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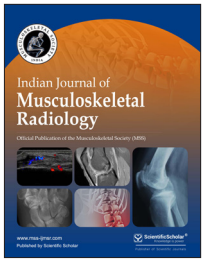

Case Report

Van Buchem disease: A rare sclerosing dysplasia

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ABSTRACT

The other name for Van Buchem disease is hyperostosis corticalis generalisata. It is an uncommon genetic bone condition characterized by aberrant metaphyseal expansion of the tubular bones and hyperostosis and sclerosis of the craniofacial bones. It is brought on by a SOST gene defect that results in increased osteoblastic activity. A 20-year-old male patient came to us with gradual hearing loss, progressive facial deformity, and breathing difficulties. Radiological examination with computed tomography and X-ray reveals sclerosis of the craniofacial bones and enlargement of the metaphyseal region of long tubular bones. Based on radiological characteristics, we suspected it to be a very rare case of sclerosing dysplasia called Van Buchem disease.

Keywords: van Buchem disease, craniofacial, hyperostosis, sclerosis, dysplasia

INTRODUCTION

In 1955, Van Buchem *et al.* were the first to publish the description of this rare autosomal recessive condition known as Van Buchem disease. The SOST gene mutation is an autosomal recessive inheritance pattern of disorder.

Sclerostin expression in osteoblasts is altered, which prevents osteoblastic bone production from being inhibited.

These individuals frequently exhibit progressive facial deformities, various neuron deficiencies due to cranial nerve involvement, and breathing problems in the latter stages. The only substantial abnormality seen during the hematological study is elevated serum alkaline phosphatase in 50% of patients. The patient's life expectancy varies depending on the degree and severity of involvement of vital structures secondary to hyperostosis.

Radiological features show gross thickening of the calvarium, particularly in the frontal and basioccipital regions. The petrous bones at the base of the skull get thickened. Due to the hyperostosis of the bones, the paranasal sinuses are significantly less pneumatized. Long tubular bones exhibit cortical hyperostosis and metaphyseal expansion.

We present a case of a 20-year-old male with typical clinical and radiological features of Van Buchem disease.

CASE REPORT

A 20-year-old male child presented to us with a progressive facial deformity, hearing loss, genu valgum, and breathing difficulty. There was no history of consanguinity or similar abnormality in

the family. On examination, the patient had protruded chin, flat nasal bridge, irregular alignment of teeth, frontal bossing, and genu valgum [Figure 1].

His blood examination revealed raised serum alkaline phosphatase and his other blood parameters were normal. Pure-tone audiometry revealed an increased gap between air and bone conduction.

A skeletal survey using X-ray showed metaphyseal widening and mild cortical hyperostosis in long bones [Figure 2], with the distal femur most affected resembling an Erlenmeyer flask. The frontal and lateral skull views showed skull base and cranial vault sclerosis with the protruded mandible. Other osseous abnormalities were the widening of the phalangeal shafts and clavicles [Figure 3]. The spine and pelvis were normal.

Subsequently, computed tomography of the skull further added sclerosis around the bilateral internal auditory canal with its narrowing [Figure 4]. Bilateral mastoid air cells showed variable amounts of sclerosis. Paranasal sinuses were relatively smaller, with hyperostosis of their bony walls [Figures 5 and 6].

Figure 1: Clinical picture of patient showing protruding chin, high forehead, and disfigured face (characteristic features of Van Buchem disease).

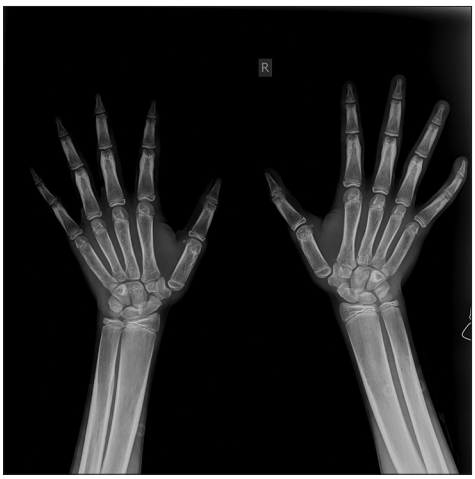

Figure 2: X-ray of both hands including both distal forearms in frontal projection reveals widening and sclerosis of tubular bones.

