Familial Superior Canal Dehiscence Syndrome

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uperior canal dehiscence syndrome (SCDS), or Minor syndrome, can present with a myriad of auditory and/or vestibular symptoms that are associated with a bony defect of the superior canal.1 High-resolution computed tomography (CT) demonstrates a dehiscent area of bone in the region of the arcuate eminence (in contact with the temporal lobe dura) or the medial (nonampullated) limb associated with the superior petrosal sinus (SPS-SCD).2

The pathogenesis of SCDS is not completely understood, and both congenital and acquired events may contribute to its etiology. One hypothesis is that patients are born with thin or absent bone overlying the superior semicircular canal3,7 and that a “second event” (eg, skull base trauma, a Valsalva maneuver, intense acoustic exposure) causes an abrupt injury to the arcuate eminence. Another hypothesis is that dural pulsations over the arcuate eminence (located higher than the surrounding tegmen) result in progressive loss of bone over the superior canal. This latter hypothesis is supported by observations that the prevalence of SCD increases among older populations. In some patients, a combination of factors, such as thin bone over the superior canal, increased intracranial pressure, or a second event, may contribute to the development of SCDS. In the case of SPS-SCD, the close proximity between the superior petrosal sinus and the medial limb of the superior canal is a relationship likely present at birth.

There have been brief mentions in the literature of SCDS occurring in family members2,8; however, these studies did not discuss the role of genetics in the context of SCD etiology. There has been little reported concerning the possible genetics of SCD, but Hildebrand et al9 proposed that SCD may be present in other patients with DFNA9 mutations (DFNA9 mutations lead to progressive hearing loss and vestibular impairment). More recently, SCD was described in 10 children aged 5 to 11 years, which does support a congenital etiology in these cases.10

To better understand the characteristics of SCDS among family members, we performed a retrospective review of more than 200 patients with SCD treated at our institution and identified 3 sets of first-degree relatives. The following is a retrospective case report of these families.

Report of Cases

Case 1: Family 1—Two Brothers With Hearing Loss

A healthy man in his 40s (brother 1) presented in 2000 with progressive hearing loss. He had no vestibular signs or symptoms (Table). His mother and brother also had hearing loss, without official diagnosis. The Rinne test had positive results bilaterally, and in the Weber test, sound was lateralized to the right. Audiometric testing showed a bilateral conductive hearing loss (more severe on the right side than on the left side) with an air-bone gap (ABG) of 60 dB at 250 Hz (Figure 1A). No tests of acoustic reflexes or cervical vestibular evoked myogenic potential (cVEMP) or imaging were obtained prior to surgery. Because of suspicion of bilateral otosclerosis, the patient underwent right suspension of bilateral otosclerosis, the patient underwent right
stapedectomy in 2001. Postoperatively, pure-tone measurements showed no improvement in conductive hearing loss on the right side, and acoustic reflexes were absent bilaterally. Given the postoperative outcomes, CT imaging was performed and demonstrated bilateral SCD (Figure 2, A and B). Testing of cVEMP was also performed, showing low thresholds on the right and elevated thresholds on the left. The patient was managed conservatively, and amplification was offered. The patient has since been lost to follow-up.

The brother of the aforementioned patient presented in 2001 as a healthy man in his 50s (brother 2) with a history of bilateral progressive hearing loss, greater on the left side. He denied vertigo or disequilibrium. His otoscopic examination had normal results. The Rinne test had positive results bilaterally, and the Weber test showed sound at the midline. Audiometric testing demonstrated a low-frequency ABG of 40 dB at 250 Hz and bilateral mixed hearing loss, with greater loss on the left side (Figure 1B). Acuity for conversational speech was diminished bilaterally. No tests of acoustic reflexes or cVEMP or imaging were obtained. As for his brother, a presumed diagnosis of bilateral otosclerosis was made, and the patient underwent a left stapedectomy in 2001. Postoperatively, the audiogram showed no change in the air-bone gap, cVEMP thresholds were low, and follow-up imaging confirmed bilateral SCD (Figure 2, C and D). Amplification was offered to the patient. Unfortunately, he was lost to follow-up.

**Case 2: Family 2—Mother and Daughter With Auditory and Vestibular Symptoms**

A woman in her 70s presented with decreased hearing, autophony, aural fullness, and pressure-associated and sound-associated dizziness (Table). Family history was significant for paternal hearing loss. Audiometric testing showed bilateral mixed hearing loss with a bilateral ABG of 40 to 50 dB at 250 Hz (Figure 1C). Acoustic reflexes were present bilaterally, but tuning fork tests and cVEMP were not obtained. Dizziness was not evoked through tragal compression, pneumatic otoscopy, or Valsalva maneuver. Computed tomography confirmed the diagnosis of bilateral SCD and demonstrated a larger arcuate eminence defect on the left side (Figure 2, E and F). Management was conservative, but the patient was subsequently lost to follow-up.

