

# CONGENITAL MALFORMATIONS OF THE INNER EAR

Malformaciones congénitas del oído interno.  
Revisión de tema

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Hearing loss  
Vestibule, labyrinth  
Cochlea  
Magnetic resonance  
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## Palabras clave (DeCS)

Oído  
Oído interno  
Pérdida auditiva  
Vestíbulo del laberinto  
Cóclea  
Imagen por resonancia  
magnética

## Summary

There are a great variety of congenital malformations that can affect the inner ear, with a diversity of physiopathologies, involved altered structures and age of symptom onset. Therefore, it is important to know and identify these alterations opportunely to lower the risks of all the complications, being of great importance, among others, the alterations in language development and social interactions.

## Resumen

Existe una gran variedad de malformaciones congénitas que pueden afectar al oído interno, con distintas fisiopatologías, diferentes estructuras alteradas y edad de aparición de los síntomas. Por lo anterior, es necesario conocer e identificar dichas alteraciones, con el fin de actuar oportunamente y reducir el riesgo de las complicaciones, entre otras —de gran importancia— las alteraciones en el área del lenguaje y en el ámbito social.

## 1. Epidemiology

Ear malformations occur in 1 in 10,000 or 20,000 cases (1). One in every 1,000 children has some degree of sensorineural hearing impairment, with an average age at diagnosis of 4.9 years. The prevalence of hearing impairment in newborns with risk factors has been determined to be 9.52% (2).

## 2. Risk Factors

- Male sex
- Ototoxic Consumption
- Stay in intensive care unit (ICU) longer than five days
- Weight less than 1.500 gr
- Gestational age less than 34 weeks
- Exchange transfusion

- Hyperbilirubinemia
- Respiratory distress from meconium aspiration
- Craniofacial alterations (3)
- Mechanical ventilation for more than five days
- TORCH Syndrome (4)

## 3. Embryology of the inner ear

The middle ear and the inner ear share an embryonic origin, but the development of the inner ear is independent, which explains why inner ear malformations tend to appear in isolation as opposed to middle and outer ear malformations, which are most often combined (2,5).

In the inner ear there is a structure called a bone labyrinth or cochlea, which develops between the fourth and eighth week of gestation from the growth of the ear capsule (5,6) which appears dorsal to the second gill arch



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during the third week and moves away from the vestibular portion of the inner ear during the first trimester forming an elongation spiral. The vestibule is fully developed by week eleven and the semicircular canals between weeks nineteen and twenty-two. The ossification of the labyrinth is completed in week twenty-three and the development of the inner ear will be complete in week twenty-six (7). Structural malformations of the inner ear that can be diagnosed by radiological studies are due to defects between the fourth and eighth weeks (5) and represent 20% of cases of congenital deafness, while subsequent lesions affecting the sensory epithelium have no image representation (5) and constitute 80% of cases of congenital deafness (8).

### 3.1 Anatomy of the inner ear

The inner ear consists of a bony labyrinth that is surrounded by a labyrinth membrane and contains in its structure the vestibule, the cochlea, the semicircular canals, the vestibule aqueduct and the cochlear aqueduct. The space in the bone labyrinth is the membranous labyrinth and inside it carries two fluids known as perilymph and endolymph (7,9).

The vestibule contains the utricle and the saccule, which are part of the membranous labyrinth. On the other hand, the vestibular aqueduct is a tubular structure that emerges from the vestibule, contains the duct and the endolymphatic sac, which are connecting to the utricle and the vestibule's sac. The vestibular aqueduct is oriented diagonally in the direction of the internal auditory canal and normally measures less than 1.5 mm in diameter. Finally, the cochlea consists of a spiral channel from 2 or  $2\frac{3}{4}$  wrapped around the modiolus, its diameter gradually decreasing as it moves towards the cochlear apex. A sheet of bone spiral runs through the cochlear canal, which is projected from the modiolus and divides this structure into an upper compartment (vestibular ramp) and a lower compartment (tympanic ramp), these two compartments communicate through the helicotrema, in the cochlear apex (7).

The internal auditory canal extends from the labyrinth to the pontocerebellar angle and contains the seventh and eighth cranial pairs. The eighth vestibulocochlear nerve is composed of three branches (lower, upper and cochlear). The upper and lower branches of the nerve occupy the posterosuperior and posteroinferior quadrant of the internal auditory canal, respectively, and the cochlear branch is located in the anteroinferior quadrant (7).

