IMAGING OF THE MIDDLE AND INNER EAR IN SYNDROMIC HEARING LOSS

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- congenital hearing loss (HL) is the most common sensory disorder, with adverse effects on the social & emotional development as well as on the quality of life

- 50% of CHL cases are due to genetic factors & 30% of them are found in the context of a multisystemic disorder, also called as SYNDROMIC Hearing Loss (SHL)

- imaging has a crucial role in patients with SHL
  - etiological analysis
  - pre operative (cochlear implantation/ossicular replacement prosthesis) information - anatomy, measurements, anatomic variants with surgical risk
  - characteristic malformations of middle & inner ear (alone or in combination with other systemic & skeletal imaging findings) can strongly suggest the diagnosis of a specific syndrome

the goals of our presentation are:

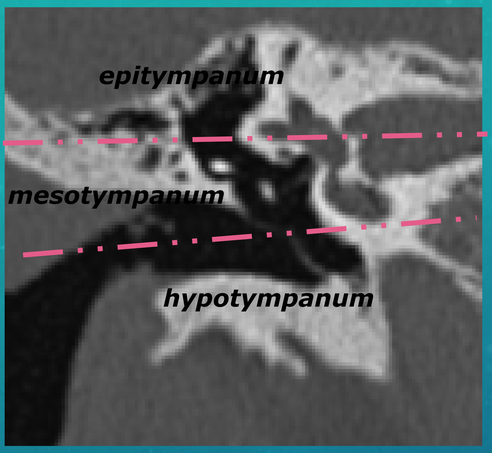
- to present a detailed, segmental imaging study of temporal bones & review the normal radioanatomy (CT & MRI) of middle / inner ear structures
- to illustrate frequent isolated or complex abnormalities of middle / inner ear structures
- to review the most characteristic radiological malformation patterns of temporal bones, suggesting the diagnosis of a specific syndrome
- to present common radiological systemic & skeletal imaging findings that in the context of SHL can lead to the diagnosis

Middle Ear (ME)

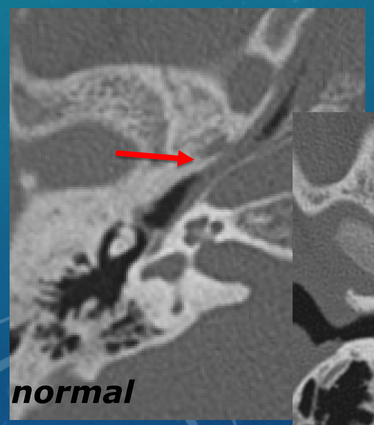
Radiological analysis CT

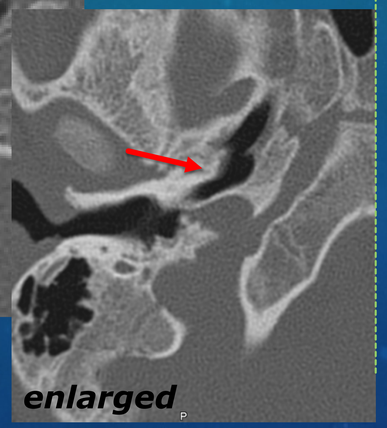
I. Tympanic Cavity (TC)

normal pneumatization



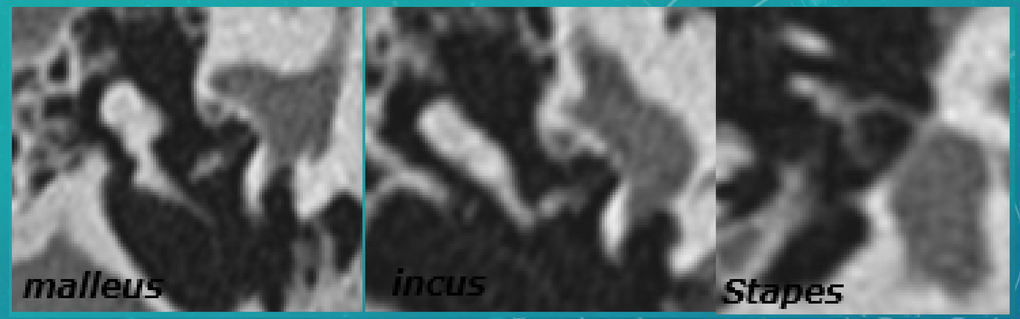
II. Eustachian Tube (ET)



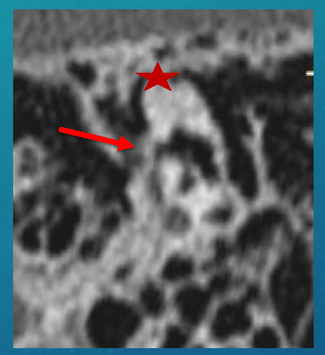


III. Auditory Ossicles (AO)

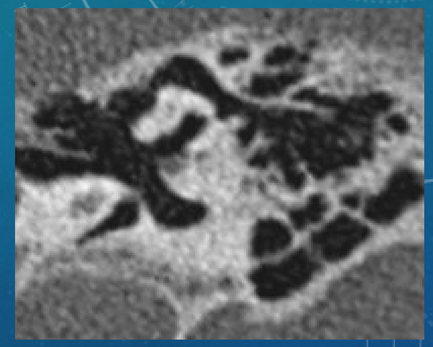
- **mineralization –density**
- **normal appearance**
- **absence of fusion or fixation**
- **joints: incudomalleolar, incudostapedial**



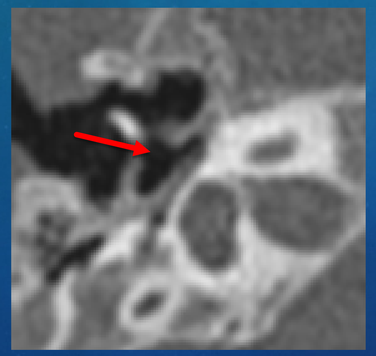
aplasia of the long process of the incus (LPI)



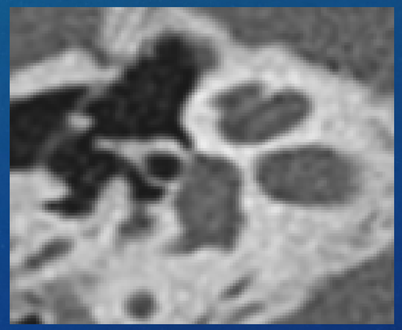
incudomalleolar fusion, aplasia of the manubrium of the malleus & fixation to the anterior tympanic wall



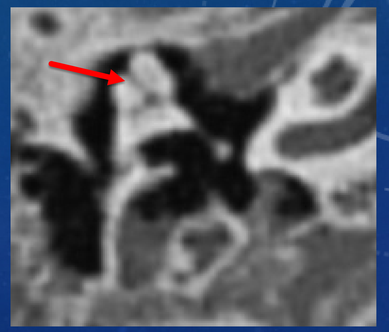
"boomerang" malleus-incus fusion deformity



