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Cochlear Implant in Charge Syndrome - A Single Case Study

Dhulase Krithega. S[1], Yuva Yoga Shree. B[2], Varsha Muthukumar[3]

[1]Professor & Clinical Supervisor, Department of Speech and Hearing, MERF Institute Of Speech And Hearing (P) Ltd, Chennai, India
dhulasesuresh[at]gmail.com

[2]Post Graduate Student, Department of Speech and Hearing, MERF Institute Of Speech And Hearing (P) Ltd, Chennai, India
yuvayogashreeb[at]gmail.com

[3]Undergraduate Student, Department of Speech and Hearing, MERF Institute Of Speech And Hearing (P) Ltd, Chennai, India
varsha.muthukumar[at]gmail.com

Abstract: ***Charge syndrome is a rare genetic disorder which arises during early fetal development due to the mutation of the CHD7 gene in the 8th chromosome. It is also known as the Hall - Hittner syndrome. It is a complex syndrome affecting multiple organs of the body symptoms include coloboma, CN dysfunction, Charge ear Chonal atresia, Congenital heart disease Congenital Heart Defects, Genital Hypoplasia, Hypotonia, TEF, Growth deficiency, CLP, CHARGE face, Renal anomalies this syndrome is often sporadic and sometimes inherited. A two years, Male was accompanied by his parents for detailed Audiological and speech evaluation and diagnosed severe to profound hearing loss Language disorder (receptive and expressive language) consequent to Hearing Impairment. Developmental delay consequent to CHARGE SYNDROME. This case study highlights on Audiological Assessment and management profile on CHARGE Syndrome.***

Keywords: CHARGE Syndrome, Cochlear Implant, Audiological Management

1. Introduction

CHARGE syndrome comprises of an intricate group of congenital abnormalities, initially described by Hall in 1979 and the acronym CHARGE was identified by Pagon in 1981 [1, 2].

‘‘CHARGE’’ stands for (C, coloboma of the eye; H, heart defect; A, atresia of the choanae; R, retardation of growth or development; G, genital hypoplasia; E, ear malformation) [3, 4]. In 1998 Blake simplified the clinical diagnostic criteria by grouping features into major and minor. The major criteria are coloboma, choanal atresia, characteristic ear abnormalities, and cranial nerve abnormalities. Minor criteria are Genital hypoplasia, developmental delay, cardiovascular malformations, orofacial clefts, trachea - oesophagel fistulae and a distinctive face. Clinical diagnosis requires three or more major criteria, or one or more major criteria with at least two minor criteria [2]. The genetic basis of CHARGE syndrome has been recently acknowledged.

A genetic abnormality, a mutation in the CHD7 gene on chromosome 8 [1, 5] is spotted in approximately two - thirds of cases clinically diagnosed with CHARGE [1].

The estimated prevalence of CHARGE syndrome is 1: 10, 000 [9, 11]. It affects equally both sexes [10, 11]. Ear abnormalities [2, 5] and hearing loss are familiar in children with CHARGE [12] and both conductive hearing loss (due to glue ear, ossicular abnormalities or ossicular fixation) and sensorineural hearing loss (due to inner ear abnormalities) may arise [2]. External ear malformations have been characterized in association with CHARGE syndrome, including short and/or hypoplastic pinna with a minimal lobule, a hypoplastic helix or an abnormal concha. The middle ear anomalies includesmal formed or absent ossicles, fixation of the ossicular chain to the wall of the tympanic cavity, absence of the stapedius muscle, absence of the oval window, and obliteration of the round window, Chronic recurrent otitis media is common may result in the conductive component [Dhooge et al 1998, Morimoto et al 2006]. The characteristic abnormalities displayed by temporal bone computerized tomography (CT) or magnetic resonance imaging (MRI) scan include hypoplastic incus, decreased numbers of turns to the cochlea (Mondini defect), and, especially, absent semicircular canals. These distinctive radiological findings are exemplary for CHARGE syndrome and can assist diagnosis in a suspected case [21, 22]. CHARGE is also related with cranial nerve abnormalities [2, 15], specifically involving the olfactory, vestibular and facial nerves, but including the cochlea nerve [2]. Roughly half of them present bilateral deafness with severe to profound severity [4, 13], thus restricting the development of communication in kids [14]. Hence there is a need to restore one impaired sense to facilitate a mode of learning.

