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# PREFACE

*Hearing Disorders Handbook* (HDH) provides descriptions of hearing and vestibular disorders, their frequency of occurrence, etiology, diagnosis, and management. It seeks the perspectives of the diverse disciplines that make up the typical hearing rehabilitation team—disciplines that include audiology, otology, speech-language pathology, and the related fields of education, genetics, pediatrics, and psychology.

HDH does not replace traditional textbooks, lectures, or practicums. While striving to be comprehensive, HDH organizes its contents—whether of a particular disorder, symptom, or diagnostic and management procedure—comprehensively and concisely. This approach fulfills the original intent of handbooks: *to contain information in a volume small enough to be held in the hand.*

The concepts of comprehensiveness and conciseness grow more timely as the flood of information continues to increase.<sup>1</sup> Because so much is written and continues to emerge, both experts and novices need the means of quickly and accurately winnowing the wheat from the chaff. In so doing, HDH guides readers through the bewildering amount of material about hearing and vestibular disorders.

## ORGANIZATION OF HDH

HDH divides into three main sections:

## **Section 1. Introduction to Hearing and Vestibular Disorders**

The first three subsections divide entries for hearing disorders by their causes: 1.1. Endogenous Etiology, 1.2. Exogenous Etiology, and 1.3. Multiple Etiology. Discussion of each disorder takes up its frequency of occurrence, etiology, diagnosis, and management.

The fourth subsection (1.4) concerns vestibular disorders, which organizes its entries by symptoms. The discussions of the cardinal symptoms replicate the order for hearing disorders; that is, frequency of occurrence, etiology, diagnosis, and management.

Any question about including a section on vestibular disorders in HDH should be allayed by considerations of the nature of the disorders and their interrelatedness. Factors affecting one system often impinge on the functioning of the other; for example, in patients with suspected cases of acoustic neuroma and those with Meniere's syndrome. Because many hearing disorders are also accompanied by vestibular symptoms, clinicians need information about both.<sup>2</sup>

## **Section 2, Management Options**

The sections augment specific discussions of management within each disorder and symptom. They present management options that apply generally to treatment of hearing and vestibular disorders. Its subsections are: 2.1, Amplification, 2.2, Cochlear Implants, and 2.3, Strategies to Compensate for Hearing Loss.

## **Section 3, Demographic, Social, and Economic Aspects**

This section addresses the principal consequences of hearing and vestibular disorders. It aims to place these disorders in their rightful position among diseases that most pervasively and sig-

nificantly affect health and quality of life. It assesses the social-economic toll that hearing and vestibular disorders levy on affected individuals and their families, highlighting information and relationships that can be useful in researching and planning.

## INFORMATION RESOURCES

For readers seeking more information about a particular topic or disorder, HDH provides references to original print and electronic sources. These supplement the material condensed to fulfill the objective of concision.

Recently, journals have appeared exclusively on the Internet. One such is Public Library of Science (PLOS Med), a nonprofit organization that supports an open-access, peer-reviewed source of articles related to medicine.<sup>3</sup> PLOS Med appears monthly. Being new, its value remains to be assessed by the professionals who access it.

For descriptions of extremely rare conditions that are not covered in HDH and for the most recent information about those that are represented, readers should consult the Internet. The National Center on Low Incidence Disabilities<sup>4</sup> and Online Mendelian Inheritance in Man<sup>5</sup> are particularly useful sources of current information. In addition, the National Institute of Deafness and Other Communicative Disorders<sup>6</sup> periodically posts notices about disorders associated with hearing loss.

Organizations specific to particular disorders are mentioned within each entry. Among organizations that address hearing and vestibular disorders broadly are the following: Association of Late Deafened Adults,<sup>7</sup> Hearing Loss Association of America,<sup>8</sup> League for the Hard of Hearing,<sup>9</sup> and National Association of the Deaf.

## EDITING NOTES

In multiauthored articles, only the first three authors are noted. If there are more than three authors, the abbreviation et al. (“and others”) follows the third author’s name. As the purpose

of the citation is to enable readers to locate a reference, this limitation should not prove a hindrance to bibliographic searches.

Terminology respects the sources cited. So, for example, the terms deaf appears in the discussion when that is the designation chosen by the referenced source. Otherwise, HDH writes hearing loss or the even broader hearing impairment as appropriate to a discussion's context.

## CAUTIONARY NOTICE

The information contained in HDH does not stand in lieu of competent professional counsel. No individual should rely on HDH to determine any medical or related professional treatment. HDH makes no claims for infallibility, though it strives to ensure that all of its contents reflect accurately the state of knowledge about hearing disorders.

Aside from organizing references for easy access, HDH is mindful that the plethora of research publications contain many that are based on weak methodology and, indeed, may present fraudulent conclusions.<sup>10</sup> With respect to such pitfalls, HDH cannot guarantee that all materials to which it directs readers are valid. Such an objective would be beyond its scope. HDH does, however, avoid citing obviously suspect publications. To that end, HDH should provide useful access to the vast literature on hearing and vestibular disorders and stimulate its appreciation.

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## Section 1.3

# *Multiple Etiology*

### **APLASIAS AND DYSPLASIAS**

Aplasia refers to lack of development and dysplasia to abnormality of development. The two conditions occur in a number of syndromes that include hearing loss. In favor of conciseness, major examples of these anomalies are gathered in this Section. However, additional variations not specifically mentioned here probably exist.

In Alexander aplasia (AA), cochlear-duct differentiation at the level of the basal coil is limited, which affects development of the organ of Corti and the ganglion cells.

Alpert syndrome (APT) is characterized by abnormal facial appearance, spina bifida, and conductive hearing loss. The name alone may cause it to be confused with Alport syndrome (see Section 1.1) with which it shares no other features except hearing loss.<sup>1</sup>

Mondini dysplasia (MonD) is also called Mondini malformations. It is characterized by hearing disturbances and cataract, branchial cleft fistulae, and preauricular pits. It is closely related to several other syndromes; for example, Branchio-Oto-Renal and Kabuki (Niikawa-Kuroki), as well as with the others listed here.

Michel aplasia and dysplasia (MichelA/D) refer to the lack of differentiated inner-ear structures. In one form, it comprises the complete agenesis of the petrous portion of the temporal bone, with the auditory nerve and inner-ear structures absent.

In that case, the external and middle ear are unaffected. The condition is named for Pierre Michel.<sup>2</sup>

Scheibe aplasia (SA) involves agenesis of the inner ear. Its more descriptive name is Cochlear-saccular dysplasia or Pars Inferior dysplasia.

### **Frequency of Occurrence**

The aplasias and dysplasias are relatively rare conditions, each of them probably occurring in fewer than 1 in 10,000 live births. Estimating the incidences of these conditions is difficult, because similar malformations have been identified in other disorders, like Pendred, Waardenburg, Treacher-Collins, and Wildervank syndromes. For that reason alone, more precise incidence rates for these conditions have dubious reliability unless the overlapping of symptoms are given careful account.

SA is thought to be the most common form of inner-ear aplasia.

### **Etiology**

The cause(s) of aplasias and dysplasias—aside from their being due to genetic defects—remain in doubt. The differentiation between aplasias and dysplasias probably depends on the time in development of the inner-ear structures when the mutation occurred.<sup>3</sup>

AA may be a phenotype rather than a genotype.

APT is assumed to be autosomal dominant.

MonD is thought to be congenital and probably X-linked.<sup>4</sup>

The vestibule and semicircular canals may develop normally, but perilymphatic fistulae commonly occur. The cochlea is deformed or missing, and the endolymphatic duct tends to be atypically large. These deformities may be unilateral or bilateral.

In MichelA/D, the defects are thought to result from an insult prior to the end of the third gestational week. Genetic studies of mouse models suggest that a number of different

genes may cause these anomalies. Autosomal dominant inheritance has been hypothesized, but recessive inheritance is also regarded as likely in some cases.

SA is usually inherited as an autosomal recessive, nonsyndromic trait. Although the bony labyrinth and the superior portion of the membranous labyrinth are usually differentiated in patients with SA, the organ of Corti, the tectorial membrane, and Reissner's membrane tend to be deformed. Some infants with congenital rubella also display these deformities.

## Diagnosis

The hearing losses in these conditions may be unilateral. Audiologic testing followed by radiologic and MRI procedures usually confirm the inner-ear defects.

In MonD, the hearing loss is in the low to middle frequencies. Hearing at 8000 Hz and above usually remains intact. When ultrahigh-frequency hearing is not checked, MonD's characteristic audiometric configuration is often missed.

