OSTEOPETROSIS – A CASE REPORT

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ABSTRACT

Osteopetrosis is a group of rare hereditary skeletal disorder characterized by marked increase in bone density. This results from a defect in bone remodeling caused by failure of normal osteoclast function. It is an extremely rare disorder affecting the bones. Osteopetrosis results because there is an imbalance between the formation of bone and the breakdown of the bones. Several types of osteopetrosis are seen which vary in severity. Symptoms can include fractures, frequent infections, blindness, deafness and strokes. The disease represents with variations in clinical presentation because of the heterogeneity of genetic defects resulting in osteoclast dysfunction. Medical treatment is based on efforts to stimulate host osteoclasts or provide an alternate source of osteoclasts. The aim of this paper is to present a 4 yr old female child’s case report with emphasis on its clinical and radiological features.


INTRODUCTION

Osteopetrosis commonly called as stone bone, also known as Marble bone disease and Albers-Schonberg disease is an extremely rare inherited disorder. In this condition the bones harden, becoming denser resulting from a defect in bone remodeling caused by failure of normal osteoclast function. Osteopetrosis can cause osteosclerosis.

Normal bone growth is achieved with a balance between bone formation by osteoblasts and bone resorption by osteoclasts. In osteopetrosis, the number of osteoclasts may be reduced, normal or increased. Most importantly osteoclast dysfunction is the main cause for the pathogenesis of the disease. The exact mechanism is not known, however deficiency of carbonic anhydrase in osteoclasts is seen. The absence of this enzyme causes defective hydrogen ion pumping by osteoclasts and this in turn causes defective bone resorption by osteoclasts, as an acidic environment is needed for dissociation of calcium hydroxyapatite from bone matrix. Hence bone resorption fails while its formation persists. Hence excessive bone is formed. Three clinically distinct forms of osteopetrosis are recognized – The infantile malignant autosomal recessive form, the intermediate autosomal recessive form and the adult benign autosomal dominant form. Osteopetrosis is generally diagnosed through skeletal radiographs. Radiographs of osteopetrosis patients will have an unusual density with a chalky white appearance. Bone density tests and bone biopsies can confirm the diagnosis while other tests such as CT scans or MRI can be performed to evaluate any potential complications.

Case Report

A 4 year old female child reported to the Department of Oral medicine , CKS Theja Dental college with a complaint of missing upper and lower teeth and difficulty in taking food. Patient complains that only a single tooth in the upper and lower jaw erupted during 2 years of age after which no other teeth have erupted. Patient is unable to chew food and has difficulty in eating. There was no relevant medical history. History of consanguous marriage of parents was present.

Her general examination revealed stunted growth, thickened wrists, frontal bossing and strabismus (squint). She has a history of consanguineous marriage of parents. Her CNS examination revealed that patient was blind. Patient lost her vision when she was one and a half year old. She speaks coherently. Hearing was impaired. All other systemic examinations were normal. Extra oral examination revealed hypertelorism of the eyes, divergent strabismus, frontal bossing of skull, increased anteroposterior dimension of the skull and flattening of nasal bridge. Intraoral examination revealed normal mouth opening and jaw movements. Only 51 and 71 were present and all other teeth had failed to erupt. No other abnormality was detected in the oral cavity.

In view of the unerupted deciduous teeth, past history of consanguinous marriage of parents, loss of vision when patient was one and a half year old, impaired hearing and features of thickened wrists, frontal bossing.
hypertelorism, increased dimension of skull, it was provisionally diagnosed as a disorder of bone (Fig.1, Fig.2 and Fig.3).

Biochemical examination revealed Serum calcium – 6.9 mg/dl, Serum Phosphorous - 5.8 mg/dl and Alkaline Phosphatase – 277 IU. Radiological findings revealed multiple unerupted teeth, Lateral and PA skull revealed frontal bossing, Radiograph of hands revealed thickening of bone above the wrists , increased sclerosis and areas of radiolucency and bands of radiopacity (Fig.6, Fig.7 and Fig.8). CT scan of skull in axial section shows thickening and increased density of skull bones (Fig.9). Correlating history, clinical findings, biochemical findings and radiographic findings final diagnosis of OSTEOPETROSIS was made.

Discussion

Osteopetrosis is an extremely rare inherited disorder of bones. In this condition the bones become hard and denser in contrast to more common conditions like osteoporosis, in which the bones become less dense and more brittle or osteomalacia in which bones soften. The exact mechanism of pathogenesis in osteopetrosis is unknown. However deficiency of carbonic anhydrase in osteoclasts is seen. Bone resorption fails while its formation continues. Though there is excessive bone formation, people with osteopetrosis tend to have bones that are more brittle than normal. Mild osteopetrosis may cause no symptoms and present no problems. However serious forms can result in stunted growth, deformity, increased chances of fractures, also patients may suffer from anemia, recurrent infections and hepatosplenomegaly due to bone expansion leading to bone marrow narrowing and extramedullary hematopoiesis. It can also result in blindness, facial paralysis and deafness due to increased pressure put on nerves by the extra bone. Infantile osteopetrosis or malignant osteopetrosis is a severe form of disease inherited as an autosomal recessive trait and leads to diffusely sclerotic skeleton. Marrow failure, frequent fractures and cranial nerve compression are common. Facial deformity manifests as broad face, hypertelorism, snub nose and frontal bossing. Tooth eruption is almost delayed. Radiographically there is increase in skeletal density with defects in metaphyseal remodeling and the radiographic distinction between cortical and cancellous bone is lost. In dental radiographs roots of the teeth are often difficult to visualize because of the density of the surrounding bone. Adult osteopetrosis is less severe in form and is inherited as an autosomal dominant trait and has been termed as benign osteopetrosis. The axial skeleton reveals sclerosis whereas long bones show little or no defect. Approximately 40% are asymptomatic and narrow failure is rare. Radiographically diffuse increased radiopacity of the medullary portions of the bone is seen. Histopathologically several patterns of abnormal bone formation have been described such as tortuous lamellar trabaculae replacing the cancellous portion of the bone or globular amorphous bone deposition in the marrow spaces or osteophytic bone formation. Numerous osteoclasts may be seen, but there is no evidence that they function, because Howships lacunae are not seen. Differential diagnosis of osteopetrosis include other disorders which can cause diffuse osteosclerosis such as hypervitaminosis D, and hypoparathyroidism, pagets disease, diffuse bone metastasis of breast or prostate.
cancer, intoxication with fluoride, lead or beryllium and hematological disorders such as myelofibrosis, sickle cell disease and leukemia. Because this disease is less severe, adult osteopetrosis is usually associated with long term survival. In contrast, the prognosis of infantile osteopetrosis without therapy is very poor, with most affected patients dying during the first decade of life. Treatment is bone marrow transplant. If complications occur patients can be treated with vitamin D, gamma interferon often in combination with calcitrol has been shown to reduce bone mass, decrease the prevalence of infections, and lower the frequency of nerve compression. Other therapeutic agents proved to be useful include administration of corticosteroids (to increase circulating red blood cells and platelets), parathormone and macrophage colony stimulating factor. Erythropoetin has been used to treat anemia. Limited calcium intake also has been advised. Fractures and osteomyelitis can be treated in the routine pattern. The infections can be routinely treated with prolonged antibiotic therapy with Fluoroquinolones and Lincomycin. Hyperbaric oxygen is useful in promoting healing of recalcitrant cases. In conclusion though osteopetrosis is a rare disorder a careful clinical diagnosis, proper investigations and appropriate treatment is very much essential.

References


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