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Bilateral Congenital Mixed Hearing Loss with Unilateral Facial Nerve Palsy with Unilateral Microtia: Tushar-Himanshu Syndrome

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Abstract

Earlier studies showed congenital facial nerve paralysis can be seen in combination with genetic syndrome, congenital hearing impairment and structural anomalies of the ear. Present study reported case with congenital mixed hearing impairment with unilateral facial nerve paralysis (left) with unilateral microtia grade 2(left). HRCT of temporal bone also revealed genu of left facial nerve is hypoplastic compare to right side. Audiological evaluation revealed bilateral moderately severe mixed hearing loss with no Middle Latency Response and Cortical Auditory Evoked Potentials in left ear. Considered it as a rare and unique syndrome with no previous literature, hence, termed as 'Tushar-Himanshu Syndrome'.

Keywords: Bilateral congenital mixed hearing loss, unilateral facial nerve palsy

Introduction

Congenital facial nerve palsy is rare disorder that can be divided into two categories i.e. traumatic or developmental. Weakness, twitching or paralysis of face are the sign and symptoms of some ailment involving the facial nerve. Pothiwala and Lateef reported 25 per 100,000 population (0.025%) is the incidence of congenital isolated unilateral facial nerve palsy. Sometimes congenital facial nerve palsy can be seen in association with genetic syndrome and congenital hearing impairment and structural malformation of middle and inner ear. Many times, facial nerve weakness go together with hearing impairment. Mason and Herrman reported 1.4 per 1000(0.1%) as the incidence of bilateral congenital hearing impairment. Hearing impairment may or may not be linked with facial nerve damage. Previous literature has reported that developmental facial paralysis is associated with other anomalies including pinna and external auditory canal from mild defect to severe microtia and atresia (Bergstrom & Baker, 1981).

As we know that the outer ear is developed from 6 hillocks, three of each of the pharyngeal arch 1 and 2 and the external auditory canal is developed from first pharyngeal cleft. It is also a well known fact that development of external auditory canal starts in the late embryo and continues through the fetal second trimester. There are variety of outer ear abnormalities related to

structure, size and position. In few cases these abnormalities is directly related to pharyngeal arch development or possibly linked with a genetic or environmental disorders. A study done by Luquetti et al., in 2012 reported prevalence of microtia from 0.83 to 17.4 per 10,000 births (0.0083% to 0.174%) and they also revealed that prevalence is measured to be higher in Hispanics, Asians, Native Americans, and Andeans. Previous literature has reported that neural crest from the first and second pharyngeal arches forms the cartilage origin of the ossicles, first pharyngeal pouch endoderm develops into Eustachian tube and mesoderm from the first and second arch leads to middle ear muscles (tensor tympani and stapedius muscles) (Ghada,2017). The inner ear is developed from a pair of otic placodes that emerges between 4^{th} and 6^{th} week of gestation period.

Need of the Case Report Study

Earlier studieshave reported congenital hearing loss with facial palsy (Bhatnagar & Prasad, 2015; Vrabec& Lin, 2010; More, Ahuja, Kulkarni, Kulkarni, 2004). There is no literature which has reported bilateral congenital mixed hearing loss with unilateral facial paralysis with unilateral grade 2 microtia(Tanzer, 1971). There is a need to coin the name for such combination of issues. Further, there is no study to the best of our knowledge that has explored detailed audiological evaluation in these population. So, there is a need to do comprehensive audiological evaluation in these population to identify the actual site of lesion.

Aim of the Study

Present case study aims to investigate comprehensive otological and audiological evaluation in a patient with congenital hearing impairment with unilateral facial nerve paralysis with unilateral microtia grade 2 (Tanzer, 1971).

Case Report

A 7 years old boy presented to Department of E N T and Audiology, Sri Jagdamba Charitable Eye Hospital, Sri Ganganagar, Rajasthan, India, with the complaint of bilateral congenital hearing loss with unilateral facial palsy (left) with grade 2 microtia in left ear (Tanzer, 1971).

