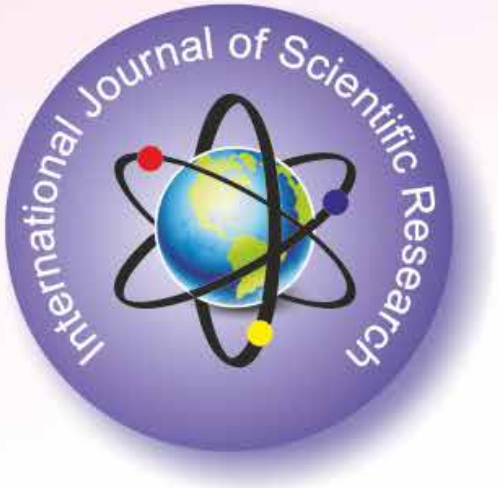
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Research Paper

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Audiological Profile In Osteogenesis Imperfecta: A Case Report

Medical Science.

KEYWORDS :Osteogenesis Imperfecta, hearing loss.

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ABSTRACT

Osteogenesis Imperfecta is a rare bone disease which affects all the bones in the body including the middle ear ossicles making them fragile. This condition is characterized primarily by frequent bone fractures and complications like respiratory, cardiac, neurological complications, and hearing loss. Hearing loss is usually believed to occur and progress in the middle ages. For audiologists, thus it is important to understand the Audiological profile observed in this population for providing effective ways of management. Current case report is an evidence of a 5 year old child with Osteogenesis Imperfecta having initial stages of hearing impairment and its management. This report is aimed at improving the existing literature and to highlight the fact that hearing loss could appear early in life in the reported population.

LINTRODUCTION

Osteogenesis Imperfecta (OI) as described by Osteogenesis Imperfecta foundation (1970) is a heterogeneous disease of the connective tissue caused by a defective gene that is responsible for the production of collagen type I, leading to defective bone matrix and connective tissue. In other words, it is a bone disease that is characterized by fragile or easily breaking bones and hence it is also known as "brittle bone disease." The incidence exploration of this condition by Osteogenesis Imperfecta foundation (2010) revealed 27,199 children to be born with OI every year in the USA and the prevalence is found to be 6-7/100,000 worldwide. Statistics of OI in India is currently unavailable but nevertheless it is a worldwide epidemic. The major causes being genetic namely, Autosomal dominant inheritance, mutation and Mosaicism. This condition is primarily characterized by easy fracturing of bones, growth deficiency, abnormal teeth, thin skin, blue sclera, overly extensible joints. The complications of this condition are cardiac problems, respiratory distress, and hearing loss. Thus, an audiologist needs to be aware of this relatively rare condition and Audiological profile that can be expected in such cases and how can it be effectively managed. Knowledge of different types of OI is also essential as hearing impairment varies in degree, onset and course in each type. When hearing loss is very common in type I and type III OI, it may or may not appear in type IV OI.

CASE REPORT

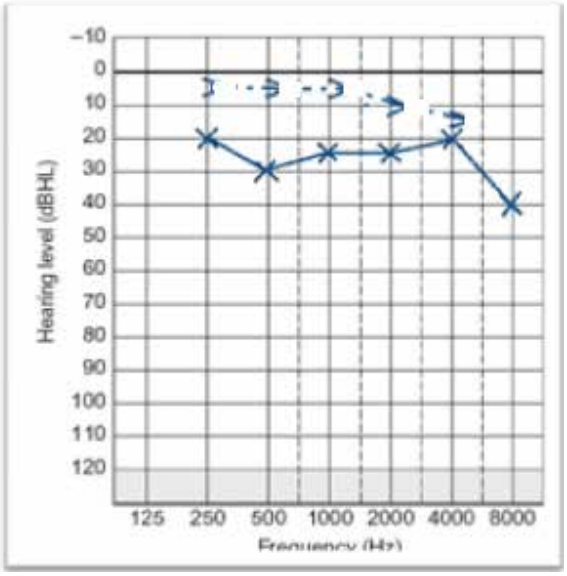
A 5 year old child diagnosed with Type I OI by the department of pediatrics in our hospital was referred to department of head & neck surgery for otological examination. Otoscopy was done following which the case was referred for Audiological consultation. The case was subjected to Pure Tone Audiometry using calibrated GSI-61 clinical audiometer through TDH-39 headphones and BC-71 bone vibrator with noise levels within permissible noise level standards (ANSI S3.1-1999) in the testing room. The child was also subjected to Tympanometry and Reflexometry carried out by a calibrated GSI-Tympstar tympanometer. Tympanometry was carried out with 226Hz probe frequency and 50dapa pump rate. Reflexometry was carried out at 500Hz, 1kHz, 2kHz and 4kHz.

RESULTS

Table 1 Pure tone Audiometry thresholds.

