CASE REPORT

Bilateral semicircular canal aplasia

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\textbf{KEYWORDS}

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\section*{Introduction}

Congenital vestibular deformity is rare; it is often discovered during assessment of congenital childhood hearing loss, although a few cases of isolated vestibular abnormality without associated hearing loss have been reported, with or without associated syndrome. Being asymptomatic, this isolated deformity is seldom investigated and thus seldom described. The present report concerns a 24-year-old man presenting with unilateral mixed hearing loss and subnormal contralateral hearing. CT and MRI found complete bilateral semicircular canal (SCC) aplasia.

\section*{Case report}

A 24-year-old male presented with right-ear hearing loss. History included esophageal atresia, managed surgically in the first day of life. The patient was free of vertigo and tinnitus, and otoscopy was normal. Pure-tone threshold audiometry found right mixed hearing loss with subnormal contralateral hearing (Fig. 1). On vocal audiometry, left ear intelligibility threshold was 35 dB, with 100% maximum intelligibility; the right ear showed no response. Weber test results showed right lateralization at low frequencies. Temporal bone CT found bilateral SCC deformity, with complete bilateral aplasia of the posterior and superior canals and severe hypoplasia of the lateral canals (Fig. 2). There was no ossicular deformity, but the oval windows were shrunken bilaterally. Over and above the absence of SCCs, the bone labyrinths showed hypoplastic distended vestibules with bilateral cochlear deformity: modiolus hypoplasia, virtual aplasia of the third turn, and cochlear nerve canal stenosis.

MRI (Fig. 3) confirmed the CT findings and, on the right side alone, found inferior vestibular nerve agenesis with no visible cochlear nerve. Vestibular functional exploration found a normal subjective vertical and normal oculography. Caloric testing found bilateral areflexia, and the video-Head Impulse Test (vHIT) found no response in any of the six canals. Otolithic evoked potentials (OEPs) were

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Figure 1  Pure-tone threshold audiometry with air (AC) and bone conduction (BC) masking value. Subnormal left-ear hearing and right-ear mixed hearing loss.

absent in the right ear; in the left ear, early P13 and N23 waves were present, with normal amplitude and latency. Genetic sequencing found no CHD7 gene mutation (underlying CHARGE syndrome) or chromosome micro-remodeling.

Discussion

Epidemiology

Dysplasia or aplasia of the SCCs, and of the lateral canal in particular, is the most frequent congenital deformity of the inner ear [1,2]. Complete bilateral SCC aplasia, however, is very rare [3]. The first report of total aplasia without cochlear abnormality was of two cases, published in 1990 [2]. Satar et al. [3], in 2003, reported a series of 15 SCC aplasia patients (the largest in the literature), including 10 with associated CHARGE syndrome; only three patients in the series showed no associated major cochleovestibular deformity, as in the present patient, but all had profound hearing loss; indeed, the available data [3,4] suggest that hearing disorder is systematically associated. There was a single report [4] of SCC aplasia with normal hearing (as in the left ear in the present case), whereas posterior labyrinth deformity is usually supposed to be associated with perception or mixed hearing loss; the present case is thus the

Figure 2  Temporal bone CT, axial slice: bilateral SCC aplasia, minor cochleovestibular hypoplasia, especially in the right ear.
second to be reported of total bilateral SCC aplasia with only right cochlear deformity and merely subnormal contralateral hearing.

In 2002, Sennaroglu [5] published a classification of cochleovestibular deformities, reporting SCC aplasia or hypoplasia in 26% of inner ear deformities (23 patients), in none of which the cochleovestibular system was normal. The deformity itself is thus frequent, but is rarely found in isolation.

**Embryology**

Inner ear development begins around the 6th week of fetal life, when the otic vesicle splits. The ventral part (pars inferior) becomes the vestibular sacculus and cochlear canal, which is fully formed by week 9; the dorsal part (pars superior) becomes the utricle then the SCCs as of week 8 [6], the lateral canal being the last to form. It is generally agreed that inner ear deformities result from a sudden arrest of development during these successive stages [2], which is why the lateral canal is the most frequently involved. In the embryonic arrest sequence underlying the classically reported deformities, canal aplasia with a normal cochleovestibular system seems chronologically impossible, as the membranous vestibular labyrinth forms before the definitive formation of the cochlea [5]; this sequential model, however, has been increasingly challenged [2], with a new molecular genetic model accounting for the range of deformities found: cochlear and vestibular development may rely on independent mechanisms, without interaction. Animal models have supplied plentiful genetic data. SCC formation is coded for by the proximal part of chromosome 4 at the 14th centromere [7]. In mouse, the Otx-1 and Nxx5-1 genes appear to be implicated, with isolated mutation inducing lateral SCC deformity without any cochlear involve-

ment seen on radiology or histology [8]; the human homologs of these genes may be mutated in isolated SCC aplasia.

**Syndromic context**

SCC aplasia may be isolated or associated with Goldenhar syndrome [9], trisomy 11 or 18 or CHARGE syndrome [10] (Coloboma, Heart defectomy, choanal Atresia, Retardation of growth and/or development, Genital hypoplasia, and Ear abnormality and/or deafness). SCC abnormality is found in 50 to 100% of CHARGE syndromes [4,11]. In a series of 13 CHARGE patients, Morimoto et al. [11] found 100% SCC aplasia, 91% cochlear dysplasia, 12% round window dysplasia, 91% oval window dysplasia, 58% vestibular deformity, 46% vestibular aqueduct abnormality, 88% facial nerve canal abnormality and 93% ossicular abnormality. Other less frequent facial (asymmetry, hypoplasia, cleft palate) or laryngeal and esophageal abnormalities may also be associated. Although there were no clinical or genetic criteria of CHARGE syndrome in the present case, the bilateral SCC aplasia was associated with esophageal atresia.

**Functional exploration**

The present audiometric findings were compatible with the literature data. In one ear, hearing was unaffected, as in one of the cases reported by Dallan et al. [4]; in the other, there was mixed hearing loss, accountable for by anterior membranous labyrinth deformity, as also described elsewhere [4]. This air-bone gap on audiometry is due both to oval window hypoplasia and to bone conduction enhanced by the labyrinthine deformity and air conduction impaired by a restriction of footplate movement under perilymphatic pressure [12]; cranial vibration transduction is improved in the labyrinthine fluid but reduces air conduction. The
right-ear mixed hearing loss can also be accounted for by cochlear abnormalities of molecular origin.

The vestibular tests corresponded exactly to the CT and MRI findings. As expected and previously described by several authors [2,11], caloric tests found bilateral areflexia, and the absence of vHIT response in all six canals corresponded to the bilateral SCC aplasia. The presence of OEPs in the left ear confirmed left saccus functionality, while their absence in the right ear confirmed the absence of the right inferior vestibular nerve (posterior saccular and ampullary nerve), as seen on MRI.

Conclusion

Isolated congenital semicircular canal deformity has been little described, being rare and asymptomatic. It is thus seldom explored for but rather discovered serendipitously during congenital hearing loss work-up. The present description of a second case of total aplasia with subnormal contralateral hearing suggests probable underestimation of incidence. The association of SCC aplasia with minor cochleovestibular abnormality supports the revision of the sequential embryopathogenic model in favor of a genetic and molecular model of abnormal inner ear embryogenesis. In case of such deformity, syndromic association should be systematically investigated and genetic analysis should be performed.

Disclosure of interest

The authors declare that they have no conflicts of interest concerning this article.

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References