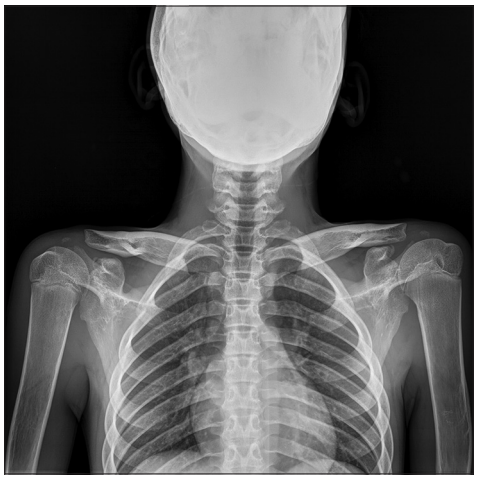

Figure 3: X-ray of chest including bilateral shoulder joints in frontal projection shows metadiaphysial widening of tubular bones with cortical hyperostosis (clavicles and humerus).

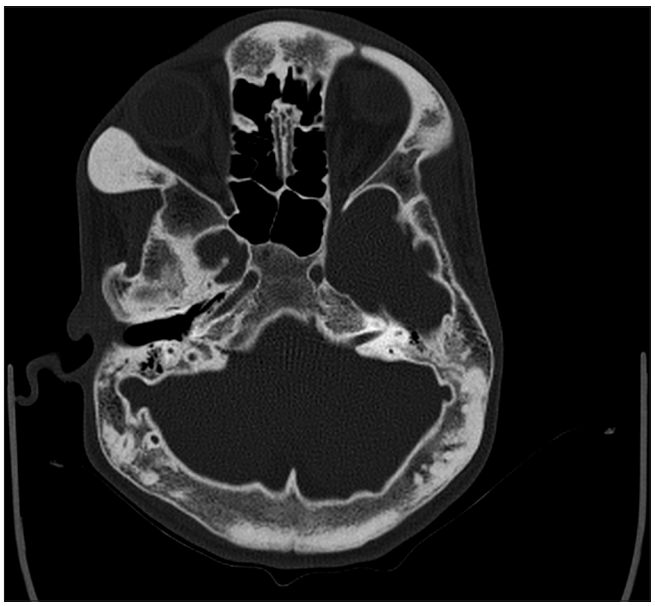

Figure 4: Axial computed tomography image of the skull base in bone window shows hyperostosis of calvarial bone with narrowing of skull base foramina including bilateral internal auditory canal and sclerosis of bilateral mastoid air cells.

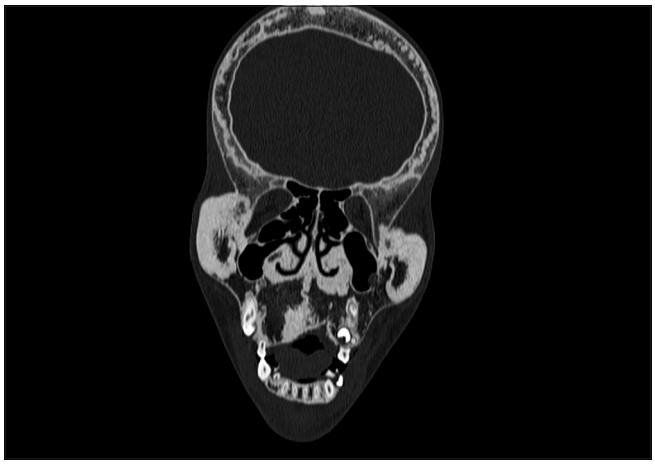

Figure 5: Coronal reconstructed computed tomography image of skull in bone window shows hyperostosis of facial bones including enlarged nasal turbinates obstructing nasal cavity.

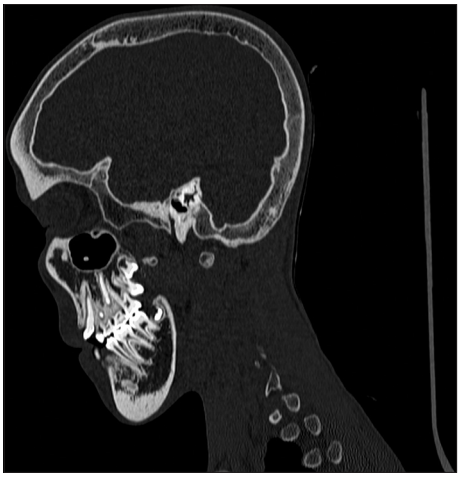

Figure 6: Sagittal reconstructed computed tomography image of skull in bone window shows generalized thickening of the skull, particularly affecting the vault, petrous bones, and mandible.

