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Case Report

Van Buchem disease: a rare case report

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ABSTRACT

Van Buchem disease, or hyperostosis corticalis generalisata, is a autosomal recessive skeletal disease which is characterized by uninhibited bone growth, especially in mandible, skull and ribs. It is a rare disorder that causes a compromised inhibitory feedback mechanism resulting in increased bone formation and overgrowth of skeleton leading to a variety of neurological symptoms. We are reporting a case of 15-year-old female child presented with short stature and dysmorphic facies along with progressive vision loss since 3-4 years.

Keywords: Van buchem disease, Hyperosteosis, Autosomal recessive, Dysmorphic facies

INTRODUCTION

Van Buchem disease is an autosomal recessive skeletal disease first described in 1955 by Prof. Franciscus Stephanus Petrus van Buchem. He found that the bone was produced faster than the body broke it down, making it much thicker as the patient got older. The first symptoms experienced by the affected were often deafness and paralysis of the face, caused by the growing bone pinching the nerve.[1] There is a deletion downstream of the SOST gene on chromosome 17q that decreases gene expression of sclerostin, which is an inhibitor of bone formation and this reduction in its expression leads to uninhibited bone formation.[2]

CASE REPORT

A 15-year-old female child product of non-consanguineous marriage 4th in order presented with complaint of short stature not gaining height along with progressive vision loss since last 3-4 years (Figure 1). She was born by normal vaginal delivery at home with no significant birth history and no history of hospitalization at the time of birth. With increasing age patient was not gaining normal height. All the developmental milestones were normal. After the age of 10 her parents noticed the short stature. Patient also developed progressive loss of vision since last 3-4 years and left her studies due to that. She studied till 5th standard, her IQ was normal according to age. There was no history of any past hospitalization. No history of similar complaints in parents or other siblings. She could comfortably do routine household activities.

Figure 1: Short stature with macrocephaly.

On examination her general condition and built was good with good appetite weighing 39 kg with raised head circumference (59 cm), length (150 cm), H/A (25th percentile), W/A (25th percentile). Her IQ was normal according to age. Patient had macrocephaly with relatively large mandible along with short stature and progressive vision loss (Figure 2). The ophthalmologic examination is found suggestive of bilateral pale optic disc with bilateral optic nerve atrophy, perception to light (PL) present, projection of rays (PR) accurate counting fingers (CF) close to face. Hearing examination was normal.

Figure 2: Macrocephaly with large mandible and maxilla.

Given the suspicion of syndromic disorder skeletal survey done. Multiple radiographs of skull, limbs and chest taken. Skull X-ray showed thickening of skull vault, maxilla and mandible (Figures 3 and 4). X ray of upper and lower limbs showed periosteal thickening (Figure 5). MRI was done which was suggestive of calvarial thickening. Gene analysis was not done due to socioeconomic reasons of patient.

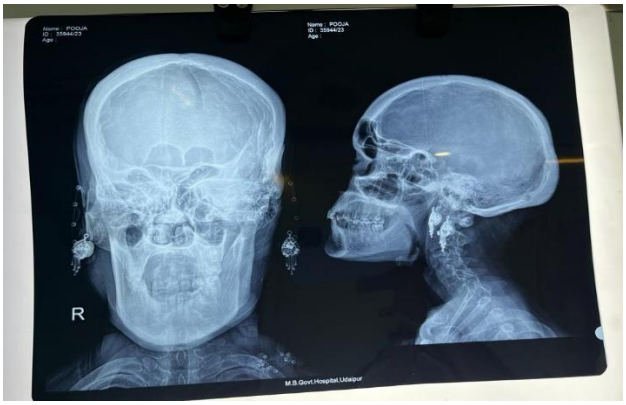

Figure 3: Skull X-ray showing thickened skull vault with large mandible.

All the routine sampling done and all parameters were within normal range. ALP was 78. Sepsis screen was negative. Endocrine profile done including mildly low T3 (1.22) normal T4 (80.98), TSH (1.42) and prolactin (186.9) and low ferritin (7.35).

