High Resolution temporal bone CT & MRI of Inner Ear in pendred syndrome- key imaging features

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Authors: P. Kala, R. Avantsa, S. Inuganti; Bangalore/IN
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Learning objectives

Sensorineural hearing loss (SNHL) in infants and children is a major cause of childhood disability worldwide.

Temporal bone and inner ear MR Imaging have crucial role in the work-up of childhood SNHL to evaluate the inner ear for anomalies, to provide clues to the etiology of the hearing loss, and associated comorbid conditions.

We review and illustrate pendred syndrome, the most common syndromic hereditary cause of congenital SNHL, emphasizing inner ear abnormalities on cross-sectional imaging with an intention of

• Familiarizing the neuroradiologist with the clinical and imaging characteristics of pendred syndrome, in order to place the correct differential diagnosis in sensorineural hearing loss in children.

• Identifying and describing the key imaging features in pendred syndrome involving temporal bone and inner ear.
Background

Congenital sensorineural hearing loss (SNHL) is generally divided into genetic and nongenetic forms. It is estimated that in 50% of patients, SNHL can be linked to a genetic cause, of which approximately 75%-80% demonstrate autosomal recessive inheritance; 15%-20%, autosomal dominant inheritance; and 1%-2%, X-linked inheritance. To date, 300 syndromic forms of hearing loss have been described.

Several of these syndromes demonstrate gross inner ear anomalies by imaging, which are common and sometimes defining features.

Pendred syndrome, an autosomal recessive disorder, has been linked to mutations in the PDS gene, which codes for the pendrin protein and is one of the most common genetic causes of SNHL representing between 4.3% and 7.5% of all causes of childhood deafness. The gene is located on the long arm of chromosome 7 leading to congenital bilateral SNHL and goiter with occasional hypothyroidism. [1]

First described by Vaughan Pendred in 1896, Pendred syndrome was established by Fraser as an important and relatively common cause of inherited deafness, estimating a prevalence of 75 cases per million population.

Fraser’s criteria for the diagnosis of pendred syndrome were congenital deafness, goitre, and a positive perchlorate discharge test.[2]

SensoriNeural Hearing Loss (SNHL):

Audio vestibular studies in Pendred syndrome typically reveal the disturbance of moderate to severe degree sensorineural hearing loss. Deafness is mostly profound and prelingual, however, slowly progressive and fluctuating hearing loss has also been documented.

It is known that the spectrum of inner ear anomalies reflects interruptions to development occurring at different junctures during embryogenesis. Insults occurring during the gestational 7th week cause abnormalities like incomplete partition type-II or Mondini dysplasia common to pendred syndrome.[3]

Thyroid dysfunction

The goiter in Pendred syndrome appears in mid-childhood which Classically tends to be diffuse initially but may become multinodular as the age progresses[fig1]. Patients are usually euthyroid, however some have variable degrees of hypothyroidism.[4]
Goiter is essential to the diagnosis, however may not be present during the time of presentation of SNHL. Identification of the defect before the development of goiter can be facilitated by the perchlorate discharge test.

**Perchlorate discharge test**

The thyroid glands of the patients cannot organify iodide efficiently, and 10%-80% of iodine taken up by the gland is discharged after administration of perchlorate. This inefficient iodide organification is the basis of the perchlorate discharge test used for the diagnosis of Pendred syndrome. Perchlorate and thiocyanate unmask defects of organification by provoking the discharge of inorganic iodide from the gland. A discharge in response to perchlorate of 10% or greater is considered abnormal.

Autoimmune thyroid disease and thyrotoxic patients treated with radioactive iodine may have similar levels of iodide discharge. It is important to correlate the perchlorate discharge data with the clinical and other investigative data.[4]

**Cross sectional imaging**

Inner ear malformations are invariably present and frequently include:

- Incomplete partition type-II (Mondini dysplasia & Modulus deficiency)
- Vestibular enlargement,
- Enlarged vestibular aqueduct & Endolymphatic sac.

**Type II Incomplete Partition.** - First described in Latin by Italian anatomist Carlo Mondini. Type II incomplete partition (Mondini deformity) represents a developmental arrest in the 7th week of gestation. The cochlea consists of 1½ turns, and the interscalar septum and osseous spiral lamina are absent in CT. The basal cochlear turn appears normal, but the middle and apical turns coalesce to form a cystic apex[fig.2,3,5,6,7] The modulus is present only at the level of the basal turn. At MR imaging distinct scalae tympani and vestibule are not visualized and the interscalar septal defects and the absence of the osseous spiral lamina from the middle and apical turns are easily distinguished on thin-section gradient-echo images obtained with heavy T2 weighting[fig.9,10]. [5-8]

Mondini deformity is the most common type of cochlear malformation, accounting for more than 50% of all cochlear deformities. Numerous syndromes are associated with the Mondini anomaly are Albinism, Alagille syndrome, Apert syndrome, Crouzon syndrome, DiGeorge syndrome, Klippel-Feil syndrome, Trisomies (including Down syndrome), Usher syndrome, Waardenburg syndrome and Wildervank syndrome. Association with goiter and positive perchlorate discharge test is classic in Pendred syndrome.
Enlarged Vestibular Aqueduct.-An enlarged vestibular aqueduct, or an enlarged endolymphatic duct and sac, are the most frequent CT or MR imaging finding in patients. Common to females, the abnormality is usually bilateral, may be asymmetric. Although enlargement of the vestibular aqueduct most likely is the result of a developmental anomaly in the 7th week of gestation, the vestibular aqueduct continues to develop throughout gestation and beyond; thus, in some cases, the malformation may actually be acquired and not congenital.