Radionuclide skeletal imaging (3 phase) with Tc-99m MDP showed increased osteoblastic activity over both nasal bones and maxillae, including the lower margins of both orbits. Increased tracer activity over the major epiphyseal plates in the body indicates young growing age [Figure 7].

Clinico-radiological findings were consistent with hyperostosis corticalis generalisata also called Van Buchem disease.

DISCUSSION

According to published research, Van Buchem disease is a hereditary sclerosing dysplasia of the bones with both dominant and autosomal recessive modes of transmission. The dominant type of this condition is a somewhat milder form, and its symptoms are often limited to enlarged and

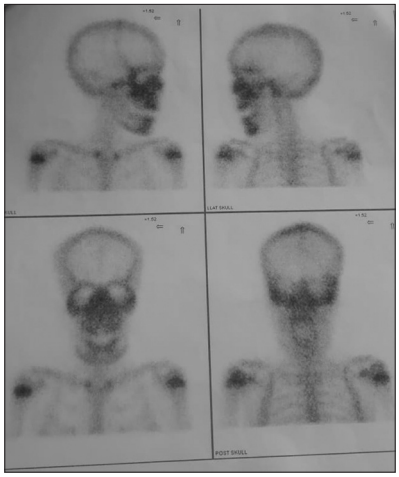

Figure 7: Radionuclide skeletal imaging with Tc-99m MDP shows increased osteoblastic activity over both nasal bones and maxillae, including the lower margins of both orbits. Increased tracer activity over the major epiphyseal plates in the body indicates young growing age.

protruded jaw bone. The hyperostotic bone at the base of the skull causes progressive compression of the cranial nerves in its much more severe autosomal recessive form.

Osteopetrosis (Albers-Schonberg disease), Camurati-Englemann disease, osteopathia striata with cranial sclerosis, and sclerosteosis are the differential for this condition.

Osteopetrosis, also known as Albers-Schonberg disease, causes marble-like bones that are prone to non-traumatic fractures.[1] Typically, there is no involvement of the cranial nerves. Segmental osteosclerosis causes the typical appearance of bone inside the bone in Albers-Schonberg disease patients.

An uncommon bone condition called Camurati-Englemann disease usually manifests in young adulthood. Muscle soreness and gait abnormalities are symptoms of this condition. It could affect any bone in the body, but the diaphyseal area of long bones is where it usually manifests. The mandible seldom becomes involved.[2]

Skeletal deformities characterized by linear striations in the metaphyses of long bones and the pelvis, in addition to cranial sclerosis, are frequently present from birth and are hallmarks of osteopathia striata with cranial sclerosis.

Sclerosteosis is a progressive bone disorder with an autosomal recessive inheritance that shows up within the 1st year of life.[3] They also exhibit frontal bossing, calvarial sclerosis, and early-onset cranial nerve palsy. These individuals also have a protruding prognathic chin. VBD and sclerosteosis have similar characteristics, although sclerosteosis has a more severe clinical presentation and is typically accompanied by elevated ICP.[4] The brain stem's impaction in the constrained foramen

magnum might result in sudden death. Another significant distinction may be the high frequency of finger anomalies (radial deviation, syndactyly, or both) and the increased height.

CONCLUSION

Less than 30 cases of this extremely rare homozygous recessive bone condition known as VBD have been documented.[5] Most of these were found among the Dutch and, to a lesser extent, among South Africans of Dutch lineage. This condition of craniotubular hyperostosis[6,7] is brought on by a 52-kb deletion downstream of the SOST gene.[8] This deleted DNA segment has been shown to influence osteoblast SOST expression, which, in turn, causes inhibition of suppression of osteoblastic bone production.[9] The cranial and tubular bones are primarily affected by progressive hyperostosis. Therefore, the most apparent clinical symptoms are face deformities and numerous cranial nerve impairments caused by cranial nerve entrapment.

In developing countries, where genetic testing facilities are not so widely available, radiological features can guide the diagnosis of craniotubular dysplasia.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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