MRI and CT Scan

HRCT of temporal bone showed maldeveloped left pinna with focus of hyperdensity seen in left auditory canal and adherent to posterior wall. HRCT of temporal bone also revealed genu of left facial nerve is hypoplastic compare to right side and distal tympanic segment of facial nerve is anteriorly located in course. A tiny suspicious sclerotic focus seen in incus bone on both side.

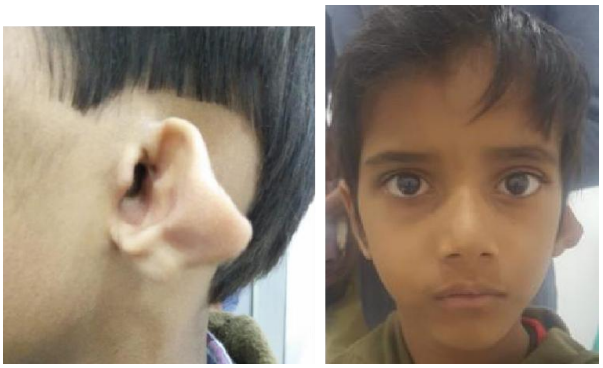

Figure 1: Above pictures of the Patient Cleary shows Grade 2 Microtia. Front View shows no features of Hemifacial Microsomia.

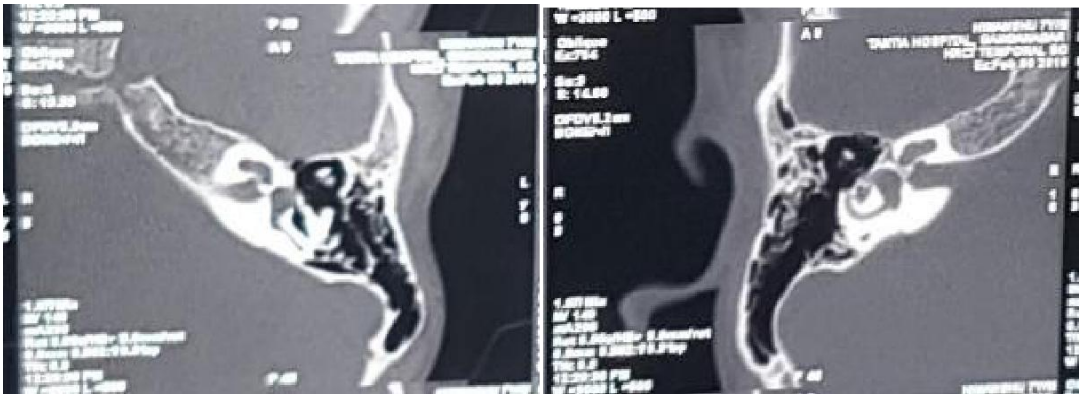

Figure 2: Above figure showing Axial Cuts of Right ear and Left Ear of the Patient at the level of Genu of Facial Nerve . Left Ear shows Hypoplasia at the Level Of first genu as compared to right ear.

Otology Evaluation

Normal and patent external auditory canal, intact tympanic membrane. Cone of light was present in both ears.

Audiological Evaluation

Pure tone audiometry, Speech audiometry, impedance audiometry, otoacoustic emission, auditory brainstem response, middle latency response and cortical auditory evoke response were done as the part of audiological evaluation for the presented case.

Pure tone audiometry, Impedance Audiometry, Speech Audiometry

Pure tone thresholds were obtained using modified version of Hughson and Westlake procedure (Carhart &Jerger, 1959) across octave frequencies from 250 Hz to 8000 Hz for air conduction and frequencies from 500, 1000, 2000 and 4000Hz for bone conduction. Middle ear analyzer (InteracousticAT235) was used to carry out tympanometry using a probe tone frequency of 226 Hz and to obtain ipsilateral and contralateral acoustic reflexes thresholds at 500 Hz, 1000 Hz, 2000 Hz, and 4000 Hz. Speech Recognition Threshold was done where patient was instructed to repeat spondee word as presented by clinician and minimum intensity level where patient can able to repeat more than 50% of the spondee words was considered as SRT. Right and left ear were tested separately.Speech Identification Score (SIS) was calculated monoaurally by presenting 25 PB words in each ear. Percentage was calculated based on total number of correct responses.

