Chapter 152: Congenital Malformations of the Inner Ear

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Development of the inner ear begins early during embryogenesis. By the end of the eighth week, the membranous labyrinth has assumed its characteristic convoluted shape (Streeter, 1906). Gradual ossification of the otic capsule develops around the membranous labyrinth and is essentially complete by birth (Streeter, 1918). Maturation of the sensory epithelium occurs long after formation of the membranous labyrinth, during the late second and early third trimester. By the twenty-sixth to twenty-eighth week of gestation, hair cell and auditory neural development are largely complete. Thus, the normal human fetus may be able to hear 2.5 to 3 months before birth.

Most inner ear malformations arise when formation of the membranous labyrinth is interrupted during the first trimester of pregnancy (Jackler et al, 1987a). This interruption may be either a result of inborn genetic error or a consequence of a teratogenic exposure during the period of inner ear organogenesis between the fourth and eighth week of gestation. Genetic errors may be either autosomal dominant or recessive and may manifest as sensorineural hearing loss (SNHL) alone or be associated with any of a number of syndromes (Beighton and Sellars, 1982; Konigsmark and Gorlin, 1976). Teratogenic influences known to affect inner ear organogenesis include in utero viral infection (for example, rubella), chemical teratogens (for example, thalidomide), and radiation exposure (Jorgensen and Kristensen, 1964). Abnormalities in otic capsule structure and deficiencies in the organ of Corti appear to arise as secondary effects of the earlier error in development of the membranous labyrinth. Derangement of the otic capsule ossification process alone does not appear to be a major mechanism in congenital hearing loss. Ossification of the labyrinthine lumen, however, is a common finding in early acquired deafness, typically arising as a consequence of meningitis.

Developmental damage to cochlear structure and function is not restricted to agents that cause gross structural malformations. Even in doses that are below those that would be ototoxic in adult species, aminoglycoside antibiotics administered in the animal equivalent of the human first trimester of pregnancy cause severe hearing losses in several species (Carlier and Pujol, 1980; Uziel, 1985; Raphael et al, 1983). This time frame corresponds to the maturation of the outer hair cells and initiation of the cochlear potentials. Human studies also document this. In 35 of the infants delivered from 72 women who received streptomycin prophylaxis for tuberculosis during the first 4 months of pregnancy, auditory deficits ranging from minor high-frequency threshold elevations to severe bilateral deafness were noted (Snider et al, 1980).

The risk is not restricted to the first trimester. Even at low doses, smaller hearing losses were seen when aminoglycoside antibiotics were administered during the human equivalent of the last two trimesters of pregnancy, which has obvious implications for use of these drugs in pregnant women and in premature infants who have not yet reached functional cochlear maturity.

Congenital anomalies of the inner ear may be considered in two broad categories: malformations with pathologic changes limtied to the membranous labyrinth and malformations that involve both the osseous and membranous labyrinth. This division has been chosen because of its clinical relevance. Only patients with malformed otic capsules have abnormal inner ear radiographs and therefore may be diagnosed during life. By inference, those children with congenital SNHL and radiographically normal inner ears may be assumed to possess anomalies limited to the membranous labyrinth or neural pathways. Although several types of membranous deformities have been described, their classification is not yet of clinical use, as differentiation requires histopathologic examination. Among deformities that affect the otic capsule, a variety of morphologic patterns may be recognized radiographically, and classification may have prognostic and even therapeutic importance.

Incidence of Congenital Inner Ear Malformations

It can be safely assumed that all children with congenital sensory hearing loss would have detectable abnormalities in their inner ears if they could be examined histologically. Because most children with profound bilateral deafness have radiographically normal inner ears, it can be inferred that malformations limited to the membranous labyrinth predominate. Jensen (1969) studied 62 children enrolled in a school for the deaf with polytomography and detected cochlear malformations in only 15%. By contrast, Unger and Shaffer (1981) evaluated a series of 104 deaf students with polytomography and found no cochlear anomalies that were clearly attributable to developmental arrest. In a recent series of 234 children who had SNHL of varying degrees of severity, Reilly (1989) found cochlear anomalies in only 4% of those evaluated by high resolution computed tomography (CT) scanning. In unilateral profound deafness, Everberg (1960) detected only a 2% incidence of cochlear malformation among the 122 children studied. In analyzing each of these studies, children with radiographic signs of labyrinthine ossification were not included in the calculation of incidence of congenital malformation, as this finding is usually a marker for acquired deafness, typically due to meningitis.

The incidence of deformities of the semicircular canals (SCCs) and inner ear aqueducts has been less well studied than that of cochlear deformities. In a recent series of patients with radiographically detectable malformations of the inner ear, the cochlea was involved in 76%, the semicircular canals in 39%, and the vestibular aqueduct in 32% of ears (Jackler et al, 1987a). These data total more than 100%, as many cases demonstrate abnormalities of more than one portion of the inner ear. In recent years, a heightened awareness of vestibular aqueduct enlargement, combined with the greater sensitivity of axiat CT scanning in demonstrating this deformity, has led to a substantial increase in its detection. The recent rapid accrual of cases by clinicians interested in inner ear malformations suggests that enlargement of the vestibular aqueduct will ultimately prove to be the most common radiographically detectable inner ear anomaly (Jackler and De La Cruz, 1989).

Among deaf children with radiographically normal inner ears, pathologic studies indicate that cochleosaccural dysplasia (Scheibe's dysplasia) is by far the most common deformity (Schuknecht, 1980b). Due to the paucity of pathologic specimens available for examination, it is impossible to estimate the relative frequency of the various membranous malformations.

