MALIGNANT INFANTILE OSTEOPETROSIS: ROENTGENOGRAPHIC DIAGNOSIS

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ABSTRACT

Osteopetrosis is the condition of increased bone density or sclerotic bone. Albers-Schoenberg disease was described as the infantile malignant osteopetrosis based on the disorders of clinical course, radiographic and genetic transmission.[2]

Osteopetrosis was classified as the generalized skeletal dysplasia[3], autosomal recessive of probably deficiency of carbonic anhydrase II with functional defect of the osteoclast resulted in nondestruction of calcified matrix during growth. This causes obliteration of marrow space in the medullary cavities and replaced by excessive calcified matrix. The thickened solid spongiosa causes little blood formation space or obliteration of marrow space within the skeletal portion.

The dysplasia presented in infantile period with anemia, hepatosplenomegaly, recurrent infection, and failure to thrive. Blindness may develop early due to narrowing of the optic canals and other cranial foramina by excessive osteopetrosis. Hydrocephalus may develop due to narrowing of the foramen magnum.

The diagnosis by radiographic examination of the skeleton reveals marked sclerosis of the skeletal structure, as dense as marble bones or osteopetrosis. The bones of osteopetrosis are made up largely of calcified cartilage and are brittle rather than strong, the fracture is not uncommonly frequent. The hematological presentation is protracted hypoplastic anemia, thrombocytopenia and reinfection.

The early dysplasia in infancy is the enlarged metaphysis resembling rickets. The failure of underconstriction of the metaphysodiaphysis or funneralization is found in long bones with the presence of alternating transverse lucency in the sclerosis. The course is early fatal due to depletion of hematopoiesis in bone marrow. The treatment was initiated by bone marrow transplantation with some favorable result.[9-11]

INTRODUCTION

Osteopetrosis is a complex disease of at least four different types. These have very distinct clinical, radiologic, and histopathologic features.[1] It is the condition of increased bone density or sclerotic bone,[2] inherited both by autosomal dominant and recessive ones.

A case report of a 5-months-old boy with malignant type (precocious, congenital or infantile type) of osteopetrosis was presented with plain roentgenographic images.

CASE REPORT

A 5 months old boy had recurrent upper respiratory tract infection for one month. He had anemia, enlarged abdomen and a hard lump at the chest wall. He breathed loudly. He was the product of normal labor and was full term. The other two siblings were healthy and there was no familial history of similar illnesses. The child appeared well-nourished and had received a normal course of immunization.

He was found to have hepatosplenomegaly. He was anemic (Hb 8 gm%), leukocytosis (WBC 23000,

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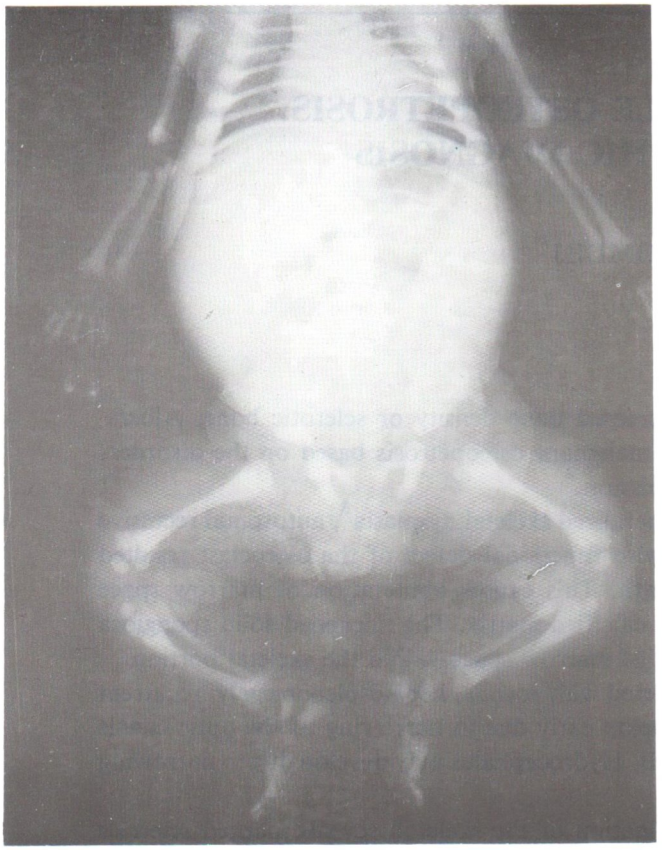