In MichelA/D, the hearing loss that accompanies this condition is usually moderate. Hearing loss is also a major component of Kabuki syndrome, in which it results in deafness due to the lack of sensorineural structures.

## Management

Audiologic testing is key for diagnosis of the aplasias and dysplasias, because counteracting the hearing loss is doable. Amplification with hearing aids and cochlear and brainstem implants should be considered.

For patients with AA who have a high-frequency hearing loss, their intact low-frequency hearing may justify use of conventional amplification or a hybrid cochlear implant.

Recognizing MonD can lead to treatment of a perilymphatic fistula that significantly helps prevent meningitis.

Conventional amplification and cochlear implants do not aid patients with MichelA/D, but vibrotactile devices, and implants

directly into the auditory portion of the brains may provide assistance.

As in all aplasias and dysplasias, early application of treatment for a hearing loss has a high probability of avoiding speech and language delays.<sup>5</sup>

## **AUDITORY PROCESSING DISORDER (APD)**

APD refers to deficits in the perceptual processing of auditory stimuli and in the neurobiologic activity underlying these processes.<sup>6</sup> It may be further designated as (Central) Auditory Processing Disorder (CAPD).

### **Frequency of Occurrence**

When estimates of APD's incidence and prevalence are available, they will appear on the Web site of the National Institute of Deafness and Other Communicative Disorders.<sup>7</sup>

### **Etiology**

What causes APD is unknown. It may be due to a number of factors acting alone or in combination. In children, it has been associated with dyslexia, attention deficit disorder, autism spectrum disorder, specific language impairment, and pervasive developmental disorder or delay.<sup>8</sup>

### **Diagnosis**

A battery of behavioral and electrophysiologic tests have been developed.<sup>9</sup> A team approach to school-age children—audiologist, neuropsychologist, otologist, special educator, speech-language pathologist—should also involve the child's parents and teachers. In arriving at a diagnosis, the team will rule out other similarly appearing problems—especially attention deficit and hyperactivity disorders, which are often confused with APD.<sup>10-12</sup>

## Management

A variety of approaches may be tried to counteract the effects of APD. In classrooms, the teacher may wear a microphone that transmits her speech to the child wearing a headset, thereby reducing the effects of competing auditory stimuli. Adjusting room acoustics and the child's seating may also improve listening conditions.

Lessons that improve language skills and enhance auditory memory have been tried, and Auditory Integration Training (AIT) has also been offered. As yet, however, sufficient research does not support any of these techniques,<sup>13</sup> and AIT has provoked a technical report and a policy statement from the American Speech-Language-Hearing Association.<sup>14,15</sup>

## AUDITORY NEUROPATHY (AN)

AN is a recently adopted term also called Auditory Dys-synchrony. Either term refers to normal otoacoustic emissions, absent or abnormal auditory brainstem responses, absent acoustic reflexes, and poor speech discrimination.<sup>16</sup>

## Frequency of Occurrence

AN is a relatively rare condition, estimated to occur at a rate of about 1 or fewer per 10,000 population.<sup>17</sup> However, the relatively new awareness of this condition suggests cases will be identified at an increased rate.<sup>18</sup>

## Etiology

AN results from a combination of disturbances of the CNS from the axon terminal of the inner hair cells to the auditory brainstem.<sup>19</sup>

The hearing loss tends to be mild at first and slowly progressive to severe and even profound levels.<sup>20</sup>

## Diagnosis

An absent or abnormal auditory-brainstem response and normally functioning outer hair cells suggests a diagnosis of AN. Additionally, middle-ear muscle response is characteristically absent, and word recognition in quiet is out of proportion to pure-tone sensitivity. The diagnosis is further bolstered by absence of auditory-nerve action potential and may be confirmed by MRI.<sup>21</sup>

## Management

First attention must be given to dealing with the cause(s). The clinician can then consider management of the residual hearing capacity.

For children with AN, conventional hearing aids give mixed results. Some success has been reported with devices that improve the signal-to-noise ratio, like FM systems. Cochlear implants also give mixed, though generally favorable, results.<sup>22,23</sup>

## AUTOIMMUNE INNER-EAR DISEASE

Autoimmune inner-ear disease (AIED) arises from inflammation of the inner ear caused by the body's immune system attacking cells in the inner ear. It is a relatively new clinical entity, first identified in 1979.<sup>24</sup>

## Frequency of Occurrence

The National Institute of Allergy and Infectious Diseases (NIAID) estimates AIED accounts for less than 1% of all cases of hearing loss and vertigo.<sup>25</sup> However, prevalence estimates for this condition may be sizable underestimates, as some cases of bilateral Meniere's disease—which NIAD categorizes as one of the autoimmune diseases—and some portion, at least, of idiopathic sudden sensorineural hearing losses and possibly some other conditions should be included.

## **Etiology**

AIED is a fluctuating cochlear disorder whose potential for auditory recovery is unpredictable.<sup>26</sup> Contrary to the belief that the inner ear cannot be attacked by its own immune system, it now appears that the perisacular tissue surrounding the endolymphatic sac can provoke an autoimmune reaction as if attacked by a virus. There may also be causal factors as yet unidentified.

AIED is a poorly understood syndrome. Persons with this condition present with sudden, idiopathic, rapidly progressive sensorineural hearing losses that may be unilateral or bilateral. They may fluctuate over a period of weeks or months, and vestibular symptoms are present in approximately half of the patients.

Research on the etiology of this condition is hampered by its low incidence, its episodic nature, failures to recognize it, and the recentness of its discovery. Furthermore, persons with AIED may have more than one type of autoimmune disease or genetic defect. As noted above, some percentage of patients diagnosed with Meniere's disease, especially those with bilateral symptoms, may instead have AIED. The diagnosis is important to treatment, as patients with AIED tend to respond well to corticosteroid therapy.

## **Diagnosis**

Diagnoses are based on the patient's history, physical examination, audiologic and vestibular examinations, and blood tests. Western Blot Immunoassay for Heat Shock Protein-70 antibodies identifies rapidly progressive sensorineural hearing loss in a sizable number of cases.<sup>27</sup>

## **Management**

When diagnosis is doubtful, a medical regimen for AIED is recommended because the condition may be reversible and

spontaneous recovery is possible.<sup>16</sup> It is usually treated initially with a variety of medications, among which steroids constitute the most favored medical response that gives relief to some patients with AIED. If the response to steroids is favorable, a cytotoxic chemotherapy, like cytoxan or methotrexate, may be used on a long-term basis.<sup>28,29</sup>

Some patients (e.g., diabetics) cannot be treated with steroids. Hyperbaric oxygen therapy is then an option, though results appear inconsistent.<sup>30,31</sup>

For patients who do not respond to drug and other medical therapy and whose hearing loss persists, amplification is recommended. Fully digital hearing aids should be considered because they can be reprogrammed as the patient's hearing loss changes.

If the hearing loss becomes profound, a cochlear implant may be prescribed. However, AIED's fluctuating nature requires that clinicians take a conservative approach to recommending cochlear implants, as the potential exists for recovery from or lessening of the hearing loss. If clinicians prescribe cochlear implants, they should advise patients with the possibility that the device will prevent the return of normal hearing.<sup>32</sup> The National Institute on Deafness and Other Communicative Disorders and the American Academy of Otolaryngology-Head and Neck Surgery Foundation cosponsor a multicenter clinical trial named Otolaryngology Clinical Trial Cooperative Group to uncover the cause of AIED and to evaluate drug therapies. As with the study of its etiology, the rarity of AIED makes research on its treatment difficult but worthwhile, as it may lead to better understanding of other sensorineural hearing losses.

## **BRANCHIO-OTO-RENAL SYNDROME (BOR)**

BOR is distinguished by branchial clefts, fistulas, cysts, and malformation of the pinnae and preauricular pits of the sinuses. Hearing impairments and kidney dysfunctions are common.

## Frequency of Occurrence

BOR probably occurs in about 1 per 40,000 live births.

## Etiology

The heritability of this congenital syndrome remains undetermined. It probably is caused by mutations within a genomic interval of 156 kB.

## Diagnosis

Three-fourths of BOR patients are estimated to have a hearing loss: about a third are conductive, a fifth are sensorineural, and the remainder mixed.

## Management

Providing appropriate amplification for the hearing loss early is the most rewarding measure in the rehabilitation of this condition. As with all syndromes, attention should be directed to the accompanying disabilities, especially renal failures that can be life-threatening. Genetic counseling should be made available to affected persons and their families.

