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Osteopetrosis and its typical Radiographic findings : A rare case report

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ABSTRACT

Background:

Marble bone disease is an inherited rare skeletal condition characterized by increased bone density on radiographs and involves not only skeletal system but also hematological,neurological systems as well as dental problems which is inherited either as autosomal dominant or autosomal recessive. Incidence 1 in 200,00LB and 1 in 2,00,000 LB respectively. These conditions vary greatly in presentation and severity ranging from neonatal onset with life threatening complications such as bone marrow failure. We describe a case of 8-year-old female with the typical radiographic and clinical features which doesnot require genetic testing for the diagnosis.

Case Presentation :

A 8-year-old female child was brought by her parents to our facility with concerns of failure to thrive and not gaining milestones as per age. She was first born child of a second degree consanguinous marriage born by normal vaginal delivery . The patient had previous complaints of right femur fracture, facial palsy, hearing loss, progressive vision loss, open mouth breathing with halitosis and reccurent respiratory tract infections. After the typical clinical features radiological investigations were organized and the diagnosis of malignant osteopetrosis was made.

Conclusion:

The case we are presenting to you has all the classical symptoms of osteopetrosis, which is a very uncommon skeletal illness that manifests as various systemic issues and has typical imaging features,which strikes the diagnosis of osteopetrosis.

INTRODUCTION:

Osteopetrosis is also known as Marble bone disease or Albers-Schoenberg disease is a disorder of bone resorption which is a rare congenital disorder which can be inherited as autosomal dominant or autosomal recessive. It is caused by failure of osteoclasts to resorb immature bone[1-3] Osteopetrosis leads to abnormal bone marrow cavity formation and leads to signs and symptoms of bone marrow failure. Impaired bone remodelling causes bone marrow narrowing and cranial nerve compression leading to facial palsy and vision loss [2]. Abnormal remodelling of primary woven bone to lamellar bone results in brittle bone that is prone to fractures. Affected children usually present in first year of life with concerns of parents regarding vision, failure to thrive ,recurrent infections with underlying anemia and bone marrow involvement. Easy brusing, nasal congestion, abnormal craniofacial appearance may be less common symptoms[4-6].Hepatosplenomegaly is present in almost all the cases. Frequently the diagnosis is almost made on the basis of radiological features and the correlating clinical features.

Case Presentation:

A 8-year-old female child first by birth order born via normal vaginal delivery throuh second degree consanguinous marriage having symptoms since infancy. Initially the baby was having developmental delay and at 11 months of age , complaints of fall from cradle resulting in right femur fracture, for which treatment was taken. At 3 years of age patient had complaints of facial deviation and progressive dimnision of vision but was not treated. Later the vision loss was progressed and lost vision in left eye at 6 years of age. She had complaints of recurrent respiratory tract infections and was being treated with oral antibiotics, but was never admitted. At 8 years of age she presented with complaints of short stature. On examining the patient she had macrocephaly, divergent squint, proptosis of right eye, pus discharge from the medial side of the right eye (Fig.2), abnormal facies, dental caries, deciduous tooth, poor oral hygiene, oro antral fistula(Fig.3), high arched palate, a polyp in the nose, open mouth breathing and halitosis was present. Short stature was observed, pectus excavatum (Fig.1) , harrisons sulcus, hepatosplenomegaly, knock knees and thrombotic nodules(Fig.4) were observed.

