COCHLEAR IMPLANT IN A CASE OF PENDRED SYNDROME- BILATERAL SENSORINEURAL HEARING LOSS WITH HYPOTHYROIDISM AND GOITRE

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ABSTRACT
INTRODUCTION
The clinical symptom of severe SNHL or deafness in children has a genetic background in about 65% of the cases, with 85% of them in a non-syndromic appearance. Concerning the pathogenesis of hearing loss in patients with proven EVA, EVA seems to not be the causative factor for the hearing loss, but more of an accompanying morphological result of an altered fluid balance in the inner ear during embryological development, due to malfunction of Pcondrin

AIMS & OBJECTIVES
We present a child of Indian origin with Pendred syndrome who underwent cochlear implant at Mehrotra ENT hospital, Kanpur, India. Patients with Pendred syndrome represent challenging cochlear implant candidate, combining goiter, severe-to-profound hearing loss, and inner-ear dysplasias.

METHODOLOGY
Cochlear implantation is the proper method for optimal hearing rehabilitation in patients with Pendred syndrome. The genetic background is a mutation of the SLC26A4 gene, coding for a transmembrane protein with anion transport function, called Pendrin [3,4,5]. Child was implanted and rehabilitated at our centre.

CONCLUSION
Outcomes in terms of hearing, speech and quality of life is comparable to cases within on syndromic hearing loss.

KEYWORDS - Pendred syndrome, Pendrin, Hypothroidism, Goiter.

INTRODUCTION
The clinical symptom of severe SNHL or deafness in children has a genetic background in about 65% of the cases, with 85% of them in a non-syndromic appearance[1]. In case of specific accompanying symptoms, the hearing loss is defined as a syndromic form. The most common syndromic form of congenital SNHL or deafness is Pendred syndrome [2]. It is defined as congenital SNHL or deafness and a malfunction of thyroid hormone synthesis. The genetic background is a mutation of the SLC26A4 gene, coding for a transmembrane protein with anion transport function, called Pendrin [3,4,5]. Pendrin is present in multiple organs, such as the kidney, the airways, the thyroid gland and the inner ear. Besides maintenance of ion composition and pH homeostasis between separated compartments in different organs[5], Pendrin plays a special role during embryological development of the inner ear[3]. However, a mutation of the SLC26A4 gene is not mandatory in case of present SNHL. The distribution of SLC26A4 mutations among patients with SNHL differs among different ethnic populations worldwide. In Asian populations
mutations are found with a much higher incidence compared to Caucasian patients [6]. Both malformations, EVA (Enlarge vestibular aqueduct) and MM (Mondini's malformation), are seen as diagnostic hints or radiologic markers for possible Pendred syndrome [7].

Concerning the pathogenesis of hearing loss in patients with proven EVA, EVA seems not to be the causative factor for the hearing loss, but more of an accompany imaging morphological result of altered fluid balance in the inner ear during embryological development, due to malfunction of Pendrin [7].

**CASE REPORT**

A 4 year male child presented in our OPD at Meherotra ENT hospital with complain of loss of hearing and speech since birth. Child also presented with diffuse enlargement in central neck suggestive of goiter and episodes of recurrent diarrhea. Child blood evaluation of thyroid hormones confirmed hypothyroid state. A thorough assessment of hearing was done including BERA (brainstem evoked response audiometry), ASSR (auditory steady state response), free field audiometry, impedance and aided audiogram tests following which child was diagnosed with bilateral profound sensor neural hearing loss along with hypothyroidism and goiter-a case of pendred syndrome.

Thyroid profile was done. Hypothyroidism was first treated with Eltroxin. Once the thyroid profile (T3, T4, TSH) were normal, the child was planned for cochlear implantation. Radiological investigations which include HRCT (high resolution computed tomography) temporal bone and MRI temporal bone was done for detailed anatomical knowledge. HRCT was abnormal and enlargement of vestibular aqueduct was found. Facial nerves, cochlear nerves, superior and inferior vestibular nerves were normal in course, caliber and signal intensity in bilaterally AC and bilateral prefrontal cistern.

CT scan of petrous bone showed enlarged vestibular aqueduct in a patient with Pendred syndrome; the black arrows showed the EVA.

On speech and language evaluation, oral peripheral mechanism (lips, teeth, tongue, hard and soft palate, uvula) were normal in appearance as well as function. The voice parameters (pitch, intensity, quality), articulation (vowel, consonant) and supraregion al aspects (intonation, stress/emphasis, pauses, rhythm, rate of speech) were inadequate. Psychological assessment suggestive of verbal IQ (intelligent quotient) in average range and overall IQ in above average range.