A Classification of Inner Ear Malformations

The traditional nomenclature used to describe congenital anomalies of the inner ear involves a confusing array of eponyms that stem from the first reports, usually by eighteenth or nineteenth century authors, of the various morphologic patterns. In this chapter, a descriptive classification system is used along with the traditional eponyms in an attempt to make this topic more logical, easier to learn, and more clinically relevant (see box). In membranous malformations this classification is based on the histopathologic changes in the inner ear, whereas in the combined osseous/membranous deformities, radiographic appearance is used to distinguish among the various entities (Jackler et al, 1987a). Correct use of the terminology used in pathoembyrology is important, as these terms are frequently applied imprecisely in the earlier literature. Key terms are 1) *aplasia*, complete lack of development; 2) *hypoplasia,* incomplete development; and 3) *dysplasia,* aberration in development.

Box: **A classification of congenital malformations of the inner ear**

I. Malformations limited to the membranous labyrinth

A. Complete membranous labyrinthine dysplasia (Siebenmann-Bing)

B. Limited membranous labyrinthine dysplasia

1. Cochleosaccular dysplasia (Scheibe)

2. Cochlear basal turn dysplasia (Alexander)

II. Malformations of the osseous and membranous labyrinth

A. Complete labyrinthine aplasia (Michel)

B. Cochlear anomalies

1. Cochlear aplasia

2. Cochlear hypoplasia

3. Incomplete partition (Mondini)

4. Common cavity

C. Labyrinthine anomalies

1. Semicircular canal dysplasia

2. Semicircular canal aplasia

D. Aqueductal anomalies

1. Enlargement of the vestibular aqueduct

2. Enlargement of the cochlear aqueduct

E. Internal auditory canal abnormalities

1. Narrow internal auditory canal

2. Wide internal auditory canal.

Malformations Limited to the Membranous Labyrinth

In this group of inner ear anomalies, which account for over 90% of congenital deafness, the bony labyrinth is normal (Altman, 1953; Lindsay, 1973; Ormerod, 1960; Schuknecht, 1974). In its severest form, membranous dysplasia involves the entire labyrinth, including the cochlea, SCCs, utricle, and saccule. Membranous labyrinthine dysplasia may also be limited, involving only a portion of the inner ear.

Complete membranous labyrinthine dysplasia (Bing-Siebenmann)

Complete membranous labyrinthine dysplasia was first described by Siebenmann and Bing (1907) and is extremely rare. It has been reported in association with the cardioauditory (Jervell and Lange-Neilsen) and Usher's (Friedmann et al, 1966) syndromes.

Limited membranous labyrinthine dysplasia

Cochleosaccular dysplasia (Scheibe)

Incomplete development of the pars inferior is the most frequent histopathologic finding in congenital deafness. It was first described by Scheibe (1892) and is commonly known as cochleosaccular dysplasia. The spectrum of its pathologic findings in this anomaly, which is confined to the cochlea and the saccule, have been well described (Beal et al, 1967; Bergstrom, 1980; Friedmann, 1974; Nomura and Kawabata, 1980; Ormerod, 1960). The organ of Corti is either partially or completely missing. The cochlear duct is usually collapsed, with Reissner's membrane adherent to the limbus. Less commonly it is distended, presumably as a result of endolymphatic hydrops. The stria vascularis is typically degenerated and may contain colloidal inclusions. Schuknecht (1974) describes characteristic strial changes consisting of aplasia alternating with regions of hyperplasia and gross deformity. Cochlear changes may be severe in the base turn and gradually lessen in intensity towards the apex, or they may be severe throughout. The saccule is usually collapsed and has degenerated sensory epithelium. In cochleosaccular dysplasia, the SCCs and utricle are normal. Auditory neuronal survival is variable, but may remain normal into adulthood, at least in some cases. Cochleosaccular dysplasia also has been demonstrated in a number of animal species, including the deaf white cat, dalmatian dogs, and various mouse mutants (Steel and Bock, 1983).

Cochlear basal turn dysplasia (Alexander)

Dysplasia limited to the basal turn of the cochlea was first described by Alexander (1904) and may be related to familial high frequency SNHL. No descriptions of membranous labyrinthine dysplasia limited to the pars superior was found in an extensive review of the literature. This outcome is not surprising as such individuals probably are asymptomatic. They would have normal hearing and, presumably, would have compensated for their congenital vestibular deficit.

Malformations of the Membranous and Osseous Labyrinth

Congenital anomalies of the inner ear that deform the otic capsule are of special interest to the clinician, as they may be recognized during life through radiographic imaging. As discussed previouly, only about 5% to 15% of congenitally deaf individuals demonstrate radiographically anomalous inner ears. The clinical manifestations and natural history of these deformities are highly variable. Although some individuals are deaf from birth, most maintain some residual hearing into adulthood. Slowly progressive deterioration of hearing during childhood, with eventual stabilization, is common. Sudden decrements in hearing are frequent and may appear to be spontaneous or may be triggered by head trauma, even minor in nature. Presumably, most of these are secondary to either internal fistulization secondary to membrane rupture within the cochlea with admixture of perilymph and endolymph, or to external fistulization to the middle ear. Fluctuant hearing loss is unusual in these patients, and endolymphatic hydrops is an atypical finding. In some patients, hearing may be best preserved in very high frequencies (> 8000 Hx), which are not measured by conventional audiometry (Feghali et al, 1985). Residual ultraaudiometric hearing should be suspected in hearing-impaired children who manifest substantially better auditory function than would be predicted by pure tone results in the speech frequencies (Berlin et al, 1978). Vestibular symptoms, which are occasionally severe, are present in about 20% of patients (Jackler et al, 1987a). A few individuals suffer vertigo with exposure to loud sounds, the so-called *Tullio phenomenon* (Kwee, 1976).

