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Large Vestibular Aqueduct and Congenital Sensorineural Hearing Loss

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The inner ear is composed of the membranous labyrinth and the osseous labyrinth (1). The membranous labyrinth has two major subdivisions, a sensory portion called the sensory labyrinth and a nonsensory portion designated the nonsensory labyrinth.

The *sensory* labyrinth lies within the petrous portion of the temporal bone. It contains two intercommunicating portions: 1) the cochlear labyrinth that consists of the cochlea and is concerned with hearing, and 2) the vestibular labyrinth that contains the utricle, saccule, and semicircular canals, all of which are concerned with equilibrium. These hollow chambers are filled with fluid, known as endolymph, that resembles intracellular fluid. The endolymph is rich in potassium ions but poor in sodium ions. The various structures considered to be involved in the production of endolymph include the specialized epithelial cells and related blood vessels of the stria vascularis of the cochlear duct, and the dark cells of the utricle and semicircular canals (1). The *nonsensory* element of the membranous labyrinth is formed by the endolymphatic duct and sac whose main function is believed to be the degradation and absorption of endolymph produced in the sensory labyrinth, ie, the cochlea and vestibule.

All of the structures of the membranous labyrinth are enclosed within hollowed-out bony cavities that are considerably larger than their membranous contents. These bony cavities assume the same shape as the membranous chambers and are referred to as the osseous labyrinth. The bony cavities of the osseous labyrinth are lined by periosteum and contain fluid, known as perilymph, that bathes the external surface of the membranous labyrinth. The perilymph is rich in sodium ions and poor in potassium ions and is roughly comparable with extracellular tissue fluid or cerebrospinal fluid (CSF). It appears to act as a hydraulic shock absorber to protect the membranous labyrinth.

The entire membranous inner ear develops from ectoderm. There are several genetic and nongenetic disorders that affect the developing labyrinth, and result in sensorineural hearing loss. The middle ear and external ear are derived from the first and second branchial arches. They develop quite distinctly from the inner ear; therefore, anomalies of the middle or external ear are not necessarily linked to anomalies of the inner ear.

Sensorineural hearing loss may be congenital or acquired, genetic or nongenetic (2, 3). It has been reported that about 50 mutant genes affect the development of the inner ear (3). Approximately 50% of all deafness can be ascribed to genetic factors (4). Congenital hereditary involvement may consist of hearing loss alone or may be associated with other abnormalities that may or may not fall into patterns of recognized syndromes (5). Agenesis or dysgenesis of the inner ear components is common in either type of congenital hereditary hearing loss (2). In a quantitative analysis of congenital anomalies of the

TABLE 1: Data for 22 patients with sensorineural hearing loss

No.	Patient Age/Year/Sex	Hearing Loss	Vertigo	Tinnitus	Audiographic Findings	Result Caloric Testing	CT Results	Tentative Otologic Diagnosis
1	8/F	AU	No	No	SNHL, AU	ND	Mondini AU	Congenital, SNHL
2	11/F	AU	No	No	SNHL, AU	ND	Mondini, AU, De-formed ossicle	Congenital, SNHL
3	16/M	AS	No	No	SNHL, AS	Abnormal	LVA, AS	SNHL
4	5/M	AS	No	No	SNHL, AS	ND	Michel anomaly, AS	SNHL
5	30/F	AD	No	No	SNHL, AD	ND	Michel anomaly, AD	SNHL
6	6/M	AS	No	No	SNHL, AS	ND	Mondini, AS	SNHL
7	26/M	AU	No	Yes	SNHL, AU AS > AD		EVA, AS	R/O retro cochlear lesion
8	7/M	AS	No	No	SNHL, AS	ND	Mondini anomaly	R/O Mondini anomaly
9	59/F	AS	No	No	SNHL, AS	ND	EVA	SNHL
10	8/F	AU	No	No	MHL, AU	ND	Mondini anomaly, AU, otomastoiditis	Congenital anomaly, otomastoiditis
11	22/M	AU	No	No	SNHL, AU AS > AD	ND	EVA AS > AD	
12	19/M	AD	Yes	No	SNHL, AD	Abnormal	EVA, AD	R/O retro cochlear lesion
13	3/M	AU	No	No	SNHL, AU	ND	Mondini anomaly, AU	Congenital inner ear anomaly, AU
14	32/F	AS	No	No	SNHL, AS	ND	EVA, AS	SNHL
15	24/F	AD	No	No	SNHL, AD	ND	EVA, AD	SNHL
16	26/F	AS	No	No	SNHL, AS	ND	EVA, AS	SNHL, AS
17	14/M	AD	No	Yes	SNHL, AD	ND	EVA, AD	SNHL, AD
18	14/M	AU	No	No	SNHL, AU AD > AS	ND	EVA AD > AS	R/O Mondini anomaly
19	7/M	AU	No	No	SNHL, AU	ND	EVA, AU AD > AS	R/O Mondini anomaly
20	19/F	AD	Yes	No	SNHL, AD	Normal	EVA, AD	R/O congenital anomalies
21	21/F	AU	No	No	SNHL, AU AD > AS	ND	EVA	
22	47/F	AS	No	No	SNHL, AS	ND	EVA	R/O retro cochlear lesion

Legend— AD = right ear   AU = both ears   LVA = large vestibular aqueduct
F = female   AS = left ear   EVA = enlarged vestibular aqueduct
R/O = rule out   M = male   MHL = mixed hearing loss
ND = not done   AC = internal auditory canal   SNHL = sensorineural hearing loss

inner ear, Sando et al (6) found that 62.3% (48/77) of the temporal bones with congenital anomalies had anomalies in the vestibular system. Of the 117 individual vestibular anomalies, 73 (62.4%) were in the semicircular canals, and among these, the lateral semicircular canal was most affected. This may be because the lateral semicircular canals develop relatively late and any organ that develops later tends to have more opportunity to become anomalous than one that develops earlier. However, from our clinical and radiologic study, we have an impression that the vestibular aqueduct, not the semicircular canals,

is the structure that is most affected in patients with anomalies of the inner ear.

Summary of Recent Experience

We studied computed tomography (CT) and magnetic resonance (MR) scans of the temporal bones of 22 selected patients with presumed, long-standing sensorineural hearing loss. All patients had noncontrast CT; five underwent additional high resolution MR. The patients' ages ranged from 3 to 59 years. The CT studies were performed on a GE CT/T 9800 unit (General Electric, Milwaukee, WI) with a scan time of 2

Surgical Findings	Comments
	Born deaf
	Patient with Wildervank syndrome
	Only a remnant of inner ear seen
	IAC was absent
	Markedly dilated lateral semicircular canal
	Family history of hearing loss
	MR showed cystic cochlea and dilated vestibule and lateral semicircular canal
	MR showed large cystic endolymphatic duct
Chronic otomastoiditis	Patient with hearing aid
	MR showed large endolymphatic duct and sacs, AU
	Hearing loss detected at age 12. Patient with hearing aid
	Patient with hearing aid
	Family history of hearing loss. Patient with hearing aid
	Prominent vestibule, seen on CT scans
	Patient with hearing aid

head coil, 3-mm section thickness, 256 × 128 matrix and a 16-cm field of view. In two patients, additional T2-weighted sagittal images were also obtained.