Ear	250 Hz	500Hz	1kHz	2kHz	4kHz	8kHz
	AC BC dBHL	AC BC dBHL	AC BC dBHL	AC BC dBHL	AC BC dBHL	eAC BC dBHL
Right	10 5	15 10	15 5	10 10	10 5	20 -
Left	20 5	30 5	25 5	25 10	20 15	40

Figure 1 Pure tone audiogram



Impression: Right ear- Normal Hearing sensitivity.

Left ear- Mild conductive hearing loss.

Table 2 Tympanometry & Reflexometry.

Ear	Static compliance	Acoustic reflexes
Right	1.1 ml	Present
Left	3.0 ml	Absent

Impression: Right ear- Normal middle ear functioning.

Left ear- Middle ear pathology present.

DISCUSSION

Hearing loss in OI is a symptom which occurs in the second and third decade of life (Pedersen, 84). Hearing loss in OI differs with the type and progressive hearing loss is a striking

characteristic and affects 50% of adults (Kuurila, Triche, Rowe & Munabi, 2000). Regarding the type of hearing loss, conductive is the most common type which accounted up to 39% of the ears examined and sensorineural loss (SNHL) was observed in 11% of the ears examined (Pedersen, 1984). Shapiro (1982) reported Sensorineural hearing loss to occur majorly in older population (older than 30 years) and conductive hearing loss to occur majorly in younger population (younger than 30 years). A mixed hearing loss can also be found.

The reported case had conductive hearing loss in left ear which was identified at the early stages and this provides an evidence for a probable onset of hearing loss much early in life. Increased compliance of the middle ear and absent acoustic reflexes are suggestive of anomalous ossicular articulation (Shapiro, 1982). When hearing loss in OI is viewed as a late appearing symptom, the early stages of the loss can be missed out. This is supported by the findings of hearing loss in 17 out of 22 children (mean age of 9 years) with OI (Imani, Vijayasekaran & Lannigan, 2003). However, the current case acquired hearing loss unilaterally quite earlier in life (5 years) than the hold of literature which says 2nd or 3rd decade. Thus, this calls for a regular monitoring of hearing starting from the point of diagnosis of the condition.

The probable explanation for the conductive hearing loss in Osteogenesis Imperfecta could be caused by Ossicular discontinuity due to aplasia or fracture, Stapedial fixation or stapedial crura fracture. Pathophysiology of SNHL in OI isn't explained in literature. However, it can be believed to be a result of bony labyrinth involvement of the inner ear.

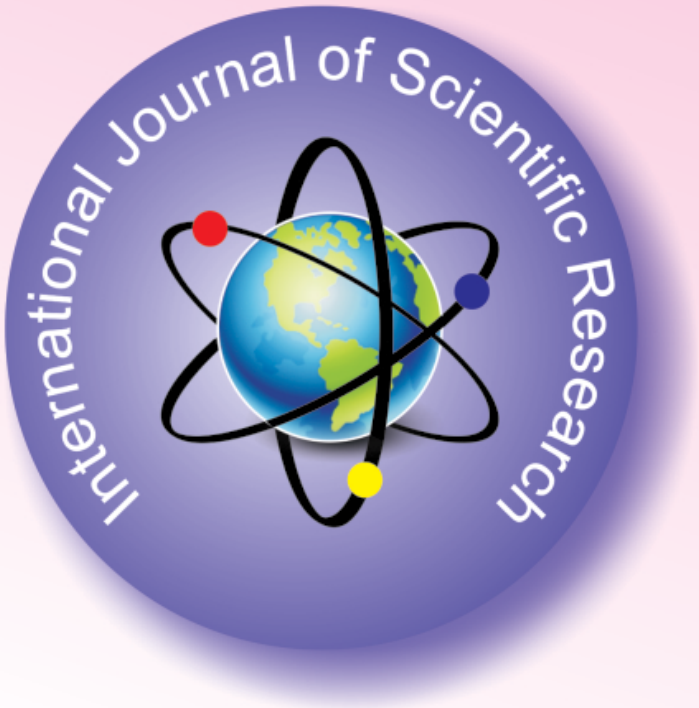
Management of hearing loss in OI should focus on preventing damage to the fragile middle ear ossicles that can be caused by loud sounds and the supportive hearing prostheses if required should be provided with noise reduction strategies and output limiting parameters clearly defined. The current case was prescribed hearing protective devices in the outside environments to prevent a probable acoustic trauma.

CONCLUSION

Hearing loss in OI could also develop early in life and thus hearing has to be periodically monitored. Osteogenesis Imperfecta foundation prescribes hearing screening early in childhood and follow up once in 3 years. The type of loss can vary but conductive type is the most common. Audiological evaluation should involve Pure Tone Audiometry, Tympanometry and Reflexometry. Management of hearing in OI should be both protective and supportive depending on the needs of the client.

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