A wide variety of morphologic patterns of inner ear malformation has been observed radiographically and may involve the cochlea, SCCs, and/or vestibular aqueduct (Fig. 152-1) (Curtin, 1988; Jackler et al, 1987a; Mafee et al, 1984; Murata et al, 1987; Olson et al, 1982; Phelps and Lloyd, 1983; Phelps, 1990). Similar diversity has been observed on histologic analysis (Johnsson et al, 1984; Lindsay, 1973; Monsell et al, 1987; Paparella, 1980; Sando et al, 1984; Schuknecht, 1980a). The majority of combined osseous and membranous malformations appears to arise from a premature arrest in the development of one or more components of the inner ear (Fig. 152-2). The strongest evidence for this theory comes from the resemblance of most malformed inner ears to the appearance of the inner ear during embryogenesis, particularly between the fourth and eighth week of gestation. As a general rule, the earlier the developmental arrest the more severe the deformity and the worse the hearing.

Other anomalies cannot be explained by a premature arrest in development alone and appear to arise from an aberrant embryologic process. An example of this type of anomaly is a cochlea of normal length but abnormal size or coiling geometry (Johnsson, 1984). In humans, the inner ear is adult size at birth and shows strikingly little variation in size among individuals. Pappas et al (1990) suggested that some children with congenital SNHL and apparently normal inner ear morphology on CT scan possess subtle abnormalities in the dimensions of inner ear structures. They propose that such dimensional variations arise from a teratogenic insult during the second or third trimester, after the membranous labyrinth has formed but before it has reached adult size. Further study is needed to determine the clinical relevance of these observations.

Whereas some inner ear malformations involve only one portion of the inner ear, many patients have a combination of anomalies involving more than one component. Between the fourth and fifth weeks of development, the spherical otocyst develops three buds that ultimately form the cochlea, SCCs, and the vestibular aqueduct (Fig. 152-2). An inner ear malformation may be limited to one of these anlages, may involve a combination of two, or may even affect all three.

The frequent coexistence of deformities involving the cochlea, SCCs, and vestibular aqueduct has several possible explanations: (1) the anomaly is genetically predetermined, (2) an insult to the embryo occurred before the fifth week, and (3) each of the buds were susceptible to some teratogenic influence at a later stage of development. A majority of inner ear malformations are bilateral and symmetric. In cases where radiographs detect an anomaly on only one side, the opposite "normal" inner ear has a hearing loss in approximately 50% of cases (Jackler et al, 1987a).

Before the evolution of high resolution imaging technology, clinicians and histopathologists alike tended to lump together these malformations under the term *Mondini's dysplasia,* after the first report by Carlo Mondini (1791). Before the Academy of Sciences of the University of Bologna, Mondini described the inner ear findings in a deaf 8-year-old boy who was struck on the foot by a wagon and later died of gangrene. The cochlea possessed only one and on half turns and had a hollow apical cavity. An enlarged vestibule and vestibular aqueduct were also present. This deformity is the most common form of cochlear anomaly (Table 152-1). Although numerous other distinct anatomic patterns of inner ear malformation are discernible radiographically and histologically, many continue to use the term Mondini's dysplasia to describe all of them. To avoid a confusing and overly broad nomenclature system, this term is best reserved for describing the particular subtype of cochlear malformation first described by Mondini, whether or not it is associated with other inner ear malformations.

Table 152-1. Relative incidence of cochlear malformations

Incomplete partition (Mondini)	55%
Common cavity	26%
Cochlear hypoplasia	15%
Cochlear aplasia	3%
Complete labyrinthine aplasia (Michel)	1%.

Complete labyrinthine aplasia (Michel)

The severest deformity of the membranous and osseous labyrinth, complete labyrinthine aplasia, was first described by Michel (1863). This malformation is exceedingly rare. Presumably, a developmental arrest occurs before the formation of an otic vesivle, resulting in a complete absence of inner ear structures. Complete labyrinthine aplasia has been reported in association with anencephaly and thalidomide exposure (Jorgensen and Kristensen, 1964; Lindsay, 1973). A recent purpoted case of Michel's dysplasia actually described a cystic inner ear of the common cavity type (Kavanagh and Magill, 1989). The incidence of complete labyrinthine aplasia is overestimated in the radiographic literature because it is confused with labyrinthine ossification. In the latter condition, which is usually acquired during life, a sizable and dense otic capsule is present radiographically. In complete labyrinthine aplasia, the otic capsule is entirely absent. Such ears are of course uniformly deaf.

Cochlear anomalies (see fig. 152-1)

Cochlear aplasia

In this deformity the cochlea is completely absent, presumably as a result of an arrest in the development of the cochlear bud at the fifth week. This morphologic pattern is rare. Radiographically, only a vestibule and SCCs (usually deformed) are present. To differentiate this anomaly from labyrinthine ossification it is necessary to assess the amount of otic capsule bone anterior to the internal auditory canal. In cochlear aplasia, the otic capsule is absent, whereas in osseous obliteration it is dense and of normal dimensions. Ears with cochlear aplasia are devoid of auditory function.

Cochlear hypoplasia

An arrest during the sixth week results in a hypoplastic cochlea consisting of a single turn or less. This deformity comprises approximately 15% of all cochlear anomalies. Radiographically, a small bud of variable length (usually 1 to 3 mm) protrudes from the vestibule (Figs. 152-3 and 152-4). The vestibule is frequently enlarged with accompanying semicircular malformations in about half of faces. Small cochlear lacking a modiolus or other internal architecture have been described histologically (Beal et al, 1967; Johnsson et al, 1984; Schuknecht, 1980a). Hearing is variable in these ears and may be remarkably good considering the minute size of the cochlea. The variability of hearing presumably is accounted for by degree of membranous labyrinthine development within the truncated cochlear lumen.