Pure tone audiometry revealed bilateral moderately severe mixed hearing loss in both ears (figure 3). Impedance audiometry revealed indication of middle ear pathology in both earSpeech recognition threshold for right ear was 60dBHL and speech recognition threshold for left ear was 6dBHL. Speech identification score for right ear was 64% and speech identification for left ear was 68%.

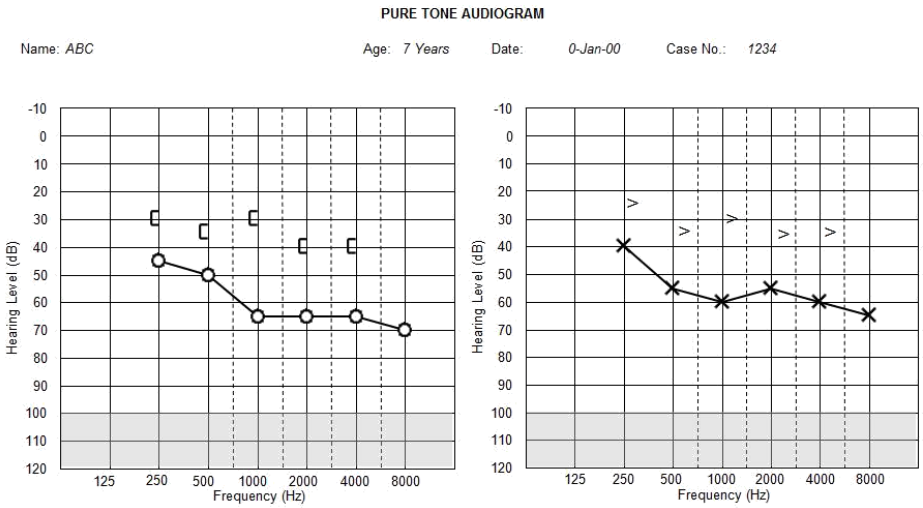

Figure 3: Audiogram of right and left ear.

Auditory Brainstem Response for threshold estimation (ABR)

Click-evoked ABR was carried out to verify the finding of pure tone audiometry. A 2-channel auditory evoked potential (IHS Duet) was used to record ABR. For ABR recording, the patient was made to sleep on a comfortable bed. The skin surface at the two mastoids (M1, M2) and forehead (Fz) was cleaned with skin abrasive to obtain skin impedance of less than 5 kΩ for all electrodes. The electrodes were placed with the help of skin conduction paste and surgical plaster was used to secure them tightly in place. To minimize artifacts, patient was instructed to relax and refrain from extraneous body movements. Testing was done monaurally. The stimulus used for ABR was click of 100 µs in duration presented at the rate of 30.1/sec at rarefaction polarity. The filter setting used was 100Hz to 3000Hz. The total number of sweeps used was 1500 for each acquisition. Insert earphone was used as transducer to present stimulus.

Finding of ABR revealed moderate hearing loss in both ears. Wave V can be traced till 60dBnHL in both ears.

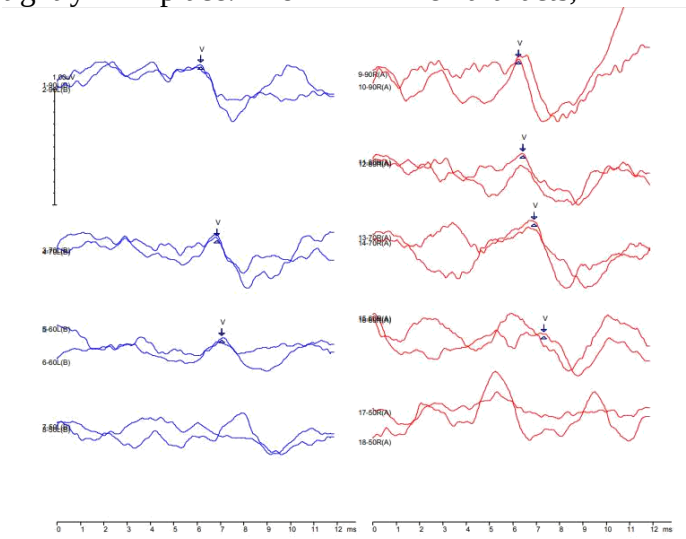

Figure 4: Waveform of ABR for right (red) and left (blue) ear.

