RFP Journal of ENT and Allied Sciences
Volume 9 Number 2, July-December 2024

Van Der Hoeve's Syndrome: A Rare Case Report

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How to cite this article:

Avinash Kumar, Ekta Yadav, Garima Sinha, *et al.*, Van Der Hoeve's Syndrome: A Rare Case Report. RFP J ENT Allied Sci 2024;9(2):107–111.

Abstract

Van der Hoeve's syndrome is an all-round variant of Lobstein's disease (late osteogenesis imperfecta) and it is characterized by the presence of osteoporosis, deafness and blue sclera. Earlier it was known as Adair Dighton Syndrome (Dighton, 1912). Vrolik in 1849 described this condition for the first time a in new-born infant who was having poorly ossified calvarium and called it osteogenesis imperfecta.The temporal bone may also be affected by this generalized abnormality of the skeletal system, which may result from a specific dominant gene abnormality. Here we discuss a case of 23yr old female with blue sclera presented to ENT OPD with chief complaint of bilateral decreased hearing from last 6yrs and unilateral ringing sensation in ear from 2yrs. Patient was also having generalised body weakness. In this article we discuss how we managed the case conservatively after proper blood and radiological investigations

Keywords: Vander hoeve syndrome, Blue sclera, Osteogenesis imperfecta, Deafness, Osteopenia.

INTRODUCTION

Van der Hoeve's syndrome is an variant of Lobstein's disease (late osteogenesis imperfecta) and it is characterized by the presence of osteoporosis, deafness and blue sclera. Earlier it was also known as Adair Dighton Syndrome (Dighton, 1912). Blue sclera occurs due to the blue hue of underlying choroidal vessels which are visible through the thin abnormal collagen tissue of sclera and it is usually non-curable and may persist throughout life. Osteogenesis imperfect a is a rare connective tissue disorder of autosomal dominant inheritance which is caused by an error in the formation of collagen type I. Mutations in chromosome 17 or 7 result in a decreased synthesis of normal collagen type 1 and synthesis of structurally abnormal collagen type 1

CASE REPORT

A 23 year old female presented to ENT OPD with chief complaint of decreased hearing in both ears from last 6yrs and ringing sensation in left ear from last 2 years. On otoscopic examination

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Received on: 21-01-2025 **Accepted on:** 26-02-2025

both tympanic membrane were intact with mild retraction. On Pure Tone Audiometry right ear had moderate conductive hearing loss and left ear had severe mixed hearing loss. **Fig. 3** is the audiogram which shows bilateral hearing loss. Facial nerve function was normal.

Sclera of both the eyes was blue in color since birth with no history of decresed or blurred vision. **Fig. 1** shows blue sclera of the patient. Ophthalmology reference was done which was suggestive of normal visual acuity.