Incomplete partition (Mondini)

Arrest at the seventh week stage yields a cochlea that has only 1.5 turns. This is the most common type of cochlear malformation, accounting for over 50% of all cochlear deformities. Radiographically, the cochlea is smaller than normal and partially or completely lacks an interscalar septum (Fig. 152-5). Although the usual cochlea measures 8 to 10 mm vertically, it is typically in the 5 to 6 mm range with incomplete partition deformity. Care must be exercised in counting the number of cochlear turns radiographically, as this may be difficult to determine even on high-resolution CT scans. The radiographic diagnosis depends more on cochlear size and the absence of a scalar septum than on the number of cochlear

turns perceived. Histologically, incomplete partition appears to be the radiographic correlate of classical Mondini's dysplasia (Fig. 152-6). In numerous reported cases a small cochlea with 1.5 turns possessing an apical scala communis due to deficiency in the osseous spiral lamina has been described (Beal et al, 1967; Johnsson et al, 1984; Paparella, 1980; Shuknecht, 1980a). Organ of Corti development is variable as is auditory neural population. As might be expected, auditory function is also variable, ranging from normal to profound deafness. The mean hearing threshold (three tone average) in a group of 41 incomplete partition ears was 75 dB (Jackler et al, 1987a). SCC deformities accompany incomplete partition of the cochlea in approximately 20% of cases.

Common cavity

A deformed inner ear in which the cochlea and vestibule are confluent, forming an ovoid cystic space without internal architecture, may be explained by an arrest at the fourth week otocyst stage. Alternatively, it may result from aberrant development at a later stage. An empty ovoid space typically longer in its horizontal dimension is seen radiographically. Although the size of the cyst may vary, it averages 7 mm vertically and 10 mm horizontally. It is quite easy to misdiagnose a dysplastic lateral SCC as a common cavity deformity. The key to differentiating between them is that a common cavity cochlea lies predominantly anterior to the internal auditory canal (IAC) on axial plane CT, and a dysplastic vestibular system lies posterior to it. Histologically, an ovoid or spherical smooth-walled cystic cavity containing a primordia of the membranous labyrinth has been described (Jorgensen and Kristensen, 1964; Paparella, 1980; Schuknecth, 1980a). Sensory and supporting cells may be differentiated into recognizable organs of Corti that are scattered peripherally around the walls of the cyst. Neural population is usually sparse or absent. Hearing is usually, but not invariably, poor.

Labyrinthine anomalies (Fig. 152-7)

Semicircular canal dysplasia

Dysplasia of the lateral SCC is a common type of inner ear malformation. Approximately 40% of ears with a malformed cochlea will have an accompanying dysplasia of the lateral SCC (Jackler et al, 1987a). Occasionally, dysplasia of the lateral SCC exists as the sole inner ear malformation. During the sixth week of development the budding SCC forms a semicircular evagination from the vestibular anlage. The central portion of the pocket-shaped protrusion adheres leaving a peripheral semicircular tube. When this central adhesion fails to occur, SCC dysplasia results (Figs. 152-2 and 152-8). SCC dysplasia occasionally takes the form of a small bud, rather than the more common half-disk shape, presumably due to a slightly earlier timing of the developmental insult. The lateral SCC is deformed more often than the posterior or superior SCC, apparently because it forms earlier in embyrogenesis. The typical radiographic appearance of SCC dysplasia is that of a short, broad cystic space confluent with the vestibule (Fig. 152-9).

There are numerous histologic descriptions of SCC dysplasia in the literature (Johnsson et al, 1984; Monsell et al, 1987; Paparella, 1980; Sando et al, 1984; Schuknecht, 1980a). The half-disk-shaped cavity may contained a rudimentary crista ampullaris. The utricle and saccule may be distended, collapsed, or entirely absent. Caloric responses in SCC dysplasia are functionally absent or reduced in most cases, but a few may have normal responsiveness (Jackler et al, 1987a). Ears with malformations that are limited to the vestibular system often habve normal or near-normal hearing. Although conclusive data are lacking, SCC dysplasia appears to have an association with conductive hearing loss due to congenital stapes fixation.

Semicircular canal aplasia

SCC aplasia is only one-fourth as common as SCC dysplasia (Jackler et al, 1987a). It is usually associated with cochlear anomalies (Paparella, 1980). Presumably, it arises from a failure in the development of the vestibular anlage before the sixth week.

Malformations of the Vestibular and Cochlear Aqueducts

Enlargement of the vestibular aqueduct

Recent experience suggests that enlargement of the vestibular aqueduct (VA) is the most common radiographically detectable malformation of the inner ear (Jackler and De La Cruz, 1989). In earlier literature its incidence was underestimated, partially due to a lack of awareness, but mostly because it could be visualized only by lateral tomography at a time when most studies were confined to the anteroposterior plane. The advent of high-resolution CT scanning in the axial plane has made assessment of the VA much easier.