Middle Latency Response (MLR) andCortical Auditory Evoked Potentials (CAEP)

A 2-channel auditory evoked potential (IHS Duet) was used to record MLR and CAEP. For MLR and CAEP recording, the patient was made to sit on a reclining chair. The skin surface at the two mastoids (M1, M2) and forehead (Fz) was cleaned with skin abrasive to obtain skin impedance of less than 5 kΩ for all electrodes. The electrodes were placed with the help of skin conduction paste and surgical plaster was used to secure them tightly in place. To minimize artifacts, patient was instructed to relax and refrain from extraneous body movements. Testing was done monaurally.The stimulus used for MLR and CAEP was click of 100 µs in duration presented at the rate of 7.1/sec and 1.1/sec at rarefaction polarity respectively. The filter setting for MLR and CAEP used was 15Hz to 250Hz and 1Hz ro 30Hz respectively. The total number of sweeps used for MLR and CAEP was 1000 and 200 for each acquisitionrespectively. Insert earphone was used as transducer to present stimulus for both the tests. Analysis window for MLR and CAEP was 50ms and 250ms respectively.

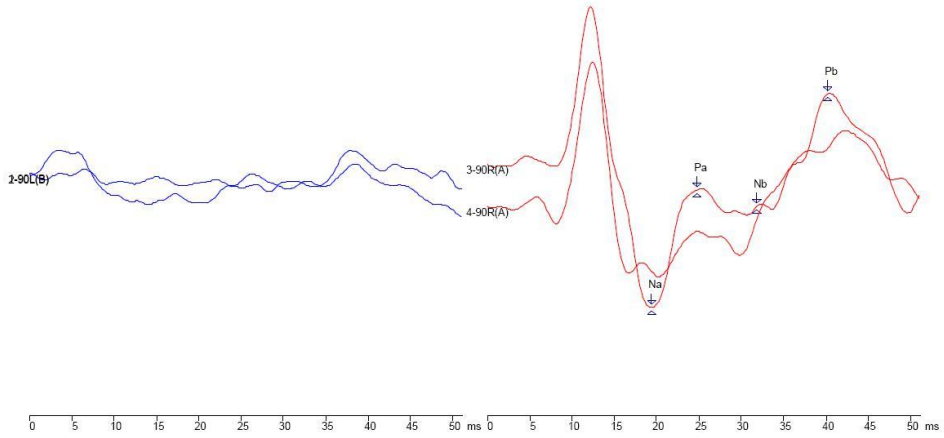

Figure 5: Middle latency response for right (red) and left ear (blue).

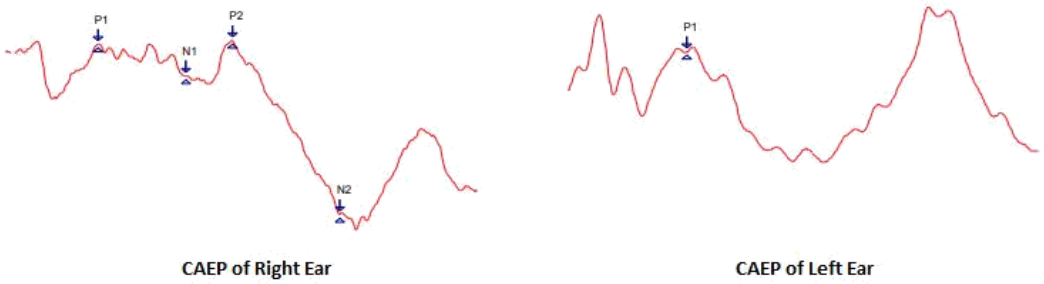

Figure 6: Cortical Auditory Evoked Potential for Right and Left Ear.

