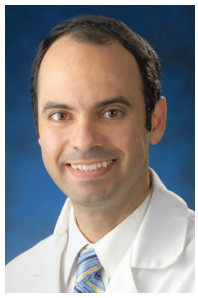

Symptom: Congenital Hearing Loss

By Hamid R. Djalilian, MD

A 6-month-old girl is brought in by her parents for having failed the newborn hearing screening. The family states that the child's mother had an uncomplicated pregnancy with no infections. The child stayed in the hospital an extra day because of being more "yellow;" however, this cleared up pretty quickly with some light therapy. No transfusions were performed. There is no family history of hearing loss. The child was not intubated nor did she stay in the intensive care unit. Sedated ABR confirmed a profound hearing loss bilaterally in the child. CT scan of the temporal bone is performed and is on the right.

What is your diagnosis? See page 28.

Dr. Djalilian is director of neurotology and skull base surgery and professor of otolaryngology and biomedical engineering at the University of California, Irvine.

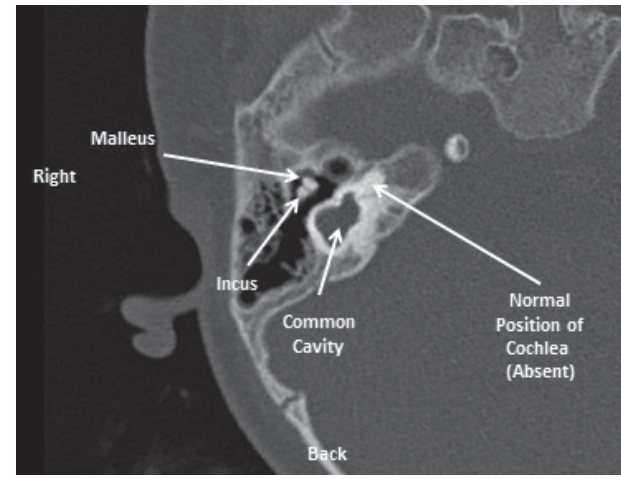

Figure 1. Axial CT image of the right temporal bone showing an absent cochlea and a cystic vestibule. No other discernible inner ear structures can be seen.

Diagnosis: Cochlear Aplasia (Non-development of the Cochlea)

By Hamid R. Djalilian, MD

Continued from p. 26

Failure of the newborn hearing screening creates a lot of anxiety for parents. In a study of over 76,000 children in Greece who had newborn hearing screening using a transient evoked otoacoustic emissions protocol, two percent were found to fail the newborn testing on two consecutive tests performed in the hospital (Korres. *Laryngoscope* 2008;118[7]:1253-6). In this study, not all children followed up for auditory brainstem response testing. One month follow-up testing showed that 36 percent of the children had hearing loss, whereas 64 percent had normal hearing. Of the children who had ABR testing, 18 percent were found to have an effusion, 28 percent had bilateral sensorineural hearing loss greater than 40 dB, and 10 percent had unilateral sensorineural hearing loss greater than 40 dB.

PROFOUND HEARING LOSS IN CHILDREN

The cause of profound hearing loss in children is multifactorial. Profound congenital hearing impairment in children is generally due to an anomaly of the inner ear, environmental, or genetic. Environmental causes include infections which can be viral (rubella, cytomegalovirus and herpes simplex virus), bacterial (e.g., syphilis), or parasitic (e.g., toxoplasma). Other causes include bacterial meningitis and high perinatal bilirubin, among others. A high perinatal bilirubin (yellow baby) will generally not cause hearing loss in a vast majority of patients. The patients at highest risk of hearing loss are those with a bilirubin level of greater than 20 or those requiring an exchange transfusion.

Genetic causes can be divided into syndromic (30%) and non-syndromic (70%). Syndromic causes include Alport, Pendred, Waardenburg, CHARGE, branchio-oto-renal and X-linked progressive hearing loss with perilymphatic gusher. Non-syndromic causes include autosomal dominant, autosomal recessive, X-linked and mitochondria.

The inner ear develops very early in gestation. At approximately the fifth week of gestation, the endolymphatic sacs are formed followed by the cochleas and vestibules. The membranous cochlea has one to 1.5 turns at the end of six weeks, and 2.5 turns by the end of seven weeks. The semicircular canals begin development between the seventh and eighth weeks of gestation. The superior canals initially form, followed by the posterior and horizontal canals. By the eighth week of gestation, all gross inner ear structures have formed in their adult form. Inner ear anomalies occur at a very early stage in gestation. One of the above processes is disrupted to cause an anomaly.

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Congenital inner ear abnormality represents approximately 10 percent of children born with hearing loss. CT of the temporal bone is the best method of imaging the inner ear, though some clinicians use the CISS sequence of the MRI. The CT scan has the advantage that it does not require anesthesia for young children. However, it is disadvantageous due to the radiation involved. The MRI, on the other hand, does not involve radiation, but requires general anesthesia for children as the image acquisition takes approximately 20 to 30 minutes.

COMMON INNER EAR ANOMALIES

There are a number of anomalies of the inner ear with eponyms attached. We will briefly review the most common inner ear anomalies below.

Michel aplasia is known as complete labyrinthine aplasia. It is an extremely rare (approximately one percent of all cochlear bony anomalies) congenital inner ear abnormality. It is defined as the complete absence of inner ear structures caused by developmental arrest of otic placode early in the third week of gestation.