Findings in our patients are summarized in Table 1. There is great variability in the inner ear findings. Two patients had the Michel anomaly; six had the Mondini anomaly or a variant of it (Figs. 1, 2, and 3). In one patient with bilateral Mondini anomaly of the cochlea, the vestibules were severely hypoplastic and there was no de-

A

B

Fig. 1. Mondini anomaly. *A*, Axial CT scan shows bilateral cystic dilation of the cochlea (*C*), vestibule (*V*), and lateral semicircular canal (*L*). Notice posterior semicircular canal (*arrow*). Note normal incus (*i*) and anterior to it the normal malleus. *B*, T2W axial MR scan shows dilated membranous labyrinth. *C*, cochlea, *V*, vestibule.

seconds, and 1.5-mm collimation. All patients underwent CT in the axial plane. In some patients, additional direct coronal and direct sagittal views were obtained. Direct sagittal images were obtained according to our protocol reported earlier (7). MR images were obtained on a 1.5-T Signa system (General Electric). Using the head coil and 5-mm section thickness, axial spin-echo, proton-weighted 2000/30 (TR/TE), and T2-weighted 2000/80 images of the posterior fossa and temporal bones were obtained. For each of these patients additional axial, coronal, and sagittal spin-echo T1-weighted images at 500–600/25/4 (TR/TE/excitations) were obtained using the
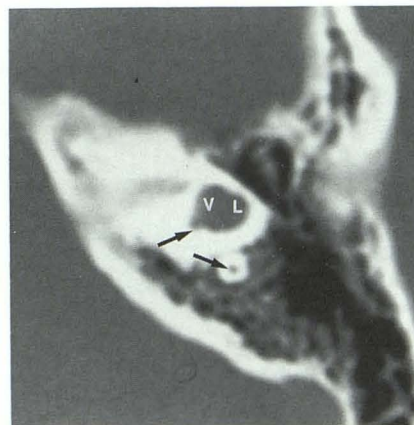

Fig. 2. Mondini anomaly. Axial CT scan shows dilated vestibule (*V*) and lateral semicircular canal (*L*). The posterior semicircular canal (*arrows*) is not dilated
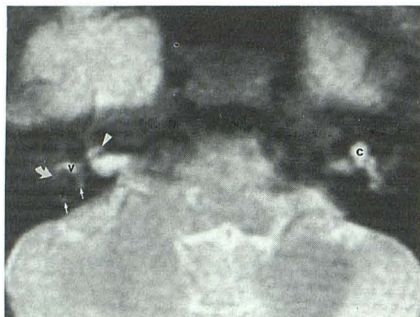
Fig. 3. Mondini anomaly. T2W Axial MR scan shows dilated left cochlea (*c*). Notice normal right cochlea (*arrowhead*), vestibule (*v*), lateral semicircular canal (*curved arrows*), and posterior semicircular canal (*arrows*).

velopment of the semicircular canals. A large vestibular aqueduct was the most common inner ear dysplasia found in this series (Figs. 4 and 5).

In patients with Mondini anomaly, MR revealed fluid-filled cystic dilation of the cochlea and vestibule (Figs. 1 and 3). In patients with a large vestibular aqueduct, MR revealed fluid-filled cystic dilation of the endolymphatic duct and sac (Figs. 4B, 4C, and 5B–5D). The dilation of the endolymphatic duct and sac was more pronounced beyond the isthmus and the endolymphatic sac was particularly involved (dilated) (Figs. 4 and 5).

Anatomy and Embryology of the Inner Ear

Embryologically, inner ear dysplasia results from arrested development during early fetal life. The rudiments of the inner ear can be found in embryos of approximately 22 days as thickened plates of surface ectoderm on each side of the developing hindbrain (rhombencephalon) (Fig. 6) (8, 9). These thickenings, termed the otic placodes, then invaginate rapidly into the underlying mesenchyme and form the otic pits, the mouth of which becomes closed, forming the otic or auditory vesicle (otocyst), the precursor of the membranous labyrinth (Fig. 6). Each otocyst then divides into a ventral, or saccular, component that gives rise to the saccule and the cochlear duct (Fig. 7), and a dorsal or utricular component that forms the utricle, semicircular canals, and endolymphatic duct (Fig. 7) (8–10). The epithelial structures so formed are known collectively as the membranous labyrinth (9). Three flat diverticula grow from the utricular portion and give rise to the semicircular canals. The development of the semicircular canals is complete by the seventh week of gestation (Fig. 8) (10). The semicir-

cular canals open into the utricle and the utricle opens into the saccule through the ductus utriculosaccularis which also joins the endolymphatic duct (1). In the sixth week of development, the saccule forms a tubular-shaped outgrowth, the cochlear duct, at its lower pole (Fig. 8). This cochlear duct penetrates the surrounding mesenchyme in spiral fashion until, at the end of the eighth week, it has completed 2½ turns (9). The duct reaches its full length of 2½ to 2¾ turns by the 10th week (10). The connection of the cochlear duct with the remaining portion of the saccule is confined to a narrow pathway, the ductus reuniens (duct of Hensen) that provides physiologic continuity in all parts of the membranous labyrinth (Fig. 8).

The mesenchyme around the otocyst condenses and differentiates into the cartilaginous shell, the otic capsule (Fig. 9). In the 10th week, this cartilaginous shell undergoes vacuolization to form the perilymphatic space. The cartilaginous otic capsule then ossifies to form the bony otic labyrinth of the inner ear. In the 10th week of gestation, as the cartilaginous labyrinth surrounding the cochlear duct undergoes vacuolization, two perilymphatic spaces are formed, the scala vestibuli and scala tympani. The cochlear duct is then separated from the scala vestibuli by the vestibular membrane, and from the scala tympani by the basilar membrane (Fig. 9). The lateral wall of the cochlear duct remains attached to the surrounding cartilage by the spiral ligament, whereas its medial wall is connected to, and partly supported by, a long cartilaginous band, the modiolus, which is the precursor of the central axis (bony modiolus) of the bony cochlea (Fig. 9). The bony canal of the cochlea takes about two turns and three-quarters round the modiolus (1)

With further development, the epithelial cells of the cochlear duct form two ridges: the inner ridge (future spiral limbus) and the outer ridge. The outer ridge forms one row of inner hair cells and three or four rows of outer hair cells, the sensory cells of the cochlear labyrinth. The hair cells are covered by the tectorial membrane, a gelatinous structure with delicate fibers (1) extending from the spiral limbus and rests with its tip on the hair cells (Fig. 9) (9).

The hair cells and other series of epithelial structures placed upon the basilar membrane are collectively referred to as the spiral organ of Corti.
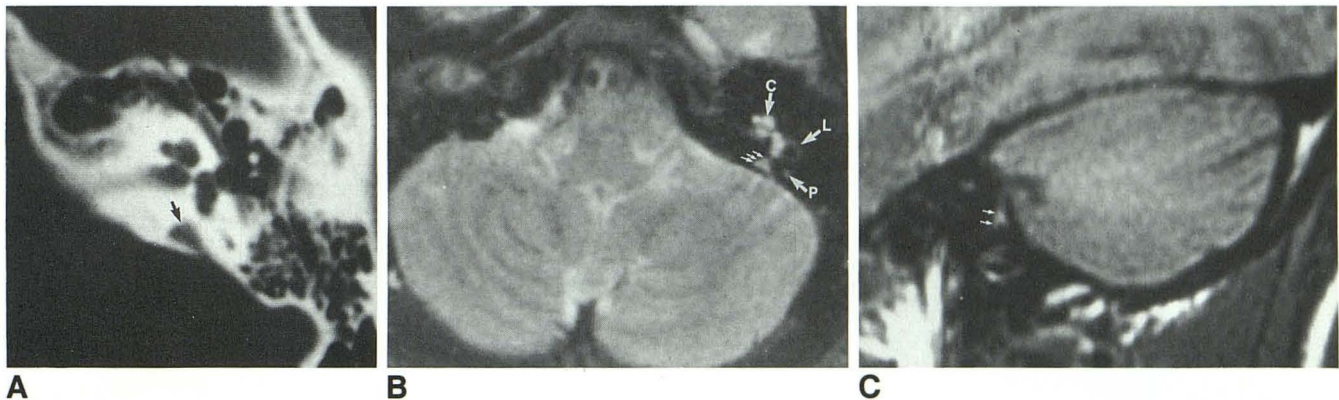
Fig. 4. Large vestibular aqueduct and endolymphatic duct and sac. *A*, Axial CT shows large vestibular aqueduct (*arrow*). *B*, T2W (2200/80) axial MR scan shows enlarged endolymphatic duct and sac (*arrows*). The cochlea (*C*), lateral semicircular canal (*L*) and posterior semicircular canal (*P*) are normal in size. *C*, T1W (600/20) sagittal MR scan shows enlarged endolymphatic duct and sac (*arrows*).