Transient Evoked Oto Acoustic Emissions/Distortion Product Oto Acoustic Emissions(TEOAEs and DPOAEs)

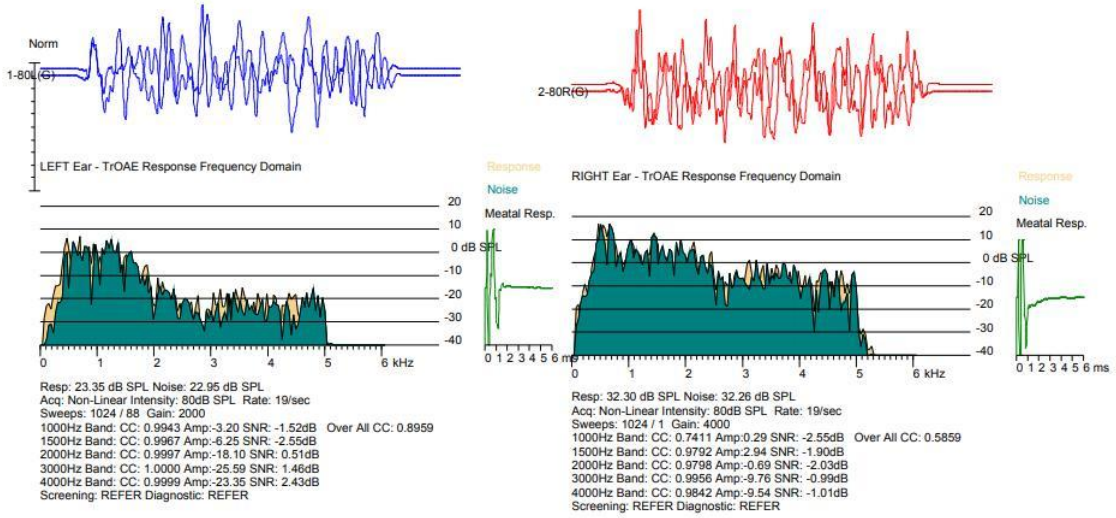

Figure 7: Transient evoked oto-acoustic emissions for both ears.

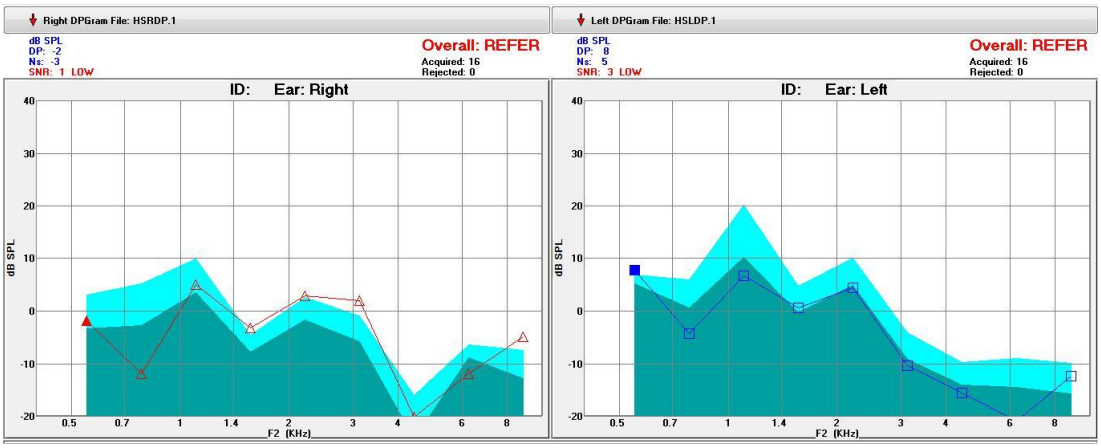

Figure 8: Distortion product oto-acoustic emissions for both ears.

