CASE REPORT

OSTEOPETROSIS WITH OSTEOMYELITIS OF MANDIBLE: A CASE REPORT

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ABSTRACT
Osteopetrosis is a rare genetic disorder caused by congenital defect in the development or function of the osteoclasts, resulting in fragile bones which are sensitive to fracture and infection. Mandibular osteomyelitis is an important and well documented complication of osteopetrosis. This case report discusses a case of osteopetrosis with chronic osteomyelitis of the mandible and increased fetal haemoglobin in a 6-year old girl.
Key Words: Osteopetrosis; Osteomyelitis; Mandible; Fetal Haemoglobin

Introduction

Osteopetrosis is a rare hereditary and familial bone abnormality characterized by dense and fragile bones which are sensitive to fracture and infection. The primary pathology is the lack of resorption of normal primitive osteochondrous tissue by the osteoclast. The persistence of this primitive tissue inhibits the formation of normal mature adult bone. This functional deficiency forms dense and fragile bones, which are sensitive to fracture and infection due to the failure of bone remodeling.[1] It was first reported by Albers-Schönberg in 1904 as delayed physical development accompanied by bone fragility. Most studies have reported that osteopetrosis is associated with hepatosplenomegaly, anaemia, increased susceptibility to infection, respiratory tract infection, cardiac disorders, multiple fractures etc.[2] The result is so severe that most patients die because of anaemia and infection before 20 years of age. Here we present a case of osteopetrosis associated with chronic osteomyelitis of the mandible and increased fetal haemoglobin.

Case Report

A 6-year old girl was referred to the department of Radio-diagnosis with chief complaints of swelling with draining sinus on left side of the jaw with dental caries on the same side and patient was advised for CT scan of mandible. On general examination, patient was short statured with large head and frontal bossing. Defective dentition and hepatosplenomegaly was present. CT scan showed an irregular lytic area with surrounding sclerosis and periosteal reaction on left side of mandibular body with associated soft tissue edema and swelling. During the scan we also observed thickening and increased density of the calvarium, base of the skull and rest of the mandible with obliteration of medullary cavity. Suspecting the case to be an osteoporosis we advised skeletal survey. Skeletal survey revealed macrocephaly, frontal bossing, increased density of base of skull, thick calvarium. Generalized sclerosis of the skeleton was present without trabeculation and little or no differentiation between cortical and medullary regions. Multiple dense curved lines paralleling the iliac crest were present in both iliac bones. The vertebrae showed dense bands adjacent to the endplates (sandwich vertebra) with accentuation of anterior vascular notches. Pathological fractures were present in bilateral femur in neck region and midshaft of right humerus. Chest x-ray (PA view) showed sclerotic ribs and cardiomegaly. Routine blood examination revealed reduced haemoglobin. Haemoglobin electrophoresis revealed increased levels of Fetal Haemoglobin. On pus culture from the region of draining sinus growth of staphylococcus was detected. Patient also underwent CT scan of brain which revealed hydrocephalus and prominence of cortical sulci and ventricles, but no calcifications.

Discussion

Osteopetrosis is a rare hereditary and familial bone abnormality that results from a deficiency in the function and differentiation of osteoclasts. The osteoclasts in the affected bone are usually devoid of ruffled borders by which they adhere to the bone surface and express their resorptive activity.[3] Thus primitive calcified cartilage persists in abundance and medullary space is never allowed to form. Estimated prevalence of the disease is 1 in 100000 to 500000.[4]

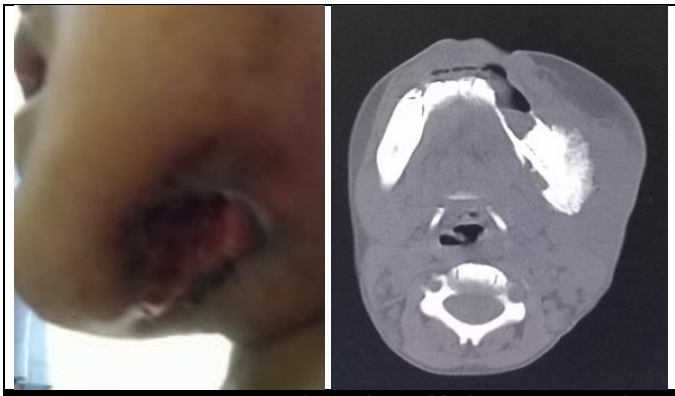

Figure-1: Draining sinus on left submandibular region. Axial CT scan of mandible shows irregular lytic area in left side of mandibular body with surrounding sclerosis, periosteal reaction and adjacent soft tissue swelling.

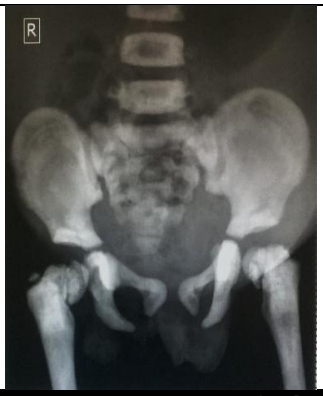

Figure-2: X-ray AP view of pelvis. Multiple dense curved lines paralleling the iliac crest and bilateral femoral neck fractures.