A woman in her 40s, the daughter of the aforementioned patient, presented in 2010 with progressive hearing loss that had started 8 years prior. She had experienced vertigo in the past and had mild dizziness during migraine headaches (which she no longer experienced at time of presentation) (Table). The Rinne test had positive results bilaterally, and in the Weber test, sound lateralized to the left. Audiometric testing showed a 25-dB ABG at 250 Hz (Figure 1D), and acoustic reflexes were present bilaterally. Testing of cVEMP showed lowered thresholds on the left side (55, 55, 50, and 60 dB normal hearing level [NHL] at 250, 500, 750, and 1000 Hz, respectively) and non-responsive thresholds on the right (90 [no response], 100 [no response], 100, and 100 [no response] dB NHL at 250, 500, 750, and 1000 Hz, respectively). No dizziness or nystagmus were evoked through tragal compression, pneumatic otoscopy, or Valsalva maneuver. Temporal bone CT revealed left SCD and a thin but intact right superior canal (Figure 2, G and H). Management was conservative, but the patient has been subsequently lost to follow-up.

**Case 3: Family 3—Mother and Daughter With Auditory and Vestibular Symptoms**

A woman in her 50s presented in 2011 with a 5-year history of dizziness provoked by sound, pressure, and exercise. She also reported aural fullness, autophony, conductive hyperacusis, right-sided pulsatile tinnitus, and bilateral hearing loss (more severe on the right side than on the left side) (Table). A previously diagnosed type 1 Chiari malformation (CM-1) was not thought to be the cause of her symptoms. Her otoscopic examination had unremarkable results. The Rinne test had positive results bilaterally, and in the Weber test, sound was at the midline. Audiometric testing showed a mixed hearing loss in the right ear (she had previously lost high-tone hearing in the right ear). The left ear showed an ABG of 30 dB at 250, 500, and 1000 Hz and bone conduction thresholds above 0 dB (Figure 1E). Acoustic reflexes were not examined preoperatively, but cVEMP thresholds were low bilaterally (60-65 dB NHL). Pneumatic otoscopy and Valsalva maneuver caused nystagmus. Temporal bone CT showed bilateral SCD (Figure 2, I and J). The patient underwent middle fossa craniotomy on the right side with occlusion of her defect with bone wax.

One year after the initial operation, the patient underwent a second middle fossa craniotomy with occlusion, this

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**Table. Overview of Symptoms Reported for Each Relative**

<table>
<thead>
<tr>
<th>Symptom</th>
<th>Family 1 Brother 1</th>
<th>Family 1 Brother 2</th>
<th>Family 2 Mother</th>
<th>Family 2 Daughter</th>
<th>Family 3 Mother</th>
<th>Family 3 Daughter</th>
</tr>
</thead>
<tbody>
<tr>
<td>Side</td>
<td>Bilateral</td>
<td>Bilateral</td>
<td>Bilateral</td>
<td>Left</td>
<td>Bilateral</td>
<td>Left thin bone</td>
</tr>
<tr>
<td>Autophony/ Hyperacusis</td>
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<td>No</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
</tr>
<tr>
<td>Hearing Loss</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
</tr>
<tr>
<td>Dizziness</td>
<td>No</td>
<td>No</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>No</td>
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<tr>
<td>ABG at 250 Hz, dB</td>
<td>60</td>
<td>40</td>
<td>40-50</td>
<td>25</td>
<td>30</td>
<td>10</td>
</tr>
<tr>
<td>cVEMP, dB</td>
<td>Low*</td>
<td>Low*</td>
<td>NA</td>
<td>≤60</td>
<td>≤65</td>
<td>Normal</td>
</tr>
</tbody>
</table>

Abbreviations: ABG, air-bone gap; cVEMP, cervical vestibular evoked myogenic potential; NA, not available. *The cVEMP thresholds were reported as low, but specific values were not available.
time on the left side. Postoperatively, she noticed improvement in autophony, conductive hyperacusis, and dizziness, whereas hearing remained stable bilaterally. The cVEMP thresholds were also within normal limits bilaterally. She continues to be symptom-free 1 year after her second operation.