The VA derives from a diverticulum formed in the wall of the otocyst during the fifth week. The aqueduct begins as a short, broad pouch, but gradually elongates and thins until it achieves its characteristic "J" shape of adulthood (see Fig. 152-2) (Anson, 1965). A premature arrest in development produces an abnormally short and broad VA (Fig. 152-10) (Becker et al, 1983; Emmett, 1985; Hill et al, 1984; Jackler and De La Cruz, 1989; Levinson et al, 1989; Valvassori, 1983). In many cases, VA enlargement accompanies malformation of the cochlea and/or SCCs. It also may be the sole radiographically detectable abnormality of the inner ear in a child with hearing loss. This condition is commonly referred to as the *large vestibular aqueduct syndrome*, after the first description of Valvassori and Clemis (1978). The diameter of the normal VA, when measured half way between the common crus and its external aperture, is between 0.4 and 1.0 mm. Enlargement of the VA is diagnosed when its diameter exceeds 2.0 mm, although enlarged VAs may exceed 6 mm wide. Histologically, the sac and aqueduct are thin-walled and lacking in both the vascularity and rugose features thought to be important for physiologic functgion (Gussen, 1985). The large VA syndrome is typically bilateral. Children are usually born with normal or mildly impaired hearing that gradually deteriorates through childhood into adolescence and early adulthood. Hearing levels are variable, although at least 40% eventually develop profound sensory hearing loss. As with other inner ear malformations, there is a tendency to suffer sudden decrements of hearing, particularly after head trauma.

Enlargement of the cochlear aqueduct

Enlargement of the cochlear aqueduct (CA) is frequently mentioned in the otologic literature because of its purported association with stapedectomy gusher and transotic cerebrospinal fluid (CSF) leak. Despite an active interest in the field and an extensive review of published radiographic images, I have never seen a radiograph that convincingly demonstrated CA enlargement. Most cases presented as enlargment of the CA are misinterpretations of the wide internal funnel that opens into the posterior fossa (Narcy et al, 1989; Phelps, 1986). In normal subjects, the radiographic diameter of this aperture averages 3 to 4 mm but ranges from radiographically invisible to over 6 mm (Fig. 152-11) (Bhimani et al, 1984; Muren and Wilbrand, 1986; Rask-Anderson et al, 1977). For enlargement of the CA to be diagnosed radiographically, the intraosseous portion coursing toward the vestibule must be enlarged beyond 1 mm, the practical resolution limit of contemporary CT scanners. If analogous criteria to enlargement of the VA are used, then an enlarged CA must have a diameter exceeding 2 mm throughout its course between the inner ear and posterior fossa.

The preponderance of evidence suggests that the human CA is functionally patent in most persons (Palva and Dammert, 1969; Rask-Anderson et al, 1977). However, in most individuals it can neither transmit sudden large pressure changes to the inner ear nor allow free flow of spinal fluid in any significant quantity, for example, when there is outflow of perilymphatic fluid from perilymphatic sac or an open vestibule. This reflects two anatomic features of the aqueduct: a narrow diameter of the bony channel and the presence of a fibrous tissue meshwork occupying and baffling the lumen. Recently, Schuknecht and Reisser (1988) evaluated over 1400 temporal bones in the Massachusetts Eye and Ear Infirmary collection and found no CA that exceeded 0.2 mm at its narrowest point. This series included 29 congenitally malformed inner ears. Notably, the CA was absent or nonpatent in 21 of these dysmorphic ears.

In some individuals there is less than normal or no fibrous tissue within the lumen, and the bony diameter is wider than normal. An opportunity could then exist for free flow of CSF to come from the oval window via the cochlear aqueduct. According to Poiseuille's law, fluid flow through a tube varies with the fourth power of the radius, suggesting the possibility of free flow if an individual had a large-diameter unimpeded channel, as discussed by Allen (1987). Several histologic studies of congenital inner ear malformations have detected slightly large CAs, particularly at its lateral orifice at the vestibule (Sando et al, 1984; Schuknecht, 1980a). Even in these malformed inner ears, however, the aqueduct diameter remained well in the sub-millimeter range. From a clinical standpoint, a CA less than 1 mm in diameter is undetectable radiographically. Despite the theoretical possibility that a widely patent CA may cause a stapedectomy gusher, abnormal connections between the internal auditory canal and vestibule appear to be etiologic in the majority of cases (Flood et al, 1985; Glasscock, 1973; Phelps, 1986; Schuknecht and Reisser, 1988). Recently, stapes gusher has been associated with an X-linked inheritance pattern (Cremers et al, 1985).

Developmental Anomalies of the Internal Auditory Canal

Narrow internal auditory canal

A narrow IAC may indicate a failure of eighth nerve development. When a patient has normal facial function and an IAC less than 3 mm in diameter, it is likely that the bony canal transmits only the facial nerve (Fig. 152-12). A narrow IAC may accompany inner ear malformations or may be the sole radiographically detectable anomaly in a deaf child. A narrow IAC has been considered a relative contraindication to cochlear implantation (Jackler et al, 1987b; Shelton et al, 1989). Experience with electrical stimulation in several patients have shown that they develop facial pain and twitching without useful auditory sensation.

Wide internal auditory canal

Unlike congenital narrowness of the IAC, a congenitally large canal may be an incidental finding in normal individuals (Fig. 152-13). When a large IAC (> 10 mm in diameter) accompanies a malformation of the inner ear, it does not, as an independent variable, correlate with the level of hearing. The importance of detecting enlargement of the IAC is its association with spontaneous CSF leak and gusher during stapes surgery (Flood et al, 1985). Because hearing after stapedectomy complicated by perilymphatic (CSF) gusher is frequently poor, a CT scan should be obtained before undertaking stapedectomy for congenital fixation. Dilatation of the IAC, especially when the partition between the lateral end of the canal and inner ear appears deficient, should contraindicate stapedectomy.