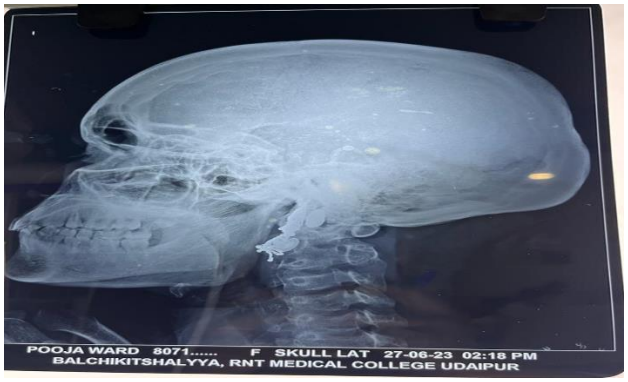

Figure 4: X-ray suggestive of skull and mandible hyperostosis.

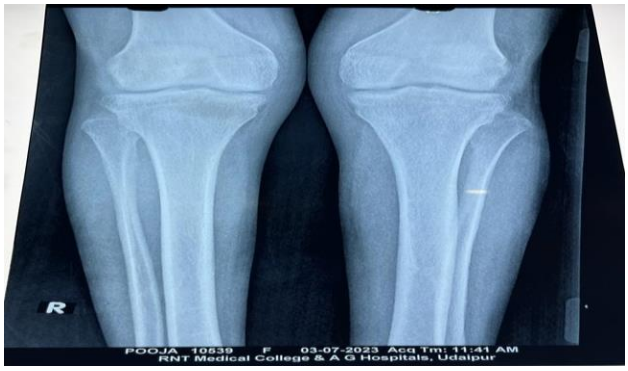

Figure 5: Lower limb X-ray suggestive of periosteal thickening.

DISCUSSION

There are less than 40 cases described. This disease usually presents with symptoms of cranial bone overgrowth resulting in macrocephaly, vision loss, hearing abnormalities, neurological pain, and progressive blindness may be due to optic nerve atrophy. The usual age of presentation is during puberty with progression of symptoms throughout the life. The most common cranial nerves to be involved are seventh and eighth causing facial palsy and hearing abnormalities including both senorineural as well as conductive hearing loss.[3] Late neurological complications are increased intracranial pressure (ICP) caused by the hyperostosis of skull with decreased intracranial space causing dizziness and recurring headaches and also spinal stenosis that occurs much later in life.[4]

Our case demonstrates only some remarkable features of this disease like presenting at the age of 15 years with macrocephaly along with enlarged mandible and short stature, progressive vision loss since 3-4 years along with signs of optic nerve atrophy but no other cranial nerve involvement has been seen so far. Signs of raised alkaline phosphate was consistent as with all other cases. However, clinical and imaging features were sufficiently specific for Van bucham disease in this case. Our case did not show

changes in ribs and single cranial nerve involvement that could be explained by the young age of our patient.

Differential diagnoses include Camurati-Engelmann disease i.e. progressive diaphyseal dysplasia characterized by sclerosis and enlargement of the diaphysis of the long bones with symptoms of pain in limbs, progressive muscle weakness and waddling gait. This is due to mutation of gene TGFB1 encoding for transforming growth factor beta.[5] Another one is osteopetrosis or Albers-Schonberg disease presenting as generalized bony sclerosis with multiple fractures a characteristic appearance of sandwich vertebra known as 'rugger jersey spine' and endobones known as 'bone within a bone' in pelvis. Gene defect is on chromosome no. 1p21.[6]

First case reported in India was from AFMC Pune, a 5-year-old male child with global delay in attaining the developmental milestones and progressive reduction in visual acuity, loss of hearing, facial dysmorphism with macrocephaly, flat prominent nasal bridge, prognathic mandible, and protruding tongue. Radiographs showed generalized thickening of the skull vault, maxilla, and mandible involving both inner and outer tables. The child had reduced vitamin D3 levels with mildly raised alkaline phosphatase levels.[7]

Another one reported from Kolkata was a 20-year-old male child presented with progressive facial deformity, hearing loss, genu valgum, and breathing difficulty. He had raised serum alkaline phosphatase and other blood parameters were normal. The skeletal X-ray showed metaphyseal widening and mild cortical hyperostosis in long bones. The frontal and lateral skull x ray views showed skull base and cranial vault sclerosis with the protruded mandible.[8]

CONCLUSION

Van Buchem disease should be suspected in children presenting with short stature, large head circumference along with large mandible, followed by neurological signs and symptoms with no intellectual impairment. These patients should undergo limb and skull X-ray followed by MRI that shows calvarial thickening. Gene analysis can be done to obtain definitive diagnosis. Steroids can be tried as treatment modality of choice. Decompressive craniotomy can be done in patients having neurological signs and symptoms and in raised intracranial tension.[9]

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Ethical approval: Not required

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