## CHARGE ASSOCIATION

CHARGE Association (CA) is a syndrome whose acronym indicates its characteristics: *Coloboma, Heart defects, Atresia of the choanae, Retarded growth, Genital hypoplasia, and Ear anomalies.*

## Frequency of Occurrence

CA is estimated to occur in 1 in 12,000 live births.<sup>33</sup> Mild to profound sensorineural hearing losses occur in about 8 of 10 CA cases.<sup>34</sup> When 4 of the 6 characteristics appear together, diagnosis is confirmed.

## **Etiology**

The initials of the syndrome indicate the conditions involved in this syndrome. The ear anomalies may include hypoplasia of the external ear, hearing loss, a Mondini-type deformity, and absence of semicircular canals.<sup>35</sup> In addition to these symptoms, CA patients may have vestibular dysfunctions.<sup>36</sup>

CA occurs in both genders, all races, and all socioeconomic groups. The nature of inheritance—if it is inherited rather than arising from damage in utero—has not been established. The tiny incidence of the condition makes aggregating sufficient cases for genetic research difficult, amplifying the problems in determining its cause.<sup>37</sup>

## **Diagnosis**

The mix of disabilities in CA complicates audiologic evaluation. With four or more of the symptoms present at birth, the diagnosis of CA is usually made. Among other conditions with which CA may be confused is Usher syndrome (deafblindness).

## **Management**

Some of CA's structural anomalies may be amenable to surgical correction. Other aspects require medical care to maintain the patient's viability and to prevent deterioration of affected functions.<sup>38</sup>

In all cases, providing early amplification for the expected hearing loss is a critical step. It should be undertaken as early as feasible.<sup>39</sup>

## **CONNEXIN 26 AND 30**

Different mutations in the same gene can be associated with both dominant and recessive hearing loss. Connexins are membrane proteins that are primarily involved in intercellular com-

munication. Connexin 26 (Cx26) and 30 (Cx30) cause the most common genetic forms of nonsyndromic hearing loss.<sup>40</sup>

## Frequency of Occurrence

The Cx26 and Cx30 mutations are heterozygous, and they are associated with several syndromes—for example, Keratitis-Ichthyosis-Deafness syndrome—which means that a single incidence-rate estimate for Cx-26 and Cx-30 defects would be misleading. They are involved in a number of syndromes.<sup>41</sup> Also, different national rates for their incidence have been reported for mutations in the GJB2 gene, which contains instructions for Cx26—mutations occurring most frequently in Caucasians, Ashkenazic Jews, and some Asian groups.<sup>42-44</sup>

## Etiology

Cx26 and Cx30 work together to form the cochlea's gap junctions and facilitate intercellular communication.<sup>45</sup> When they function properly, they encode gap-junction channels that connect adjacent cells and allow passage of cytoplasmic ions and small molecules.<sup>46,47</sup> More than 20 connexin subtypes have been identified in humans. Some connexin mutations induce hearing loss and may be responsible for a large amount of nonsyndromic congenital hearing loss in children.<sup>48</sup>

Nonsyndromic cases with hearing loss are not free of other disorders. But as other disorders are not due to the gene causing the hearing loss, they do not appear to be a part of a syndrome. Thirty-eight loci for dominant, nonsyndromic deafness have been mapped and 11 genes have been identified.

The recessive nature of these connexin disorders—when they are recessive—impairs permeability of cochlear structures.<sup>49</sup> Exogenous factors influence some cases.<sup>50</sup>

## Diagnosis

Genetic testing provides the basis for distinguishing aspects of this syndrome.<sup>51</sup>

## Management

As more molecular mechanisms are discovered for the effects of deafness-linked Cx26 mutations, the distinctions may assist clinicians to tailor therapeutic strategies to achieve best outcomes.

When Cx30 is missing, research with mice suggests that hair-cell depletion may be overcome and hearing restored by adding extra Cx26. Supplying a drug to boost production of Cx26 protein might provide similar results in the human instance. Although the majority of these genetic mutations cause recessive hearing loss, a few have been found to be dominant, in which case the hearing loss may be of any degree, ranging from moderate to profound. Such phenomena illustrate the nascent state of genetic research.

Research suggests a rationale for future therapeutic strategies to rescue cell death by G45E mutation by altering the  $[Ca^{2+}]$  and/or binding affinity within the cochlea. Identifying more molecular mechanisms for the effects of deafness-linked Cx26 mutations may assist therapeutic strategies that achieve maximum clinical outcomes based on Cx26 mutations.<sup>52</sup>

## DOWN SYNDROME AND THE TRISOMY ABERRATIONS

Down syndrome (DS) is marked by developmental delays, characteristic features of the face, head, neck, and hands, and a mild congenital hearing loss.<sup>53</sup>

DS is named for J. L. H. Down, the physician who first described it.<sup>54</sup> DS is also known as Trisomy 21, the best known of the chromosome disorders. Trisomy 13 (Edwards' syndrome) and Trisomy 18 (Patau syndrome) are two related, but much less frequent variants of DS.

## Frequency of Occurrence

Estimates of the overall incidence of DS hover around 1 per 800 live births. This rate is affected by the age of the mother at

conception: before 35 years of maternal age, the estimated incidence rate is about 1 per 1,000; from 35 to 40 years it rises to about 1 per 385; from 40 to 45 years, it reaches 1 per 106; and from 45 years of age and older it balloons to 1 per 30 live births.

The most common form of DS—about 90 to 95%—is Trisomy 21. The remaining 5 to 10% divide between the chromosomal anomalies Mosaicism and translocation.

The incidence of Patau is estimated to be about 1 per 7,600 and Edwards' to be slightly higher at an estimated 1 per 7,500 live births.<sup>55</sup>

## **Etiology**

DS results from extra genetic material. Instead of inheriting 23 chromosomes pairs from each parent, the neonate with DS has an extra chromosome (the “tri” in trisomy), which affects cell division and CNS development in utero.<sup>56</sup>

The hearing loss is frequently conductive and is associated with anomalies of the external ear and atresia of the auditory canals. Intellectual slowing commonly occurs.

## **Diagnosis**

Diagnosis of DS can be made in utero by ultrasound. DS is readily identified in newborns by noting its prominent physical characteristics.<sup>57</sup>

A variety of malformations of the ossicular chain, outer ear, middle ear, and nasopharynx, often with concomitant conductive hearing loss, characterize patients with DS.<sup>58</sup> They tend to have pinnae that are smaller and lower set than usual.<sup>59</sup> Ear canals are often stenotic, and malformations of the middle-ear structures have been reported.<sup>60</sup> DS also features excessive cerumen.<sup>61</sup>

Inner-ear malformations that involve both the cochlear and the vestibular portions do occur, but are infrequent.<sup>62</sup> However, when early onset sensorineural hearing loss similar to presbycusis occurs, it is considered significant and illustrative

of DS. Its occurrence suggests that patients with DS exhibit accelerated aging, not only of the auditory systems, but also of the visual system and cognitive functions as well.<sup>63,64</sup>

## Management

Periodic monitoring is essential to patients with DS to detect and treat ear infections and to adjust any treatment for hearing losses they may have. Their tendency to conductive hearing losses is due to the narrowed eustachian tube and ear anomalies that characterize babies with DS and make them unusually susceptible to otitis media and serous otitis media. To avoid acquired hearing losses, management steps are the same as those for otitis media (see Section 1.2).<sup>65,66</sup>

The March of Dimes maintains a list of support groups on its Web site.<sup>67</sup>

## IDIOPATHIC SUDDEN SENSORINEURAL HEARING LOSS

Idiopathic, sudden sensorineural hearing loss (ISSNHL) ranks high among the difficult clinical conundrums. Patients with ISSNHL usually present with demands for relief, and much of the literature urges prompt treatment.

### Frequency of Occurrence

The incidence of ISSNHL is about 1 per 5,000 annually. Studies suggest that between 5 and 25 per 100,000 persons in the United States will suffer ISSNHL in any year.<sup>68</sup>

ISSNHL is predominantly unilateral. In about 7 in 10 cases, it is accompanied by tinnitus. An estimated 5 in 10 patients also suffer vertiginous attacks.<sup>69</sup>

Its incidence is far greater in adults than among children, with average age at onset between 40 to 50 years, and the majority of ISSNHL cases occurring in persons over 40 years old.<sup>70</sup>

Although the estimated incidence rates are relatively small, the number with ISSNHL amounts to about 65,000 persons annually in the United States. These estimates are for *reported cases*, which probably underestimate the actual rate, because many people do not seek medical assistance when the hearing loss is mild and their cases may not be brought to the attention of professionals when hearing recovers spontaneously after an attack.

## Etiology

The cause or causes of ISSNHL usually remain idiopathic despite assiduous efforts to identify etiology. Conductive losses from sharp changes in atmospheric pressure (aerotitis media), buildup of obstructive cerumen, ossicular-chain disruption, and swimmer's ear—all of which are usually self-limited, easily repaired, quickly reversed, and readily identified; thus, they are neither idiopathic nor sensorineural.