Outcome of MLR revealed absent MLR in left ear whereas MLR was present in right ear with all peaks (Na, Pa,Nb,Pb). Similarly, finding of CAEP revealed poor morphology for left ear, whereas, better morphology was observed for right ear.

TEOAEs and DPOAEs revealed indication of outer hair cells dysfunction in both ears.

Discussion

Microtia states to abnormal growth of external ear leads to deformed auricle. It can varies between mild deformity to complete agenesis of ear. Previous literature has reported similar case study (Bhatnagar & Prasad, 2015; Vrabec& Lin, 2010; More, Ahuja, Kulkarni, Kulkarni, 2004). Congenital facial nerve palsy is a rare condition with incidence of 0.8 to 2.1 per 1000 live births (Falco & Eriksson, 1990). Syndromes which encompass congenital facial nerve palsy are Moebius, Poland's and Goldenhaar's syndrome. Few cases of congenital facial nerve palsy have been accredited to agenesis of the petrous portion of the temporal bone, with resulting agenesis of the facial and auditory nerves, the external ear and the mastoid region (Jemec, Grobbelaar & Harrison,2000).Earlier studies have also reported congenital facial neve

palsy with hearing impairment (Carvalho, Song, Vargervik&Lalwani, 1999; Gathwala, Singh &Dalal, 2013). A case study done by Carvalho, Song, Vargervik&Lalwani in 1999 reported auditory and facial dysfunction in patients with hemifacial microsomia. Similarly, Jankauskiene and Azukaitis in 2013 reported a rare case with congenital facial nerve palsy in association with branchio-oto-renal (BOR) syndrome. Gathwala, Singh and Dalal in 2013 reported a case of congenital facial palsy with bilateral anotia and external auditory canal atresia. Stanley et al. in 1988 reported mixed hearing loss in Larsen syndrome. They also reported that Larsen syndrome, characterized by multiple joint dislocations and bony malformations, the ossicular joints may also be affected. No literature till date reported facial nerve palsy and outer ear abnormalities in Larsen syndrome. Kumar in 1990 revealed the characteristics features of Moebius syndrome i.e. facial paralysis, paralysis of sideways (lateral) movement of the eyes, short, malformed tongue and/or an abnormally small jaw (micrognathia), cleft palate and missing teeth. Similarly, Feingold and Baum discussed the Goldenhar's Syndrom mainly affects the development of the eye, ear, and spine. The characteristics features are facial asymmetry, microtia or anotia, ocular dermoid cysts and spinal abnormalities. Goldenhar syndrom may also affect the heart, lungs, kidneys, and central nervous system. Vento et al., 1991 stated Hemifacial microsomia is a condition in which the structures on one side of the face are smaller or underdeveloped relative to the other side. The other sign and symptoms of hemifacial microsomia are abnormalities in the outer ear, small maxillary, temporal and malar bones, hearing impairment due to middle ear abnormalities, cleft lip and palate, eyes abnormalities, skeletal abnormalities including problem of the spine and rib. The present case was not matching with the cases reported in previous literature and any syndroms. The case presented at out department was having congenital grade 2 microtia in left ear with bilateral moderately severe mixed hearing loss with left side facial palsy. These finding reveals pathology is located at both conductive and sensory portion of the ear. The finding of auditory brainstem response showed intact cochlear nerve, cochlear nucleus and nucleus of lateral lemniscus in both ears. Middle latency response revealed presence of MLR in right ear indicates normal functioning of sub cortical regions of auditory system and superior temporal gyruswhereas MLR was absent for left ear Similarly, cortical auditory evoked potentials (CAEP) were present for right ear whereas morphology of CAEP was poorer for left ear. Normal CAEP for right ear indicates normal functioning of primary auditory cortex, heschl's gyri, cingulate gyrus planum temporale, auditory association areas in the lateral temporal and parietal lobe. TEOAEs and DPOAEs were absent in both ears revealed outer hair cells dysfunction.