Fig.1: Humanogram including the long bone of upper and lower limbs, spine, ribs and pelvis reveals the increased bone density as sclerosis of the entire bone with obliteration of normal marrow cavity, sclerosis involved the epiphysis, spine and tarsal bone. The sclerotic bone in bone is seen in the pelvic bone, talus, calcaneum, and short bones of the hand and feet. The healing fracture with periosteal new bone as the result of brittle osteopetrosis presented at metaphyso-diaphyseal region of proximal and distal parts of femurs, tibia, humeri, radius and ulna.

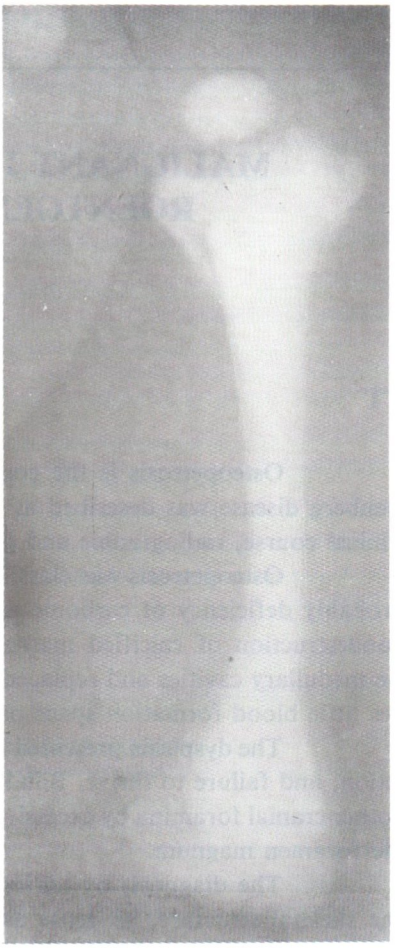

Fig.2: Long bone of left femur revealed the characteristic bone changes of infantile malignant osteopetrosis. The sclerotic clubbing enlarged and fraying metaphyseal end with sclerotic diaphysis and obliteration of normal medullary marrow cavity is noted. The periosteal reaction was disclosed at the metaphyso-diaphyseal region which indicated the healing fracture of the fragile bone disorder. The epiphysis was sclerotic which was the opposite finding seen in ricket.

PMN 29 Band 2, blast 1, promyelocyte 1, metamyelocyte 2, Eos 8, L 49, atypical L8), NRBC was 21/100 RBC, the reticulocyte count was 8% polychromasia 1+.RBC morphology was of hypochromic normocytic pattern and the platelet count was 48000. The LFT, UA and the stool examination was within normal limit. The BUN and creatinine ratio was 5.4 to 0.11. The blood picture was defined as leukoerythroblastosis. The TORCH and VDRL titer were negative. The bone marrow biopsy was obtained from the metaphysis of the tibia and revealed myelofibrosis.

The roentgenographic study of the skull, long bones, spine and flat cuboid bones revealed generalized bony sclerosis with obliteration of the marrow space (Fig.1,2). The metaphyseal ends of the long bones were enlarged and clubbed, having fraying appearance. The long bones of the limbs, were affected. The anterior end of the ribs had the rachitic rosary pictures (Fig.3). The striking findings are the sclerosis of the diaphysis or shafts of the long bones with obliteration of the normal medullary cavity. The fracture of the metaphysis was present with evidence of healing process by the periosteal new bone reaction at the metaphysodiaphyseal region of the proximal femurs. The epiphysis were sclerotic which was in opposite to the findings in ricket. The vertebral bodies and the

small bones of the tarsal bones were also sclerotic (Fig.4). The similar involvement of the skull, and orbits were seen (Fig.5).