Exposure to a brief loud noise can cause a temporary threshold shift that is sensorineural (see NIHL in Section 1.2). Like conductive losses, such hearing losses are usually temporary, but with further noise exposure may result in permanent sensorineural hearing loss that a reasonably careful history correctly diagnoses. Nonetheless, their etiology can often be readily identified, so they are not idiopathic.

## Diagnosis

When a loss is sudden and sensorineural, its causation may be difficult or impossible to establish definitively, hence the *idiopathic* in ISSNHL. The published research emphasizes the difficulties in establishing an accurate diagnosis and prognosis for sudden hearing loss in about one of three cases.<sup>71</sup>

In addition to thorough audiologic and otologic examinations, other aspects of the patients' health should be examined and potential sources of hearing loss investigated; for example, brain scans and blood analyses. If these tests uncover a cause,

the ISSNHL no longer is idiopathic, so treatment indicated by the new diagnosis shifts to management of that cause.

When the patient's hearing measures 70 dB HL or better, the remission possibilities are high.<sup>72</sup> Similarly rising and mid-frequency audiometric curves predict spontaneous recovery more frequently than sloping or flat configurations.<sup>73</sup>

Although most cases of ISSNHL are unilateral, bilateral cases have been reported.<sup>74,75</sup> When sudden deafness is bilateral, specific causes often have been diagnosed, typically instances of systemic disease.<sup>76</sup> A retrospective analysis of 324 cases found 16 patients (4.9%) with bilateral losses, of whom 6 were diagnosed with diabetes mellitus, 7 with hypertension or cardiovascular disease, and 3 remained idiopathic.<sup>77</sup>

## **Management**

Spontaneous recovery complicates rehabilitation planning in the interval between onset of the condition and beginning remediation. To be accepted as salutary, treatment must succeed better than the rates for spontaneous recovery, which occur in about one-third of cases. The proportion of cases enjoying spontaneous recovery declines after 30 days, with little hope for regaining hearing spontaneously after 6 months.<sup>78</sup>

Results of an 8-year prospective study of 225 patients concluded that the longer treatment is delayed, the poorer the recovery.<sup>79</sup> However, to date, no single treatment for ISSNHL has achieved success greater than that for spontaneous recovery.

## ***Medical and Surgical Treatment***

In a study of short-term oral steroid treatment, hearing in the treated group improved by 29 dB HL compared to 11 dB HL for untreated cases. However, the expected rate of spontaneous recovery was unchanged.<sup>80</sup>

Magnesium added to steroids, but not antiviral drugs, were found to increase the rate of recovery somewhat.<sup>81</sup> A double-blind study that added vitamin E to the magnesium, steroid,

and carbogen-inhalation therapy reported the combination significantly improved hearing for a majority of the patients.<sup>82</sup> Despite success of steroid therapies, alone or in combinations with other chemicals, questions about their use remain.<sup>83</sup>

### ***Amplification***

Once medical or surgical treatments are deemed inappropriate, the patient should be offered amplification and auditory training. When the normal or better-hearing ear of an individual with unilateral loss is exposed to environmental noise, the ability to communicate is seriously reduced; in effect, under such conditions, the person is functioning with a significant bilateral hearing loss.

The time to amplify may depend on patient resistance and economics. The odds favoring spontaneous recovery justify a delay of 60 to 90 days, but a longer wait would be inadvisable in view of the handicap imposed and its isolating effects on social and occupational interactions.

Patients may resist suggestions to try a hearing aid, if it means to them they will not recover their hearing. Most patients will accept a hearing aid when told that its gain can be adjusted as hearing improves and its use discontinued if hearing returns to normal.

The style of hearing aid selected for patients must be consistent with their ability to manage the components easily and comfortably. Very small, completely in-the-canal instruments that use tiny batteries may not be appropriate for elderly persons with limited dexterity and poor vision. For them, a full-shell in-the-ear or an open-mold behind-the-ear hearing aid may be preferred.

Should the affected ear receive the aid? Or should the choice be among contralateral-routing-of-offside-signals aid (CROS), bone-anchored hearing aid (BAHA), and FM systems?<sup>84</sup> The indications and limitations of each system should be presented to the patient. If the affected ear has usable residual hearing, the audiologist should consider a hearing aid for that

ear. If the affected ear has a profound hearing loss and word-recognition scores poorer than 20%, a CROS, transcranial CROS, or BAHA may offer a better solution.

In a CROS fitting, a microphone on the side of the affected ear electronically routes sounds originating on that side to an amplifier and receiver mounted near the normal or better-hearing ear, thus directing the sounds into the normal or better-hearing ear by tubing or a nonoccluding earmold that extends into the open ear canal. The object is to pick up sounds on the side of the affected ear and route them to the good ear, thus overcoming the head-shadow effect.

Some patients have reported mixed satisfaction with CROS amplification, although the majority of responses have been negative. The originator of the transcranial version reported a success rate of 25% in patients with severe to profound unilateral hearing loss.<sup>85</sup>

Personal FM systems that provide an improved signal-to-noise ratio in difficult listening environments are alternatives or additions to CROS aids. The transcranial CROS may be another option for persons with ISSNHL who have sustained severe-to-profound unilateral losses but have excellent hearing in the better-hearing ear. If the better-hearing ear has only a mild high-frequency hearing loss, conventional CROS aids may be recommended.<sup>86</sup>

The U.S. Food and Drug Administration has approved the BAHA for persons with single-sided deafness. BAHA is a cochlear stimulator that transmits auditory stimuli via bone conduction to the contralateral cochlea. This requires the surgical implantation of a 4-mm titanium fixture behind the affected ear. A multi-institutional study has shown greater patient satisfaction and improved communication with a BAHA than with a CROS.<sup>87</sup>

### ***Counseling***

ISSNHL patients need counseling. They should have full explanations of the condition, its potential for spontaneous recovery and, if the loss persists, the treatment options.

The clinician should discuss fully and empathetically patients' desires to recover lost hearing. If told by one expert that hearing cannot be restored, many patients will shop for other more optimistic opinions. That is why clinicians should advise patients that, once the permanence of the hearing loss becomes likely, rehabilitation becomes an option and should counter any fears that rehabilitation may prevent later recovery of hearing.<sup>88</sup>

### ***Monitoring***

Periodic audiologic examinations should not be overlooked. Partial recovery can occur in small steps that might be missed by patients. Further changes in hearing, both in the originally affected and the contralateral ear, can be useful in determining treatments formerly ignored and to identify possible causes originally rejected. For those reasons, serial pure tone audiometry and word-recognition testing appear wise.

## **KERATITIS-ICHTHYOSIS-DEAFNESS SYNDROME (KID)**

KID comprises ocular defects, including progressive blindness due to inflammation of the cornea (keratitis), and congenital hearing loss. Various other manifestations have been reported in association with KID, depending on the type of inheritance and degree of penetrance.

### **Frequency of Occurrence**

KID is a rare condition. No significant gender, racial, or ethnic influences have been reported.

### **Etiology**

KID appears to cover two modes of inheritance—one autosomal dominant and the other recessive—and one in which the

genetic mutation leading to this cluster of conditions appears to be spontaneous, as other similarly affected relatives are not found in several of the reported cases.

## **Diagnosis**

The dominant characteristics, keratitis and deafness, present no difficulty in diagnosing. There are additional ectodermal defects in most cases—like dry, scaly skin—defects that are evident.

## **Management**

The hearing loss requires prompt attention, relying on amplification and other strategies to overcome potential impediments to speech, language, and social development.

## **MENIERE'S DISEASE**

Meniere's disease (MenDis) consists of a combination of hearing loss, vertigo, and tinnitus. It may also be accompanied by a sensation of fullness in the affected ear. It is named for the physician who first recognized it in 1861.