Cerebrospinal Fluid Leakage and Meningitis

Congenitally malformed inner ears may be a source of CSF otorhinorrhea (Phelps, 1986; Quiney et al, 1989). For CSF leakage to occur, two abnormal connections must be present: one between the subarachnoid space and the inner ear, and a second between the inner ear and middle ear. The most common pathologic interconnection between the subarachnoid space and the inner ear is through the fundus of the IAC. Histologically, large defects in the modiolar end of the IAC have been shown to exist in 10% of dysplastic ears (Schuknecht and Reisser, 1988). Leakage may occur less often around the facial nerve at the lateral end of the IAC (Barcz et al, 1985; Gacek and Leipzig, 1979). The junction between the IAC and the inner ear may be evaluated with CT (Fig. 152-13). A well-defined bony plate normally partitions the distal IAC from the labyrinthine lumen. In a number of well-documented cases of CSF leakage, radiopaque contrast placed into the cerebellopontine angle cistern could be traced through the IAC into the inner ear (Barcz et al, 1985; Burton et al, 1990; Kaufman et al, 1969; Park et al, 1982; Rockett et al, 1964). A second potential pathway between the subarachnoid space and inner ear is the CA. As discussed previously, little evidence suggests that this pathway is a source of vigorous CSF leakage. In one published report, a CT metizamide cisternogram is said to have demonstrated a large CSF leak via the CA; however, the published image showed the defect to be far anterior and superior to the region of the CA (Park et al, 1982). A direct route between the subarachnoid space and middle ear, bypassing the inner ear, may exist in rare cases. Hyrtl's fissure, a congenital cleft that runs between the hypotympanum and posterior cranial fossa, passing in proximity to the jugular bulb, has been reported to be a source of CSF leakage (Gacek and Leipzig, 1979).

The pathologic interconnection between the inner ear and middle ear most frequently is at the oval window. Numerous reports have described defects in the central portion of the footplate with a protruding membrane, most probably arachnoid (Fig. 152-14) (Barcz et al, 1985; Herther and Schindler, 1985; Luntz et al, 1986; MacRae and Ruby, 1990; Ohlms et al, 1990; Parisier and Birken, 1977; Quiney et al, 1989). In other cases the defect is adjacent to the footplate, particularly just anterior to it. Much less frequently, leakage may occur via the round window or a fissure on the promontory (Quiney et al, 1989). Schuknecht and Reisser (1988) evaluated the oval and round windows in 29 ears with congenital malformations of the inner ear. On histologic examination, the stapes were absent in 4, fixed in 6, and normal in 15 cases. The footplates in 4 cases were noted to have defects that were bridged by thin membranes only. The round windows were normal in 28 and abnormal in only 1 (bony closure). No deficiencies in the round window membranes were noted.

The primary clinical importance of CSF leakage is the risk of meningitis. There have been numerous reports of meningitis complicating congenital malformation of the inner ear (Barcz et al, 1985; Herther and Schindler, 1985; MacRae and Ruby, 1990; Ohlms et al, 1990; Parisier and Birkin, 1977; Park et al, 1982; Quiney et al, 1989). Recurrent bouts of meningitis are typical and recognition of the inner ear as the source frequently comes only after several episodes have occurred. Acute otitis media appears to be the bacterial source in most cases. All children with recurrent meningitis without an obvious cause should undergo CT scans of the inner ear to exclude inner ear malformation. Even children with a single episode of meningitis should have a postrecovery audiogram, followed by a CT scan if unilateral or bilateral SNHL is discovered. The causative organisms in a review of 24 reported cases have been *Streptococcus pneumoniae* (71%), *Haemophilus influenzae* (33%), and beta-hemolytic streptococci (8%). The total exceeds 100% due to the incidence of polymicrobial infection.

Surgical closure of transotic CSF leakage may be attempted at four anatomic levels: (1) the posterior cranial fossa, (2) the dysplastic inner ear, (3) the middle ear window, and (4) the eustachian tube. In an ear with residual hearing, tympanotomy and overlay grafting of the site of leakage with a connective tissue graft is indicated as a first attempt. Unfortunately, recurrent leakage is common. When this technique fails and a useful amount of hearing persists, posterior fossa craniotomy with placement of a muscle plug in the IAC or connective tissue graft over the fistulous tract may prove successful and spare hearing (Park et al, 1982). When the hearing is poor, a direct approach to the dysplastic inner ear is indicated. After tympanotomy and removal of the footplate, an oversized piece of muscle may be used to obliterate the vestibule (Herther and Schindler, 1985; Schuknecht, 1980a; Quiney et al, 1989). If the anatomy is unfavorable for this maneuver, a postauricular translabyrinthine approach to the IAC should prove effective. Farrior and Endicott (1971) proposed a hypotympanic apprach to ablation of the cochlear aqueduct. However, this aqueduct has never been convincingly demonstrated to cause a high volume CSF leak. The final option, closure of the eustachian tube, is not a good choice for children. Following this procedure the anomalous CSF pathway is still open to the middle ear, a disadvantageous situation during an age when acute otitis media is common. Subsequent to the repair of any CSF leakage, the patient is placed on bed rest, with the head elevated 30% for several days. Acetazolamide (Diamox) is administered to reduce CSF production and fluids are restricted. Placement of a lumbar drain is a useful adjunctive measure, although it may be difficult to insert and maintain in young children.