Crista Ampullaris, Macula Utriculi, Macula Sacculi, Spiral, and Scarpa Ganglia

During the sixth week of gestation, the semicircular canals appear as three flat diverticula of the utricular part of the otic vesicle. The epithelial cells of the membranous labyrinth form a crest in the ampullae of the semicircular canal. This crest, known as the crista ampullaris, contains the sensory cells of the semicircular canals. Similar sensory areas develop in the walls of the utricle and saccule, where they are known as the maculae acusticae (macula utriculi and macula sacculi) (9). Impulses received by the organ of Corti are transmitted by peripheral (afferent) cochlear nerve fibers to the cell bodies of the spiral ganglion, and then to the central nervous system by the central auditory fibers of the cochlear nerve. The cochlear nerve fibers arise from bipolar cells (primary neurons of the auditory system) of the spiral ganglion which is situated in the spiral canal of the modiolus of the cochlea (Fig. 9). Impulses generated in the sensory cells of the cristae and maculae as a result of a change in the position of the body are carried to the brain by the vestibular fibers of the eighth cranial nerve. The vestibular nerve arises from bipolar cells in the vestibular ganglion (Scarpa's ganglion), situated in the trunk of the nerve at the lateral end of the internal auditory canal.

Statoacoustic Ganglion

During formation of the otic vesicle, a small group of cells breaks away from its wall and forms the statoacoustic ganglion (Fig. 6). Other cells of this ganglion are derived from the neural

crest. The ganglion subsequently splits into a cochlear portion that supplies the sensory cells of the organ of Corti, and a vestibular portion that supplies the sensory cells of the saccule, utricle, and semicircular canals (9).

Endolymphatic Duct and Sac

The endolymphatic duct and sac are the non-sensory part of the membranous labyrinth (11). The duct continues into a blind-ended sac, the endolymphatic sac which expands under the dura mater on the posterior surface of the petrous bone (1). The vestibular aqueduct that houses the endolymphatic duct (ED) and part of the endolymphatic sac (ES) is a bony canal related to the bony labyrinth of the inner ear. There is a great variation in the size and shape of the nonsensory ES, unlike the sensory regions of the membranous labyrinth, ie, the cochlear and vestibular end organs (11). Friberg et al (12) studied a large number (29 specimens) of human extraosseous ES specimens. They found that the size and shape of the extraosseous ES region varied widely. The ED-ES system is partially embedded in the temporal bone and partially surrounded by a duplication of the posterior fossa dura. In 1869, Boettcher (13) published a detailed description of the anatomy of histologic serial sections of the vestibular aqueduct. Since then, the concept that the ED and ES constitute a single-lumen, tube-like structure has prevailed in the literature (11). However, the work of Antunez et al (14) and Linthicum and Galey (15) revealed that the ES does not consist of a single lumen; rather it is a system of parallel interconnected tubules. These authors concluded that the lumen of the ES
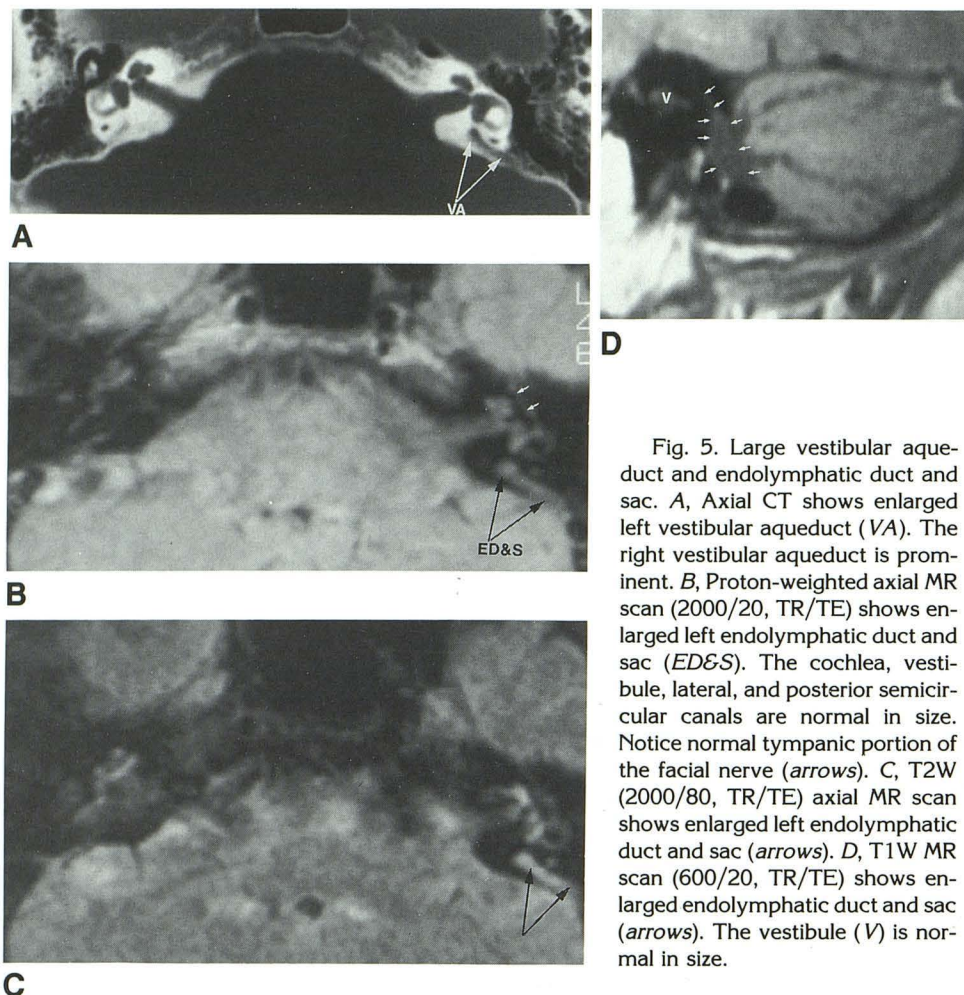
Fig. 5. Large vestibular aqueduct and endolymphatic duct and sac. *A*, Axial CT shows enlarged left vestibular aqueduct (*VA*). The right vestibular aqueduct is prominent. *B*, Proton-weighted axial MR scan (2000/20, TR/TE) shows enlarged left endolymphatic duct and sac (*ED&S*). The cochlea, vestibule, lateral, and posterior semicircular canals are normal in size. Notice normal tympanic portion of the facial nerve (*arrows*). *C*, T2W (2000/80, TR/TE) axial MR scan shows enlarged left endolymphatic duct and sac (*arrows*). *D*, T1W MR scan (600/20, TR/TE) shows enlarged endolymphatic duct and sac (*arrows*). The vestibule (*V*) is normal in size.

consists of a series of parallel tubules, or canaliculi, rather than folds and rugae. Bagger-Sjöback et al (11) reported their study with the use of computerized and graphic three-dimensional reconstruction of a human ED and ES. They showed that the ES is a fusiform and flattened structure with marked tubularity, especially in the extraosseous region. Their study made it possible to estimate, for the first time, the volume of the ED and ES. The ED and ES of their specimen was 18.2 mm long. It measured 60×200 μm at the isthmus portion of the ED and 200×7000 μm at the broadest part of the ES. The volume of the ED was 0.03 mm^3 and of the ES, 1.85 mm^3. The specimen in Bagger et al's study represented a well-developed ES. They concluded that, in general, most ES volumes are probably somewhat less than 1.85 mm^3. The extraosseous ES volume represents more than two-thirds of the total ES volume (11, 16). The ES is important to inner ear function. The finding of a stainable substance in the lumen, sometimes containing

foreign material, indicates that the ES tubules probably participate in the final degradation and absorption of endolymph (11).