## **Frequency of Occurrence**

The prevalence of MenDis has been estimated to be the third most common inner-ear disorder after presbycusis and noise-induced hearing loss. Its occurrence has been variously estimated to be between 50 and 218 per 100,000.<sup>89-91</sup>

In about 70 to 80%, the hearing loss initially occurs unilaterally. Involvement of the unaffected ear—reaching about 40% after 15 years—often occurs as the disease progresses.<sup>92</sup> A higher prevalence of MenDis occurs in populations screened for migraine.<sup>93</sup>

## Etiology

MenDis has been attributed to many etiologies—food allergies, endocrine insufficiencies, vascular disease, syphilis, viral infection, and genetic factors—but none has gained consensus. It is now considered a disease of the membranous inner ear, attributable either to excessive endolymphatic fluid, causing Reissner's membrane to become distended, or to an injury of the fluid-absorption system. It has been postulated that Reissner's membrane may perforate during the acute attack and reattach itself between attacks.<sup>94</sup>

Because of the excess production or poor absorption of fluid, MenDis is often called endolymphatic hydrops, the assumed histopathologic hallmark of MenDis. However, Danish scientists have made an intriguing discovery (as yet unconfirmed) that the endolymphatic sac produces saccia, a hormone that plays a role in regulating the sodium level in the bloodstream.<sup>95</sup> This finding has led to the possibility of blocking the hormone's release with medication.<sup>96</sup>

## Diagnosis

The characteristic audiometric configuration in the early stages of MenDis is a rising curve. As the frequency increases, the hearing loss decreases. This contour has also been called a "reverse slope," as high-frequency hearing losses are by far the more frequent among persons suffering hearing loss. A much lower than expected word-recognition score often accompanies the pure-tone loss. High-frequency sensitivity tends to be normal in the early stages, but more lower and middle frequencies are involved as the disease progresses, leaving the patient with a "flat" or sloping audiometric configuration.

In MenDis, the hearing loss may vary from mild to severe. As MenDis progresses, the hearing loss usually becomes severe, ranging from 70 to 90 dB, and is accompanied by a roaring tinnitus with pronounced low-frequency components. These

symptoms occur paroxysmally, with their duration varying from 20 minutes to several hours or days. Patients with MenDis usually have a premonition of vertigo that enables them to cease dangerous activities (such as driving) before its onset.

A complete audiologic evaluation of the patient suspected of having MenDis should include electrocochleography and electronystagmography.<sup>97</sup> A glycerol test is believed to aid diagnosis, but patients find it uncomfortable so it has largely been abandoned. The electrical responses of the cochlea and the assessment of vestibular function, with both positional and caloric stimulation, also provide important information for diagnosis and monitoring of treatment.<sup>98</sup>

In the early stages, MenDis usually presents as a unilateral, primarily low-frequency sensorineural hearing loss. However, lesions of the auditory nerve must be ruled out. Auditory brainstem responses and MRIs with contrast are necessary to rule out space-occupying lesions before the diagnosis of MenDis is established.

Differential diagnosis requires ruling out migraine-associated dizziness, as there is a higher prevalence of MenDis in a population screened for migraine than in the general population and vice versa.<sup>99-101</sup> The overlapping symptoms—vertigo or dizziness, tinnitus, and hearing loss—suggest an underlying link between the two pathologies.<sup>102</sup>

Audiologic and vestibular tests—such as electrocochleography and rotational-chair stimulation—provide limited diagnostic assistance.<sup>103</sup> Serial audiometry over time, along with vestibular findings and the case history combine to provide the strongest basis for a differential diagnosis.<sup>104</sup> Abnormal vestibular symptoms help to reinforce the differential diagnosis, but a clear pattern of progressive hearing loss is the principal criterion for differential diagnosis.<sup>105</sup>

The possibility that both conditions coexist further complicates diagnosis, especially in their prodromal stages. The expanded treatment options that arise from the differentiation, which include vestibular and balance rehabilitation, improve greatly the management outcomes.

## Management

The management of MenDis remains problematic. Initial treatment is conservative and a variety of medications provide symptomatic relief, but its severity usually prompts an immediate response. However, no single medical or surgical treatment has gained wide acceptance.<sup>106</sup>

A program of vestibular and balance rehabilitation training can have positive results for both MenDis and migraine-associated dizziness. The program is most useful in patients with MenDis following aggressive, destructive surgical or chemical procedures.<sup>107</sup>

This MenDis's paroxysmal character creates a conundrum for research and treatment. Emotional and/or physical stresses are known to trigger individual attacks of vertigo, hearing loss, and tinnitus. During acute attacks, patients' markedly reduced ability to discriminate speech and the accompanying vertigo challenge efforts to prescribe amplification and direct hearing rehabilitation.<sup>108</sup>

Between attacks in the early stages, auditory function may be normal or near-normal, but the fear of another attack often leaves the patient psychologically compromised. Further complicating management, the hearing loss is accompanied by loudness recruitment, narrowed range of comfortable loudness, and severe acoustic distortions. Digital amplification provides needed flexibility to counter hearing fluctuations.

Surgery may be undertaken for patients whose vertiginous attacks become disabling; these include the endolymphatic shunt.<sup>109</sup> A study compared this procedure to a sham operation and concluded that the 70% improvement in both treated and control groups was probably due to a placebo effect.<sup>110</sup> In extreme cases, severing the vestibular portion of the eighth nerve might be an option, if no usable hearing remains.<sup>111</sup>

Chemical ablation of hair cells in the vestibular labyrinth with intratympanic injections of gentamycin is another approach that claims success with low side effects.<sup>112,113</sup>

The Meniett device, a portable low-intensity alternating pressure generator, has been tried to alleviate the symptoms of MenDis.<sup>114</sup> In a study of 67 patients with unilateral MenDis assigned randomly to treatment or a control group, treatment was effective for at least 4 months in controlling severity and number of vertigo attacks.<sup>115</sup>

## OTOSCLEROSIS

Otosclerosis (OTO)—also known as otospongiosis—is a common cause of gradual hearing loss in adults, first identified by Politzer, in 1893.<sup>116</sup> It is progressive, with an unpredictable course.

### Frequency of Occurrence

OTO is one of the commonest causes of hearing loss in adults. Its frequency varies by age, race, and sex.<sup>117</sup> Clinical OTO increases with age, affects Caucasians more than other racial groups, and develops in women more than men.

### Etiology

The cause of OTO remains obscure, though family histories suggest an autosomal dominant transmission. Furthermore, there may be several genetic types. Exogenous causes of OTO, like measles virus infection, have also been suggested.<sup>118</sup>

Two types of OTO have been identified: histologic and clinical. The former has no apparent symptoms, being diagnosed by sectioning of the temporal bones. Hearing loss marks the clinical type, which is usually conductive, affecting the ossicles, and less frequently is sensorineural.<sup>119</sup>

OTO is bilateral in about 9 of 10 females and about 8 of 10 males. Clinical onset during pregnancy has been reported to be between 10 to 17% of females with OTO. It becomes more clinically apparent in females after pregnancy, which seems to accelerate its progression.<sup>120</sup> The hearing loss may worsen

during periods of hormonal-endocrine changes, which may explain, at least in part, the discrepancy in gender incidence.<sup>121</sup>

In most cases, the onsets of hearing loss and tinnitus occur between 15 and 45 years. As it progresses, sensory involvement may be secondary to the footplate lesion or to a primary otosclerotic lesion in the cochlea. The latter may result from toxic metabolites released during the otosclerotic process, reduced blood supply to the lateral wall of the cochlea, or extension of the otosclerotic lesion to the cochlear duct, causing disruption of electrolyte composition and alteration of the basilar membrane biomechanics.<sup>122</sup>

Tinnitus is present in a majority of otosclerotic patients—the reported occurrence of which varies between half to over three-quarters of those with the clinical form. When the inner ear is involved, bouts of dizziness and imbalance may also occur.<sup>123</sup>

## Diagnosis

The onset of OTO is subtle and insidious. Because the hearing loss develops slowly, patients are often unaware of its initial onset. They may experience tinnitus long before they recognize a lessened hearing ability. The air-bone gap arouses suspicion of OTO, especially in patients emerging from their teens. At first, the loss occurs more in the lower frequencies. As the disease progresses, all frequencies are affected to some degree. Bone-conduction audiograms of patients with OTO typically show a greater loss at 2000 Hz—called a Carhart notch.<sup>124</sup> When OTO is limited to the stapedial footplate, a conductive hearing loss up to 65 dB may result. A loss greater than 65 dB would indicate that the cochlea has become involved.