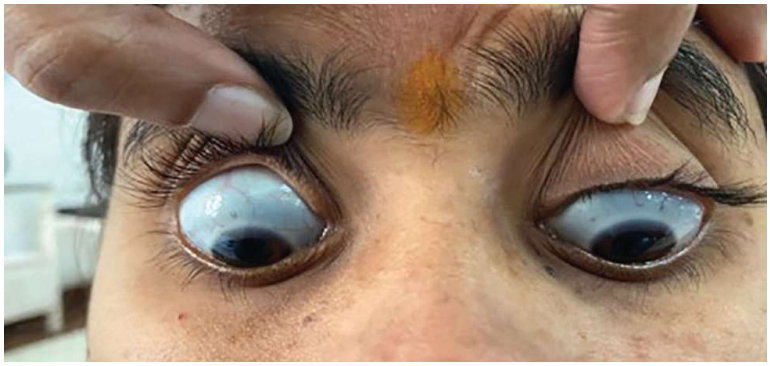

Fig. 1: Showing blue sclera

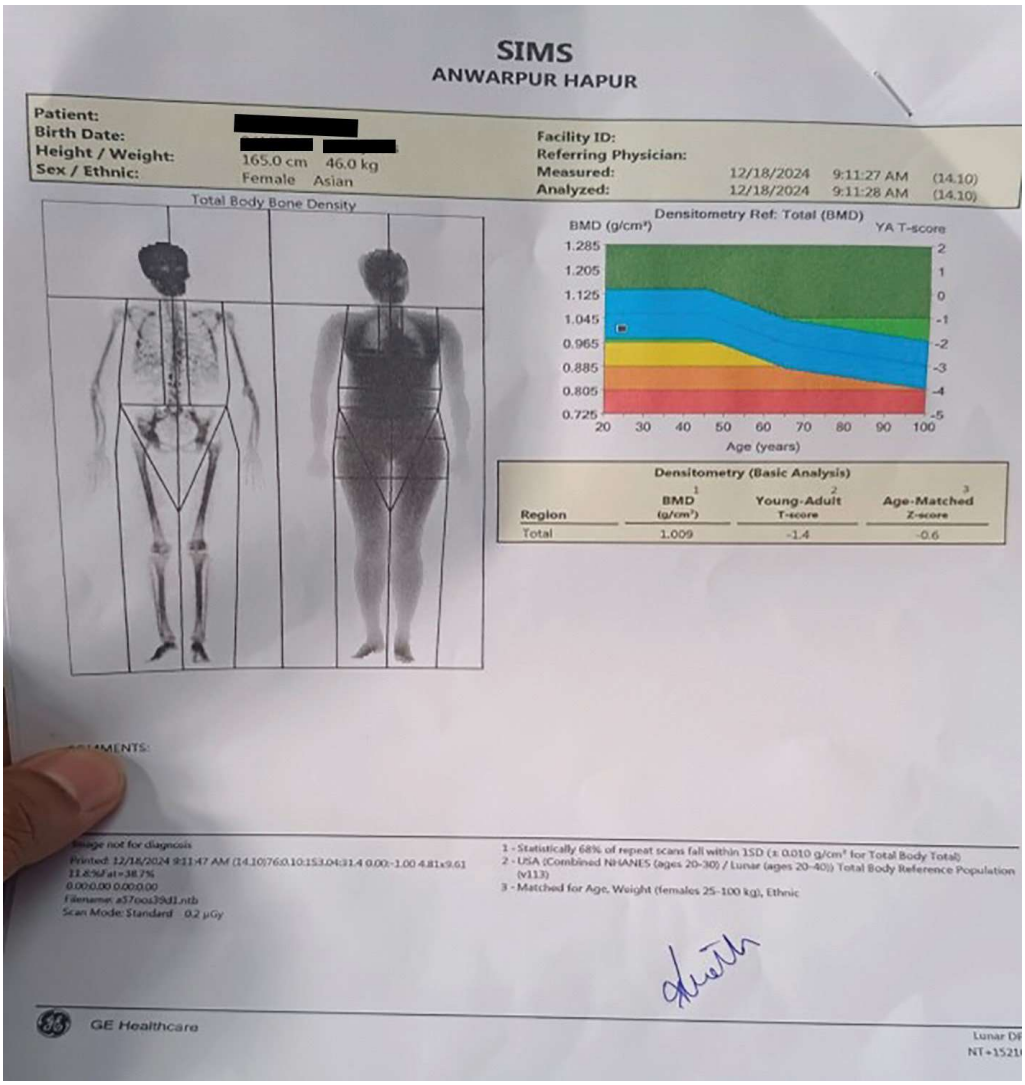

Fig. 2: Bone densitometry showing osteopenia

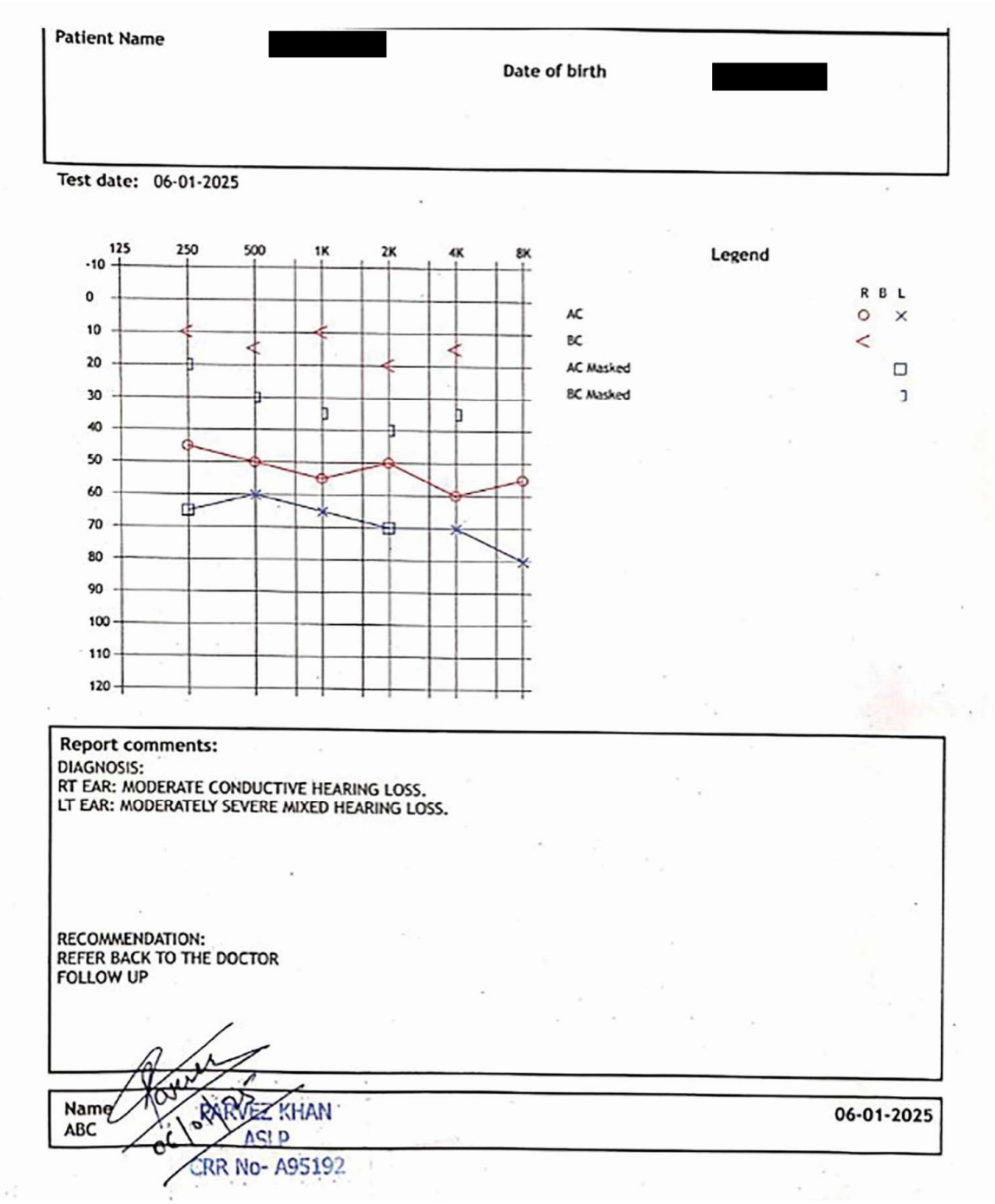

Fig. 3: Audiogram showing bilateral conductive hearing los

She also complained of weakness in joints especially lower limbs with history of fracture of right lower limb 2 yearsyrs back after a trivial trauma. DEXA scan was suggestive of BMD 1.009g/cm^2 and xray of both wrist showed no features of malunion or non union of bone.

Patient was managed conservatively with Topical drugs, calcium and vit. D supplements and multivitamins

INVESTIGATIONS DONE

Complete blood picture: low Hb 8gm/dl

RBC count - 5.19m/cumm

Increased RDW - CV 16.00%

PTA: Right ear - Moderate conductive hearing loss

Left ear - severe mixed hearing loss

Ultrasound B-scan: Normal for both eyes

DEXA Scan: BMD: 1.009g/cm2

T-score: -1.4

Z score: -0.6

Fig. 2: shows the bone densitometry with the Tscores and Z scores depicting osteopenia.

DISCUSSION

The disease is characterized by abnormal fragility of the bones, defective dentition, osteopenia, ligamentous laxity blue color of sclerae and pre-senile hearing loss[1]. The early evolution of the nomenclature is under Ekman-Lobstein syndrome. In 1896 John Spurway emphasised on the association of skeletal fragility with the presence of blue sclerae[2]. In 1900 Alfred Eddowes of London addressed the cause for scleral blueness and he proposed that this blue color of sclera may be due to a defect of mesenchyme, whereas in 1912 Charles Adair-Dighton of Liverpool drew attention on the dominant mode of transmission of blue sclerae and its association with adult-onset deafness[3]. In 1918 van der Hoeve and his colleague de Kleyn of Utrecht, emphasised on the syndromic relationship of brittle bones, deafness and blue sclerae in osteogenesis imperfecta tarda[4]. The most characteristic feature of the condition is the occurrence of multiple fractures resulting from trivial trauma. In some patients, it was also associated with facial disproportion often requiring surgical interventions for esthetic reasons[5]. Hearing loss may also occur in childhood which results in additional disability in education and psychosocial development which can be prevented by appropriate otological and audiological treatment on time (Kuurila, 2000)[6]. Disease risks to other family members is totally dependent upon the inheritance pattern[7].

