Measurement of cochlear implant outcomes in goldenhar syndrome: A case report

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ABSTRACT

Goldenhar Syndrome is a rare syndrome which affects most of the characteristic features of that particular individual ranging from inner ear anomalies to renal anomalies. As some individuals have inner ear anomalies, it is important to document the habilitation done for hearing loss. Our article focusses on documenting the outcome of Cochlear Implant over a period of one year in an individual with Goldenhar Syndrome. The paper documents a case of a Goldenhar Syndrome child of two years of age who has undergone Cochlear implantation and outcomes of implantation was measured over a period of one year. Auditory habilitation, behavioural MAPing and parent training were given at regular intervals. Conditioned play audiometry was done at regular intervals to monitor the progress of the child. At the end of one year, speech and language skills and auditory verbal skills were assessed. It was found out that, there was an improvement in all parameters but at a lesser rate, hence concluding there should be adequate stimulation to yield better results.

Keywords: *Goldenhar syndrome, Cochlear implant, MAPing, Auditory habilitation.*

Goldenhar syndrome is a hereditary condition characterized by preauricular appendages, fistulas, epibulbar dermoids and also includes renal, genitourinary, cardiac and skeletal anomalies. In 1952, Goldenhar reviewed these symptoms in 3 patients and named it as Oculoauriculo Vertebral Dysplasia. Later, in 1963 Gorlin et al included vertebral anomalies [1] and in 1978, Smith used the term facio-auriculovertebral sequence that includes Goldenhar syndrome and Hemifacial microsomia [2]. The incidence of this syndrome was found to vary from 1:3500 to 1:5600 live births and 1:1000 in children with congenital deafness, with a male to female ratio of 3:2 [3]. This report provides pre and postoperative profiles of a patient with Goldenhar Syndrome who had undergone cochlear implantation.

CASE REPORT

A two-year-old girl child was brought to the Otorhinolaryngology department with the complaint of not responding to different sounds and delay in speech and language. Birth history reveals neonatal jaundice and birth asphyxia, family history reveals second-degree consanguinity. The child has multiple papillomas, especially in the preauricular areas, limbal dermoid in the left eye and thus diagnosed as Goldenhar syndrome based on the presence of triad features with history of hard of hearing too.

The child had undergone a detail preoperative evaluation which included radiological evaluation (Computed tomography and Magnetic resonance imaging), audiological evaluation, speech and language evaluation, cardiology evaluation, occupational therapy tests, ophthalmological evaluation, biochemistry examination and psychological evaluation. CT revealed small-sized internal auditory canals (left>right) with absent vestibulocochlear nerves bilaterally, thinned out left facial nerve, bilateral otomastoiditis, and normal bilateral cochlea, vestibule and semicircular canals. Three-Tesla MRI was done and on a cross-sectional view revealed the presence of cochlear nerve in the right side which is the same size as that of the facial nerve.

Audiological evaluation included a test battery of pure tone audiometry, immittance audiometry, Otoacoustic emission test, Auditory brainstem response test and Hearing aid trial revealed severe to profound hearing loss in pure tone audiometry, 'B' type tympanogram in both ears in impedance audiometry, bilateral outer hair cells dysfunction in Otoacoustic emissions, severe to profound hearing loss in brainstem auditory evoked potentials and aided responses out of the speech spectrum for hearing aid trial. Speech and language evaluation reveals mixed receptive and expressive language disorder due to hearing impairment with delayed milestones. Electrocardiogram (ECG) was done as a part of cardio electrophysiology testing and reveals normal ECG.

Occupational therapists opinion revealed mental retardation, psychomotor delay, poor comprehension of gestures, poor social and play behaviours with predominance of solitary play and poor social skills, the report also mentions that the child does not

indicate her needs with behaviours like gushing of teeth, hand waving in front of face, temper tantrums with parents reporting left side twitches. Ophthalmologist revealed normal facial symmetry, orthophoric alignment, full ocular movements with normal pupillary reaction, absence of nystagmus and presence of limbal dermoid in the left eye. A psychological evaluation revealed poor eye contact and inadequate attention, poor communication and inadequate self-help skills with 'borderline intelligence'.

The child was advised to undergo cochlear implantation. Surgery was done at the age of four on 17/12/2017 and the child was fitted with MEDEL sonata T_1 STANDARD in the right ear and switch on was done on 05/01/2018 with Interferential therapy (IFT) satisfactory and ARTs measured with an initial program of P_1. The outcomes were analysed over a period of one year. A coding strategy of FS4 was used. Initial responses in January revealed aided responses well below the speech spectrum, MAPing was done on 19/01/18 (1st month) with a new MAP #4 created with 100% volume set in program P_1 and old MAP #2 set in program P_2 at 95% volume. Following aided audiometry on 31/01/2018 revealed aided responses below the speech spectrum but better than previous responses.