His CAP (categories of auditory perception) level before implantation was 0 (unaware of environmental sounds). SIR (speech intelligibility rating) score was 1 and GCBI (Glasgow children benefit inventory) poor. Comprehension and expression of ling sound were not very good, discrimination of environmental sounds like bird chirp, door knock were very poor and comprehension and expression of vehicles sounds like horns of traffic were poor too. Child was unable to speak family members name like mama, baba.

After all the routine and specific investigation, right ear implantation was done. Pericranial flap
was elevated, mastoid bone drilled, an trum
opened. Posterior tympanotomy done. Round
wind own iiche identified and widened, cochlear
implant receiver placed in bony skull base and
screwed. Round window insertion was done. CSF
gusher was present but was managed and
controlled. Successful impedance testing done
and all electrodes were found working. Flap
closed and wound sutured. Digisonic implant was
used. Post op was uneventful. X ray skull. A P view
was done. Electrodes were in cochlea. Switch on
was done after 10 days.

After 1 year of follow up and regular rehabilitation
at our center by our rehabilitation team, child
attained CAP level of 5, SIR score of 3 and GCB
Index of 51.23suggestiveoof moderate benefit.
Child achieved 80% comprehension and
expression of ling sound. Discrimination of
environmental sound achieved 70% and
comprehension and expression of vehicles sounds 70%.
Child speech of mono and bisyllabic
words improved by 40%. Comprehension of
rhymes improved by 60%

The auditory performances before and after CI
and the comorbid abnormalities of the patients
were analyzed. During their follow-up, the
auditory performance analysis scores improved
nearly the same as the patients in the same ages
without any abnormality.

<table>
<thead>
<tr>
<th>FREEDMPLANT</th>
<th>POSTIMPLANTAND REHABILITATION STATUS</th>
</tr>
</thead>
<tbody>
<tr>
<td>CAP</td>
<td>0</td>
</tr>
<tr>
<td>SIR</td>
<td>1</td>
</tr>
<tr>
<td>GCB</td>
<td>Poor</td>
</tr>
<tr>
<td>Comprehension and expression of ling sounds</td>
<td>Poor</td>
</tr>
<tr>
<td>Discrimination of environmental sounds like dog’s bark, bird songs</td>
<td>Very Poor</td>
</tr>
<tr>
<td>Comprehension and expression traffic sounds</td>
<td>Poor</td>
</tr>
<tr>
<td>Child speech of mono and bisyllabic words</td>
<td>No able to speak</td>
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</tbody>
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DISCUSSION

Pendredsyndromeisinheritedinaautosomalreces
tive manner, meaning that one would need to in
her itan abnormal gene from each parent to
develop the condition. This all so means that a
sibling of a patient with Pendred syndrome has
25% chance of also having the condition if the
parents are unaffected carriers. It has been linked
to mutations in the PDS gene, which codes for the
pendrin protein (solute carrier family 26, member
4, SLC26A4). The genes is located on the long arm
of chromosome 7q31. [9][10] Mutations in the
same gene so cause enlarged vestibular
aqueduct syndrome (EVA or EVAS), another
congenital cause of deafness.

SLC26A4 can be found in the cochlea (part of the
inner ear), thyroid and the kidney. In the kidney, it
participates in the secretion of bicarbonate.
However, Pendred syndrome is not known to lead to
kidney problems.[11] It functions as an
iodide/chloride transporter. In the thyroid, this
leads to reduced organification of iodine (i.e. its in
corporation into thyroid hormone).[9]

People with Pendred syndrome present with a hearing
loss either at birth or during childhood. The
hearing loss is commonly progressive. At thyroid
goitre maybe present in the first decade. MRI
scanning of the inner ear may show widened
large vestibular aqueduct with enlarged
dolymphatic sac and may show abnormalities of
the cochleae that are known as Mondini’s
dysplasia [8]. Not every one with Pendred
syndrome, however, has an abnormal cochlea. All
of those inner ear malformations, including
Enlarged vestibular aqueduct and mondini’s
malformation, might be a result to a defect within
the development of the bony otic capsule.

Genetic testing to identify the pendrin gene
usually establishes the diagnosis.

CONCLUSION

Cochlear implantation is the proper method for
optimal hearing rehabilitation in patients with
Pendred syndrome. Cochlear implant can be an efficient resource for acquiring oral language and reaching complex stages related to hearing abilities, despite the peculiar difficulties caused by the syndrome, it promotes access to sounds, minimizes auditory sensory deprivation, favors interaction with the environment and undeniable quality of life for the child and his family. Although the precise pathogenetic mechanism and the genetic background of deafness in Pendred syndrome have not been completely understood, the pre existing hearing experience represents a positive factor for satisfying hearing outcome. The inner ear malformations if present can cause mild surgical difficulties and extended Surgery duration. The cochlear implant surgeon should be aware of these Difficulties to avoid complications

DECLARATION
Ethics approval and consent to participate: The study was approved by Institutional Ethics committee.

Conflict of Interests- The authors declares that there are no conflicts of interest.

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