Cochlear aplasia is when the development of the cochlea is arrested in the third week of gestation. In these patients, the vestibule and semicircular canals may be normal, dilated or underdeveloped.

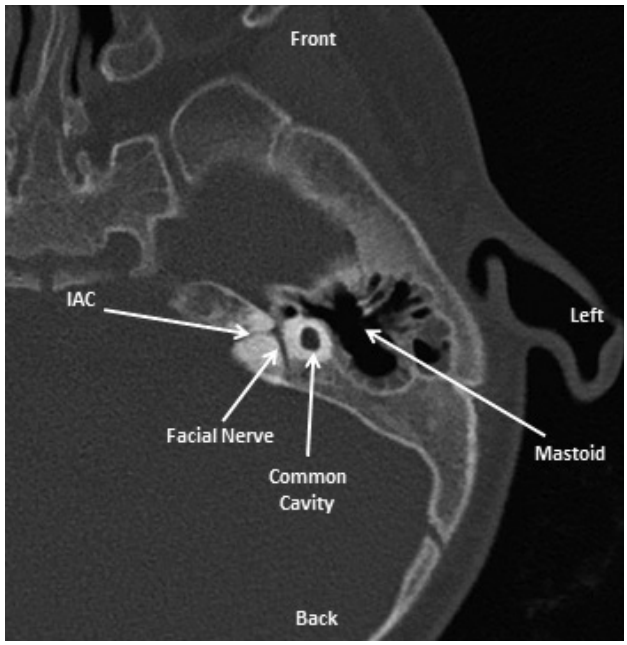

Figure 2. Coronal CT image of the right temporal bone showing a very narrow internal auditory canal (IAC) and a rudimentary superior canal attached to the cystic vestibule. The stapes is connected to the oval window but no round window is present.

Common cavity is a result of an arrest in the fourth week of gestation and occurs when there is a single cavity that contains the structures of the cochlea and vestibule.

Incomplete partition I is defined as a cochlea which has no bony modiolus and the vestibule is generally dilated or cystic. It represents an arrest in development during the fifth week of gestation.

Cochlea hypoplasia occurs when there is a disruption in embryogenesis at the sixth week of gestation. The cochlea and vestibule are separate but the size of the cochlea is smaller.

Incomplete partition II includes the Mondini deformity of the cochlea. The cochlea has 1.5 turns: the basal turn is intact, but the apical and second turns are fused into a single space. The vestibule is generally normal in appearance

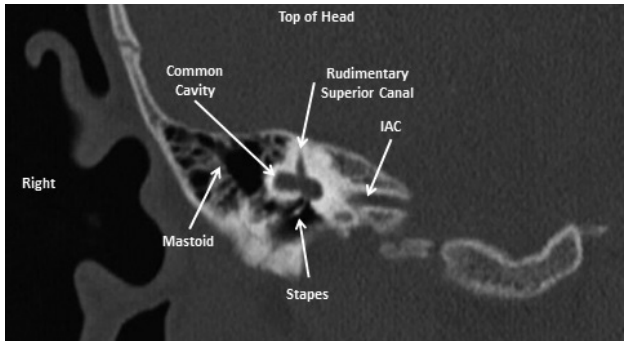

Figure 3. Axial CT image of the left temporal bone showing a common cavity which is not connected to the internal auditory canal (IAC).

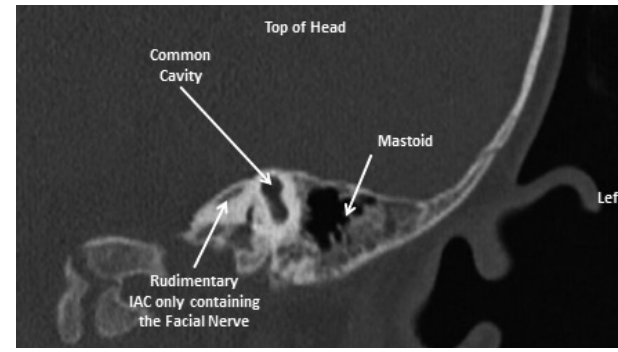

Figure 4. Coronal CT image of the left temporal bone showing a common cavity not connected to the narrow internal auditory canal (IAC). The IAC was only continuous with the facial nerve.

but the vestibular aqueduct is enlarged. This condition occurs due to a disruption in development in the seventh week of embryogenesis.

Semicircular canal abnormalities can either be small, absent or dilated. The horizontal canal is the most common anomalous semicircular canal. It is most commonly seen as a horizontal canal that is fused with the vestibule with no bony island.

Enlarged vestibular aqueduct is most commonly defined as a vestibular aqueduct that is larger at its midpoint than the nearby posterior canal. Others have used a width of larger than 1.5 mm, measured in the same location. An enlarged vestibular aqueduct is associated with Pendred syndrome which causes a goiter around puberty age.

While cochlear implants can be done in a majority of patients with inner ear anomalies, the results are mixed for the more severe anomalies such as common cavity.

Our patient suffered from a right-sided cochlear aplasia and a left common cavity in the position of the vestibule with no connection to the internal auditory canal (IAC). There was no discernible structure in the position of the cochlea on the left. While cochlear implants can be done in a majority of patients with inner ear anomalies, the results are mixed for the more severe anomalies such as common cavity. Common cavity patients can get implants if a separate cochlear nerve can be seen on MRI. Cochlear aplasia and Michel's aplasia patients cannot have a cochlear implant as no cochlea is present. Auditory brainstem implant may be a better option for these patients.