## 4. Diagnosis: CT and MRI images

Computed tomography (CT) and magnetic resonance imaging (MRI) are used to study pathologies in the inner ear. CT is the imaging study of choice for assessing the bony structures of the ear, while MRI allows better assessment of soft tissues, such as the membranous labyrinth, and also the representation of cranial pairs (5). As mentioned, the prevalence of detectable radiological alterations in patients with congenital hearing loss ranges from 7 to 20 % (2).

## 5. Classification

The classification of congenital malformations of the inner ear is described in different ways in the literature, for this review the 2002 classification by Sennaroglu and Saatci (10) will be used.

### *Cochlear malformations*

- Michel's deformity: complete absence of all cochlear and vestibular structures.
- Cochlear aplasia: Complete absence of the cochlea.
- Common cavity deformity: cystic cavity replacing the cochlea and vestibule.
- Cochlear hypoplasia: The dimensions of the cochlea and vestibule are smaller than normal.
- Incomplete Partition Type 1 (IP-I): The cochlea lacks a modiolus and is accompanied by a large cystic vestibule.
- Incomplete partition type 2 (IP-II) or Mondini malformation: The cochlea consists of 1.5 turns, in which the middle and apical turns join to form an apex of cystic aspect, with the dilated vestibule and the enlarged vestibular aqueduct.

### *Vestibular malformations.*

Malformación de Michel, cavidad común, vestibulo ausente, vestibulo hipoplásico y dilatado.

### *Semicircular canal malformations.*

Semicircular canal absent, hypoplastic semicircular canal, dilated semicircular canal.

### *Malformations of the internal auditory canal.*

Internal auditory canal absent, narrow internal auditory canal, dilated internal auditory canal.

### *Vestibular and cochlear aqueduct findings.*

#### *Dilated or normal cochlear vestibule and aqueduct.*

This review briefly describes the main imaging characteristics of some of them and shows CT and MRI images of some malformations taken in the radiology service to which the authors belong.

### *5.1 Malformation of Michel. Complete Aplasia of the Membranous Labyrinth*

It was first described by Siebmann and Bing in 1907. It may be associated with cardiac abnormalities. It represents 6% of cochlear malformations and is the most serious (8).

It is due to an alteration in the development of the otic capsule, which occurs before the third week of gestation (7).

Radiologically, there is a total absence of the membranous labyrinth and the bone labyrinth and, clinically, the patient will have total sensorineural hearing loss or deafness, which it is not possible to treat with hearing aids (1).

### *5.2 Cochlear aplasia and hypoplasia*

Cochlear aplasia accounts for 5% of cochlear malformations (8). Embryonic development stops at the end of the third week of gestation and is usually of unknown etiology (5). Cochlear hypoplasia represents 12% of cochlear malformations (8) and is due to an alteration in the development of the cochlear canal during the sixth week of gestation. Clinically, patients show unilateral or bilateral sensorineural hearing loss from birth.

Both pathologies are diagnosed through diagnostic imaging, in cochlear aplasia, the absence of the cochlea and vestibule is confirmed, the semicircular canals are often malformed, globular or dilated and the cochlear nerve and its conduit are absent; in cochlear hypoplasia, on the other hand, a small cochlea (1-3 mm) with a single primitive spiral is observed; the vestibule and semicircular canals may be normal or have some type of malformation (5).

### 5.3 Incomplete partition type 1 (PI-1).

#### Cochleovestibular cystic anomaly

It represents 20% of cochlear malformations and is characterized by a cystic cochlea and the dilated vestibule (8). It originates during the fifth week of gestation and affects the development of the internal cochlear structure. There are different degrees of anomalies, the mild degree presents the malformation only at the level of the cochlea with vestibule and normal semicircular ducts and the absent modiolus, unlike the more advanced degree, in which the cochlea, vestibule and horizontal semicircular duct are globularly enlarged and form an outline. Radiological findings are detected by CT scan (5) (Figure 1).

### 5.4 Incomplete partition type 1 (PI-1).

#### Cochleovestibular cystic anomaly

Mondini's aplasia is the second most frequent cause of congenital deafness (5). It accounts for 19 % of cochlear malformations (8) and 30 % of congenital malformations of the inner ear (5). It is characterised by a triad consisting of a cystic cochlear apex, minimal dilation of the vestibule and a long vestibule aqueduct (8). This anomaly is caused by a developmental arrest during the seventh week (11) or a developmental arrest of the ear between days 58 and 70 of gestation, which causes the interruption of cochlear development at one and a half turns producing this deformity and being associated with complete deafness and vestibular malformations (12). In terms of diagnosis, some patients with Mondini malformation have mild sensorineural deafness while others have profound deafness. In CT, it is the congenital malformation with the greatest variability, since it can be a single cystic cavity or the basal spiral can be differentiated (11), but the classic finding in CT is a normal basal turn with a pseudocystic cavity that replaces the middle and apical turns; the rest of the ear is normal (2,11). It is detected in 20% of children with congenital sensorineural hearing loss; of these, 65% affect both ears, and 35% are unilateral (2) (Figure 2).