DISCUSSION

Infantile malignant osteopetrosis is a rare autosomal recessive disorder characterized by the presentation of diseases in the infancy. It occurs as the result of the osteoclastic functional defect. There is generalized osteopetrosis or dense sclerosis of the bones of the calvarium, spine, long and short small bones. The obliteration of the marrow cavity in the bone is characteristic which results in the profound anemia, hepatosplenomegaly, myelofibrosis or leukemia as were noted in this case. Most patients of the reported cases presented with anemia, bleeding tendency, susceptible infection of the upper respiratory tract or of skin due to the functional defect of the osteoclast.[3,4,5]

The metaphyseal dysplasia is seen as enlarged metaphyses, clubbing and fraying; the sclerosis is less, compared with the denser sclerotic diaphysis. The epiphysis is also sclerotic. These abnormalities are definitely not similar to what is seen in ricket. In ricket, the metaphysis is osteoporotic though the fraying and cupping appearance is present. The diaphysis and epiphysis is osteoporotic and the diaphysis may have the picture of osteomalacia. Congenital metastaes of neuroblastoma and syphilis would be in the differential diagnosis.

The defect of the osteoclast is responsible for the under constriction of the tubular bone at the metaphysodiaphyseal part. The sclerotic bone is fragile and the fracture is noted at the metaphysis with the periosteal new bone formation. The appearance of the dense bone in the bone is mostly observed in the small bones and in the epiphysis of the mandible and iliac bone. The rachitic rosary noted in this case was shown at the anterior end of the ribs of the enlarged metaphysis with sclerosis[2,6].

The thickening of the bone at the cranial foramens causes the regional neurological deficit such as blindness, hearing loss and facial palsy. The thick calvarium later would present as the hair on end on radiographic film, MRI and CT scan with the perpendicular striation of the active marrow. The thickening of the paranasal sinuses was reported and appeared as the poor or nonpneumatized air sinuses, including small nasal cavities, which might be responsible for recurrence of the upper respiratory tract infection.

The dense orbits and skull base are quite unique for this entity. The bone marrow scintigraphy of the cranium revealed extensive marrow activity in the expanded bone marrow cavity.

Hydrocephalus, brain stem compression and the obstruction of the CSF flow is caused by impingement at the foramen magnum, demonstrated clearly by MR and CT imaging in osteopetrosis[6,7,8].

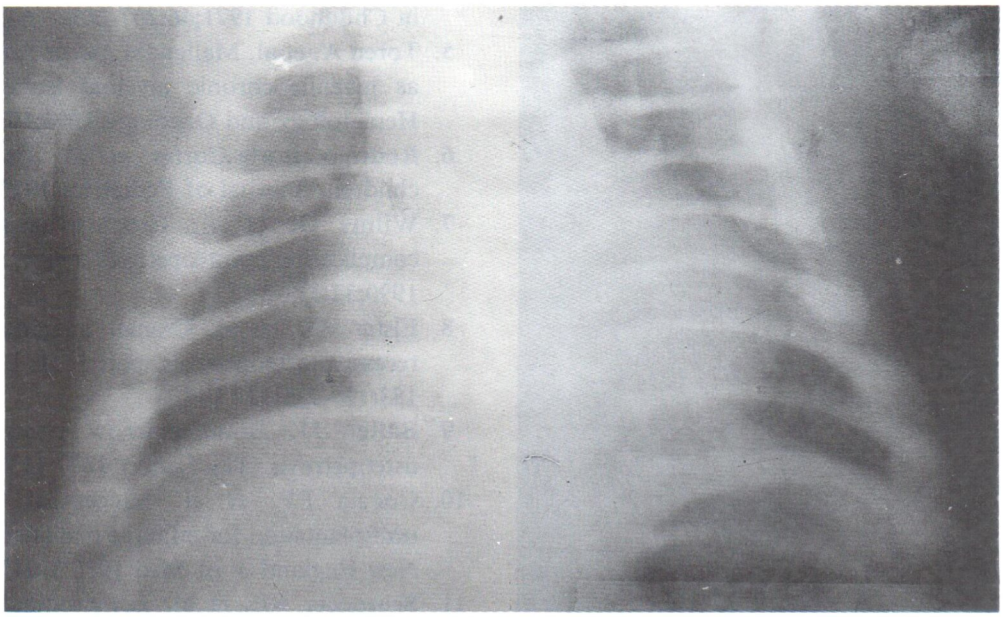

Fig.3: Dense rachitic rosary of the anterior end of the ribs is seen at the lateral part of thoracic cage, as the dense enlarged metaphysis of the sclerotic ribs. The thoracic spine was osteopetrotic.

