ABSTRACT

Osteogenesis imperfecta (OI) is a genetic disorder that affects the synthesis of collagen in the body. It is also known as 'Brittle Bone Disease'. It is heterogenous in its clinical presentation. The commonest presentation is a history of frequent fractures, joint deformities and blue sclera. Secondary deformities of the extremities, spine, skull as well short stature frequently observed.

Hearing loss has been well documented to occur in OI. It is most commonly seen in types I, II and III. Hearing loss forms part of the diagnostic criteria for these types. Depending on the study, the prevalence of hearing loss in children with OI is between 6.7 % and 77.3%

The estimated prevalence of OI is 1 in 20000. In South Africa, the commonest type of OI was found to be Type III. The prevalence of OI Type III has been estimated to be between 1:125000 and 1:200000 . Hearing loss is a common feature of OI Type III. Studies such as this will contribute to the body of knowledge of OI and will in future help researchers identify the cause of the hearing loss.

METHODS

This study was a prospective cross-sectional study. Ethics Approval was obtained from the University of Witwatersrand Ethics committee (Ethics number M190975). Children with OI attending the Metabolic Bone Clinic at Chris Hani Baragwanath Academic Hospital were the target group. The patients and their parents or guardians were recruited at the clinic after a consent and or an assent was obtained. An otoscopy followed by tympanometry and a hearing screen based on the age of the patient was done.

DPOAE (Distortion Product Oto-Acoustic Emission) tests were also done as a screening test to confirm the pure tone audiogram findings. The results were given to the patients and their parents/ guardians immediately.

RESULTS

The paediatric patients with OI who consented to take part in the study had their hearing screen done at the Audiology Department at Chris Hani Baragwanath Academic Hospital. All of the children were found to have normal hearing. On tympanometry, all except 2 were found to have type A curves in bilaterally. Two patients had a type As curve in one ear with an A curve on the other side.

CONCLUSION

Hearing loss in OI forms part of the diagnostic criteria for certain types of this genetic disorder. Hearing loss in the paediatric patients does not seem to be as prevalent as previously thought. All the patients involved in the study received the bisphosphonate therapy (Zoledronic acid) for OI. This may possibly cause a delay in the onset of hearing loss but long term follow-up studies and bigger sample sizes will be required to prove this hypothesis.