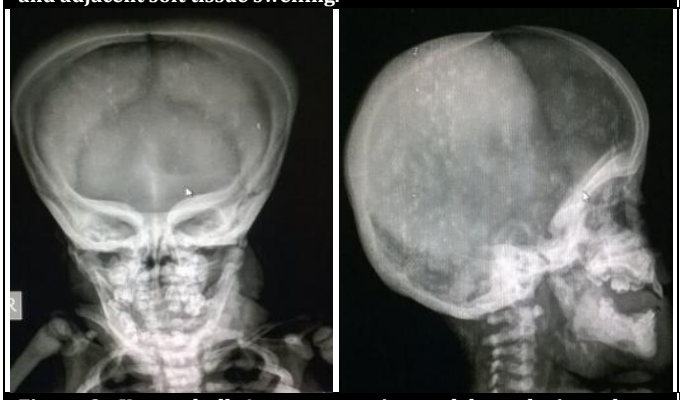

Figure-3: X-ray skull Antero-posterior and lateral view shows Macrocephaly, sclerotic calvarial bones with obliteration of diploic space, sclerosis of base of skull and mandible.

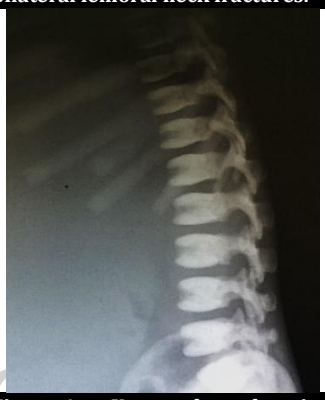

Figure-4: X-ray lateral view lumbar spine shows Sandwich vertebra with accentuation of anterior vascular notches.

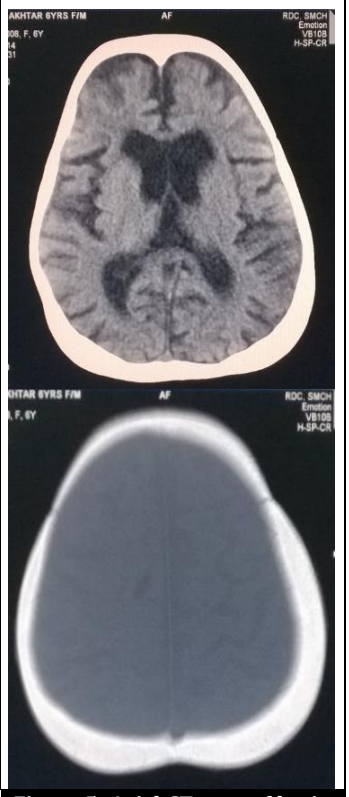

Figure-5: Axial CT scan of brain shows prominent cortical sulci, sylvian fissures and mildly dilated lateral ventricles. Bone window shows sclerotic skull.

The diagnosis of osteopetrosis remains radiographic and is supported by CT scan. Generalized increase in bone density is the cardinal feature. Sclerosis and thickening of skull base is present which is more prominent in the floor of the anterior cranial fossa. Skull changes may produce macrocephaly and hydrocephalus. Underpneumatization of paranasal sinuses and mastoids may occur. Anaemia and extramedullary hematopoiesis leads to hepatosplenomegaly. The anemia is typically macrocytic and the fetal haemoglobin is increased.[5] The vertebrae show dense bands adjacent to the endplates with normal appearing midbody, giving rise to "sandwich vertebra" or "rugger jersey" appearance. Thickening of long bones with failure of metaphyseal remodeling results in expansion of metaphyseal regions[3] and gross undertubulation leads to the Erlenmeyer flask deformity. Ilium show multiple, dense curved lines paralleling the iliac crest.[6] The "bone within a bone" or "endobone" appearance is commonly seen in the vertebral bodies and in the small bones of the hand. Endobones represent fetal vestige and contains embryonic strata which is normally removed.

Most common complication of osteopetrosis is pathological fracture which heals with normal callus, but the onset of callus formation after injury is variable.[2] Bowing of long bones and coxavara may be present due to multiple fractures. Neural foramina are encroached upon and blindness results in serious cases.

Oral features of osteopetrosis are important, since this disease is often diagnosed as a result of oral changes. In patients with osteopetrosis, decreased bone vascularity and damaged white cell function, may cause the development of osteomyelitis after dental extractions. Constriction of canals housing the neurovascular bundles, supplying the teeth and jaws is the most likely contributing factor. 10% of osteopetrosis cases develop osteomyelitis that usually involves the mandible.[7]

Management of the patients with osteopetrosis requires a comprehensive approach to characteristic clinical problems. Medical management is based upon stimulation of host osteoclast with calcium restriction, calcitriol, steroids, parathyroid hormone, and interferon.

Bone marrow transplantation is the only permanent cure for osteopetrosis. As patients are more prone to infection, so dental procedures, especially the extraction of mandibular teeth, must be assessed and carried out with great caution.

Conclusion

Taking clinical and radiological findings into consideration, our case was diagnosed as osteopetrosis with chronic osteomyelitis of mandible. The patient was referred to Department of dental medicine for further management.

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