Sillence *et al* stated that there are four major types of OI, which range from mild to severe[8]

- Type I (autosomal dominant inheritance),it is the mildest form— also frequently known as "OI tarda"— it is characterized by distinctly blue sclerae, nondeforming fractures and hearing impairment.
- Type II (autosomal-recessive inheritance)it is the most severe form and is almost always very lethal in the fetal period or perinatal period.
- Types III and IV are considerd to be moderate to severe forms which present with bone deformities, whereas hearing impairment is quite less common in both these types[9].

As knowledge for the disease increased, many different names were applied to it. Lobstein's disease (*maladie de Lobstein*) or, as he called it, idiopathic osteopsathyrosis, was first renamed by Vrolik as osteogenesis imperfecta[11]. Since that time it has been given many other names: fragilitas ossium (Klebs and Hochsinger), blue sclera, brittle bones and otosclerosis (Eddowes's syndrome), osteitis parenchymatosa chronica, dystrophia periostalis , periosteal dysplasia and periosteal aplasia, have all been applied to one form or another form of this disease[12].

CONCLUSION

There are many rare dysmorphic syndromes and clinical syndromes including genetic syndromes and sometimes it becomes very difficult for many clinicians to well equip themselves with adequate professional knowledge that might help them to make an early diagnosis for many such rare syndromes they may encounter in their clinical practice.

This is a dominant form of the syndrome. It is a combination of blue sclera, deafness and fragile bones. The course is usually progressive, but many patients tend to live to an advanced age. The hearing loss can progressively increase with age and patient can also have tinnitus in future The most characteristic feature of this condition is the occurrence of multiple fractures resulting from trivial trauma

The disease is a rare dominant form of syndrome hence it should be diagnosed properly by various otological, audiological, radiological and clinical examination. The main management is aimed at improving the quality of life of the patient with a combination of multidisciplinary medical and surgical modalities.

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