2. Case Report

A 2.2 year old, male was brought to the Department of Audiology and Speech Language Pathology with the concern of not speaking age appropriately and not responding to sounds and name calls. He was accompanied by his parents for the Audiological and Speech and Language Evaluation. The child predominantly communicates through differential cry. Genetic testing was done and the results show that the child is a known case of CHARGE syndrome. Natal histories revealed, the child was born of pre - term cesarean delivery with birth weight of 2.4kgs, normal birth color and immediate birth cry (not sure) also the child had respiratory and swallowing difficulties, and was admitted in NICU for 25 days. Motor milestones were observed and reported to be delayed, with head control,

turning over are not yet attained. The child recognizes parents, strangers and other objects through sensation of smell and touch. OPME could not be assessed. The child's intake is predominantly through Gastrostomy and also partially through oral feeding (swallowing) with absence of aspirations. Occupational therapy assessment was done which revels the child has the sensory processing difficulties and emotional behaviors. Psychological evaluation reveals emotional skills and self - help skill is not age adequate and recommended for the physiotherapy. Cochlear implant was suggested for him as he had inadequate benefit with hearing aids and he had hypoplastic nerve. Trans tympanic EABR responses were present so we went ahead for implanting him with cochlear implant.

Table 1: Audiological Evaluation

Test	*Result*
Behavioral Observation Audiometry	Both ears: severe to profound hearing loss.
Immitance Audiometry	Both ears: 'B' type tympanogram Indicative of middle ear pathology
Oto - Acoustic Emission	Both ears: Distortion Product Oto Acoustic Emissions are absent Suggestive of OHCs dysfunction. * Noise floor is too high due to child's noisy breathing
Auditory Brainstem Response	Both ear: ABR V peak could not be obtained even at 90 dBnHL at the rate of 19.3/s using click stimulus in both rarefaction and condensation polarity Indicative of Severe To Profound Hearing Loss
Hearing Aid Trial Hearing Aid Trial: Hansaton Flow Up	Both ears: Aided responses are out of the speech spectrum for both unilateral and bilateral fitting of hearing aids.

Constrains during audiological assessment

- Poor ability to understand requirements of test
- Inability to communicate
- Poor physical mobility: head and neck movement
- Vision problems
- Tactile defensiveness
- Chronic otitis media
- Conductive/mixed hearing loss may be indicated in tympanometry
- Acoustic reflexes absent: cochlear and conductive hearing loss
- children have noisy breathing and swallowing difficulties which would add to the physiologic noise obscuring OAE and ABR results
- Some children with CHARGE syndrome would show resistance to sedation hence testing may be difficult.

Diagnostic evaluation reveals severe to profound hearing loss in both ears. Hearing aid trial shows limited benefit with hearing aid for the development of speech and language, vowing to the candidacy assessment for CI.

Table 2: Radiological Findings

Tests	Findings
Pre - Operative: High Resolution CT Temporal Bone	• Bilateral cochlea appears mildly hypoplastic with less than 2¾ turns. • Bilateral with hypoplasia of superior, posterior and lateral semicircular canals with hypoplasia of vestibule. • Bilateral internal auditory canal and meati appears small in calibre.
Ultra High Field 3 T MRI – Brain & Cochlea	• Right cochlear nerve agenesis. • Left cochlear nerve hypoplasia. • Partial agenesis of bilateral superior semicircular canals and complete agenesis of bilateral posterior and lateral semicircular canals.
Intra Operative Report	• CI surgery was done • The electrode insertion was done with Med - El Sonata Form 19 • Complete Insertion Achieved • Intra - OP Impedance was measured to be satisfactory (<15KΩ), Ground Path Impedance: 1.27KΩ with Integrity and Coupling: OK and ART was present for electrodes (5, 6, 10, 11) • Post - OP facial nerve clinically normal
Post Operative - CT Scan of Temporal Bone	•

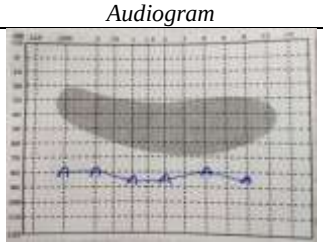
The child had undergone CI Surgery on 26.07.019 implanted with Med - EL SONATA FORM 19 on the LEFT SIDE. Switch on was done on 19.08.2019 IFT's were measured and observed to be satisfactory (<15KΩ), Integrity and coupling was observed to be satisfactory was done and the responses were obtained on electrodes 5, 7, 10, 11, 12 at high stimulation levels, 'M' levels were set based on behavioural responses and ART

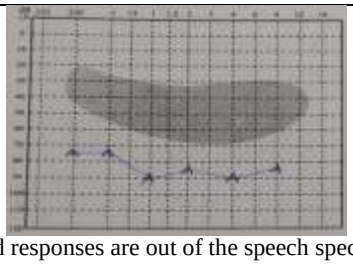
Post operative CT Temporal bone

CT features in the post left cochlear implant status reveals

- Tip of cochlear implant electrode at the apical turn of the cochlea on the left side
- Expected post - operative changes in the left ear
- Bilateral absent semi - circular canals
- Complete soft tissue density opacification of right middle ear cavity and mastoid air cells