## Management

A variety of treatments are available at each stage of the disease—amplification, surgery, and medication. But OTO presently has no cure.<sup>125</sup>

## *Surgery*

Primarily, the conductive form of OTO is treated surgically usually with stapedectomy. This procedure has several versions, most of which restore hearing when the disease is confined to the stapedial footplate and when cochlear function is normal.<sup>126</sup>

When OTO is conductive below 1000 Hz and sensorineural above 1000 Hz, surgery that reduces or eliminates the low-frequency component will not provide serviceable hearing and will make the patient a poorer candidate for amplification. Postoperatively, such patients will experience a drop in ability to discriminate speech in noise, a narrowed dynamic range, and the absence of a “cushioning” or dampening effect provided by the conductive component.<sup>127</sup>

When bone conduction is depressed by 30 to 35 dB across the frequency range but significantly better than air conduction, surgical intervention has less deleterious effect on speech recognition, but the patient still lacks serviceable hearing in many listening situations and requires amplification. In far-advanced forms of the disease—in which the joint between the stapedial footplate and the oval window is obliterated—aggressive drill-out surgery using microdrills to thin out the footplate creates an opening for a prosthesis.<sup>128</sup>

Once there is significant inner-ear involvement, surgery does not reverse the hearing loss and may not eliminate the tinnitus. Studies so far find that tinnitus is either relieved or eliminated in the majority of patients undergoing stapes surgery, but about 1 in 10 patients report that tinnitus remains the same or worsens postoperatively.<sup>129</sup>

## *Amplification*

If surgery is unsuccessful, hearing aids remain an option.<sup>130</sup> A mixed hearing loss complicates the choice between amplification and surgery. The conductive component tends to improve suprathreshold tolerance and to “flatten” the audiometric configuration, contributing to greater and earlier adjustment to amplification.

Hearing aids may vary in a patient's lifetime from completely in-the-canal hearing aids to powerful behind-the-ear instrumentation. Some patients have or continue to use conventional bone-conduction aids. Others become candidates for cochlear implants when their hearing losses become profound or total.

### ***Medication***

Sodium fluoride has been prescribed to retard progression of the disease. It has fairly innocuous side effects, such as mottling of teeth, so it can be prescribed with impunity, especially in cases in which there is a rapid decline in bone conduction, but its use has aroused some controversy.<sup>131-133</sup>

### ***Monitoring***

Annual audiologic examinations are recommended to check the disease's course and to provide benchmarks for treatment evaluation. Patients should be advised to have an evaluation when there is any subjective change in their hearing.

Periodic audiometry for family members of patients with OTO who do not seem affected by it appears justified by the likely genetic origin of the disorder. Such a precaution would ensure that intervention begins as early as possible, if subsequently found.

## **PRESBYCUSIS**

Presbycusis derives from the Greek *presbys* (old) and *kousis* (hearing), signifying the complex relationship between age and hearing loss.

### **Frequency of Occurrence**

It is the most frequently reported cause of hearing loss among older adults.<sup>134,135</sup>

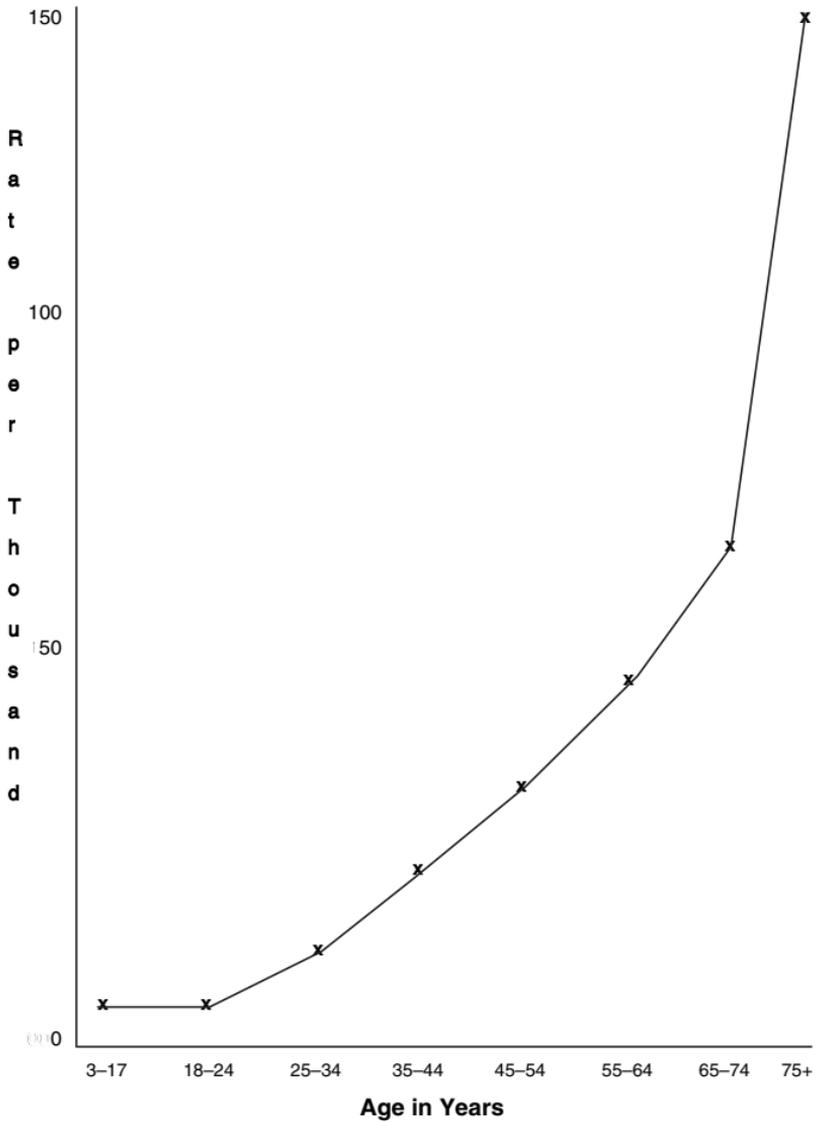
A gender difference in the prevalence of this condition appears to favor females in the frequencies above 1000 Hz with superior retention of hearing in the low frequencies for males.<sup>136</sup> The overall severity of presbycusis is greater in males than females, which may result from (a) genetic factors and/or (b) greater exposure for males to high-intensity noise in work and recreation.<sup>137</sup>

## **Etiology**

The name seems to imply that hearing loss is a consequence of aging, as shown by the high correlation between age and hearing loss (Figure 1.3.1). Some studies challenge the notion that aging causes hearing loss.<sup>138</sup> Others assume the correlation indicates a genetic component because it appears to affect some persons and their parents and grandparents as early as the third decade of life and others retain excellent hearing (at least as measured by pure-tone sensitivity) into their eighth and ninth decades.<sup>139</sup> Many factors probably contribute to the hearing impairments found in the elderly; for example, prior infections, noise exposure, otosclerosis, and ototoxic drugs. Central auditory processing disorder (CAPD) may account for some “cognitive deficits.” As people age, deterioration occurs at all levels of their auditory system.<sup>140</sup> Decrements in sensitivity, particularly for the higher frequencies, characterize much of the sensorineural hearing loss in the elderly.<sup>141</sup>

Although aging can affect any portion of the auditory system, presbycusis is viewed as involving primarily the cochlea and retrocochlear structures up to and including the CNS. Two additional forms of presbycusis have been identified: vascular presbycusis—resulting from deterioration of the blood supply to the lateral wall of the cochlear duct and the spiral lamina—and hyperosmotic presbycusis due to abnormal bone growth in the modiolus or internal auditory meatus compressing auditory nerve cells causing them to degenerate.<sup>142,143</sup>

### Number of Persons with Impaired Hearing per Thousand, by Age



**Figure 1.3.1.** Number of persons with impaired hearing by age. From Ries PW. Prevalence and characteristics of persons with hearing trouble: United States, 1990-91. *Vital and Health Statistics*. 1994;Series 10(188):1

## **Diagnosis**

Most elderly persons enter their senior years with a lifetime of diseases, insults, and injuries to their auditory system. Although different types of presbycusis have been identified, the assertion that the types are accompanied by specific audiologic findings is often difficult to confirm audiometrically.<sup>144,145</sup>

Cognitive factors make necessary the inclusion in the initial audiologic evaluation of measures to detect CAPD. It may complicate diagnosis and management in some seniors who have greater difficulty hearing than would be expected from their audiograms.<sup>146</sup> Because CAPD is believed to affect up to three-fourths of the elderly population with hearing loss, testing for this condition should be required for all patients with presbycusis.<sup>147</sup>

## **Management**

Because of the gradual onset of presbycusis—possibly over several decades—elderly patients often come to rehabilitation reluctantly. They have been adapting to their hearing loss for so long they often tell clinicians, “I don’t miss anything; it’s just that people mumble.” By the time elderly patients finally decide (or are driven by others) to seek help, the clinician will need patience convincing them that they are not too old to be helped.<sup>148</sup>

## ***Amplification***

Real-ear measurements are used in the initial and postfitting visits.<sup>149</sup> Putting audiologic results in percentage terms rather than decibels can assist in convincing patients of their loss and its extent.

In recommending hearing aids, clinicians must be aware of elderly patients’ possible physical limitations, like arthritis. They must have sufficient dexterity to change batteries, adjust volume controls, and properly insert ear molds and the hearing aid.