The characteristic imaging feature seen at CT is enlargement of the osseous vestibular aqueduct[fig4]. At MR imaging, the characteristic feature is enlargement of the endolymphatic duct and sac and has been reported on 100% of patients with Pendred syndrome[fig.8]. The vestibular aqueduct is considered enlarged when its diameter midway between the common crus and the external aperture is greater than 1.5 mm on CT or MR images.[9]

Vestibular enlargement---Various abnormalities of the vestibule are noted with Type II Incomplete Partition. Most common vestibular abnormality found in pendred syndrome is enlarged vestibule. There is no definite measurement given in literature, but subjective enlargement has been reported[fig.7].
Images for this section:

**Fig. 1:** One of the three siblings who presented to us with SNHL and goiter. Audiometry and ultrasonography for thyroid was done. All three underwent HRCT & MRI for temporal bone pathology. All the three siblings had similar imaging findings and positive Perchlorate discharge test.

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Fig. 2: Type II incomplete partition seen in a case of Pendred syndrome presenting in our hospital with SNHL and goiter. Axial CT image shows deficient modulus from a cystic cochlear apex (red arrow shown in fig.3) formed by coalescent apical and middle turns.

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Fig. 3: Type II incomplete partition seen in a case of Pendred syndrome presenting in our hospital with SNHL and goiter. Axial CT image shows deficient modulus from a cystic cochlear apex (red arrow) formed by coalescent apical and middle turns.

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Fig. 4: Type II incomplete partition: case in Fig.1 seen in case of Pendred syndrome presenting in our hospital with SNHL and goiter-Axial image at another level shows dilatation of the vestibular aqueduct (orange arrow)

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Fig. 5: Type II incomplete partition: case in Fig.1 seen in case of Pendred syndrome presenting in our hospital with SNHL and goiter-Axial CT image obtained at a slightly lower level shows a normal basal turn of the cochlea.

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**Fig. 6:** Type II incomplete partition: case in Fig.1 seen in case of Pendred syndrome presenting in our hospital with SNHL and goiter.-Coronal CT image shows the cystic cochlear apex (arrow).

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**Fig. 7:** Type II incomplete partition: case in Fig.1 seen in case of Pendred syndrome presenting in our hospital with SNHL and goiter- Vestibular enlargement was noted on the left (*).  

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Fig. 8: Axial heavily weighted T2 gradient-echo MR images of the same case in Fig 1, shows marked dilatation of vestibular aqueduct (red arrow) and endolymphatic sac (yellow arrow).

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Fig. 9: Axial heavily weighted T2 gradient-echo MR images of the same case in Fig 1, shows deficient modulus and a cystic cochlear apex. (compare with normal modulus in MR image in Fig.10)

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Fig. 10: Axial heavily weighted T2 gradient-echo MR images of a normal subject shows normal modulus and cochlea.

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Findings and procedure details

Temporal HRCT and inner ear MR imaging provide useful information about inner ear malformations typical for the syndrome.

HRCT Temporal bone Examinations

High-resolution CT of the temporal bone is required followed by image reconstruction in both the axial and coronal planes to evaluate the inner ear and its malformations. Helical scanning is performed from the top of the petrous apex to the inferior tip of the mastoid bone, in planes parallel to the hard palate to obtain isometric dataset.

The raw image data set can be reconstructed with a section thickness of as little as 0.3 mm to obtain high-quality coronal reformatted images. Usually a 512 × 512 matrix and a small field of view (9 cm) is used to review images with a high-resolution bone algorithm separately for documentation of the right and left ears. Coronal reformatted images are obtained from the anterior margin of the petrous apex to the posterior margin of the mastoid.

Internal ear MR Imaging Examinations

The use of a 1.5- or 3-T MR imaging system is preferred for inner ear examinations. Sedation is used in most children. A thin-section heavily T2 weighted gradient-echo sequence is best for evaluation of the fluid-filled spaces of the membranous labyrinth and the eighth cranial nerve. Acquisition is performed in the axial plane with a small field of view, slice thickness of less than 3mm and 0 gap. Routine axial T2-weighted imaging of the brain is performed to exclude central nervous system causes of sensorineural hearing loss. Submillimeter thickness allows high-quality multiplanar reformations.

Relative strengths of modalities

MR imaging provides superior soft-tissue contrast without ionizing radiation. High resolution images of the inner ear and brain are possible now due to advanced technology, high-field-strength units, improved coil technology, and parallel imaging.

High-resolution temporal bone CT is better for assessing osseous details. As compared with MR imaging CT is lower in cost and time, frequently not requiring sedation or anesthesia.
Conclusion

Neuroradiologists who routinely evaluate temporal bone studies performed for childhood SNHL can make a reliable diagnosis of pendred syndrome when equipped with knowledge about typical HRCT and inner ear MR imaging imaging appearances of the syndrome as well as accurate clinical information.

Genetic testing (homozygosity of c.1151A>G) confirms the diagnosis.

No specific treatment exists for Pendred syndrome. Such patients should be diagnosed at the earliest after birth and intervened with cochlear implant and auditory verbal therapy. Genetic counselling is further required for prevention of the disorder in the offsprings.
Personal information

Dr Prachi Kala, MD.
Neuroradiology Division, Department of radiology,
Vydehi institute of medical sciences, Bangalore, India.
prachi_kala@yahoo.com

Dr Rohini Avantsa, MD.
Department of radiology, Vydehi institute of medical sciences, Bangalore, India.
rkgayatri5@gmail.com

Dr Srikar Inuganti, MBBS.
Resident, Department of radiology, Vydehi institute of medical sciences, Bangalore, India.
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