Perilymphatic Fistula in Congenital Malformations of the Inner Ear

In contrast to CSF leakage, a perilymphatic fistula requires only one abnormal pathway: an interconnection of the inner ear with the tympanic cavity. In all probability, perilymph fistula and CSF leak are part of a spectrum of related disturbances of inner ear fluid homeostasis. Although separation of the two topics may be artificial pathophysiologically, it can be justified by the different clinical settings of the two problems. With CSF leakage, the cardinal clinical issues are gross fluid leakage and recurrent meningitis, whereas with perilymph fistulization, sudden or progressive hearing loss and vertigo are the primary manifestations. Much controversy surrounds the diagnosis and management of perilymphatic fistula (PLF) in children. The candidate group for exploratory tympanotomy is children with progressive or sudden SNHL. CT scan of the inner ear may demonstrate pneumolabyrinth, although this is rare (Fig. 125-15). Conservative surgeons have advocated exploring only children with radiographically abnormal inner ears who have a clear antecedent event of head trauma or barometric pressure change (Pappas et al, 1988). Others have espoused exploring all children with unexplained SNHL (Parnes and McCabe, 1987; Reilly, 1989; Supance and Bluestone, 1983). There is great variability in the number of fistulas "confirmed" at surgery. Pappas et al (1988) found only 4 fistulas in 36 ears (11%) explored for progressive SNHL during childhood. It is important to note that 50% of these ears had radiographically malformed inner ears. By contrast, Parnes and McCabe (1987) found fistulas in 20 of 26 children's ears (77%), only 6 of whom had inner ear anomalies radiographically. When performing a perilymph fistula exploration it is important to perform provocative maneuvers to render subtle perilymph flows more visible. These include performance of the Valsalva maneuver, using the Trendelenburg position, and compression of the jugular veins. Depending on one's point of view, many surgeons are either missing subtle or intermittent PLFs during explorations or overdiagnosing them by misinterpreting middle ear secretions or local anesthetic as perilymph. Most surgeons patch both oval and round windows with connective tissue whether or not a PLF was identified during exploration.

Rather than dwell on the controversy about how many fistulas are found, the critical observer should judge the effectiveness of the therapeutic intervention by analyzing its outcome relative to the natural history of the disease. In this regard, results of PLF exploration in children have been disappointing. In one series of 36 patients, hearing was better in 3, unchanged in 21, and worse in 12 (Pappas et al, 1988). Similar results have been obtained in other childhood PLF series. It would be difficult to argue that these data represent a significant improvement over the natural history of the inner ear disease. Congenital progressive SNHL is often characterized by long periods of stability interspersed with periods of rapid deterioration. Even a relative improvement in hearing may occur after an episode of sudden loss. It is apparent that most children with sudden or progressive SNHL have suffered from cochlear diseases other than PLF. Examples include hereditary progressive loss, viral labyrinthitis (for example, measles, mumps, cytomegalovirus), autoimmune inner ear disease (for example, Cogan's syndrome), and endolymphatic hydrops. Even in the situation of highest suspicion - sudden loss triggered by head trauma in a child with radiographically malformed inner ears - only a minority of patients demonstrate oval- or round-window fistulization. Many of these children probably lose hearing as a result of internal fistulization between the endolymphatic and perilymphatic space due to deficiencies of the osseous spiral lamina rather than due to external fistulization to the middle ear. A minority of children with sudden or progressive SNHL have prominent vestibular symptoms. Results with PLF repair in the relief

of vertigo have been quite good, although spontaneous recovery from vestibulopathy is common in this age group.

Inner Ear Malformations in Congenital Aural Atresia

The development of the external and middle ears is embryologically distinct from that of the inner ear. Anomalies of the lateral portion of the temporal bone typically result from derangement of the first or second branchial arches. Despite their embryologic separateness, congenital aural atresia may coexist with malformation of the inner ear (Jahrsdoerfer, 1978). In one study of atresia patients, 12% (8/66) had a radiographically detectable inner ear abnormality (Naunton and Valvassori, 1968). In this group, SCC abnormalities were most common, including some ears with normal bone conduction hearing. Cochlear anomalies were present in 5% (3/66) of these patients, all of whom had poor bone conduction hearing. It is an embryologic enigma that one patient with unilateral atresia had a malformed inner ear only on the contralateral side. In another polytomographic study of atretic ears, 8% (4/48) had SCC anomalies, and 2% (1/48) had a deformed cochlea (Potter, 1969). Based on a comprehensive review of the relevant literature, about 5% of atresia patients have cochlear deformities, whereas about 10% possess malformed SCCs. As a general rule, it is not advisable to undertake surgical reconstruction of atretic ears when they are associated with malformation of the inner ear. The deformed cochlea is more vulnerable to injury from vibratory trauma and there is an increased risk of inducing a sensory hearing loss. This surgery also carries with it a heightened risk of CSF leakage.

Evaluation and Management

High-resolution CT scan of the inner ears is recommended for all children with suspected congenital malformation of the inner ear, including all children with otherwise unexplained SNHL, regardless of whether it has existed from birth or occurred later. Scans are usually obtained at the time hearing impairment is first recognized. To adequately image the minute inner ear structures, we use thin slices of 1 or 1.5 mm thickness with 0.5 mm overlap (Jackler and Dillon, 1988). If only one plane of view is to be obtained, the axial orientation is preferred. Axial scans are recommended because they provide superior images of the vestibular aqueduct, a frequently malformed structure, which is difficult to visualize on coronal scans. When possible, both axial and coronal views should be obtained, as certain subtle deformities, such as deficiency of the interscalar septum, are best seen in the coronal orientation. It is important to examine thoroughly all portions of the inner ear, as anomalies are frequently multiple (Fig. 152-16).

No medical or surgical therapy has been shown to prevent the progressive hearing loss associated with congenital malformation of the inner ear. After a sudden decrease in hearing, a short course of oral corticosteroid (for example, prednisone) is administered. Although no studies have assessed the efficacy of corticosteroids in children with inner ear deformities, studies with idiopathic sudden SNHL in adults suggest some benefit. Furthermore, autoimmune mechanisms may play a role in a few children with rapid hearing deterioration, a possibility that justifies a therapeutic trial. Of course, many patients have a spontaneous partial recovery after sudden losses, even without therapeutic intervention.