MAPing and Aided Audiometry were done at regular fifteen days intervals initially. The Aided audiometry done on 24/04/2018 (3rd month) reveals responses at the lower border of speech spectrum; follow up MAPing was done post aided audiometry. After several follow up MAPing and Aided Audiometry, responses were checked on 18/06/2018 (6th month) which shows similar findings of a below speech spectrum response. After adequate Stimulation and several follow up MAPing, responses were found to be within the speech spectrum for certain frequencies during the 9th month. Current investigations during the month of January 2019 revealed an improvement in speech spectrum response for all frequencies.

Under auditory rehabilitation Category of Auditory Perception (CAP), Meaningful Auditory Integration Scale (MAIS), Speech Intelligibility Rating (SIR) and Meaningful use of speech scale (MUSS) were used to assess the auditory skills of the child. The assessment was done in three-month intervals upto one year. CAP scores reveal a score of 4 in one year from a baseline score of 1, SIR reveal a score of only 2 in one year from a baseline score of one, scores of 16 and 8 were got in MAIS and MUSS respectively in one year from a baseline score of one each. These results revealed that there has been an improvement in scores with respect to auditory skills in one year, but comparatively less progress has been seen. In the context of speech and language, the child's expressive language age has been improved from 3-6 months to 9-12 months, which also shows progress in speech and language development, but at a lesser rate.

DISCUSSION

Goldenhar syndrome is a rare but phenotypically variable condition, with a complex and heterogeneous condition. Origin of this syndrome is unclear but two pathophysiological mechanisms have been proposed – one a reduced blood flow and other is a focal hemorrhage in the developmental region of the first and second branchial arches around 30 to 45 days of pregnancy in the Blastogenesis period, thus explaining outer ear abnormalities as first branchial arch gives rise to anterior ear primordium and second branchial arch gives rise to posterior ear primordium, thus resulting in outer ear, middle ear and inner ear anomalies [4]. Trisomy of a large number of autosomes might be the cause for Goldenhar syndrome, especially trisomy 9 [5].

Temporal bone abnormalities include poorly pneumatised mastoid antrum, enlarged cartilaginous portion of Eustachian tube, absence of cartilaginous lateral lamina of Eustachian tube and lengthened mastoid antrum, outer ear malformations include preauricular appendages, pits, atresia/stenosis of the ear canal, undeveloped tympanic membrane, microtia/anotia, middle ear anomalies include incomplete development of the tympanic cavity, immature and malformed ear ossicles, absent oval and round window, absence of the tympanic tensor muscle, abnormal path of the facial nerve and absence of chorda tympani nerve, inner ear malformations include distorted and hypoplastic cochlea, absence of cochlear aqueduct, immature vestibular system and absence/fusion of semicircular canals, displaced endolymphatic duct, widened vestibular aqueduct, abnormally facial nerve canal, small or duplicated inner ear canal and agenesis of the inner ear canal [4]. Goldenhar syndrome often has a hearing loss of a conductive origin, but a sensorineural component is suspected in some cases, evident through malformations of the inner ear.

Goldenhar syndrome is difficult to differentiate with Hemifacial microsomia and hence a differential diagnosis must be made. Miller in 2004 [6] reported the presence of multiple accessory tragi in a linear distribution from the preauricular skin, along the mandible, to the lateral neck, to be a clue to diagnose Goldenhar syndrome. Feingold in the year 1978 [7] considered lipodermoid, epibulbardermoid or upper eyelid coloboma and two of the three: (a) small size or abnormal shape of the ears or preauricular skin tags or both; (b) unilateral aplasia or hypoplasia of the ramus of the mandible; and (c) vertebral abnormalities to be the features to diagnose Goldenhar syndrome.

Radiological findings in Goldenhar syndrome have been documented by various authors. Pane in the year 2004 [8] did MRI for two patients with Goldenhar syndrome and results revealed severe abnormalities in pons along with moderate cerebellar hypoplasia. Morse [9] in the year 1986 reported absence of portal vein in MRI findings. Berker in 2004 [10] reported congenital facial nerve palsy as a rare symptom. Goldenhar syndrome is mostly present with ear anomalies with most of them presenting with external and middle ear anomalies, very few individuals present with inner ear anomalies and hence there is no correlation amongst external, middle and inner ear anomalies [11].

Two studies have shown cochlear implants fitted in individuals with Goldenhar syndrome. Skarżyński [12] in 2009 documented two case reports where cochlear implantation was done and reported auditory responses at the level of 50 dB and 40 dB respectively in preliminary free field audiometric testing from Implant rehabilitation clinic. MacArdle in 2002 [13] documented a study where they had done Cochlear Implantation in 3 craniofacial anomalies

subject amongst which one was of Goldenhar Syndrome, a full electrode insertion was done and subject demonstrated improvement in detection, recognition, and identification of environmental sounds. Supporting these studies, our findings also had an auditory response level well within the speech spectrum.

CONCLUSION

The outcome of cochlear implant has proven to be good for individuals with Goldenhar syndrome, but in our case prognosis shown was at a slower rate. Adequate stimulation by parents, auditory habilitationalist and regular follow-ups can prove beneficial and speedup the prognosis.

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