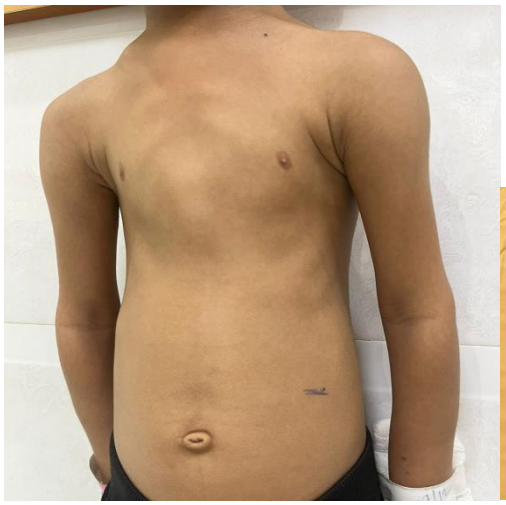

Fig.1 Pectus excavatum

Fig.2 Proptosis of right eye

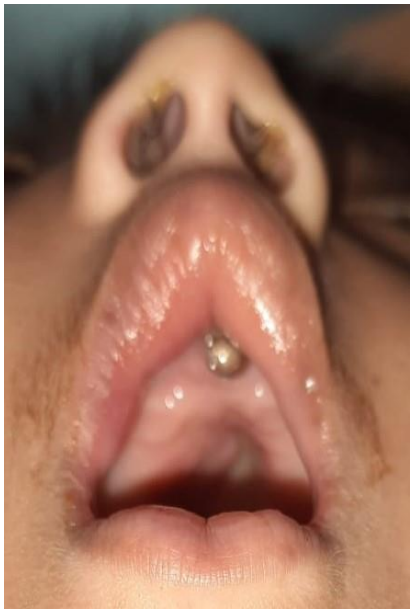

Fig .3 High arched palate with

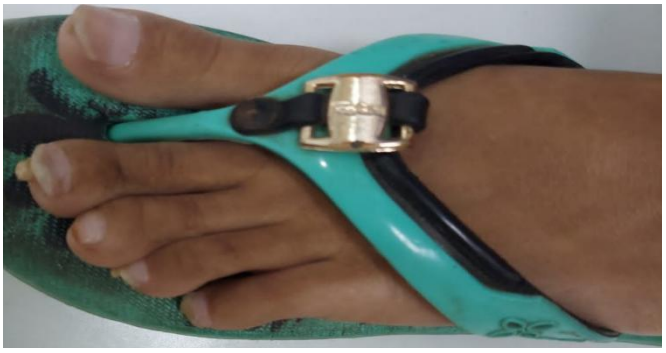

Fig.4 Thrombotic Nodules

Oro antral fistula

Then the blood investigations and the radiological investigations were organized.
The CBC showed anemia, thrombocytopenia and lymphocytosis suggestive of underlying bone marrow involvement leading to dimnished marrow space and chronic infections with anemia.
Urea and creatinine were increased and rest all the lab investigations were normal.
CT Brain and PNS were done which was suggestive of osteomyelitic features and chronic sinusitis with ventriculomegaly and increased bone density.
Xray Upperlimb was done suggestive of bone in bone appearance and spurting at the end of radius and ulna, and bone modelling defects at the metaphyses of long bones ,such as funnel-like appearance also known as Erlenmeyer flask deformity(Fig.6). Xray spine sugegstive of fish shaped vertebrae (Fig.5)and xray skull shows increased density of long bones[7].

Fig.5 Fish Mouth vertebrae

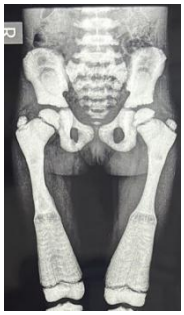

Fig.6 Erlenmeyer flask deformity

Conclusion:
Osteopetrosis encompasses a group of highly hetrogenous conditions, ranging in severity from asymptomatic to fatal in infancy. The more severe form tend to have autosomal recessive inheritance, which is caused by failure of osteoclast differentiation or function and mutations in atleast 10 genes have been identified as causative in humans. The most important clues for diagnosis the radiological evidence and associated clinical features.

Discussion:
The prescence of fractures, short stature, compressive neuropathies, hypocalcemia, life threatening pancytopenia. Prescence of neurodegeneration, mental retardation, skin and immune system involvement or renal tubular acidosis may point to the rarer osteopetrosis variant, while primary skeletal manifestations such as fractures and osteomyelitis in late childhood or adoloscence is typical of Autosomal Dominant Osteopetrosis. Diagnosis is largely based on clinical and radiographic evaluation and is confirmed by genetic testing where applicable and paves way to understand natural history, specific treatment where available. Treatment of the condition constitutes an integrated approach of all the departments including Paediatrician, Orthodontist, Orthopaedician, Neurologist, Ophthalmologist and Endocrinologist to improve the quality of life.

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