On phylogenetic, developmental, structural and functional grounds, the ear is divided into external, middle, and internal ears (11). The bony cochlea is a conical, snail-shaped structure. The bony canal of the cochlea consists of a spiral canal, also called cochlear canal, that makes roughly 2¾ turns about a central pillar called the modiolus (1). The modiolus is the conical, central pillar of the cochlea (Fig. 9). Projecting laterally from the modiolus is a delicate bony shelf or ridge, termed the osseous spiral lamina, that projects from the modiolus into the canal, similar to the flanged thread of a screw (Fig. 9). Extending from the outside of the spiral canal is a thick ligament known as the spiral ligament (Fig. 9). The spiral lamina and spiral ligament are connected by the relatively thin basilar membrane (Fig. 9). The bony spiral cochlear canal is divided by the basilar membrane and the vestibular mem-
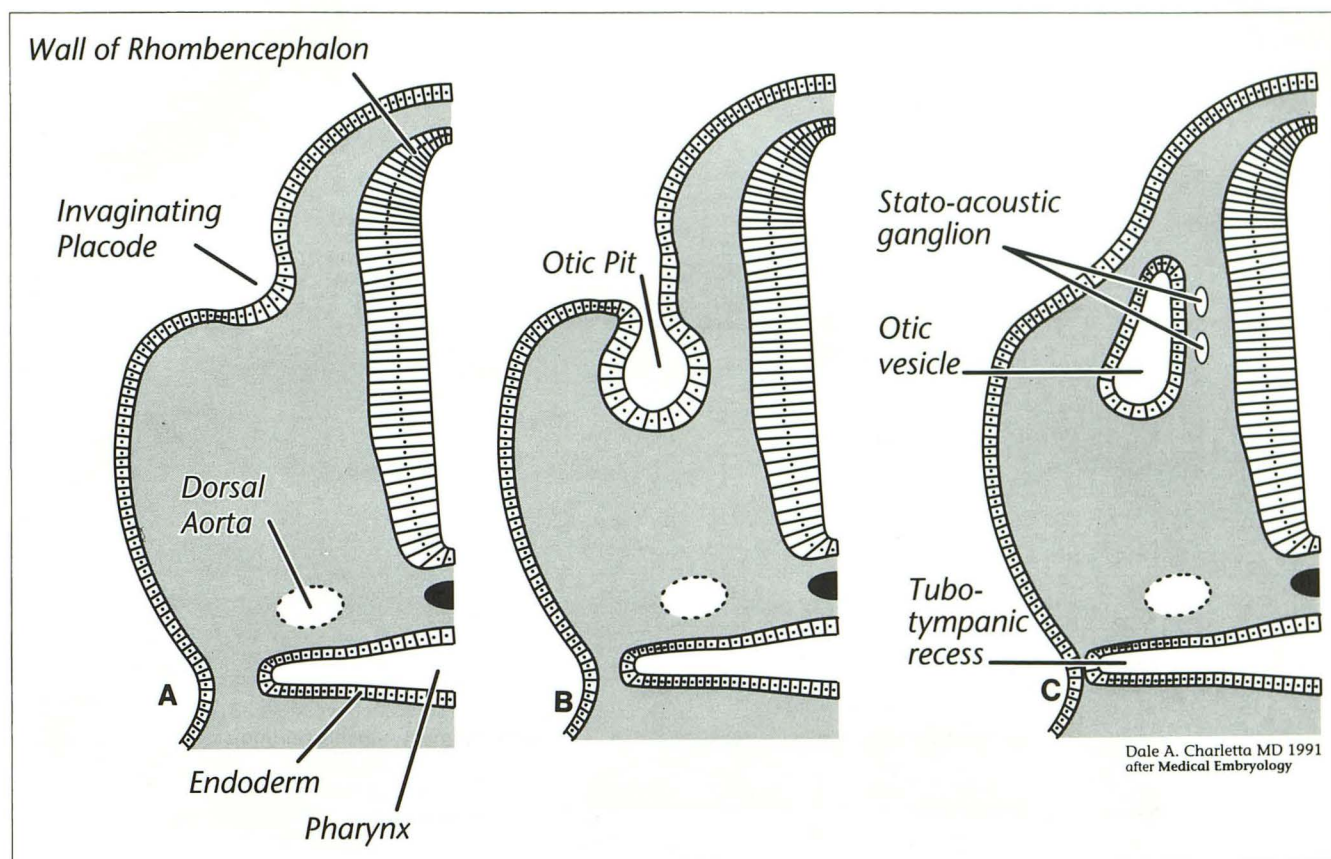
Fig. 6. Schematic transverse sections through the region of the rhombencephalon showing the formation of the otic vesicles. *A*, 24 days; *B*, 27 days; *C*, 4½ weeks. Note the appearance of the statoacoustic ganglion. (Reprinted with permission from Sadler (9).)

brane (Reissner's membrane) into three compartments (Fig. 9). An upper compartment called the scala vestibuli (Fig. 9) connects with the vestibule (Fig. 10). The lower compartment, the scala tympani (Fig. 9), connects with the middle ear through the round window membrane (Fig. 10). The middle compartment, the scala media (Fig. 9), is the cochlear duct (which should not be confused with cochlear canal and cochlear aqueduct). The cochlear canal is the perilymphatic space (lumen) of the osseous cochlea. The cochlear duct is basically the endolymphatic space of the membranous cochlea. The cochlear aqueduct serves as connection between the perilymphatic space and the subarachnoid space (Fig. 10). The cochlear duct is separated from the scala vestibuli by vestibular or Reissner's membrane and from the scala tympani by the basilar membrane (Fig. 9). It is filled with endolymph and the greatly specialized epithelium of the cochlear floor which is the spiral organ of Corti. The function of endolymph is nourishment of the organ of Corti and freeing it from the vibrations that its own intrinsic blood supply would produce. The fibers

of the auditory nerve terminate on this structure which is the peripheral sense organ of hearing.

The cochlear aqueduct terminates medially as a funnel-shaped aperture medial to the jugular fossa. It serves to connect the subarachnoid space with the perilymphatic space in the scala tympani (Figs. 10 and 11). From the internal aperture, the cochlear aqueduct runs in a slightly medial and posterior direction inferior to the internal auditory canal. The average length of the aqueduct measures 6.2–12 mm. Unlike the vestibular aqueduct, the cochlear aqueduct does not contain a well-defined membranous duct. It is filled by loose connective tissue that becomes continuous with the periotic lining of the scala tympani at the inner opening (Fig. 10) and with the dura mater at the outer aperture (Fig. 11).

The cochlear aqueduct is wide during fetal life. However, it narrows normally with growth. The fibrous tissue within the aqueduct is connected with the subarachnoid space of the posterior fossa. The perilymph can flow through the cochlear aqueduct from the scala tympani of the inner

Fig. 7. Schematic transverse section of a 7-week embryo in the region of the rhombencephalon, showing the development of the endolymphatic duct and utricular and saccular portions of the otic vesicle. (Reprinted with permission from Sadler (9).)
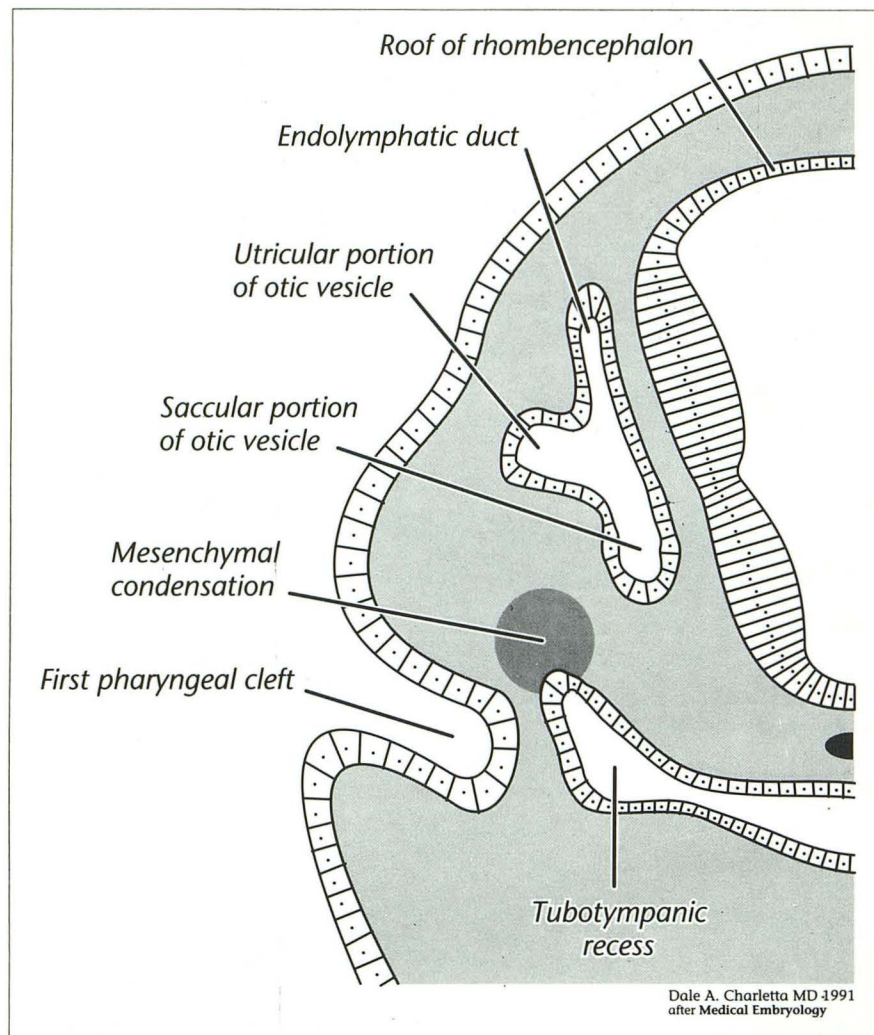
ear into the CSF adjacent to the sigmoid sinus (17).