Table 3: Shows the Behavioral Observation Audiometry post - surgery

Aided Audiometry	*Audiogram*
First Aided Report Implant Age: 26 Days	 Aided responses are out of the speech spectrum SAL: 75 dBHL Ling sounds: CNT

Current Aided Report - Implant Age: 3 Months 25 days	 Aided responses are out of the speech spectrum SAL: 75 dBHL Ling sounds: CNT

3. Constrains During Fitting Map Levels Using Behavioral & Objective Method

There are many reasons why children with charge can be very difficult to evaluate behaviourally (Thelin, 1999)

- Poor ability to understand requirements of tests because of developmental delay due to illness, sensory deprivation, and /or cognitive delay
- Inability to speak or communicate otherwise
- Poor physical mobility - especially head and neck movement
- Voids in visual fields due to retinal coloboma that makes head movements difficult to interpret in localization task
- behavioral observation during mapping requires the consideration of nonauditory stimulation, which can be subtle and difficult to detect
- In case of objective method there can be challenges faced in fitting based on ART there can be no response in all the electrode or, response obtained at higher thresholds levels or can be mixed responses.
- In EABR there can be no response or poor waveform with facial nerve stimulation observed, ESRT can be affected by middle ear problems, and cause discomfort sometimes
- Balkany et at states that in congenital deaf children there is an aberrant course of facial nerve within the temporal bone
- Cochlear implantation in children with anomalous cochleovestibular anatomy –Blake C. Papsin (2005) which says
- There were numerical programming difficulties (ranging from 1 [easy] to 5 [difficult]) was given to the audiologists, and the results were significantly higher (increased difficulty)
- Children with common cavity deformity and hypoplastic cochlea had reduced dynamic range and increased incidence of facial simulation and were judged to be more difficult to program.
- They required wider pulse width. Older oral children are able to tell the MAP is adequate and often report frequent changes in their ability to hear.
- But there is no way to estimate in case of non - oral young child with abnormal cochleovestibular anatomy because the child is unable to communicate the percept and might be unaware

Challenges Faced during Mapping in this child *on Behavioural Method*

- In BOA there is no consistency of the response exhibited by the child during the stimulus present
- The child is typically delayed in development - motorically, intellectually, and physically – head and neck control and VRA could not be done due to vision problem

On Objective Method

- In ART because of the hypoplastic nerve so responses are obtained in only few electrodes
- In ESRT could not be used because in the left ear there in removal of middle ear structures during surgery and in the right ear cochlear nerve agenesis is present
- Because of cardiac arrhythmia there is limits of establishing of the dynamic range.
- Intermittent usage of the device can be one reason

Overcoming this challenges, the follow up maps are set based on AVT responses and the parents reports.

- Triphasic pulses are provided to eliminate the FN stimulation and it was recommended to use it and follow up.
- EABR is recommended.

Auditory Hablitation

- The child attended 16 AVT sessions in MERF
- The child was observed to be less attentive and requires assistants & support for sitting

Goals worked on:

- Detection of Environmental & ling sounds
- Awareness of LTL sounds
- Provided with multimodality stimulation (Auditory, visual, tactile)

Responses: loud sounds (drums) & LTL - Smiling, sometimes becomes silent, leg movements (leg swinging) and gets excited (loud rhymes are played)

4. Discussion

Intervention of hearing loss in patients suffering from CHARGE syndrome is often complex, also because of other associated diseases, like mental retardation, visual loss, and other severe congenital malformations. Furthermore, the multiple surgical procedures to which these children need to be submitted influence the approach to hearing loss. Cochlear Implant can be a remedy in these patients if conventional hearing aids or bone - anchored hearing aids fail to provide them with optimal hearing abilities [19, 20]. Moreover, if the placement of a cochlear implant (CI) is decided, the frequent manifestation of multiple anatomical anomalies of the middle and inner ear, in association with aberrant course of the facial nerve, can make surgery exceedingly challenging. CI in CHARGE syndrome is not without challenges. The first feature seen in CHARGE syndrome is the cochlear nerve deficiency [23, 20]. Moreover the development of speech and language after implantation can be difficult due to cognitive disabilities or physical handicaps [17]. In spite of reduced speech and language development, most CHARGE patients with cognitive disabilities exhibit more responsiveness and receptiveness to the environment around them after implantation [24.20]. Many children with CHARGE receive

cochlear implants to aid their sensorineural hearing loss. Most also have balance problems (vestibular abnormalities) associated with absent semicircular canals, which is a key finding in making the diagnosis of CHARGE syndrome. [25]There is an increasing number of studies suggest that a Cochlear Implant is a valuable method for the treatment of deafness in ‘‘CHARGE syndrome’ [4, 16 - 18].

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