### 5.5 Vestibular and semicircular canal malformations

The embryological development of the semicircular canals begins in the sixth week of gestation and ends in week 22 (11). The clinical presentation is based on vestibular alteration and may cause different degrees of hearing loss, depending on the association it has with cochlear malformations. In both cases, the diagnostic method of choice is CT (2). High-resolution magnetic resonance imaging with multiplanar reconstructions also detects them (Figures 3 and 4).

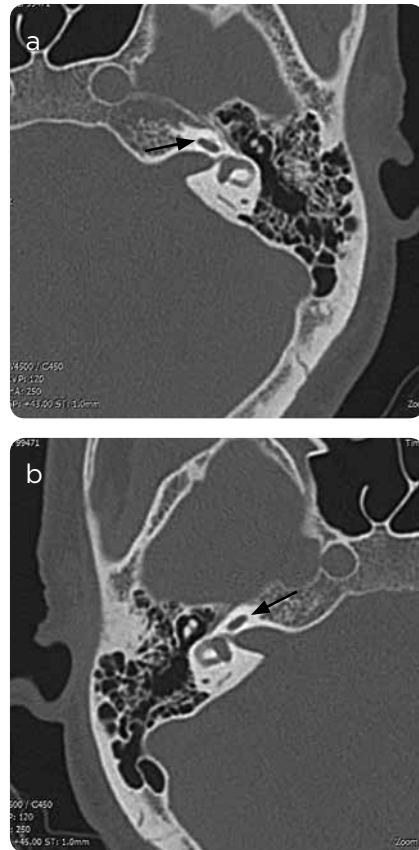


Figure 1. a and b) Left and right ear. Incomplete partition type 1 (PI-1) or cochleovestibular cystic anomaly is observed: The arrow in each figure indicates a slight degree of incomplete partition, in this case the malformation is only in the cochlea, since there is no interscalar septum or modiolus, the vestibule and the semicircular ducts are normal.

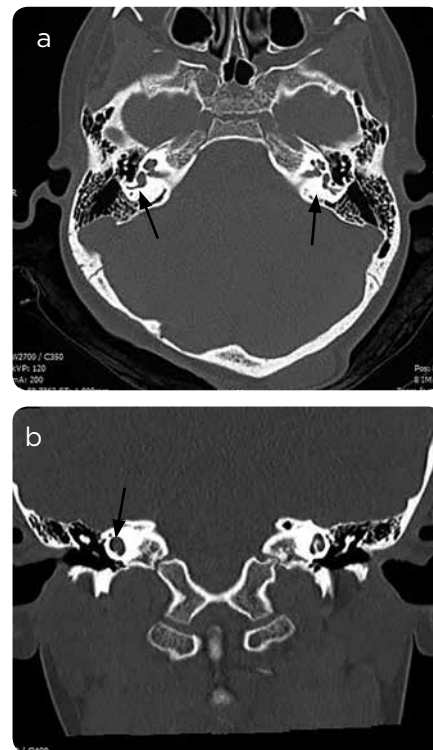


Figure 2. Mondini malformation. a) CT axial cut: dilation of the right vestibule, on the left side there is a vestibule with a normal appearance. The arrows indicate the dilated right vestibule and the normal left vestibule. b) CT in coronal cut: the Right cochlea consists of 1.5 turns, in which the middle and apical turns join to form a cystic apex.