The cochlear aqueduct is best evaluated by CT scanning. The inner or medial portion of the aqueduct is seen as a funnel-shaped bony canal along the undersurface of the petrous bone (Fig. 11). Its outer or lateral portion is very narrow and is seen as a filiform line. It may not be visualized at all. A large or a widely patent cochlear aqueduct is pathologic. In these situations, CSF freely communicates with the perilymphatic space (Fig. 10), inducing a perilymphatic hydrops (17). This condition may be seen in patients with congenital fixation of the stapes footplate (18), resulting in mixed hearing loss with progressive sensorineural component. An enlarged cochlear aqueduct is also responsible for the perilymphatic "gusher" during stapedectomy (19), as well as in patients with unexplained progressive sensorineural hearing loss.

Michel and Mondini Anomalies

Interruption in the orderly development of the inner ear during fetal life results in a continuum of inner ear malformations. Four major patterns of congenital inner ear malformation are commonly recognized. These patterns represent points along a continuum, often making specific "classification" of a given anomaly difficult (8, 10).

1) The Michel anomaly is the most severe type of aplasia of the inner ear. It is characterized by complete failure of inner ear development and results from an insult in the 4-week embryo. In some cases, even the petrous portion of the temporal bone is not developed; in others, the petrous temporal bone is present but underdeveloped. The external and middle ears may be normally formed and capable of functioning.

2) The Mondini anomaly is the incomplete formation of the bony and membranous laby-
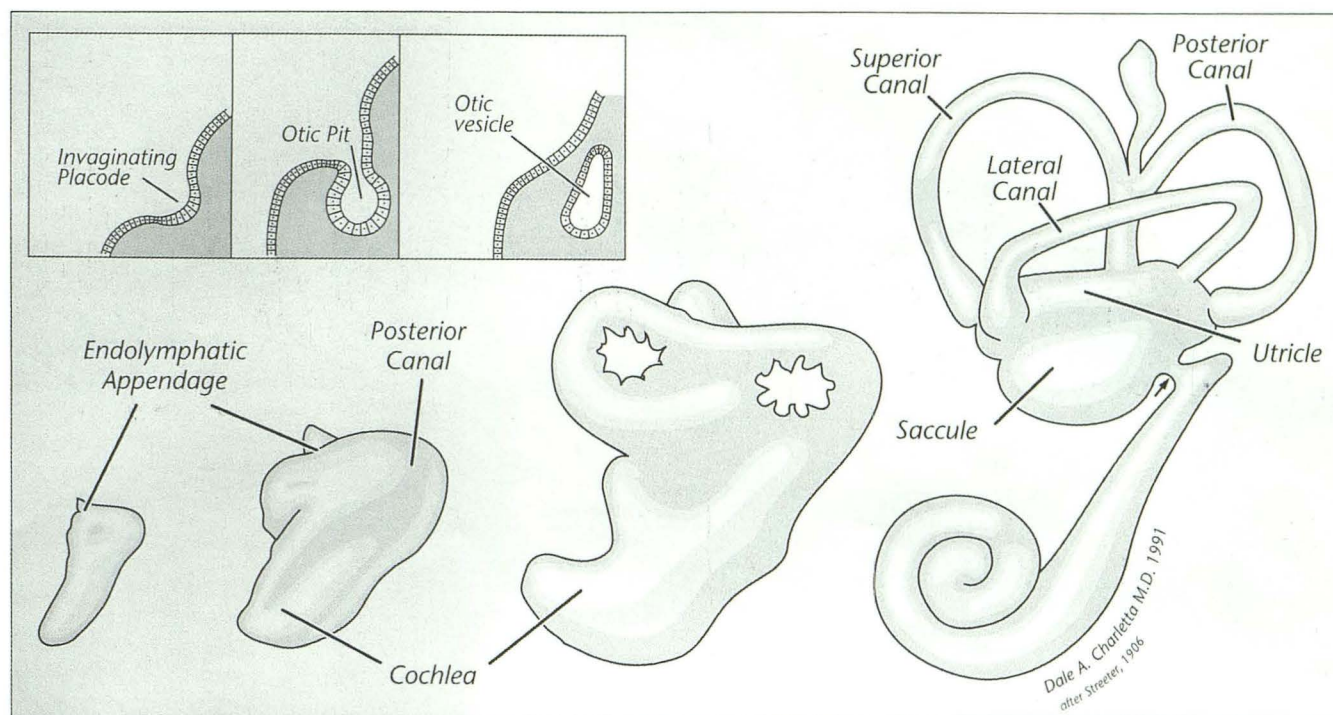
Fig. 8. Lateral view of developmental stages of otocyst. Notice the communication between the saccule and cochlea through the duct of reuniens (*arrow*). (Reprinted with permission from Valvassori (37).)

rinths and results from developmental arrest at the end of the seventh week (10). This type of malformation shows only a primitive single curved tube representing the cochlea, presumably due to interruption of its development at the stage in which it is only a short curved tube (2, 5). The vestibular structures may also be underdeveloped, but at times the vestibular structures including ED and ES may be markedly dilated (6, 8).

Developmental arrests at later times lead to milder malformations (10): 3) The Scheibe malformation, involves only the cochlea and saccule (6, 8). It is the most common form of hereditary congenital hearing loss. 4) The Alexander dysplasia is the least severe type and consists of aplasia of the cochlear duct. Usually only the basal turn is affected. In Scheibe and Alexander malformations, the bony labyrinth is completely formed, so these types do not routinely demonstrate radiographic abnormalities (8).