stapedius aplasia

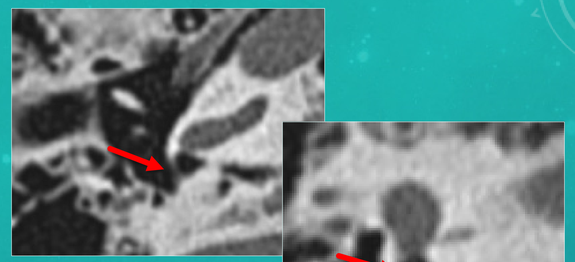


hyperdense aspect of stapes crura thickening of the footplate

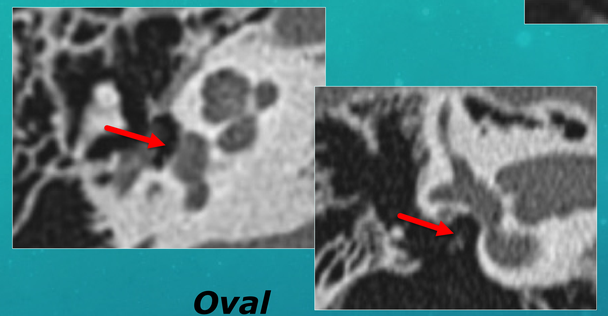


large incus body with a cleft

IV. Windows



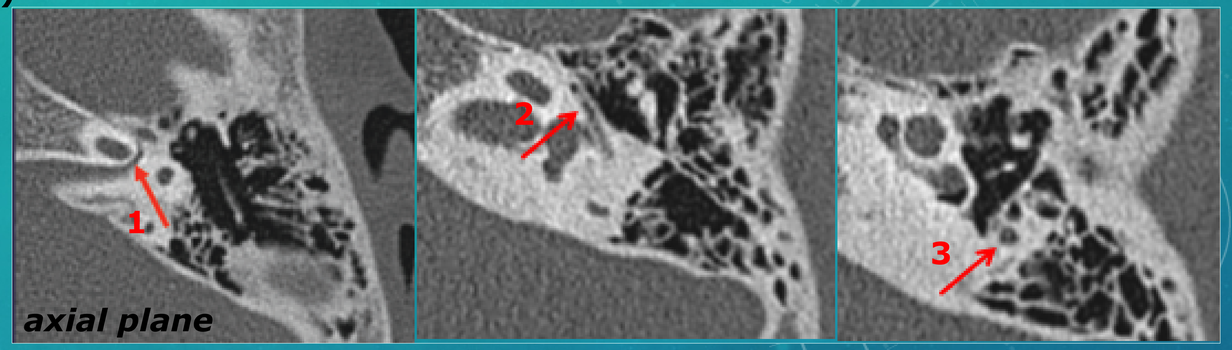
Round Window (RW)

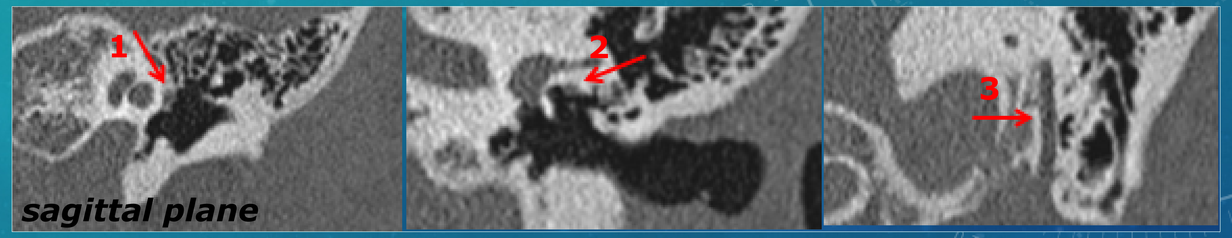


Oval Window (OW)

V. Facial Nerve Canal (FNC)

- **symmetric size**
- **normal situation**
- **3 segments:**
  1. **labyrinthine**
  2. **tympanic**
  3. **mastoid**



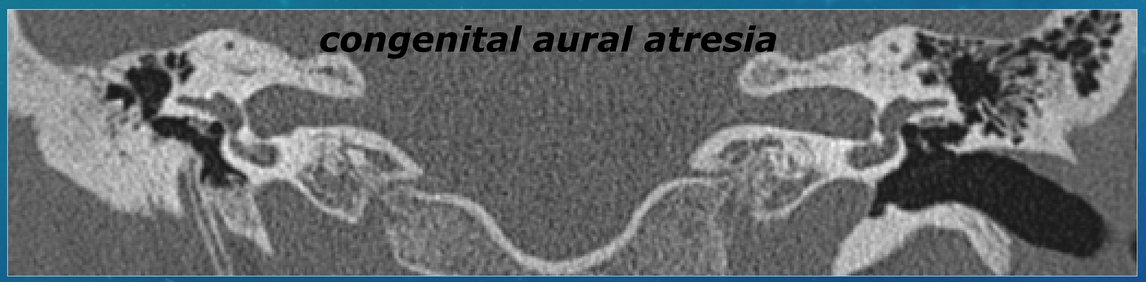


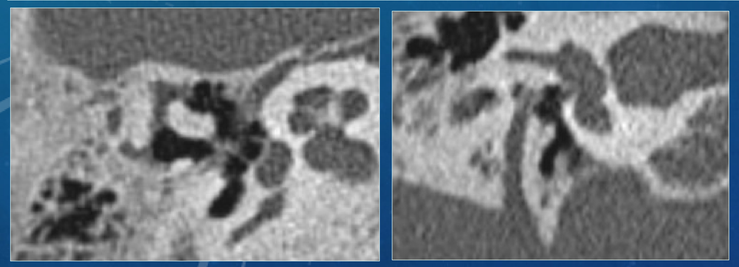
The embryology predicts the malformation pattern !!!!

1st branchial arch: mastoid process, External Auditory Canal (EAC), malleus, incus, FN

2nd branchial arch: manubrium of malleus, stapes, round & oval windows

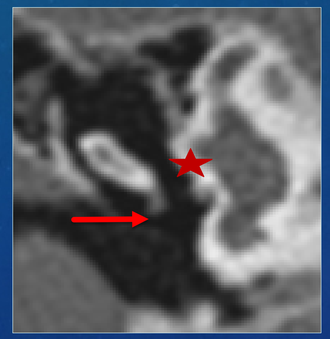
1st branchial arch mlf





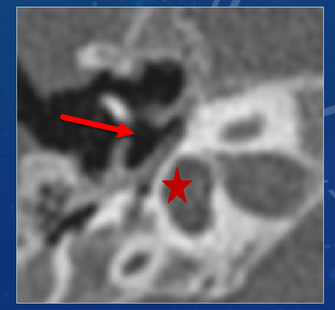
1. ***aplasia of EAC & mastoid hypopneumatization***
2. ***hypoplastic tympanic cavity***
3. ***incudomalleolar dysplasia & fusion***
4. ***aberrant position FNC***

2nd branchial arch mlf



LPI aplasia

OW atresia



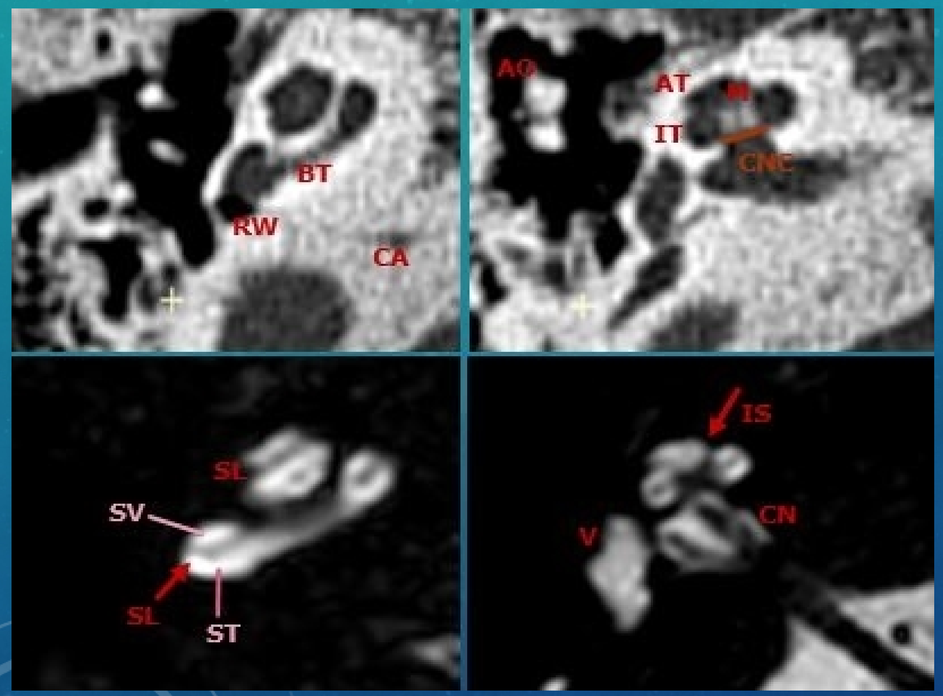
stapes aplasia

OW atresia

Inner Ear (IE) - Radiological analysis CT / MRI

I. Cochlea (Co.)

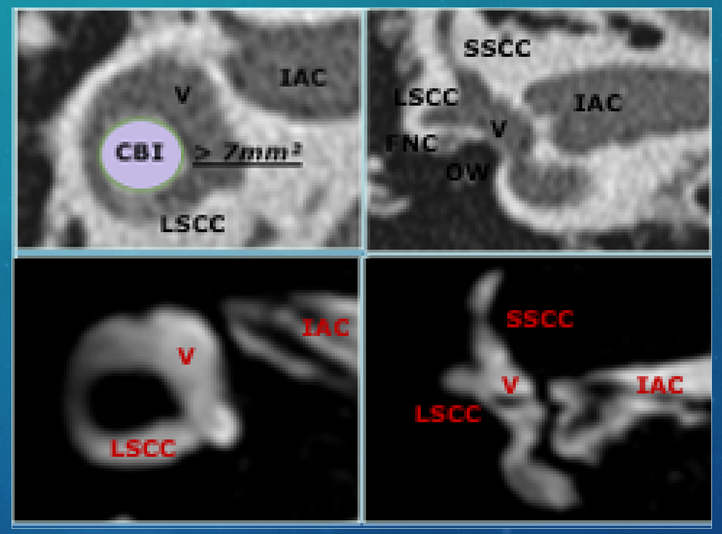
- size
- shape / internal architecture
  - ✓ normal partition into 2½ or 2¾ turns
  - ✓ intra cochlear structures
- cochlear nerve canal (CNC)
- cochlear aqueduct (CA)
- round window (RW)