**Figure 1. Audiometric Testing**

A, Family 1, brother 1; B, family 1, brother 2; C, family 2, mother; D, family 2, daughter; E, family 3, mother; F, family 3, daughter. Despite similarities in symptom presentation, audiograms vary considerably between family members. Family 1 (A and B) has substantial air-bone gaps at all frequencies, but the magnitude of the air-bone gap differs. B also shows supranormal conduction (bone conduction of approximately −10 dB at 250 and 500 Hz), whereas A does not. Family 2 (C and D) has largely symmetric hearing loss in both relatives. However, C shows a moderate to severe mixed hearing loss, whereas D only shows a mild mixed hearing loss. Finally, family 3 (E and F) share little in common on audiogram. E shows air-bone gaps at all frequencies, whereas F does not show an air-bone gap. E also shows moderate hearing loss in the high frequencies (60 dB at 4 kHz and 65 dB at 8 kHz), whereas F shows normal hearing. AC indicates air conduction; ANSI, American National Standards Institute; BC, bone conduction; PTA, pure-tone average.
A woman in her 30s, the daughter of the aforementioned patient, presented with dizziness and headaches in 2011. The daughter’s symptoms started during pregnancy (1 year prior to presentation). She had symptoms similar to those of her mother, but they were less severe. The patient reported imbalance; dizziness provoked by sound, pressure, and exercise; aural fullness; hyperacusis; pulsatile tinnitus; and headaches. She reported no hearing loss (Table). The Rinne test had positive results bilaterally, and in the Weber test, sound was lateralized to the right. Audiometric testing showed a 10-dB ABG at 250 Hz on the right side (Figure 1F). Acoustic reflexes were not examined, but cVEMP testing showed normal thresholds bilaterally. The Rinne test had positive results bilaterally, and in the Weber test, sound was lateralized to the right. Audiometric testing showed a 10-dB ABG at 250 Hz on the right side (Figure 1F). 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dition with an increased prevalence in older populations. They
cesses.
In this case report, we present 3 families with symptomatic SCD among
first-degree relatives. Our findings suggest that genetics may play a role in some patients with SCD (in the absence of
head trauma or skull base fracture). Interestingly, first-degree relatives present with similar complaints: the 2 broth-
ers experienced only conductive hearing loss, whereas both mother and daughter sets experienced vestibular and audi-
tory symptoms. Furthermore, CT imaging of relatives showed similar skull base topography and anatomic SCD defects. It is
possible that these features are passed down genetically to off-
spring, which may explain the resemblance of symptoms in first-degree relatives. A larger cohort is needed to formalize a
radiologic classification system based on the anatomic charac-
teristics of the canal defect and the surrounding skull base topography.

Although similar symptoms were reported in families 2 and 3, both mothers presented with more severe symptoms than
described a patient who reported initial SCD symp-
toms in adulthood has been described before; Hegemann and Carey\textsuperscript{13} describe a patient who reported initial SCD symp-
toms at age 10 years with progressively worsening symptoms until surgical repair at age 40 years. They hypothesize that the
dehiscence began to transmit more pressure between the in-
er ear and intracranial space as the patient aged. Con-
versely, Nadgir et al\textsuperscript{14} speculate that SCD is an acquired condition with an increased prevalence in older populations. They
found no association between temporal bone thinning and aging and also no association of thinning with contralateral de-
hiscence (ie, thinning occurs independently of dehiscence de-
velopment). On the basis of the present case report, we agree with Hegemann and Carey\textsuperscript{13} and additionally hypothesize that an increase in bony defect size over time may correlate with
symptom progression. Nevertheless, more research is needed to further elucidate the pathophysiologic mechanism of such
processes.

Of note, the mother of family 3 had an established diag-
nosis of CM-1 before she was given the diagnosis of SCD. Kuhn and Clenney\textsuperscript{15} have shown that the prevalence of CM-1 is increased in patients with SCD compared with the prevalence of CM-1 in the general population, 23% vs 0.6% to 1%, respectively. The pathogenesis of CM-1 has been attributed to neuroectodermal developmental abnormalities and over-
crowing of the hindbrain by an underdeveloped posterior
fossa.\textsuperscript{16} As a result, the cerebellum obliterates the
cerebrospinal fluid space surrounding the cervicomedullary
junction, and exaggerated cerebrospinal fluid pressure waves may have a cumulative erosive effect on the surrounding
bone. Because neuro-otologic symptoms tend not to occur until adulthood, this slow, bony erosive process may affect a preexisting developmental bony abnormality, thus leading to the
development of SCD in patients with CM-1. However, the
authors suggest that it may be premature to have all patients with SCD undergo magnetic resonance imaging to detect
underlying CM-1.

Finally, the daughter of family 3 had very thin bone over the
superior canal on the left side (Figure 2L) rather than a frank
dehiscence. Nevertheless, she did present with auditory and vestibular symptoms suggestive of SCD. Vertigo can be seen
in 1 of 10 patients with thinning of the superior canal.\textsuperscript{7} In ad-
dition, patients with a “near-dehiscence” and signs and symp-
toms of SCD have recently been described.\textsuperscript{12} The authors sur-
gically managed these patients by plugging and/or resurfacing
the superior canal, and improvements in SCD signs and symp-
toms were reported. Additional studies are needed to deter-
mine the benefit of surgery in this unique group of patients with
SCD, as well as an objective, validated methodology for defin-
ing “thin” bone.

Conclusions

Our observations suggest that (1) SCD signs and symptoms in
members of the same family were similar, (2) skull base topography relative to the bony defect of the superior canal
was similar among first-degree relatives with SCD, and (3) symptoms seemed to be more pronounced in older
patients (mothers) compared with their younger counterparts (daughters). Although SCD etiology is still debated, a
genetic basis seems plausible. Additional genetic and cohort studies are needed to examine potential contributions for
this condition.
Conflict of Interest Disclosures: None reported.

REFERENCES