In 1791, Carlo Mondini (20) presented a paper before the Academy of Sciences of the University of Bologna entitled *An Anatomical Dissection of a Boy Born Deaf* (10). He described an 8-year-old boy, born deaf, who developed gangrene after he was struck on the foot by a wagon; in a few days, he died. Mondini examined very carefully the ears of the child and noted that the external and middle ears appeared normal. However, the cochlea was abnormal and completed only 1½ turns, with the final spiral at the apex missing. The vestibule was larger than normal size. This was the first description of combined membranous and bony dysplasia based on the anatomical dissection of the inner ear. Since then, several temporal bone studies have detailed the histologic and pathologic changes first described by Mondini (21, 22). Characteristically, the cochlea is flattened and shortened, with 1 to 1½ turns instead of the normal 2½ to 2¾ turns. Only the basal turn of the cochlea may be developed (10). The modiolus and osseous spiral lamina are hypoplastic (Fig. 12), and the cochlear duct is at times dilated and shortened. The stria vascularis may be atrophied. The organ of Corti is often absent and the number of spiral ganglion cells is decreased (10). The endolymphatic duct, vestibule, and semicircular canals can be enlarged (Figs. 1–3). In extreme cases, the vestibule has a cyst-like appearance (Fig. 1). Many patients also have an anatomical defect in the stapes footplate; occasionally, the stapes is totally absent (10). Patients with Mondini dysplasia are predisposed to developing CSF leak and meningitis (Fig. 13) (23–25). The cochlear aqueduct and internal auditory canal are both connected to the subarachnoid space. Therefore, abnormalities of either can
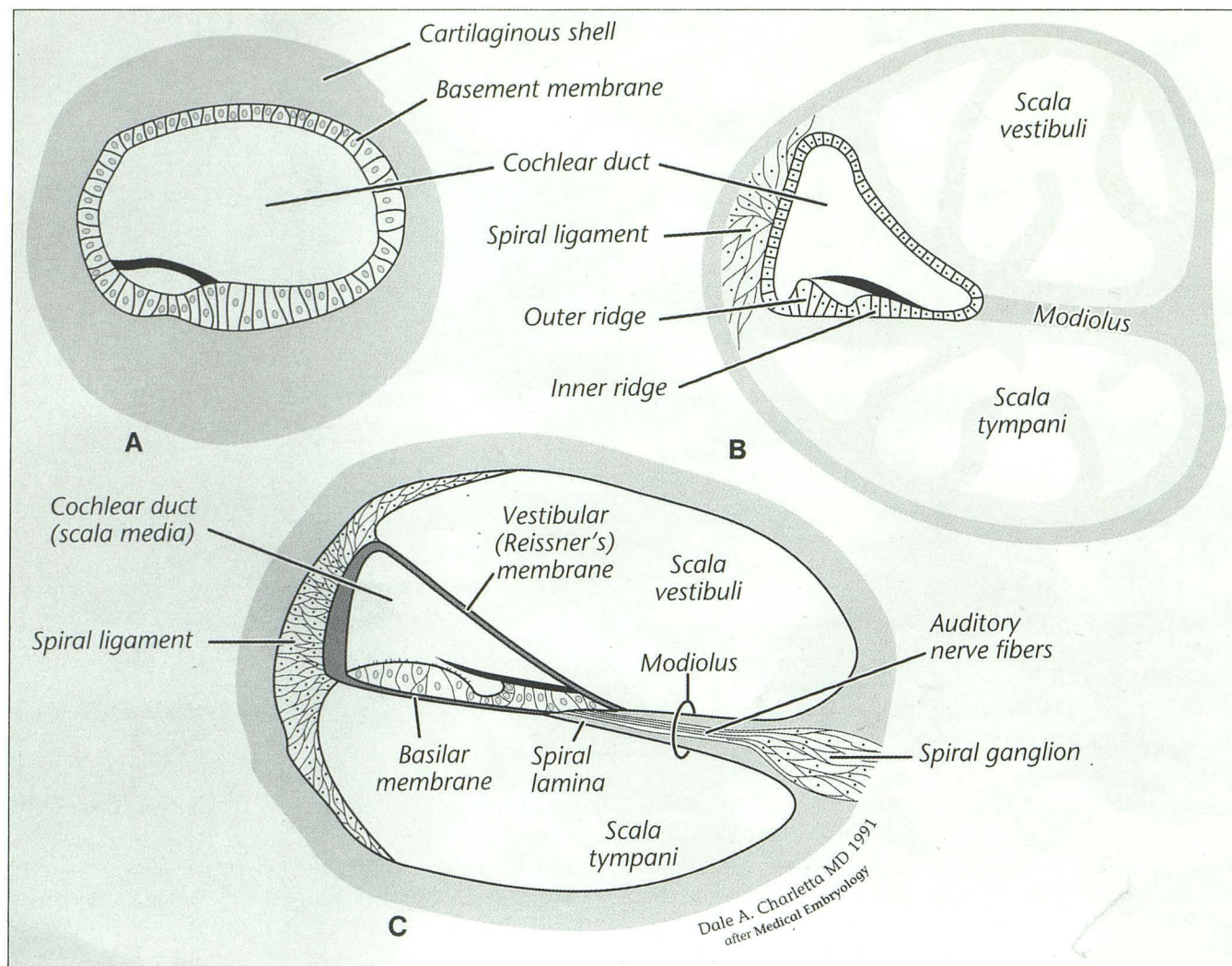
Fig. 9. Schematic representation of the development of the scala tympani, scala vestibuli, and scala media or cochlear duct. *A*, The cochlear duct is surrounded by a cartilaginous shell. *B*, During the 10th week, large vacuoles appear in the cartilaginous shell. *C*, The cochlear duct is separated from the scala tympani and the scala vestibuli by the basilar and vestibular membranes respectively. The tectorial membrane overlies the spiral organ of Corti which lies over the basilar membrane. Note the auditory nerve fibers and the spiral (cochlear) ganglion. Note that the hair cells lie at top of the basilar membane, and the tectorial membrane rests on the hair cells. (Reprinted with permission from Sadler (9).)

increase CSF flow into the labyrinth (26). A widely patent cochlear aqueduct allows CSF to pass directly to the scala tympani of the malformed cochlea, and cyst-like vestibule. Flow through the internal auditory canal is possible if the lamina cribrosa is thin and fragile as noted in some cases of Mondini dysplasia (22). From the inner ear, CSF can leak through a defect in the otic capsule into the middle ear (Fig. 13). The most common site of leakage is the oval window or stapes footplate, although defects have been reported in the round window, promontory, fallopian canal, hypotympanum, and eustachian tube (10).

The stapedial footplate defect also has a developmental origin: the footplate consists of two layers: an outer lamina derived from Reichert's cartilage; and an inner lamina, the vestibular layer, or lamina stapedius, formed from the otic capsule (10). The initial cartilaginous form of the stapes is replaced by three layers of bone: outer and inner periosteal layers, with an endochondral layer in between. The bone is then remodeled in a resorptive fashion (23 to 26 weeks) to thin the crura and to enlarge the obturator foramen (10). The vestibular layer of footplate cartilage does not undergo bony replacement, but is preserved as a base on which the outer bony layer of the footplate is deposited. If the vestibular layer of cartilage does resorb, a defect results.