BT: basal turn, IT: intermediate turn AT: apical turn
SL: spiral lamina, SV : scala vestibuli, ST: scala tympani,
IS: interscala septum, AO: auditory ossicles, CN: cochlear nerve

II. Vestibule (V.)

- size
- shape / ovoid cavity
- oval window (OW)
- vestibular aqueduct (VA)

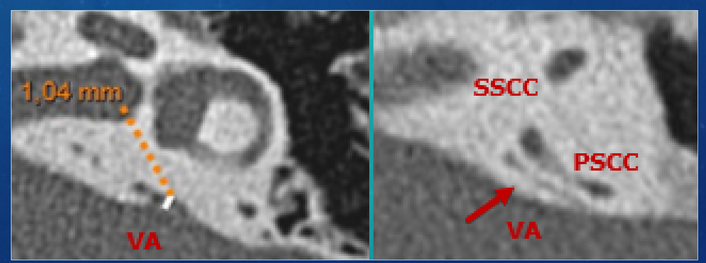


III. Semicircular Canals (SSC)

- anterior (ASCC), posterior (PSCC), lateral (LSCC) ***is the last to be formed !!!***
- size, shape, ampoula (Amp)
- central bony island (CBI)

IAC: internal auditory canal
FNC: facial nerve canal

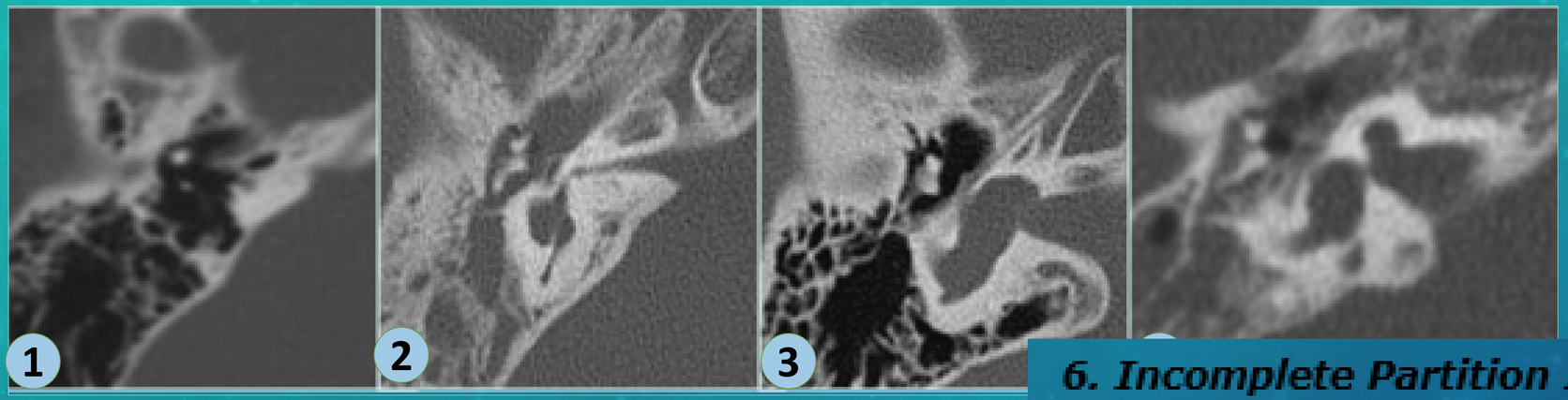
IV. Vestibular Aqueduct (VA)



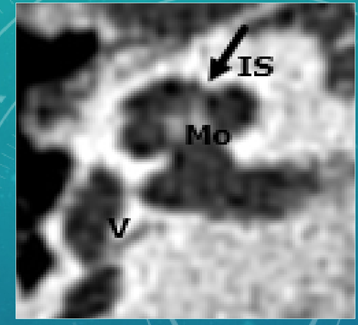
- VA width range: 0.3-1.5 mm
- *or* < width of PSCC

CT based classification of Congenital Internal Ear malformations with emphasis on cochlea

Sennaroglou, 2002

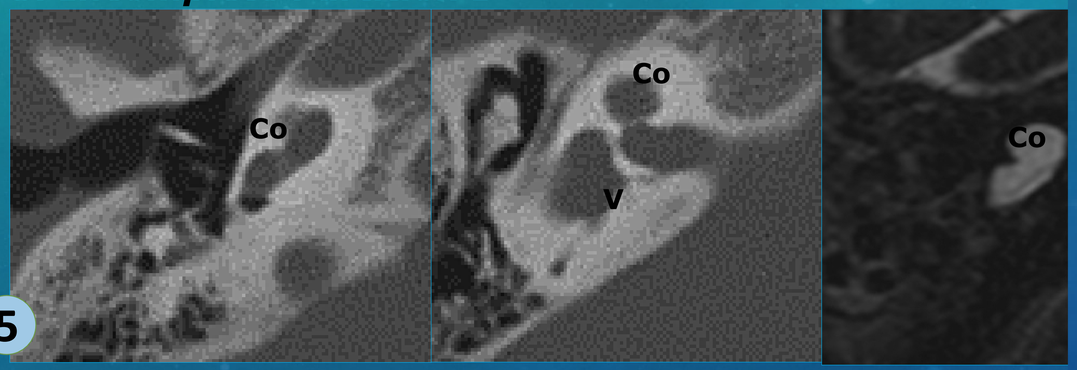


normal cochlea CT



1. ***Michel deformity***
2. ***cochlear aplasia***
3. ***common cochleo -vestibular cavity***
4. ***cochlear hypoplasia***

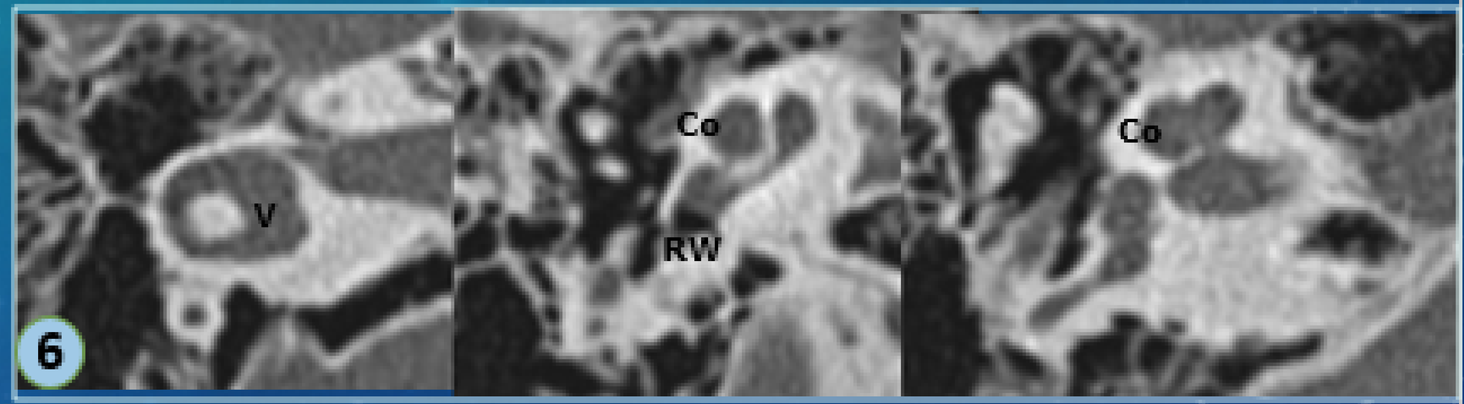
5. Incomplete Partition I



V & Co: dilated BUT separated ➡ ***figure '8' morphology***

Co: altered internal architecture, absent Modiolus, Interscala Septum & Spiral Lamina

