Case report

OSTEOPETROSIS WITH LEUKOERYTHROBLASTIC ANAEMIA

1Dr. D. G. KULKARNI *, 2Dr VRUSHALI C KULKARNI, 3Dr. JYOTI KUDROMOTI

1,3 ASSOCIATE PROFESSOR, 2 LECTURER
PATHOLOGY DEPARTMENT, BJGMC, PUNE, INDIA
CORRESPONDING AUTHOR *

ABSTRACT
Osteopetrosis is a rare inherited disorder of osteoclastic function, resulting in presence of too much bone. It has a variety of clinical manifestations including visual and hematologic impairment. We recently came across a case of osteopetrosis with leukoerythroblastic anaemia in our hospital. The patient was a two & half years old child who presented with dysmorphic features, blindness & bone pain. X rays of limbs showed ‘bone within bone appearance’ & peripheral blood smear showed leukoerythroblastic anaemia.

Key words: Osteopetrosis, Leukoerythroblastic anaemia

INTRODUCTION
Osteopetrosis is a rare autosomal recessive disorder characterized by defects in osteoclastic function, resulting in a failure of removal of bony trabeculae and decreased marrow space. It is conventionally subclassified into a benign autosomal dominant adult form and a malignant autosomal recessive variety. The severe form is usually detected in infancy or earlier because of macrocephaly, hepatosplenomegaly, deafness, blindness and severe anaemia. X rays show diffuse bone sclerosis and a characteristic bone within bone appearance. Later on these patients have failure to thrive and psychomotor delay. These individuals rarely survive beyond second week.

The autosomal dominant form of osteopetrosis is also known as Albers- Schonberg disease or marble bone disease. It usually presents during childhood or adolescence with fractures and mild anaemia. X rays show increased bone density and a sandwich appearance to vertebral bodies. The clinical presentation pertains to developmental delay, ocular involvement, hepatosplenomegaly, neurodegenerative changes and spontaneous fractures. The hematological abnormalities include hypoplastic anaemia, hemolytic anaemia or leukoerythroblastic anaemia.

Recently we came across one such case of malignant osteopetrosis presenting with leukoerythroblastic anaemia which we thought worthy of reporting.

CLINICAL SUMMARY
A two and half years old male child, second issue of third degree consanguineous marriage presented with complaints of bone pain & tenderness aggravated since one week and dysmorphic features with tower head, slanting forehead, retrognathism, craniosynostosis, depressed bridge, flat occiput, double malleoli, small chest, wrist widening and tender long bones since birth and mild hepatosplenomegaly. There was history of physical
and mental developmental delay & the child was blind since birth. 
There was no family history of similar illness. 
Multiple X rays appendicular skeleton showed osteosclerosis with ‘bone within bone appearance’. 
His serum calcium was 8.8 mg/dl & serum phosphorous was 5.3mmol/l.

PATHOLOGICAL FINDINGS

Patient’s automated complete blood count revealed:
HGB-8.4g/dl RBC indices & platelet count within normal limits, WBC count of 19x10^3/µL, with Lymphocytes% 29.6%, Mixed population% 8.8%, Neutrophils% 61.6%.Peripheral blood smear showed predominantly normocytic mild hypochromic RBCs, and leukoerythroblastic picture with 8 to 10 nucleated RBCs/ 100WBCs, and leukocytosis with shift to left upto promyelocytes with differential count as follows: promyelocytes- 1%, Myelocytes – 4%, Metamyelocytes -6%, Band forms -6%, Polymorphs -50%,Lymphocytes -28%,Eosinophils -3%,Monocytes -2%. Platelets were adequate, Bone marrow aspiration was deferred due to severe bone pain. The liver and spleen biopsy revealed foci of extramedullary hemopoiesis.

DISCUSSION

Osteopetrosis is a rare inherited disease with an incidence of 1:500000 in north American population. [6] Extensive literature hunt yielded few stray (19) case reports from India. In osteopetrosis, due to defective bone resorption, bone encroaches on marrow leading to leukoerythroblastic anaemia. [7] Due to marrow failure, patient has extramedullary hemopoiesis in liver and spleen causing hepatosplenomegaly. Patients with early hematologic impairment, especially when combined with visual impairment have a very poor prognosis. [8]

Fig 1: X ray of hand showing bone in bone appearance
Fig 2 :Peripheral smear showing leukoerythroblastic anaemia
Fig 3: Liver biopsy showing extramedullary hemopoiesis
References
2. Wintrobe’s clinical haematology 11th edition: 1482
7. de Gruchy’s clinical hematology in medical practice. fifth edition: 274