The child with Mondini malformation may present to the otolaryngologist in several ways. If the malformation is bilateral, profound sensori-
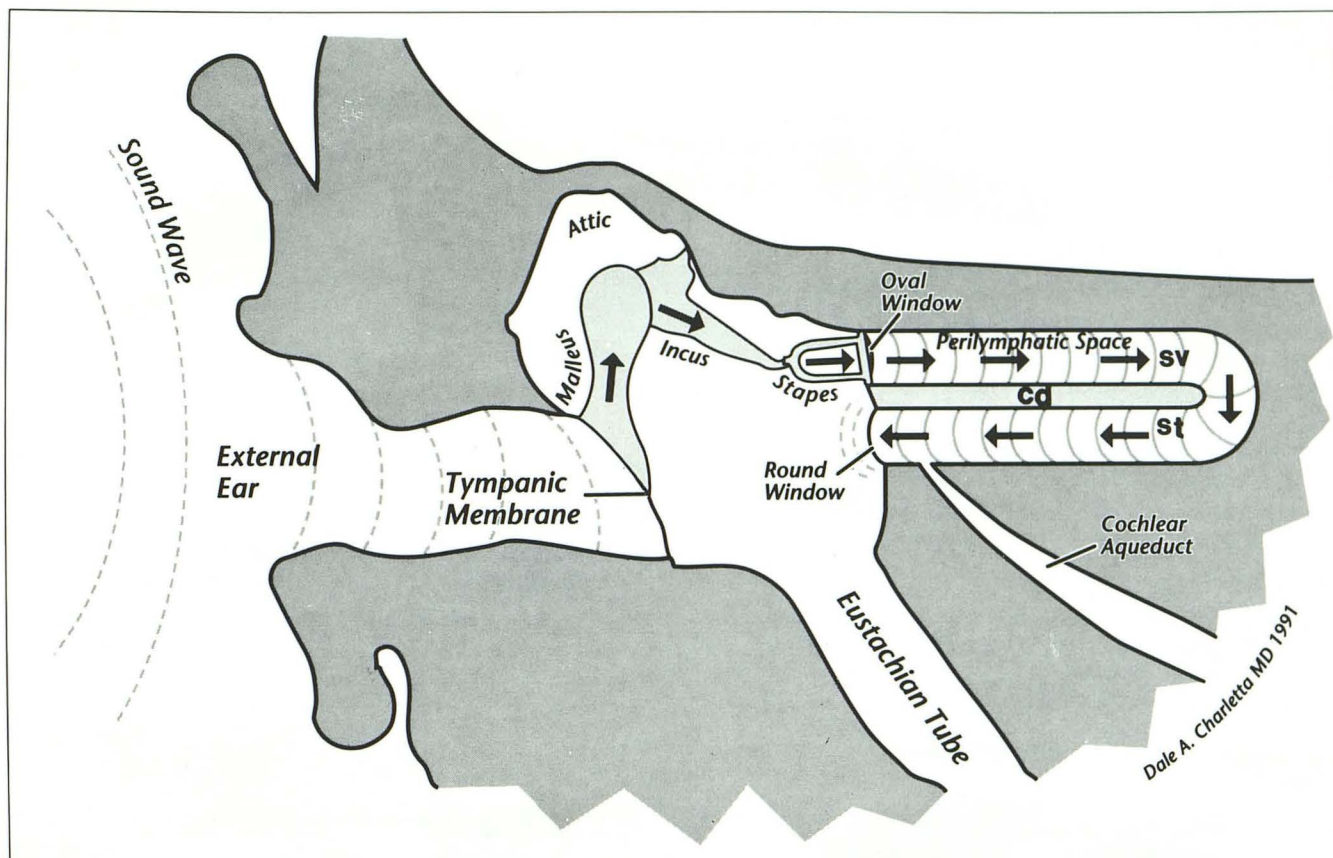
Fig. 10. Schematic representation of the three subdivisions (external, middle, and inner) of the ear. Vibrations of the sound-waves are transmitted into the scala vestibuli (*sv*) by the footplate of the stapes at the oval window. The waves are transmitted across the cochlear duct (*cd*) to the scala tympani (*st*), causing the round window membrane to move in the opposite direction to the oval window. The cochlear aqueduct connects the subarachnoid space to the scala tympani of the perilymphatic space. A widely patent cochlear aqueduct results in a stapes gusher during stapedectomy.
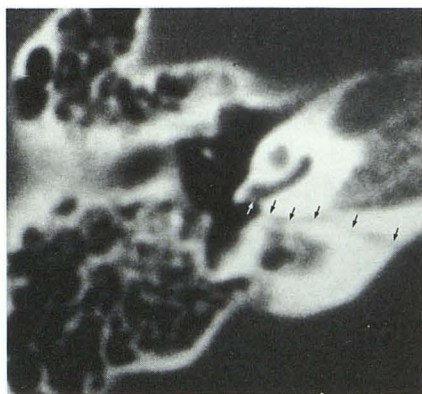

Fig. 11. Cochlear aqueduct. Axial CT shows the round window membrane (*white arrow*) and cochlear aqueduct (*black arrows*). Normal cochlear aqueduct has a filiform appearance in its outer portion. In this case, the cochlear aqueduct is slightly prominent because its outer portion is well visualized.

neural hearing loss may be the initial sign of a congenital defect. If the malformation is unilateral, the dysplasia may go undiagnosed for years (10). Patients with normal hearing have been

reported occasionally, but more frequently, the Mondini ear is clinically unreactive, having neither cochlear nor vestibular functions (10). When a CSF leak develops, findings depend on the functional status of the involved ear. If there is residual auditory function, a sudden or fluctuating sensorineural hearing loss may occur. With residual vestibular function, the patient may experience vertigo and have a positive fistula test (10). A CSF effusion in the middle ear may cause rhinorrhea or otorrhea. More commonly, however, the effusion is asymptomatic and does not cause a perceptible conductive hearing loss, because the ear is nonfunctional. The gravity of the problem remains undetected until a myringotomy is performed, releasing a profuse flow of clear fluid. Alternatively, the CSF effusion may remain undiagnosed until meningitis develops as a result of retrograde infection through the defect in the otic capsule. Ohlms et al (10) reported three infants with CSF otorrhea and recurrent meningitis due to Mondini dysplasia. They reviewed an additional
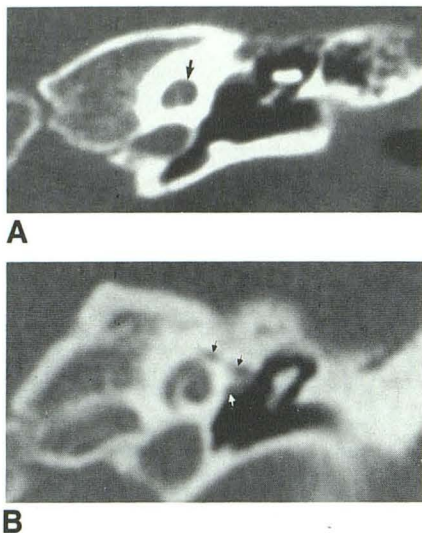
A

B

Fig. 12. Mondini anomaly. A, Coronal CT shows an "empty" cochlea (*arrow*). B, Normal cochlea for comparison. Notice that, in A, the bony partitions between apical, middle, and basal coils are missing. B, notice the geniculate portion of the facial canal (*black arrows*) and the semicanal for the tensor tympani (*white arrow*).
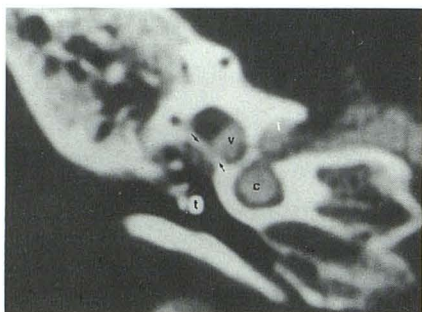

Fig. 13. Mondini anomaly. Coronal CT obtained following intrathecal injection of iodinated contrast agent, shows contrast in the dilated cystic cochlea (*c*) and cystic vestibule (*v*). Notice the abnormally wide oval window (*arrows*) with contrast present at the oval window niche. *t*, ventilating tube. (Reprinted courtesy of Edward M. Burton, MD.)
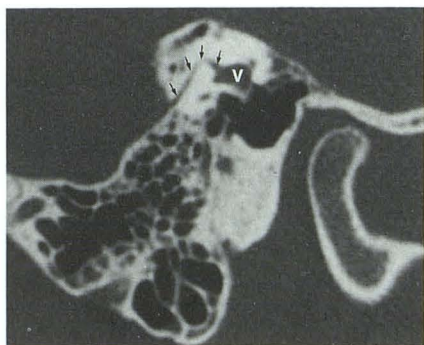
Fig. 14. Normal vestibular aqueduct. Direct sagittal CT shows the entire course of the vestibular aqueduct.
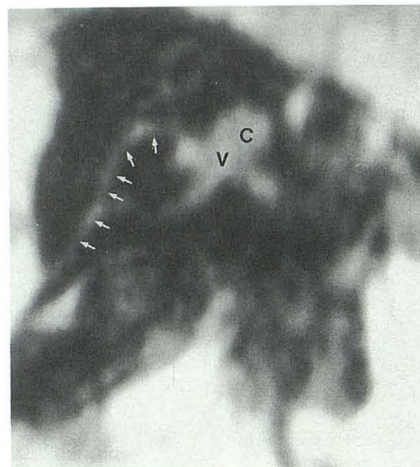
Fig. 15. Normal endolymphatic duct and sac. Sagittal T1W MR scan shows the cochlea (*C*), vestibule (*V*), and endolymphatic duct and sac (*arrows*).

Fig. 16. Enlarged left vestibular aqueduct. Axial CT shows the normal right vestibular aqueduct (*small arrows*), common crus (*curved arrow*), and the superior semicircular canal (*long arrows*). Notice enlarged *left* vestibular aqueduct. The left superior semicircular canal is also enlarged.

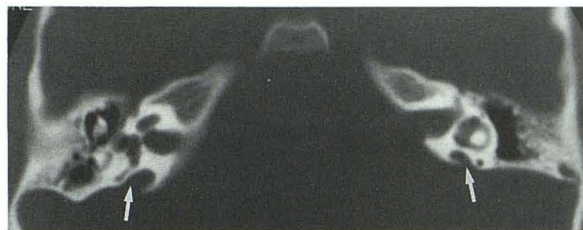Fig. 17. Enlarged vestibular aqueduct. Direct sagittal CT scan shows enlargement of the vestibular aqueduct (*arrows*).