6. Incomplete Partition II



Co 1.5 turns: normal basal turn with cystic apex

Deficient Mo

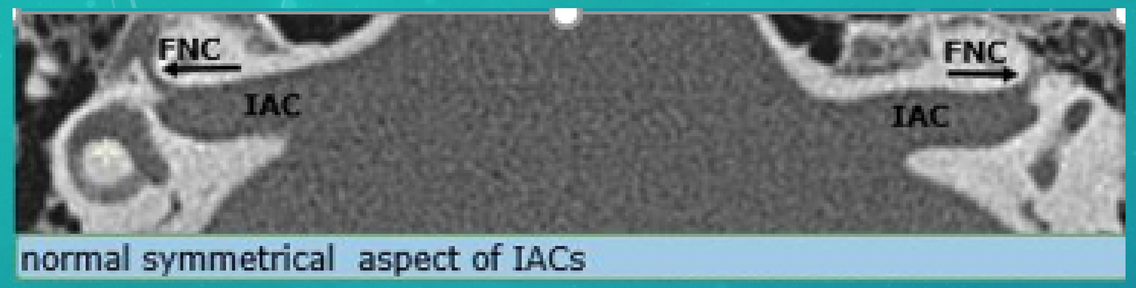
Absent IS & SL

+ extracochlear findings: large VA & enlarged V

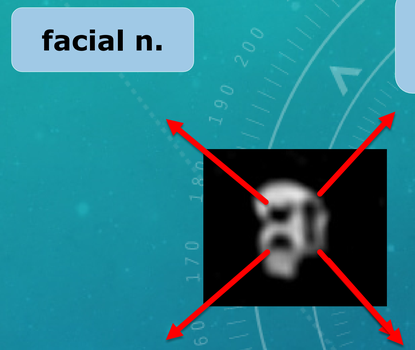
MONDINI malformation

V. Internal Auditory Canal (IAC)

- **symmetric aspect**
- **width, orientation**
- **fundus : CNC & FNC**
- **content: VII & VIII *(MRI)***



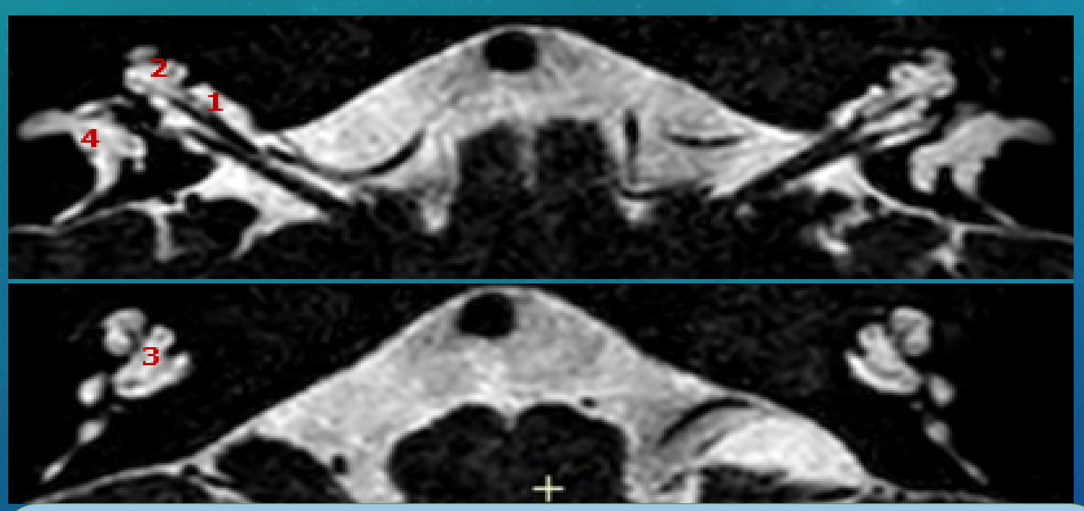
normal symmetrical aspect of IACs



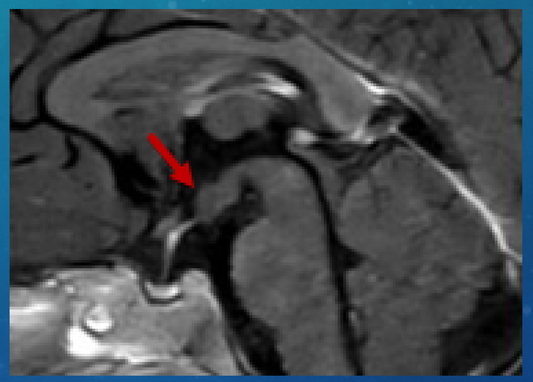
reconstruction oblique sagittal

Incomplete Partition III

X- LINKED DEAFNESS with stapes gusher (mixed: conductive & SN hearing loss)



- **bilateral widening of the fundus of IAC (1), absent cribriform plate & modiolus (2)**
- **present interscala septa** ➡ ***corkscrew cochlea (3)***
- **irregular vestibule (4)**


interhypothalamic adhesion

Bilateral IP-III ➡ ***X-linked deafness***

+/- hypothalamic malf.

(POU3F4 g. mutation)

C.H.A.R.G.E. syndrome *(CHD7 g. mutation)*

- ✓ **hearing loss is prevalent**
- ✓ **usually: asymmetrical mixed hearing loss, ranging from severe to profound**

- ✓ **various ear malformations : inner , middle & external ear**
- ✓ **more characteristic: SSC dysplasia / vestibular dysplasia / ossicular malf./ abnormal auricle**