Few patients with congenitally malformed inner ears have a fluctuating pattern of hearing loss suggestive of endolymphatic hydrops. When fluctuations are encountered, however, a low-salt diet and diuretics are prescribed. Surgical intervention is indicated in a few well-defined circumstances. Obviously, CSF leakage and meningitis require intervention. Exploration for perilymphatic fistula is reserved for patients with radiographically abnormal inner ears who have suffered a sudden hearing deterioration temporally related to a precipitating event such as head trauma, rapid barometric pressure change, vigorous exercise, or sneezing. As discussed previously, even in these patients only a minority have a fistula, and those that do frequently continue to lose hearing despite repair. House and House (1978) and later others have advocated endolymphatic sac surgery for congenital malformations of the inner ear (Goin et al, 1984; Mangabeira-Albernaz et al, 1981; Mitchell and Rubin, 1985); however, a careful analysis of the outcome in a large series revealed no benefit of this surgery in terms of hearing stabilization (Jackler et al, 1988). In fact, nearly 30% suffered a significant postoperative hearing deterioration. Patients with large vestibular aqueducts appear to be at particularly high risk for poor outcome after sac surgery.

Preventive measures are important in the management of patients with congenitally malformed inner ears. The risk of CSF leakage may be estimated by CT of the inner ear. Anatomic features of concern include a wide IAC, deficient partitioning between the IAC and inner ear, and enlargement of the vestibule in the horizontal plane. Parents should be made aware of the risk of meningitis and instructed in recognizing its early symptoms and signs. As most episodes of meningitis associated with inner ear malformation are pneumococcal, use of the pneumococcal vaccine is recommended. Children with malformed inner ears should avoid head trauma and rapid barometric pressure changes, as these risk provoking a sudden hearing loss or even CSF leakage. Activities such as contact sports and scuba diving are discouraged. Finally, in a hearing-impaired child, it is useful to establish a prognosis for future auditory function, as this serves as a guide for educational and rehabilitative efforts. Recognizing an inner ear malformation on CT scan may be helpful, as certain morphologic patterns have more favorable prognoses than others.

Cochlear Implantation

Electrical stimulation of the auditory nerve has proven useful in many forms of sensory deafness, including those associated with congenital malformations of the inner ear. The fundamental premise of cochlear implantation is that some of the cochlear neural population survives despite the absence of hair cells. In congenital malformations of the inner ear, the auditory nerve population is typically less than with other forms of sensory deafness. The normal human spiral ganglion cell count is usually 25.000 to 35.000. In eight congenitally dysplastic ears, Schimdt (1985) reported cell counts that ranged from 7677 to 16.110, with an average of only 11.478. In contrast, ears profoundly deaf due to ototoxicity, otosclerosis, and sudden loss have ganglion cell populations in the 18.000 to 22.000 range (Nadol et al, 1989). This observation suggests that malformed inner ears could be electrically stimulated, but that improvements in speech discrimination would fall below average for the deaf population at large. A very narrow IAC on preimplant CT (< 3 mm diameter) is a strongly adverse predictor of neural survival (Shelton et al, 1989).

Cochlear implantation in deformities that involve both the membranous and osseous labyrinth raises several special considerations. Cochlear implantation in malformations limited to the membranous labyrinth is equivalent to implantation in the prelingually deaf child (see Chapter 180). Results of single-channel implantation in patients with cochlear hypoplasia, common cavity, and incomplete partition deformities appear in the literature (Jackler et al, 1987b; Miyamoto et al, 1986). In successful cases, sound detection levels are similar to those in other prelingually deaf children. In one report of multichannel implantation (nucleus device), a patient with an incomplete partition deformity obtained a beneficial, although not spectacular, degree of speech rehabilitation (Silverstein et al, 1988). The design of multichannel electrodes makes certain assumptions about the geometry of the scala tympani that affect the anticipated orientation of stimulating electrodes to neural elements. Although standard electrodes are not optimized for dysplastic cochleas, fabrication of custom electrodes for each malformed cochlea is not feasible with current technology. One complication of unpredictable electrode positioning is stimulation of the facial nerve at current levels lower than the threshold for auditory perception. Cross-stimulation of the facial nerve has been reported with both single and multi-channel designs and has prevented some patients from using the device. Multichannel systems have a distinct advantage in this regard, as the offending electrode pair or pairs may be programmed out of the stimulation map, thus preserving the ability to provide auditory stimulation. A malformed inner ear may be a warning that the facial nerve is malpositioned. In a patient with cochlear hypoplasia, an anomalous facial nerve was encountered immediately above the round window niche, a highly vulnerable location during implant surgery (Luxford, 1990).

Another complication of implantation in malformed cochleas is inadvertent penetration of the IAC by the electrode (Novak, 1990). When such a malpositioned electrode is activated, vigorous facial twitching may occur at an extremely low stimulation threshold. To avoid this mishap, a postimplantation CT scan is advisable before undertaking stimulation to assess electrode position after implantation of a malformed cochlea.

CSF leakage also has been a frequent complication during implantation of the malformed cochlea. This complication is typically evident immediately on opening the round window and presumably arises due to deficiencies in the partition between the modiolus and the dural envelope at the lateral end of the IAC. Leaks have been successfully handled by packing connective tissue around the electrocde, but may require temporary CSF diversion to obtain a lasting seal. In one unpublished case, bacterial meningitis occurred several days after a multichannel implantation complicated by CSF leakage (Daspit, 1990). Fortunately, the infection resolved with antibiotics, and the device did not require removal.