The greater the CNS involvement, the greater the time and effort required to derive benefit from amplification and the greater the need for expanded hearing rehabilitation often on an individual basis.

### ***Counseling***

Counseling designed to address elderly patients' concerns about activities of daily living should be undertaken (also see Section 2.3). Repetition and reinforcement of instructions are essential. An empathic clinician is a more important determiner of success than the specific brand or style of hearing aid. Also important are arranging for a quiet place in which to talk with patients, because so many have difficulty understanding what is said against a noisy background.

Sending printed information home with patients can enable them to check with others, to ensure they have absorbed the information and instruction provided. The positive attitudes toward rehabilitation of family members and other significant persons can encourage patients to seek assistance and accept treatment.

The high correlation between aging and hearing loss arouses a dangerous attitude that may lead some elderly patients to accept as inevitable further damage to their hearing and to abandon hearing-conservation efforts. Instead, they need to be strongly urged to avoid prolonged, intense noise exposure and ototoxic drugs, to seek prompt treatment of ear infections, and to protect in all possible ways whatever hearing remains. Such counseling will enhance the hearing-aid use that has been prescribed by forestalling further hearing loss.

### ***Medication***

A Dutch study provided evidence for slowing presbycusis by adding oral folic acid to the diets of older men and women. Although hearing continued to decline in the treated and

untreated groups, the losses in the treated group's speech frequencies though statistically significant were clinically insignificant, pointing to the need for cross-validation.<sup>150</sup>

## **STICKLER SYNDROME AND PIERRE-ROBIN SEQUENCE**

Stickler syndrome (SS)—also known as Marshall-Stickler syndrome and Hereditary Arthro-Ophthalmopathy—consists of cleft palate, ocular defects (myopia, retinal detachment, cataracts), flattening of the facial profile, and arthropathy. SS is named after Gunnar B. Stickler, who described it in 1960.

SS has a closely related form designated Pierre-Robin sequence (PRS).<sup>151</sup> PRS symptoms typically comprise orofacial defects (cleft palate, unusually small lower jaw (micrognathia and glossoptosis) and otic anomalies (otitis media, auricular dysplasias, external auditory canal atresia, and conductive hearing loss). In addition to SS, PRS is related to Velocardiofacial syndrome, Möbius syndrome, and CHARGE association.

### **Frequency of Occurrence**

The incidence of SS is probably in the range of 1 to 3 per 10,000 births. PRS has been estimated at 1 per 8,500 live births, but the underlying estimate may include SS rather than being for PRS alone.<sup>152</sup> SS affects both genders.<sup>153</sup>

### **Etiology**

Although generally familial, the mode of inheritance of SS remains undetermined in some cases and open to question in others. A precise explanation of its relatively frequent concurrence with PRS lacks a consensus.

SS affects the connective tissues' collagen. Several genes that govern collagen synthesis may cause SS. As it is a progressive condition, not all aspects may appear at birth. The number of symptoms and the developmental time of their appearance depend on the genes involved: some may only cause the joint

and hearing problems, whereas others give rise to different symptoms, as in Oto-spondylo-megaepiphyseal dysplasia.

In SS, hearing loss occurs in a sizable proportion of cases. The hearing loss is usually secondary to eustachian-tube malformations and palatal anomalies and is typically conductive.<sup>154</sup>

Independent of SS, PRS's etiology is heterogeneous. It has been considered, in different cases, to be an autosomal dominant and an autosomal recessive condition. Although PRS generally does not appear to favor males or females, an X-linked variation that includes clubfeet and heart problems has been reported. Three explanations have been advanced to account for PRS: mechanical, neurologic-maturation, and rhombencephalic anomaly. The mechanical explanation is most widely accepted.<sup>155</sup>

Cleft palate occurs in about 9 of 10 cases of SS. The potential ophthalmic disorders are myopia, a high risk of retinal detachments, cataracts and glaucoma, and conductive hearing loss.<sup>156,157</sup> Stiff and overly flexible joints, with late onset of osteoarthritis, also occur in about half the cases.

## Diagnosis

SS's symptoms and its severity are variable, making it somewhat difficult to diagnose. Differential diagnosis increases the difficulty because of its similarity to the several additional syndromes mentioned above. DNA analysis and familial history are essential to determining the cause(s) of SS and PRS.

Hearing losses often occur, so audiologic examination is essential, though not conclusive with respect to its etiology. Otitis media and auricular anomalies account for the major share of conductive hearing losses. A relatively small number of cases also have auditory-canal atresia and anomalies of the ossicles.<sup>158</sup>

## Management

Treatment begins with repair of the cleft palate, when present, and amplification, when there is a hearing loss. If middle-ear

surgery to correct ossicular-chain defects does not yield serviceable hearing, amplification should be prescribed promptly.

Monitoring on an annual basis should be undertaken because there is a high risk of retinal detachment and late-onset hearing impairment. Early indications of such problems enable patients who experience any of SS's and PRS's numerous symptoms to seek appropriate care for them.

Treating the numerous features of SS and PRS requires a multidisciplinary team: audiologists, pediatricians, otolaryngologists, plastic surgeons, orthodontists, speech-language therapists, and social workers.<sup>159</sup> Referring the family to genetic counseling is recommended.

## TINNITUS

Tinnitus is a sensation of sound perceived in the absence of external stimuli. Patients localize it in one or both ears or "in the head." They describe it with a variety of adjectives, like "throbbing," "hissing," "ringing," or combinations of such sounds. These sensations are almost always subjective, but objective forms of tinnitus that are audible to others do occur, though rarely.

### Frequency of Occurrence

The American Tinnitus Association (ATA) estimates that 40 to 50 million persons in the United States experience tinnitus—a number that far exceeds the prevalence of hearing loss.<sup>160</sup> The number of people whose quality of life is adversely impacted by severe, intractable tinnitus is probably closer to 2.5 million.<sup>161</sup> Among U.S. veterans, 3 to 4 million report they have tinnitus, with up to one million of them seeking clinical intervention.<sup>162</sup> Although some prevalence estimates assume tinnitus may occur in the absence of hearing loss, an auditory basis for virtually all cases of significant tinnitus might be identified if given thorough audiologic evaluation.

About 80% of persons with chronic tinnitus say they are not overly annoyed by it. However, approximately 1 in 5 say it bothers them to the extent they require some form of relief.<sup>163</sup> For the latter persons, tinnitus can be a dominating force in their lives. Anxiety and depression are present in many of these individuals, and suicidal tendencies are not unusual.<sup>164,165</sup> Nearly one-fourth (669) of 2,800 older adults said tinnitus diminished their quality of life, referring to it in terms of physical pain and stress rather than mental or emotional effects.<sup>166</sup>

## **Etiology**

Like a headache or a cough, tinnitus is a symptom not a disease. Any otic or auditory disorder may give rise to tinnitus.

It is a significant early indicator of a hearing loss due to ototoxic medications or to an acoustic neuroma. Much inner-ear tinnitus also results from exposure to prolonged, intense noise levels.

When tinnitus arises from damage to the cochlea, it presents a more challenging condition—identified by air- and bone-conduction thresholds that are in close agreement, tests of word-recognition tests that reveal deficiencies, and measurements of otoacoustic emissions that indicate damage to the outer hair cells. In all such cases, tinnitus elimination typically cannot be achieved, and even its reduction is more difficult, although relief and some control may be obtained.

## ***Objective Tinnitus***

Although the vast majority of cases of tinnitus are subjective, objective tinnitus heard by the patient and audible to others does occur. Some of the causes of objective tinnitus involve muscular, structural or vascular abnormalities. It has been observed in some degenerative diseases, like amyotrophic lateral sclerosis, causing a repetitive flutter of the middle-ear muscles and consequent mechanical sounds. It is even possible

that elevated blood pressure and thyroid dysfunctions can cause objective tinnitus.<sup>167</sup>

### ***Pulsatile Tinnitus***

Pulsatile tinnitus overlaps with the vascular pathologies causing objective tinnitus. Often unilateral, the condition may be the result of vascular pathology; for example, transmission of atrial or intratemporal carotid-artery pulsations. Even serous otitis media can give rise to this condition. Patients with pulsatile tinnitus need a comprehensive radiographic evaluation and consultation with an otologist skilled and experienced in managing patients with this and related conditions.<sup>168</sup>

### **Diagnosis**

The patient who presents with tinnitus requires thorough audiologic and otologic examinations. Because tinnitus may be present at frequencies not customarily tested in pure-tone audiometry, clinicians should use full-sweep Bekesy audiometry that tests all frequencies between 100 to 10,000 Hz and, with the necessary special equipment, above 10,000 Hz.