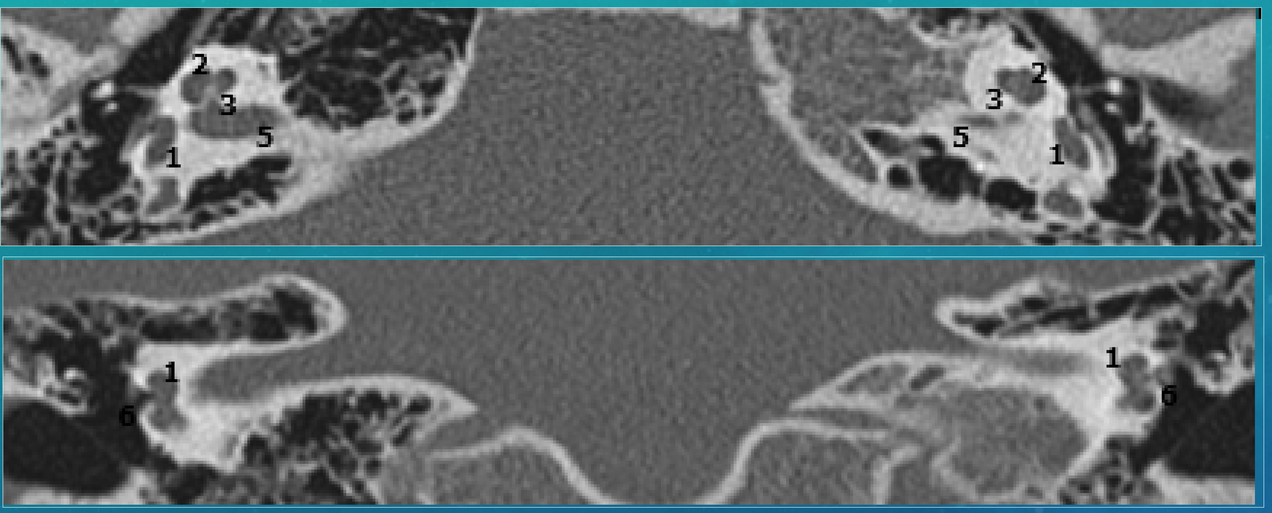
Major Criteria : 4 C

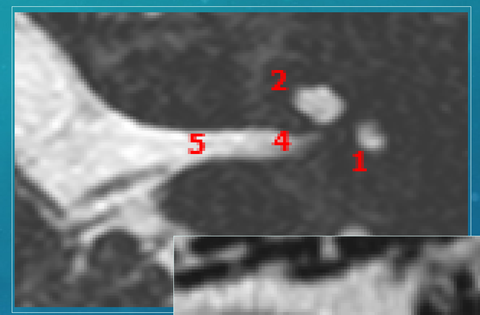
Coloboma

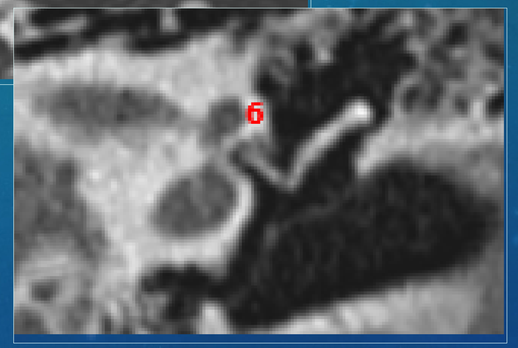
Choanal atresia

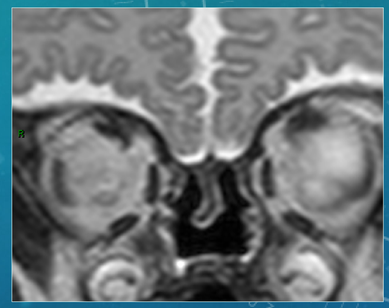
Cranial nerve abnormalities (olfactory ++)

Caracteristic ear mlf (SCC dysplasia/aplasia)

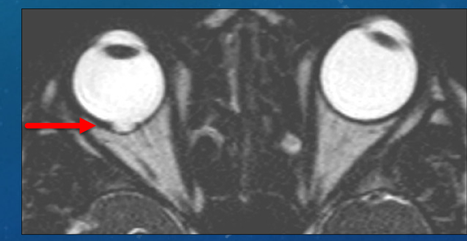




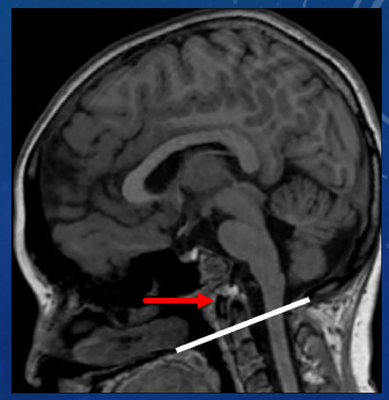




agenesis of olfactory bulbs & sulci



ocular coloboma



basilar invagination

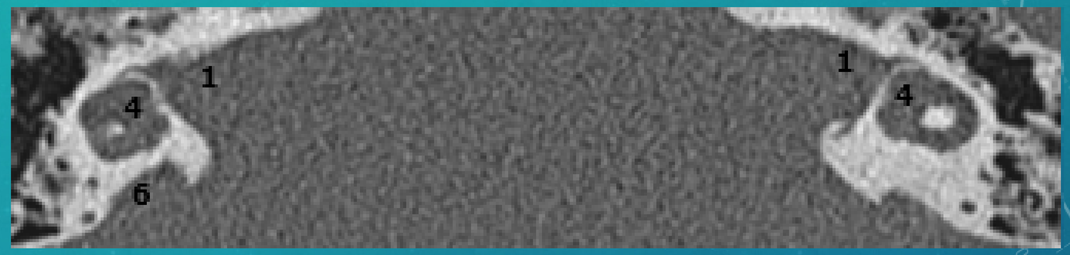
- **<u>bilateral: vestibular hypoplasia & agenesis of SCC</u> (1)**
- **hypoplastic cochlea & deficient modiolus (LT>RT) (2)**
- **narrow FNC (LT>RT)(3) ➡ facial nerve aplasia / hypoplasia (4)**
- **narrow IAC (5)**
- **aplasia of oval window (6)**

BRACHIO – OTO- RENAL (BOR) syndrome

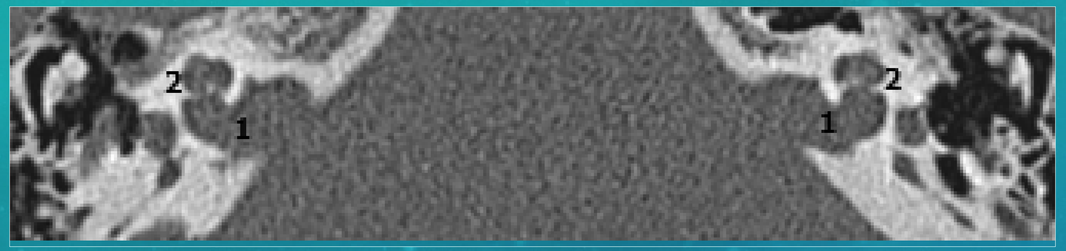
2nd branchial arch malformation

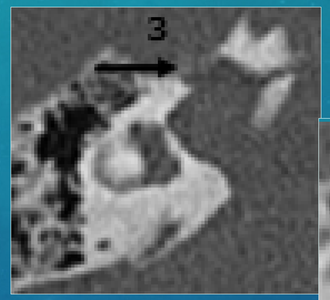
- ❖ branchial arch fistulas or cysts, preauricular pits & tags, auricular malf.
- ❖ ear malformations with hearing loss
- ❖ anomalies of the kidney

✓ conductive hearing loss / SNHL or mixed

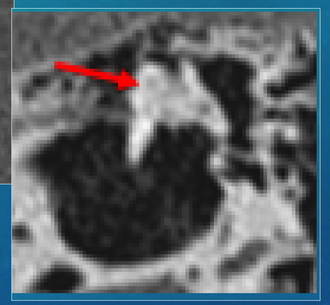


- bilateral, asymmetric vestibular & LSSC dilatation with small CBI
- enlarged vestibular aqueduct (6)

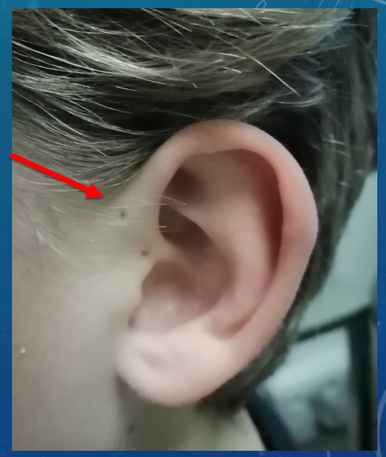




enlarged FNC



indunomalleolar fusion



preauricular pit

- enlarged, funnel shaped IAC (1)
- 'unwound' cochlea: middle & apical turns anteromedial rotated (2)
- cochlear apical turn hypoplasia & deficient modiolus (2)
- medialized course & dilated labyrinthine segm. of facnial n. canal (3)
- posterior labyrinthine mlf (4) + middle ear mfl (5)
- enlarged Eustachian tube

BOR syndrome:
(EYA1 g. – SIX1 g.)