Conclusion

The present case study was unique and there is no literaturewhich has reported bilateral congenital mixed hearing loss (but with unilateral cortical deafness) with unilateral facial paralysis with unilateral grade 2 microtia (Tanzer, 1971). The features of the patients suggest that patient had incomplete development of first and second pharyngeal arche s but the presentation does not fit into any of the syndroms related to the above arches reported in the literature. We have considered it as a rare syndrome and termed as 'Tushar Himanshu Syndrome'. Current case study also showed significance of detailed audiological investigation in similar syndrome. The way forward is to work toward the genetic causes and environmental factors underlying this presentation.

Conflict of Interest: None reported

Acknowledgement

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Reference

Baum, J. L., & Feingold, M. (1973). Ocular aspects of Goldenhar's syndrome. *American Journal of Ophthalmology*, *75*(2), 250-257.

Bergstrom, L., & Baker, B. B. (1981). Syndromes associated with congenital facialparalysis. *Otolaryngology--Head and Neck Surgery*, *89*(2), 336-342.

Bhatnagar, R., & Prasad, P. L. (2015). Rare Association of Bilateral Anotia with Congenital Facial Palsy. *Journal of Nepal Paediatric Society*, *35*(2), 195-197.

Carhart, R., &Jerger, J. (1959). Preferred method for clinical determination of pure-tone thresholds. *Journal of Speech & Hearing Disorders*, *24* (4), 330-335.

Carvalho, G. J., Song, C. S., Vargervik, K., &Lalwani, A. K. (1999). Auditory and facial nerve dysfunction in patients with hemifacial microsomia. *Archives of Otolaryngology–Head & Neck Surgery*, *125*(2), 209-212.

Falco, N. A., & Eriksson, E. (1990). Facial nerve palsy in the newborn: incidence and outcome. *Plastic and reconstructive surgery*, *85*(1), 1-4.

Gathwala, G., Singh, J., &Dalal, P. (2013). Congenital facial palsy with bilateral anotia. *Indian J Clin Pract*, *23(2)*, 628-34.

Ghada, M.W.F. (2017). Ear Embryology. Global Journal of Otolaryngology, 4(1), 178-183.

Jankauskienė, A., &Azukaitis, K. (2013). Congenital unilateral facial nerve palsy as an unusual presentation of BOR syndrome. *European journal of pediatrics*, *172*(2), 273-275.

Jemec, B., Grobbelaar, A. O., & Harrison, D. H. (2000). The abnormal nucleus as a cause of congenital facial palsy. *Archives of disease in childhood*, *83*(3), 256-258.

Kumar, D. (1990). Moebius syndrome. *Journal of Medical Genetics*, *27*(2), 122.

Luquetti, D. V., Heike, C. L., Hing, A. V., Cunningham, M. L., & Cox, T. C. (2012). Microtia: epidemiology and genetics. *American Journal of Medical Genetics Part A*, *158*(1), 124-139.

Mason, J. A., & Herrmann, K. R. (1998). Universal infant hearing screening by automated auditory brainstem response measurement. *Pediatrics*, *101*(2), 221-228.

More, V., Ahuja, S. R., Kulkarni, H. V., & Kulkarni, M. V. (2004). Bilateral anotia with congenital hypothyroidism. *The Indian Journal of Pediatrics*, *71*(4), 369-370.

Pothiawala, S., & Lateef, F. (2012). Bilateral facial nerve palsy: a diagnostic dilemma. *Case reports in emergency medicine*, *2012* (2), 234-239.

Stanley, C. S., Thelin, J. W., & Miles, J. H. (1988). Mixed hearing loss in Larsen syndrome. *Clinical Genetics*, *33*(5), 395-398.

Tanzer, R. C. (1971). Total reconstruction of the auricle: The evolution of a plan of treatment. *Plastic and reconstructive surgery*, *47*(6), 523-533.

Vento, A. R., Labrie, R. A., & Mulliken, J. B. (1991). The OMENS classification of hemifacial microsomia. *The Cleft Palate-Craniofacial Journal*, *28*(1), 68-77.

Vrabec, J. T., & Lin, J. W. (2010). Inner ear anomalies in congenital aural atresia. *Otology & neurotology*, *31*(9), 1421-1426.