Audiologists should be cautious about testing the tinnitus patient's acoustic-reflex thresholds at high intensity because it might cause acoustic trauma or worsen the tinnitus. Likewise, clinicians should avoid measuring acoustic-reflex decay at 10 dB above the auditory-reflex threshold for the same reason.<sup>169</sup> When tinnitus accompanies asymmetrical and unilateral sensorineural hearing losses, auditory-brainstem testing and MRI should be performed before considering tinnitus management, to rule out auditory tumors and other pathology.<sup>170</sup>

### ***Tinnitus Inventories***

A tinnitus questionnaire and a stress inventory should be administered to all patients with the complaint of tinnitus. Upon their completion, the clinician reviews their responses and dis-

cusses treatment options with them in a relaxed, supportive environment. Among such instruments, the Tinnitus Handicap Inventory has been subjected to statistical analysis and provides encouraging evidence for its validity.<sup>171,172</sup>

## **Management**

Management of tinnitus falls into three categories: direct intervention to eliminate or ameliorate it, masking to overcome its effects, and education to assist in the process of adaptation to it.<sup>173</sup>

### ***Direct Intervention***

Some treatments may relieve or even eliminate tinnitus; for example, removal of impacted cerumen or excision of an acoustic tumor. When such procedures are successful, the tinnitus often abates or ceases altogether.

### ***Hearing Aids***

Instrumentation, such as maskers and hearing aids, are virtually free of any side effects, when they are professionally prescribed and monitored. Clinicians who instruct their patients in the use of these instruments provide the added advantage of giving patients a means by which they can react positively to tinnitus; that is, they can cease to be its victims and are empowered to exert a degree of control over an unwelcome visitor.

The patient whose tinnitus occurs in the range above 1000 Hz should be evaluated for hearing-aid use. Hearing aids can facilitate the intermittent suppression of tinnitus after auditory stimulation (residual inhibition). It may also direct patients' attention away from their tinnitus.<sup>174</sup>

Fitting hearing aids for the tinnitus sufferer with minimal high-frequency hearing loss may be recommended even when, without the tinnitus, the use of amplification would not be advised.

To gain full benefit from instruments that contain both a masker and an amplification circuit, clinicians should guide their use and they should include a volume control that enables the patient to have some control over their tinnitus.

Phase-shift treatment uses sound cancellation to induce partial or complete inhibition to the frequency of the patient's tinnitus for an extended period of time.<sup>175</sup> A study of 61 patients with tinnitus found a high correlation between treatment and decrease in tinnitus intensity.<sup>176</sup> A Swedish study reported similarly favorable results.<sup>177</sup>

Periodic monitoring of hearing status while hearing aids or maskers are in use is essential.

### ***Masking***

For some tinnitus sufferers, masking provides relief. To aid sleep, for example, a speaker placed under the pillow connected to a sound source plays throughout the night. During waking hours, wearable maskers that provide sound to counter the tinnitus may be prescribed.<sup>178,179</sup>

The maskability of the patient's tinnitus needs to be determined by having patients match their tinnitus to an audiometric frequency or frequencies and determining its loudness. The clinician then presents the patient with a narrow band of noise centered at the frequency of the tinnitus for one minute via earphones at a level rendering the patient's tinnitus inaudible. When the masking noise is removed, the clinician asks the patient whether the tinnitus has become better, worse, unchanged, or different in quality. The presence of residual inhibition is considered a favorable indicator for the use of masking.<sup>180</sup>

Patients with cochlear implants sometimes report it improved or eliminated their tinnitus. Successful stapedectomy for otosclerosis also may affect tinnitus.<sup>181,182</sup> Exceptions have been noted in which tinnitus remains the same or worsens after surgery or is noticed more in the nonoperated ear.<sup>183</sup>

### ***Tinnitus Retraining Therapy (TRT)***

TRT also known as habituation or desensitization therapy has become an increasingly popular method of treating severe, intractable tinnitus.<sup>184</sup> Although a tinnitus masker is used, the level of the masking is below the point at which the tinnitus is obscured. Instead, patients are repeatedly exposed to audible signals from the masker and their own tinnitus in a controlled, supportive setting.<sup>185</sup>

TRT is reported to be successful in over 70% of such patients. However, it is unclear how much of the improvement is a result of habituation and how much is the result of the intensive one-on-one counseling, which is a critical component of the therapy and which takes most patients 18 months before success is realized. Long-term follow-up is essential to determine TRT's efficacy, as it is in all forms of tinnitus therapy.<sup>186</sup>

### ***Cognitive Rehabilitation Therapy (CRT)***

CRT aims to correct the patient's misperceptions about their tinnitus. Adapted from a treatment for depression, it is short-term and involves a contract with the tinnitus patient. The clinician addresses these maladaptive cognitions in terms of scientific data relevant to patients' concerns and appropriate to their educational levels and coping mechanisms.<sup>187,188</sup>

A study of groups receiving education only, education combined with CRT, and a no-treatment waiting list found no significant benefit for the control group.<sup>189</sup> Another study found reduced self-perceived tinnitus impairment following a single group session.<sup>190</sup> When group counseling based on TRT principles was combined with masking, it provided statistically more benefit than either traditional support or no treatment.<sup>191</sup>

### ***Neuromonics Tinnitus Treatment (NTT)***

NTT was formerly called Acoustic Desensitization Protocol. It combines acoustic stimulation with a program of counseling and support. The acoustic component provides stimulation to

auditory pathways and to the limbic system that is presumed to be deprived by hearing loss. Intermittently associating the tinnitus with pleasant, relaxing sound desensitizes the patient's reactions to the tinnitus. Patients are provided with a personal sound player and a recording matched to their audiometric profiles.<sup>192</sup>

NTT has received FDA clearance. A study reported that 32 of 35 subjects with moderate to severe tinnitus attained relief.<sup>193</sup>

### ***Medication***

Numerous medications, herbal extracts, and vitamins have been prescribed for tinnitus relief. Most have not had scientific studies and, when studied, have not been shown to be effective. As for positive anecdotal accounts, the powerful placebo effect provides a likely explanation of such positive results. When these agents seem to work, they probably relieve the anxiety and/or depression rather than the tinnitus.<sup>194,195</sup>

Melatonin supplementation showed a statistically, but not clinically, significant decrease in their scores on the Tinnitus Handicap Inventory.<sup>196</sup> These results may encourage studies with larger doses and, perhaps, over longer periods.<sup>197</sup>

A double-blind, randomized study of the herbal remedy *Ginkgo biloba* found no significant differences in tinnitus alleviation between the 478 placebo and treatment pairs.<sup>198</sup> As with studies of Melatonin, further research using higher dosages of *Ginkgo biloba* over a longer time might obtain results favoring it. Such investigations are underway.<sup>199</sup>

One medication that may provide relief from tinnitus is Xanax (Alprazolam). A double-blind study showed 76% of the treated subjects experienced a reduction of tinnitus, whereas only 5% of the placebo subjects reported improvement.<sup>200</sup>

### ***Other Therapies and Lifestyle Modification***

Among the other treatments that have been suggested are acupuncture, biofeedback training, transcutaneous nerve stimulation, craniocervical massage, and various forms of relaxation

therapy. Most of these techniques have not been subjected to systematic study, and reports of success are based primarily on anecdotal reports.<sup>201</sup>

A comprehensive, integrated approach to tinnitus control and management focuses on the overall emotional well-being of the patient, using management and coping strategies. It postulates a common genetic cause of tinnitus and depression—possibly the serotonin transporter gene *SLC6A4*—and includes informational counseling, sound therapy, and other management strategies.<sup>202</sup>

Clinicians recommend that tinnitus sufferers avoid alcohol, aspirin, caffeine, tobacco, and ototoxic drugs, such as aminoglycosides. Persons at risk should advise their physicians of their vulnerability, seek their advice with respect to any less ototoxic treatments, and report the onset of tinnitus and any changes in their hearing when initiating a new medication.<sup>203</sup>

### ***Surgery***

In several degenerative diseases of the head and neck, loss of control of the tensor tympani or stapedius muscle occurs, causing tinnitus that can be surgically relieved. A dentist specializing in the treatment of temporomandibular joint (TMJ) syndrome can provide relief from tinnitus that often accompanies TMJ.<sup>204</sup>

### ***Monitoring***

Audiologic monitoring on a regular basis is essential for all patients fitted with maskers, hearing aids, or a combination instrument. Even those who choose no treatment should be followed to detect significant alterations in hearing and/or tinnitus and to continue to provide counseling.<sup>205</sup>

## **REFERENCES**

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