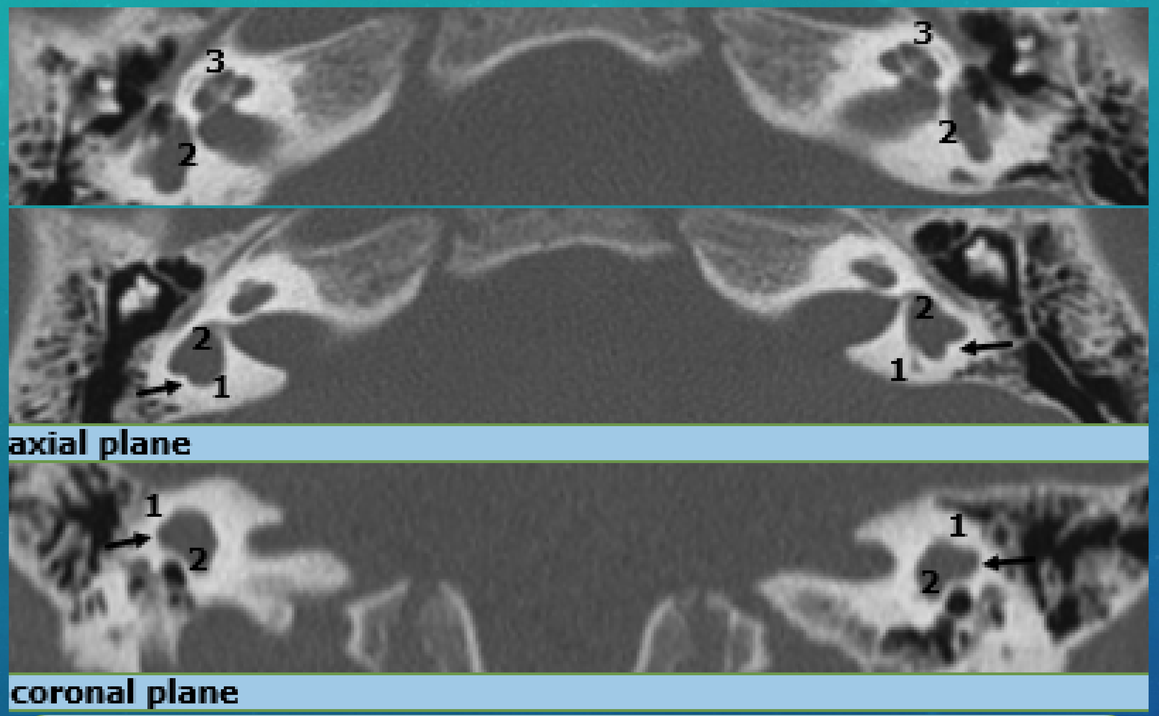
unwound & hypoplastic cochlea
medialized course & dilated FNC
enlarged, funnel shaped IAC

WAARDENBURG syndrome

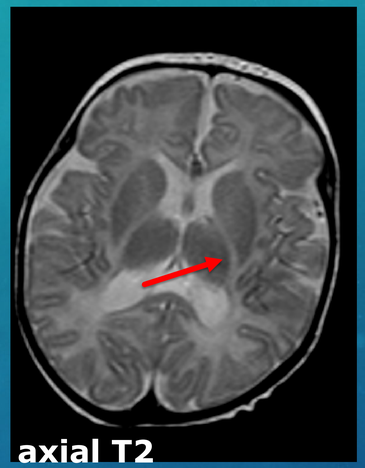
- clinically and genetically heterogeneous (7 GENES)
- 4 clinical distinct subtypes, WS 1-4 : all characterized by deafness (SNHL) and pigmentary disturbance

- SOX 10 mutation : WS 2 (15%) & WS 4 (> 50%)

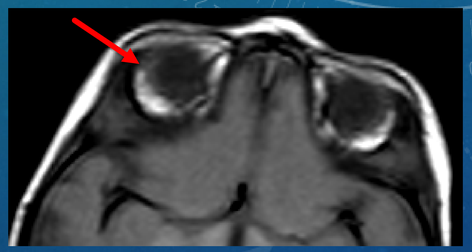
+ neurologic abnormalities <impaired myelination ➡ (PCWH) Peripheral demyelinating neuropathy-central dysmyelinating leukodystrophy-Waardenburg syndrome-Hirschsprung disease



- bilateral aplasia/ hypoplasia of all SCC (1)
- enlarged vestibular cavity with evaginations representing SCC anlages (2)
- flattened apex and midturn of the cochlea (3)



WM lesions in a 3-month-old baby:
absence of normal myelination in posterior limb of internal capsule



Lack of visibility of lacrymal glands

WAARDENBURG Syndrome

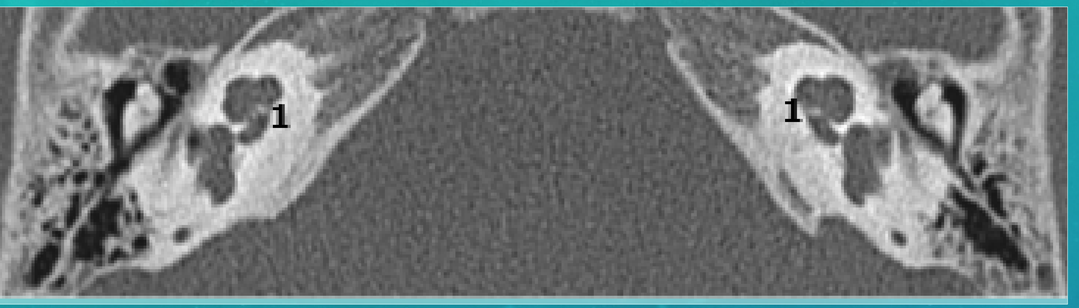
(SOX 10 mutation)

- agenesis/hypoplasia >/ 1 SCC
- *enlarged vestibular cavity*
- *cochlear deformity*

(reduced size, abnormal shape but usually normal partition)

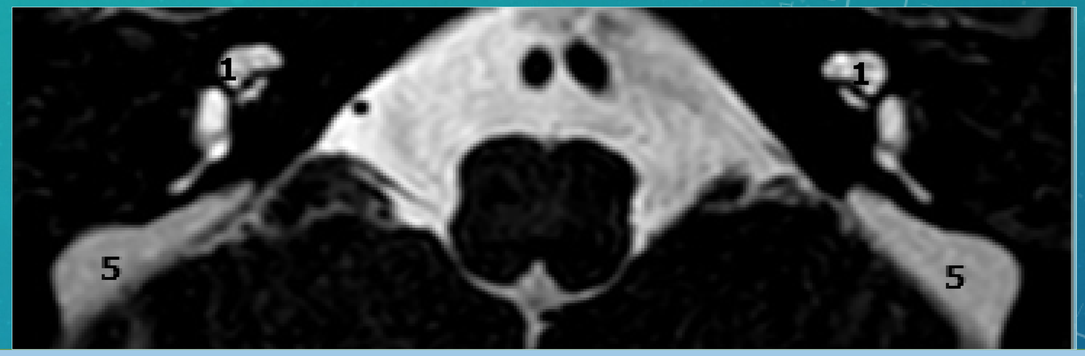
PENDRED SYNDROME

- most common syndromic form of congenital hearing impairment
- SNHL

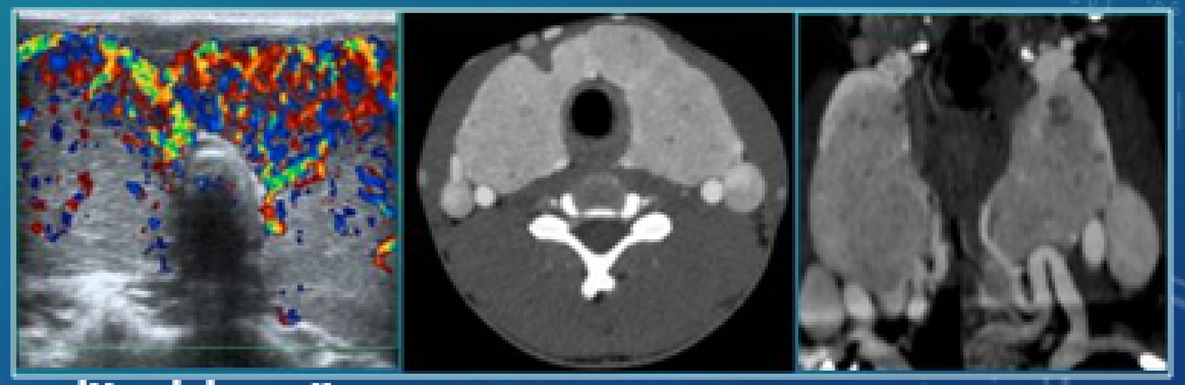


Bilateral MONDINI malformation:

IP-II with cystic cochlear apex (1) & extracochlear findings: enlarged vestibule (2) - LSSC with small central bony island (3) - large vestibular aqueduct (4)