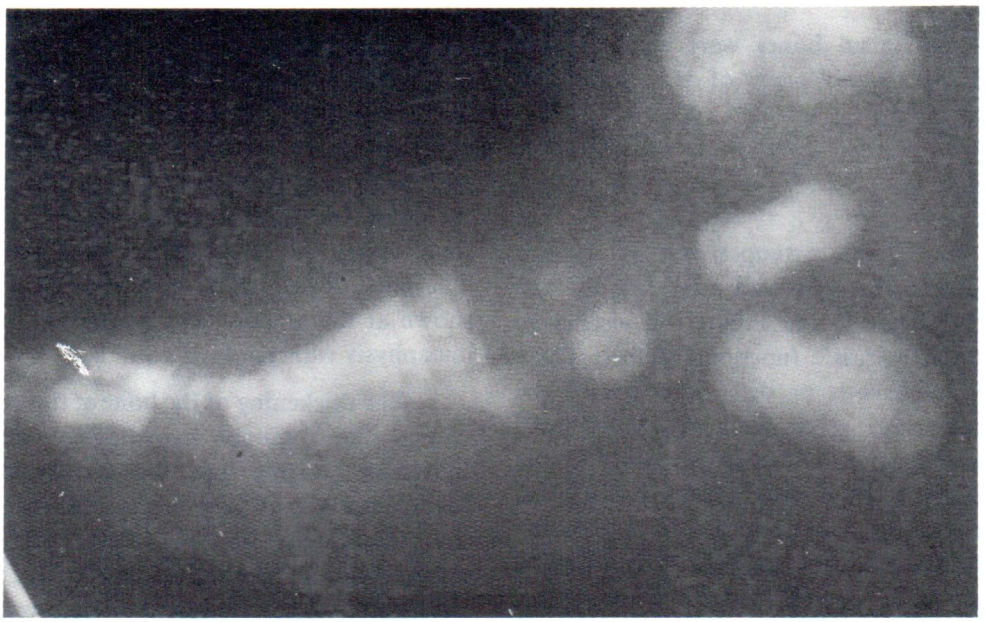

Fig.4: The sclerotic bone in bone revealed at the Talus, calcaneum and metatarsal bones.

The infantile malignant osteopetrosis shows the malignant course of the disease. It has extremely poor prognosis, the patients died in early childhood. The recent treatment was successful in the aspect of remission of the hematologic abnormalities and decreased bone abnormalities by bone marrow transplantation.[9,10,11]

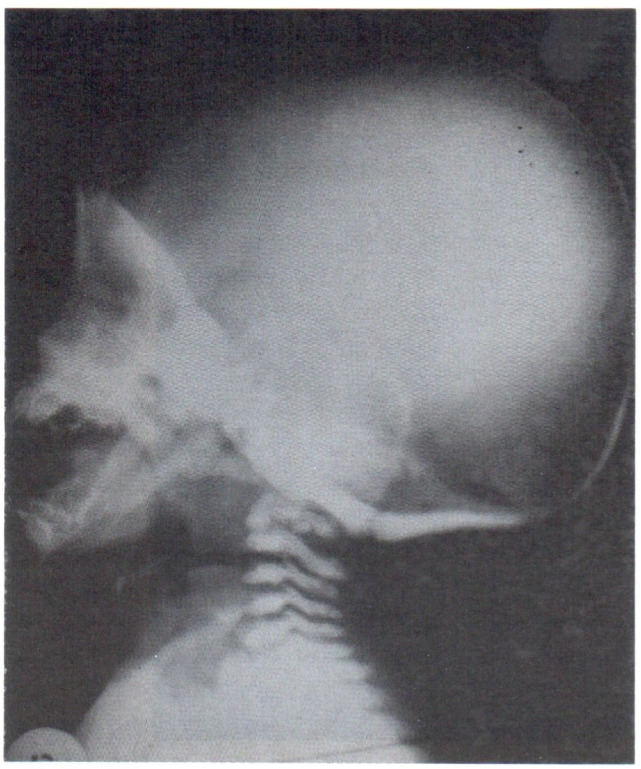

Fig.5: Lateral view of the skull revealed the marked sclerosis of the skull base, occipital bone, orbits, mandible and cervical spine.

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