displayed minor imbalance, nausea and vomiting but was ambulatory within a few days and appeared to compensate well over the next few weeks. There were no reported episodes of vestibular decompensation for 4 months, but he then suffered at least two episodes of acute vestibular decompensation with left-beating nystagmus. These episodes were

markedly shorter in duration than before the first operation (<48 hours as opposed to 3 - 5 days previously). Behavioural testing confirmed inadequate assistance from the hearing aid, and he received his second implant at age 1 year 10 months. A full insertion was obtained with a curved electrode array. Postoperatively he again displayed minor imbalance, but was ambulatory within a few days.

Genetic testing for mutations associated with Pendred syndrome was offered to the family, but was declined. The results of thyroid function testing were normal in both children.

### Discussion

Inner ear malformations are present in approximately 20% of cases of congenital hearing loss.<sup>1,2</sup> However, the exact anatomical defect can often be difficult to ascertain from the literature owing to the use of eponyms.<sup>1</sup> In 1987 Jackler *et al.* proposed a new classification system for congenital inner ear malformations based on CT imaging, which divided inner ear malformations up into those of labyrinthine aplasia (Michel deformity), cochlear aplasia, cochlear hypoplasia, incomplete partition (Mondini dysplasia) and common cavity defects.<sup>1</sup> The pathogenesis of the various defects were explained by arrested development of the inner ear at different stages. Sennaroglu and Saatci have proposed an amendment to this classification in order to more accurately classify the incomplete partition malformations by dividing them into three types:<sup>2</sup>

Type I – the presence of a cystic cochlea which lacks a central modiolus or interscalar septae; the vestibule is separate and usually dilated; the outer dimensions of the cochlea are normal; and the endolymphatic duct and sac are usually normal.

Type II – a cochlea with a normal basal turn and basal modiolus while the second and apical turns form a cystic space which lacks interscalar septae; the vestibule is minimally dilated; enlargement of the endolymphatic duct and sac is present.

Type III – a cochlea where interscalar septae are present but the modiolus is completely absent; the cochlea lies directly lateral to the internal auditory canal instead of the usual anterolateral position.

Large vestibular aqueduct syndrome (LVAS) was first described by Valvassori and Clemis in 1978 after reviewing polytomograms of the inner ear.<sup>3</sup> They found that only 60% of patients with enlarged vestibular aqueducts (EVAs) demonstrated other abnormalities of the inner ear and coined the term LVAS for the 40% of cases in which the EVA was the only abnormality found. Subsequent studies based on high-resolution CT imaging have shown that up to 88% of cases of LVAS are associated with other malformations of the inner ear.<sup>4</sup> The presence of an EVA on CT imaging should therefore prompt a detailed examination of the other inner ear structures.

Although the literature contains many references to LVAS or EVAs in children, there are very few reports of children diagnosed with these conditions under the age of 3 years, and even fewer that mention vestibular symptoms. Griffith *et al.* reported on a family of 7 children; 5 with profound SNHL, 1 with moderate SNHL and 1 with normal hearing.<sup>5</sup> Four of the children demonstrated Mondini malformations on CT imaging; 2 children demonstrated episodic head tilt and emesis and another 2 had perilymphatic fistulas. No further mention was made of vestibular function deficits. The profound hearing loss was detected in all cases within the first year of life. Chan *et al.*

reported on 2 children and their mother, all of whom had Mondini dysplasia.<sup>6</sup> The children were 5 and 9 years old, and demonstrated abnormal findings on vestibular testing but were otherwise asymptomatic for vestibular function impairment. Oh *et al.* reported on two unrelated children with childhood diagnoses of LVAS with vestibular symptoms.<sup>7</sup> These symptoms ranged in duration from 15 minutes to 3 hours and were not apparent until after 2 years of age. Grimmer and Hedlund reported on vestibular symptoms in children with an EVA;<sup>8</sup> 10 out of 21 children aged under 18 years reported vestibular symptoms, including 3 children under the age of 3 years. Two of the 3 were reported as having vertigo, while the other was reported to have a motor delay. All 3 demonstrated cochlear dysplasia as well as progressive hearing loss.

Most reports of LVAS/EVA report the diagnosis of the anatomical abnormality many years after diagnosis of early childhood hearing loss with progression of the hearing loss into late childhood or adolescence.<sup>4-7</sup> The hearing loss is also reported to be fluctuant with a gradual decline, but subject to rapid deterioration after minor head trauma.<sup>9</sup> Glucocorticosteroids administered after sudden hearing loss may help to preserve residual hearing.<sup>9</sup> The hearing loss associated with an enlarged vestibular aqueduct may be the presenting or only clinical feature of Pendred syndrome.<sup>10</sup> Most patients with Pendred syndrome remain euthyroid, as dietary iodine intake is usually sufficient to compensate for the iodide organification defect.<sup>10</sup>

### Conclusion

Gross anatomical defects of the inner ear are visible on CT and MRI imaging in approximately 20% of cases of congenital hearing loss. These defects have been well described, but the correct terminology is often not applied. Incomplete partition defects of the cochlea are strongly associated with progressive hearing loss, which may result in profound deafness in the first year of life. Vestibular function impairment in young children, which may manifest as an acute episode of nystagmus, emesis, acute loss of an ability to stand or even sit, delayed motor milestones or regression of motor milestones, should prompt urgent assessment of inner ear morphology with high-resolution CT and MRI as well as a thorough assessment of the hearing.

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