MRI: enlarged endolymphatic sac (5) into the vestibular aqueduct



multinodular goiter

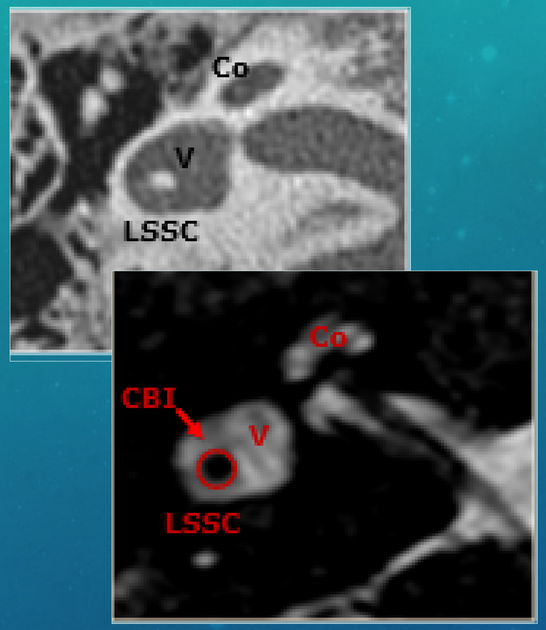
bilateral MONDINI malf. + euthyroid GOITER

PENDRED Syndrome

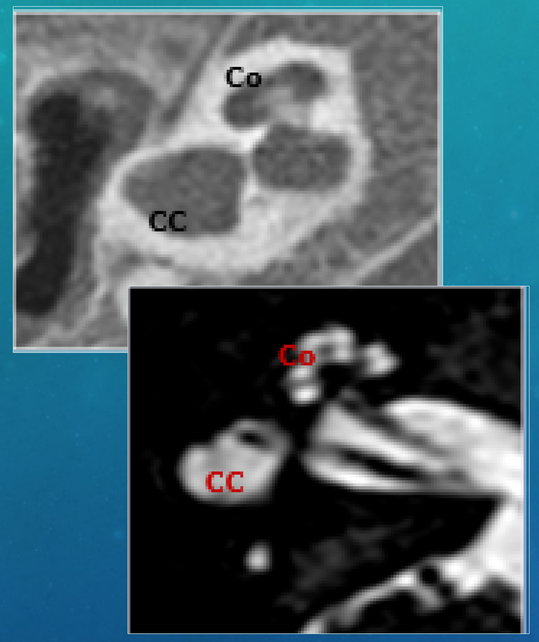
(SLC26A4 g.)

22q11 deletion syndrome (DiGeorge syndrome or velocardiofacial syndrome)

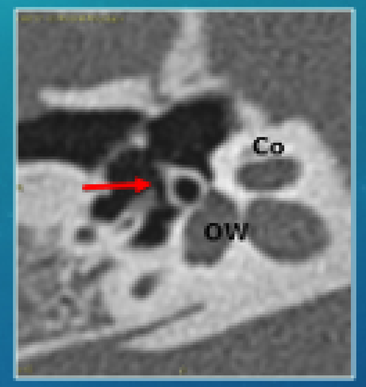
- ❖ cleft lip /palate
- ❖ congenital heart disease (conotruncal anomalies)
- ❖ facial malposition.
- ❖ defective development of parathyroid & thymus
- ❖ mild hearing loss is highly prevalent - 40% (middle & inner ear malf. + repetitive otitis media)
- ❖ mixed conductive & SN hearing loss



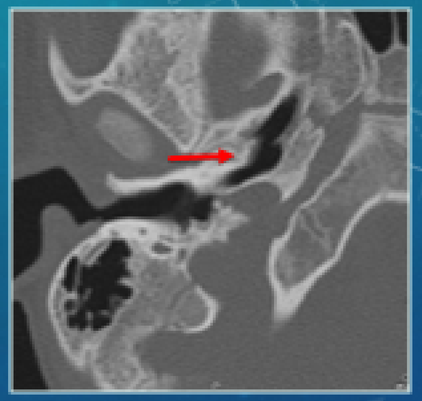
enlarged vestibule & LSCC
with small CBI < 7 mm²



common cavity CC (fusion) between
vestibule & LSSC, absent CBI



thickening & hyperdense
aspect of stapes crura



enlarged Eustachian tube

22q11 deletion

- ▪ posterior labyrinth malf. (***vestibule & LSSC***)
- ▪ middle ear malf. (***stapes***)

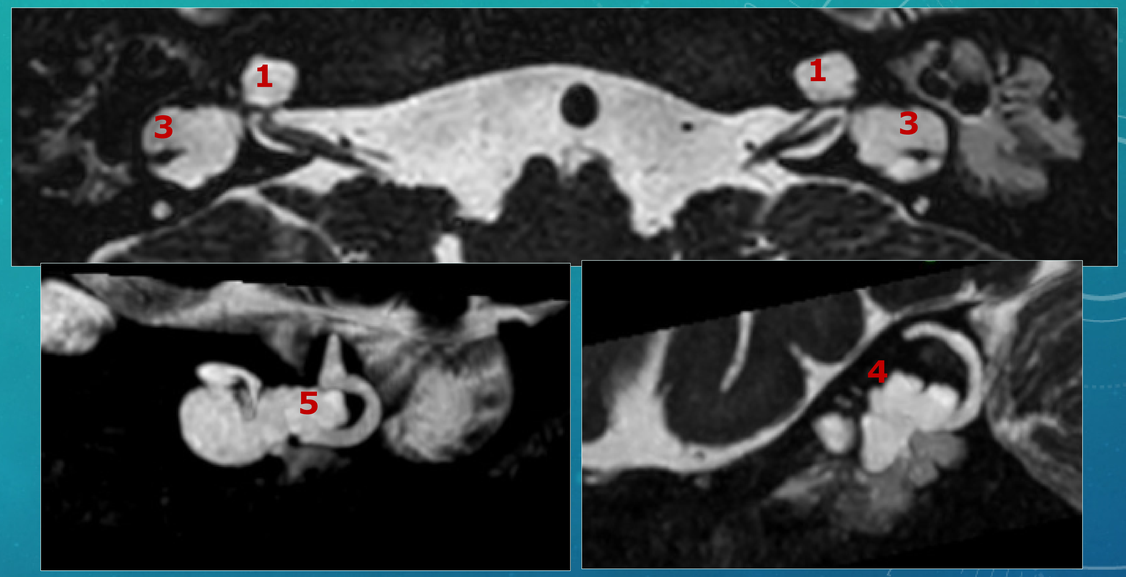
!!! common cavity between vestibule & LSSC or enlarged vestibule with hypoplastic CBI is a common ear abnormality *(also seen for example in Down Sd (T21)*

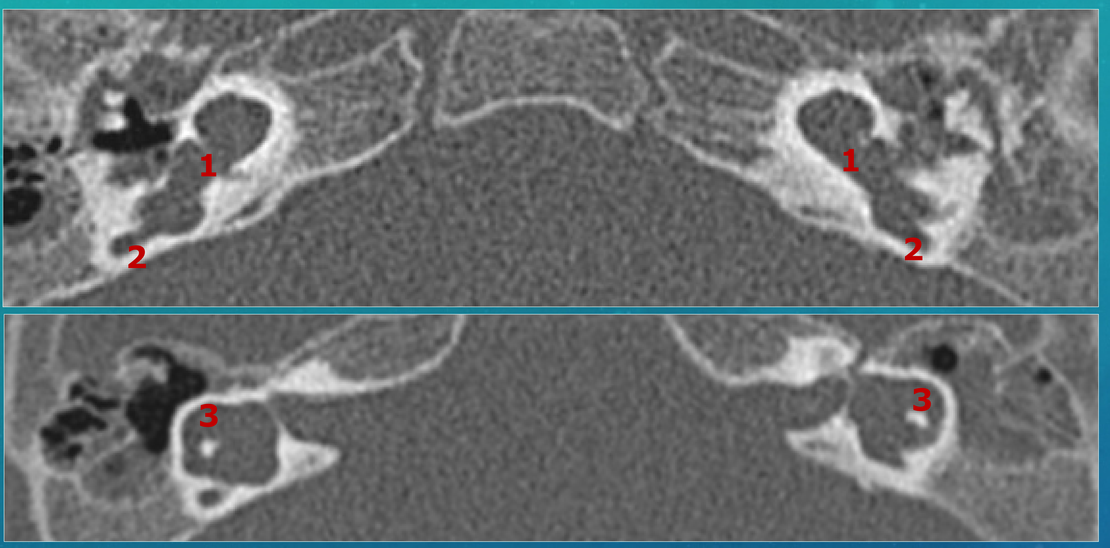
KLIPPEL FEIL syndrome

❖ skeletal & spinal cord malformations (cervical vertebral fusion)

