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Osteopetrosis: A Case Report

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ABSTRACT

Osteopetrosis also known as marble bone disease or Albers-Schonberg disease, is an extremely rare inherited disorder whereby the bones harden, becoming denser, in contrast to more prevalent conditions like osteoporosis, in which the bones become less dense and more brittle, or osteomalacia, in which the bones soften. Osteopetrosis is one of the hereditary causes of osteosclerosis it is considered to be the prototype of osteosclerosing dysplasia [1]. Here is a 75-year- old gentleman who was presented with paraesthesia on the right side, right sided weakness, slurring of speech, right UMN (Upper Motor Neuron) facial palsy and right upper and lower limb ataxia. Hence MRI (Magnetic Resonance Imaging) brain with angiography was done which revealed Left thalamic acute infract and Right vertebral artery narrowing with visualisation of distal part of V4 segment. He was closely monitored and was treated with statins, anti-platelets and neuroprotective agents. During the course his complaints improved and was discharged with same medications. He revisited again after 7 years with complains of Type 2 Diabetes Mellitus and numbness on both feet and back pain. His routine blood investigations showed high calcium levels, low red blood counts levels and low levels of Vitamin D. His Bone Mineral Density test (BMD) was done and values were more than +2, and X-ray results revealed anterior spinal ligament calcification.

Keywords: MRI (Magnetic Resonance Imaging) [1], BMD (Bone Mineral Density) [2], UMN (Upper Motor Neuron) [3]

INTRODUCTION

[2]Osteopetrosis is marked by increased bone density due to a defect in bone reabsorption by cells called osteoclasts. This leads to accumulation of bone with defective architecture, making them brittle and susceptible to fracture and to skeletal abnormalities. Although symptoms may not initially be apparent in people with mild forms of this disorder, trivial injuries may cause bone fractures due to bone fragility. The underlying condition can be categorized based on whether sclerosis or defective skeletal modelling predominates. There are different types of osteopetrosis which include craniotubular dysplasia, craniotubular hyperostosis, and osteosclerosis. They are familial but have different inheritance patterns. Some types are comparatively benign others are progressive and can be fatal. This condition mainly occurs due to its genetic predominance, based on genetics they have been categorised into 4 types which includes autosomal dominant, autosomal recessive, intermediate autosomal osteopetrosis, and adult delayed-onset osteopetrosis. Autosomal dominant osteopetrosis, Albers-Schonberg disease, is the most common type of osteopetrosis, with an onset during adolescence or adulthood. Bone is in a dynamic state and is dependent upon a healthy balance between osteoclast-mediated resorption and osteoblast-mediated

deposition. In osteopetrosis, defective osteoclast development or function leads to a disruption in normal bone homeostasis. Osteoclasts that have defective proton pumps, chloride channels, or carbonic anhydrase II proteins are unable to resorb bone effectively. Consequently, the unorganized, overly dense bone that is prone to fracture develops unchecked. While osteopetrosis is a disease of primary bone sclerosis, many conditions can lead to similar osteosclerosis in a secondary way, thus the differential diagnosis observed on radiographic evaluation may include Beryllium, lead, and bismuth poisoning, Fluorosis, Myelofibrosis, Paget disease, Cancer (lymphoma or osteoblastic bony metastases). The diagnostic measures considered for the disease include X- ray scans, BMD (Bone Mineral Density) tests that are useful in confirming the diagnosis. Patients and their families should be educated about the natural history and common complications of the disease. Management of patients with osteopetrosis must be tailored to the individual patient. Treatment is predominantly supportive with no known cure, and interprofessional care and surveillance are treatment mainstays.

CASE REPORT-

A 75- year- old male was brought to the emergency department in 2018 with complains of paraesthesia over the right side for 2 days, slurring of speech and right sided weakness for 1 day. His vitals were normal. But on physical examination showed right upper motor neuron facial palsy, slurring dysarthria and right upper limb and lower limb ataxia (grade 4+/5) to be present. On doing his MRI- Brain with angiography the scan showed left thalamic acute infract and right vertebral artery narrowing with visualisation of distal part of V4 segment. His Echo was done which showed normal reports and LVEF of 68%. Cardiology opinion was obtained in view of underlying CAD and irregular pulse and their medication advice were followed. The patient was under close neuro monitoring and was thus started on statins, antiplatelet and other neuroprotective agents. On examination he was found to have high sugars thus endocrine opinion was considered and was initiated on insulin and was also known to have diabetic retinopathy on screening, thus the endocrine advices were followed. Over the course of hospital stay, the complains improved and he was discharged with same medications, and was advised to review in OPD with reports. After 7 years this gentleman came again for review in the Endocrinology department with complains of high sugars and numbness on both feet. His blood investigations were done which showed low corrected calcium levels (10.21), low RBC levels (3.99), high ESR levels (23) and low Vitamin D levels (29). He was also advised to do the bone mineral density (BMD) test and an X-ray of lateral spine. His BMD thus showed

Table 1 Bone Mineral Density

REGION	BMD	T-SCORE
L1	1.485	+3.0
L2	1.555	+3.0
L3	1.712	+4.3
L4	1.500	+2.5
L1-L4	1.572	+3.3
NECK LEFT	0.959	-0.8
NECK RIGHT	1.001	-0.1

As his BMD is +2, he was diagnosed with osteopetrosis/marble bone disease. Also, his X- ray was done of thoracolumbar spine (AP AND LAT) which showed multiple osteophytosis in the vertebrae thus pro-

ving to have anterior spinal ligament calcification.

DISCUSSION-

Osteopetrosis is most frequently diagnosed based on the patient having the typical clinical and radiographic findings of the disease. The first choice of treatment in osteopetrosis is advising a bone mineral density test and doing an X- ray scan. Radiographs will show diffuse osteosclerosis throughout the skeleton with a "marble bone" appearance. There will generally be increased cortical thickness with associated decreased medullary canal diameter. The "Erlenmeyer flask" deformity can be found at the metaphysis of long bones, particularly at the proximal humerus and the distal femur. A "bone-in-bone" or "endobone" appearance most frequently is noted in the bones of the spine or the hand phalanges.

[3]A case report done by Mauro Grinfelder on osteopetrosis concluded that – This disease is a highly impacts the quality of life and if not treated may lead to multiple complications thus, increasing the risk of fractures. [4]Another study conducted by Simon Lampart et.al in 2018 on Special Form of Case Report in 53- year old man was concluded as this disorder is a type that affects both the connective tissue and the bone structure, it is rare and its estimated incidence is 1 in 20,000 adults, mainly caused by autosomal dominant inherited mutations in genes encoding alpha-1 and alpha-2 chains of type1 and type 2 collagen, its rarer forms include mutations in genes encoding proteins that promote post-translational modifications of type 1 collagen and osteoblast maturation.

Male osteoporosis often remains unrecognized. Osteoporotic fractures occur approximately 10 years later in men than in women due to higher peak bone mass. However, 30% of all hip fractures occur in men. Risk factors of osteoporotic fractures can be grouped into primary and secondary causes based on their etiology.

CONCLUSION

Osteoporosis causes bones to become weak and brittle, so brittle that a fall or even mild stress such as bending over or coughing can cause a break. Osteoporosis-related breaks most commonly occur in the hip, wrist or spine. Certain drugs, physical and occupational therapies and bone care helps reduce the progression of the underlying condition.

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