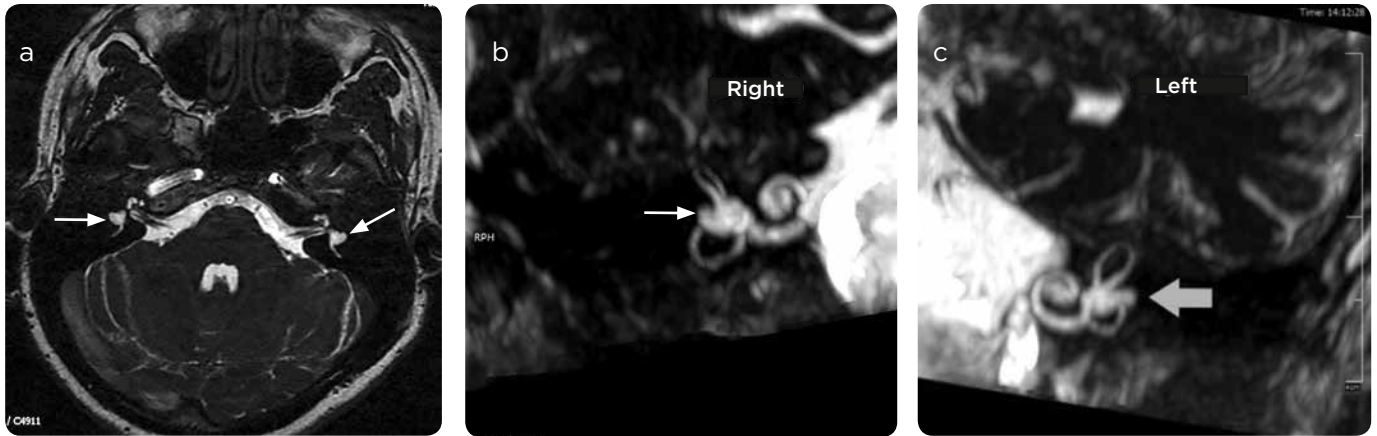


Figure 3. Hypoplastic lateral semicircular canal. MR T2 FIESTA sequence. a) Axial cutting of both ears. b and c) Reconstruction (three-dimensional) coronal oblique. A marked decrease in the size of the lateral semicircular canal is evident, with a small sack with loss of the tubular appearance of the lateral semicircular canal (arrows).



Figure 4. a, b and c). Contiguous axial images of the right rock: hypoplasia of the vestibule, with absent semicircular canals (arrows). Images courtesy of Dr. Eliana Bonfante. UT Health Houston.

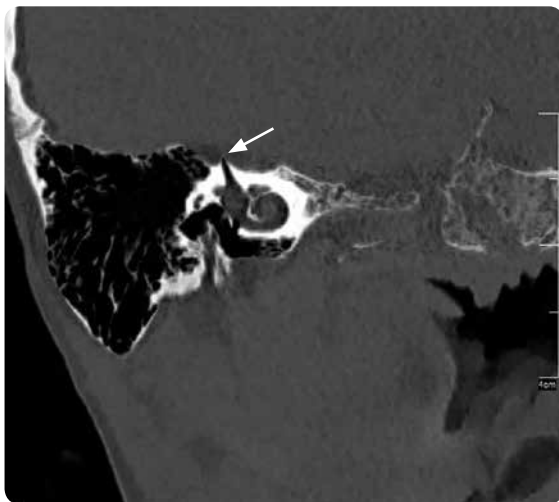


Figure 5. Upper semicircular canal dehiscence: Asymmetric thinning of the superficial ridge of the upper semicircular canal (arrow).

### 5.6 Dehiscence of the upper SCC

It was first described in 1998 by Minor and colleagues as a condition in which the dura separates the upper semicircular canal from the middle cranial fossa because the thick sheet of bone that should serve this function is absent due to incomplete development during early childhood. Clinically it manifests itself with vestibular and auditory signs and symptoms such as vertical rotary nystagmus, hearing loss, tinnitus and recurrent vertigo following manoeuvres that increase pressure in the inner ear or intracranially, for example, Valsalva manoeuvre or a very loud noise (Tullius phenomenon) (13,14) (Figure 5).

### 5.7 Dehiscence of the posterior SCC

The first cases described in 2003 have a very low incidence (15) compared to the alteration in the upper semicircular canal; however, they can be found together in some patients (16). They may be caused by a partial loss of bone coverage separating the posterior semicircular canal from the posterior cranial fossa (15), or acquired due to erosion of the jugular bulb. Clinically it is similar to dehiscence of the upper semicircular canal because it also presents Tullium phenomenon and pressure changes in the ear, so they are differential diagnoses and the only clinical variation between them is the direction of rotational nystagmus (16).

### 5.8 Dehiscence of the lateral SCC

Unlike the previous ones, this alteration is generally acquired and associated with eroded cholesteatomas and mastoidectomies (17). Clinically it manifests as noise-induced vertigo, orthogonal nystagmus towards the lateral semicircular canal and no vertical movements are observed (18).

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