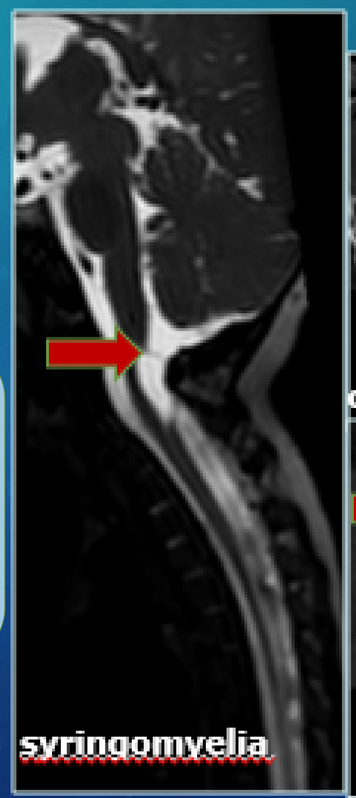
❖ deafness 2nd most common abnormality associated - *30%*

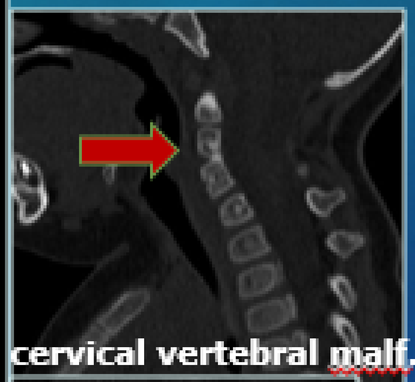
inner ear >>middle ear >external ear

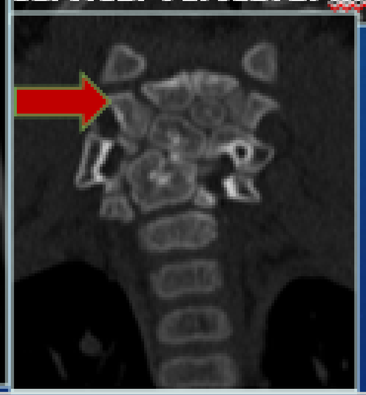




- **bilateral IP-I:** cystic cochlea-vestibular malformation (1)
- **bilateral SSC dysplasia :** enlarged arch of PSSC (2) & LSSC with small CBI (3), partial agenesis of SSCC (4), absence of common crus of S. & P. SSC arches (5)







inner ear malf.

+

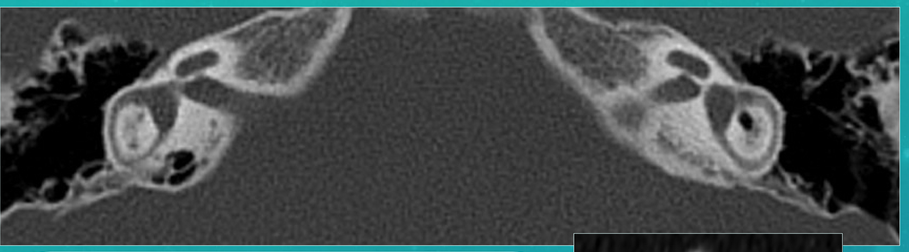
vertebral malf. (++cervical)

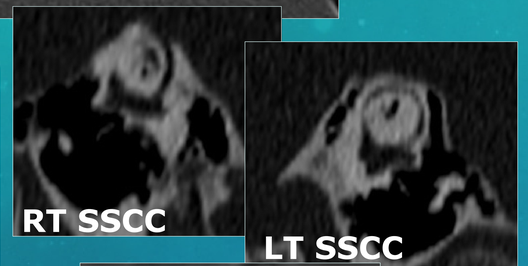
KLIPPEL FEIL

Syndrome

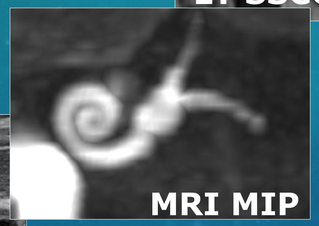
ALAGILLE syndrome (arteriohepatic dysplasia)

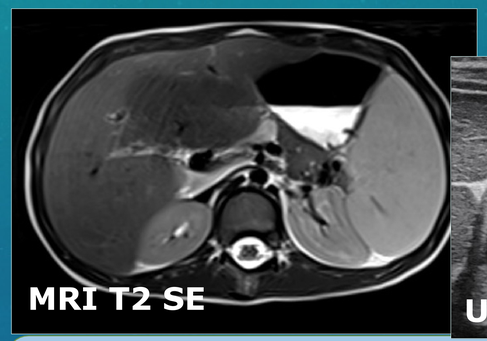
- typically, conductive hearing loss
- dysplasia of PSSC but normal LSSC





- bilateral absence of PSCC
- LSCC & SSCC: thin arch with large CBI
- partial agenesis of the right SSCC





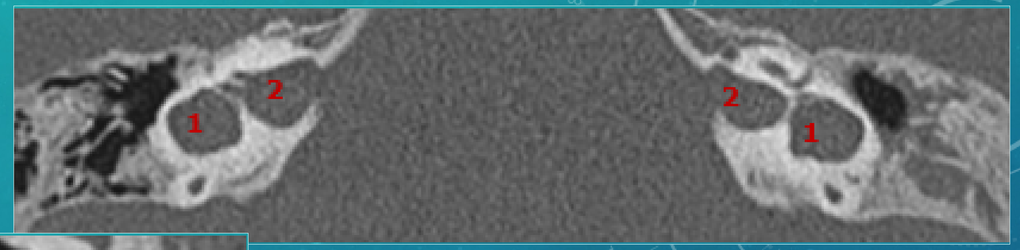
- regenerative hepatic nodules
- thickened gallbladder wall with polyps & echogenic debris

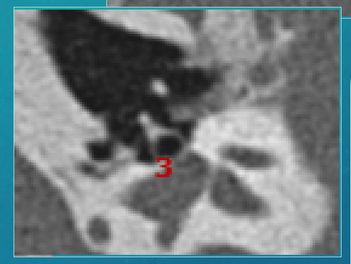
ALAGILLE Sd *(JAG1g. - 90%)*

- dysplasia of PSSC *BUT* normal LSSC
- *hepatic disorders / vertebral malf. …*

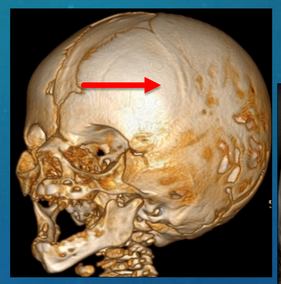
APERT syndrome

- conductive hearing loss associated
- Malf.: vestibule >> LSSC > cochlea

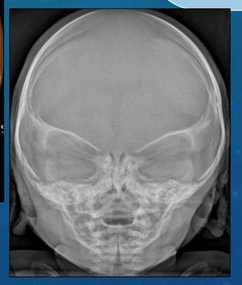




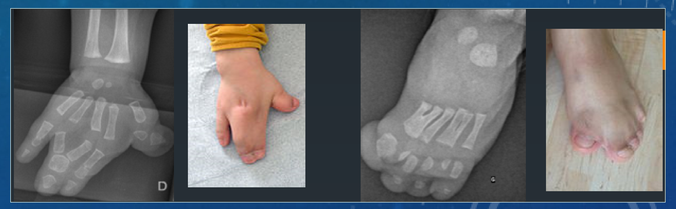
- bilateral fusion between vestibule & LSSC, absence of CBI (1)
- enlarged IAC (2)
- hyperdensity of stapes crus & stapes footplate thickening (3)



premature closure coronal suture



'harlequin orbit'



syndactyly of fingers & toes

APERT Sd *(FGFR2g)*

- vestibular dysplasia
- *craniofacial malf. (craniosynostosis)*
- *syndactyly*

CONCLUSION

- **RADIOLOGICAL SEGMENTAL ANALYSIS**

  Middle ear structures - CT

  Inner ear structures - CT & MRI

- **think of *EMBRYOLOGY***

  1st & 2nd branchial arch malf.

  classification labyrinthine malf. / *Sennaroglou, 2002*

- **CHARACTERISTIC RADIOLOGICAL MALFORMATION PATTERNS**

- **ASSOCIATED FINDINGS, OTHER SYSTEMS**

  - **X- LINKED Deafness**
  - **CHARGE Sd**
  - **BOR Sd**
  - **WAARDENBURG Sd**
  - **PENDRED Sd**