Fig. 18. Bilateral enlarged vestibular aqueducts. Axial CT shows enlargement of both vestibular aqueducts (*arrows*).
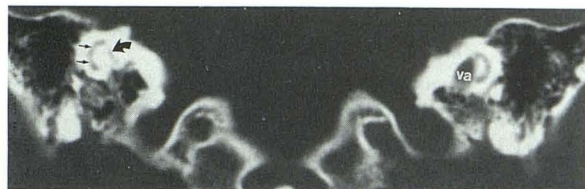

Fig. 19. Enlarged vestibular aqueduct. Coronal CT shows normal right posterior semicircular canal (*arrows*) and normal right vestibular aqueduct (*curved arrow*). Note the enlargement of the left vestibular aqueduct.

39 patients with Mondini dysplasia and meningitis reported in the literature. Sixty-seven percent had a unilateral malformation and profound congenital unilateral sensorineural hearing loss. Most had no vestibular symptoms, although formal vestibular testing was rarely performed. Two patients had 20 episodes of meningitis prior to surgery. All patients required surgical procedures to seal the CSF leak. Mondini dysplasia should be suspected in a child with congenital (often unilateral) profound sensorineural hearing loss or peripheral vestibular weakness, and recurrent meningitis. Associated abnormalities such as Klippel-Feil syndrome, Wildervanck syndrome, Alagille syndrome, Waardenburg syndrome, Pendred syndrome, chromosomal trisomies, and DiGeorge syndrome may be present. Mondini dysplasia can also result from treatment with teratogenic drugs, such as thalidomide, or can occur as an isolated finding (10). Contrast studies may identify the location of the leak. Surgical intervention is required to avoid further complications, and to diminish morbidity and mortality (10). The method of closure of the fistula is dictated by the functional status of the ear. If hearing loss is present, it may be possible to stop the leak with a fascial graft (10). If a stapedectomy is performed, a stapes gusher may occur and preclude a favorable outcome. When the ear is nonfunctional, the vestibule should be packed with fascia, muscle, or fat, and the oval window corked with bone (10). Alternatively, the middle ear cavity

can be obliterated. Occasionally, an intracranial approach is required to pack the meatus of the internal auditory canal or the cochlear aqueduct (27–29).

Congenital deafness may be associated with other systemic abnormalities, sometimes as a recognized syndrome and sometimes not (5, 8). Craniofacial dysostosis (Crouzon disease) and acrocephalosyndactylism type I (Apert disease) can be associated with hearing loss. In these two conditions, there is marked derangement of the middle cranial fossa with a peculiar upward and medial angulation of the petrous portion of the temporal bone (30). The middle ear and ossicles are usually normal, but ossicular deformity can occur. In most cases, the inner ear structures appear radiographically normal, although malformations in the form of large vestibules and short, stubby lateral and superior semicircular canals can be found in a few patients (30).

In Usher syndrome, the temporal bone pathology is thought to be of the Alexander type, and, therefore, no tomographic abnormalities are seen. The main characteristics of this syndrome are progressive retinitis pigmentosa, congenital severe to moderate sensorineural hearing loss, and mental retardation and epilepsy (4, 6, 8).

Patients with Waardenburg syndrome may have congenital sensorineural hearing loss (4, 8). The primary features of Waardenburg syndrome include lateral displacement of the medial canthi of the eyes (telecanthus), producing the appearance of hypertelorism, flat and broad nasal root, partial or total heterochromia of the iris, partial albinism in the form of a white forelock, and congenital deafness (4, 8). The pathologic condition of the temporal bone has been described as atrophy of the organ of Corti and stria vascularis, with reduction of the number of cells in the spiral ganglion (4, 8). One would not expect any radiographic findings, so it is noteworthy that in our previous study (8), we could not identify the posterior semicircular canal in any of the three patients studied (8). Bilateral severely hypoplastic cochleas were found in one of our patients.

Patients with Klippel-Fell syndrome may also have vestibular dysfunction and sensorineural hearing loss (4, 8). Klippel-Feil syndrome consists of skeletal defects that may include cervicothoracic scoliosis with various degrees of cervical vertebral fusion, Sprengel deformity (congenital upward displacement) of the scapula, vestibular dysfunction, and profound sensorineural hearing loss (4, 8). On the basis of our five cases studied

tomographically (8), we may state that this syndrome can be associated with severe, but not characteristic or consistent dysplasia of the inner ear.

The primary features of Wildervanck syndrome (cervico-oculoacoustic syndrome) include facial asymmetry, torticollis, short neck with fused vertebrae, high scapula, ocular motility problems in terms of abducens palsy often with limited adduction, and congenital deafness (8, 31). Dysplasia of the inner ear (Mondini type) and ossicular deformity have been reported in these patients (8).

Alagille syndrome, first reported in 1975 (32), is a congenital condition characterized by seven main clinical features: 1) chronic cholestasis, 2) characteristic facial appearance, 3) cardiovascular abnormalities, 4) vertebral arch defect, 5) growth retardation, 6) mental retardation, and 7) hypogonadism. Recently, ocular and otologic abnormalities have also been reported (33). Severe anomalies have been observed in temporal bones of four individuals with Alagille syndrome (33). In all cases, both the bony and membranous structures of the posterior semicircular canal were partially or totally absent. In three ears, the bony and membranous structures of the anterior semicircular canal were also partially absent. The cochlea was observed to be shortened in only one case (33). The cochlear aqueduct may be absent, and the vestibular aqueduct may be enlarged (33). In the middle ear, anomalies observed in the ossicles included a bulky incus with bone marrow. The incudomalleal and incudostapedial joints were underdeveloped in most cases (33).

Large vestibular aqueducts are the most common congenital inner ear anomalies demonstrated on radiographic study. The vestibular aqueduct (Fig. 14) is a bony canal that extends from the medial wall of the vestibule to an outer opening in the posterior surface of the petrous bone. Through this canal courses the endolymphatic duct (Fig. 15). The best projections for visualization of the vestibular aqueduct by complex motion tomography are straight lateral and modified lateral (34–37). For visualization of the vestibular aqueduct by CT, we obtain high-resolution 1.5-mm axial sections of the temporal bone (Fig. 16), as well as direct sagittal (Fig. 14) or reformatted straight sagittal images and para-axial images along the long axis of the vestibular aqueduct. Nonvisualization of the aqueduct does not necessarily mean that it is not present. It is considered enlarged when its anterior-posterior diameter or lateral-medial dimension is 1.5 mm or greater.

Valvassori and Clemis (34) reported 30 cases with enlarged vestibular aqueducts; in 13 of these, the enlarged aqueduct was the only radiographic finding. In the other 17, there were also anomalies of one or more of the other inner ear structures. The most common associated anomaly was an enlarged and rounded vestibule; next, was abnormal appearance of the semicircular canals. Seven cases had hypoplastic cochleas. Valvassori and Clemis concluded that the enlarged vestibular aqueduct represents an arrested phase of inner ear development (34, 35).

In this series, the anteroposterior diameter of the enlarged vestibular aqueducts ranged from 2 to 7 mm, with a mean of 4 mm (Figs. 17–19). It is likely that other anomalies of the labyrinth are present in these cases but are limited to the membranous labyrinth and thus are not visible by CT and MR.

In seeming contradiction to the widely held belief that, because the outer ear develops completely separately from the inner ear, there is no correlation between middle and inner ear abnormalities, we have reported eight cases in which there were anomalies of both the middle and inner ears (8). Half of these patients with microtia and sensorineural hearing loss also had congenital facial nerve paralysis. It is well known that, in many microtic ears, the facial nerve has an aberrant and unpredictable course (8, 23). However, in these cases, apparently the nerve cannot even exit the cranium due to severe inner ear dysplasia. We suggest that the coexistence of microtia and inner ear anomalies is not as infrequent as has been thought, and we emphasize that radiographic evaluation of patients with microtia should be done before any reconstructive surgery is attempted.

Acknowledgments

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