



Case Report

Osteopetrosis Masquerading As Anemia: A Case Report

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ABSTRACT

Osteopetrosis, also called as marble bone disease or the Albers Schonberg disease; is an extremely rare inherited disorder. The basic pathology is the defective resorption of bone by the osteoclasts. This results in an increased bone mass along with a decreased hematopoietic tissue, leading to normocytic normochromic anaemia. We recently encountered a case in a 4 month infant presenting with delayed milestones, pallor and mild hepatosplenomegaly. On hematological examination, the smear showed a leukoerythroblastic picture. The bone marrow biopsy and radiological features were instrumental in the diagnosis of this rare disease. This case highlights osteopetrosis as a rare cause of anemia.

Key words: Osteopetrosis, leukoerythroblastic picture, anemia.

INTRODUCTION

Anemia remains a widely prevalent problem in medical science. Among the morphological types of anemia, normocytic normochromic anemia could be a result of either marrow production defect, red cell maturation defects (ineffective erythropoiesis), and reduced red cell survival (blood loss/hemolysis). Osteopetrosis is a rare, inherited disorder responsible for several morbidities pertaining to skeletal system. But a presentation of anemia hardly evokes a thought of such a rare disorder. We present here one such rare incident of osteopetrosis in an infant presenting with anemia.

A 4-month old male infant presented to the hospital with presenting complaints of poor sucking and pallor since one month. He also had a history of noisy breathing. He was born out of a non-consanguineous marriage. His developmental milestones were mildly delayed. His weight and height were 3.88 kilograms and 58 cms respectively. The child was immunized for age.

On examination, the infant was found to have pallor and frontal bossing. There was rotatory nystagmus and puffiness of eyelid. Multiple mongolian spots were noticed all over the body. Significantly, the child had mild hepatosplenomegaly, both palpable 5cms beneath the respective costal

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margins. The rest of the systems were within normal limits.

The laboratory investigations revealed anemia (hemoglobin; 7.3gm %), leucocytosis (WBC count: 33.6×10^3 / cu.mm) and thrombocytopenia (platelet: 72×10^3 / cu.mm). The peripheral smear showed a leukoerythroblastic profile with a normocytic normochromic anemia, polychromasia and nucleated red cells. The leucocytes showed left shift of granulocytes upto myelocytes (figure 1). A hemolytic work up comprising of a negative osmotic fragility test, a normal hemoglobin electrophoresis, a negative Coomb test and normal glucose 6 phosphate dehydrogenase level (31U/gm) essentially ruled out a hemolytic disorder.

The biochemical tests showed mild hypocalcemia (7.9mg/dl) and hypomagnesemia (2.1mg/dl). Serum alkaline phosphatase was elevated (788U/L). The ophthalmic examination showed bilateral partial optic atrophy devoid of cherry red spots.

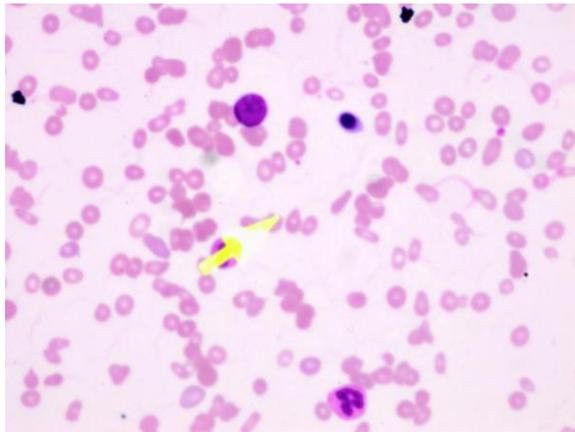


Figure 1: Peripheral smear showing a leukoerythroblastic picture. Note the normocytic normochromic anemia and nucleated RBCs (Leishman, x 200).

In view of significant organomegaly, a bone marrow study was requested by the clinician. The bone marrow aspirate was dilute and showed only minimal cellularity. Erythroid series was mildly increased and

showed normoblastic maturation. Myelopoiesis showed predominantly mature forms, with mild dyspoiesis. Megakaryocytes were adequate in number. However, no abnormal cells were seen. The imprint smear was also inconclusive. The pediatrician was requested to wait for the ensuing bone marrow biopsy.

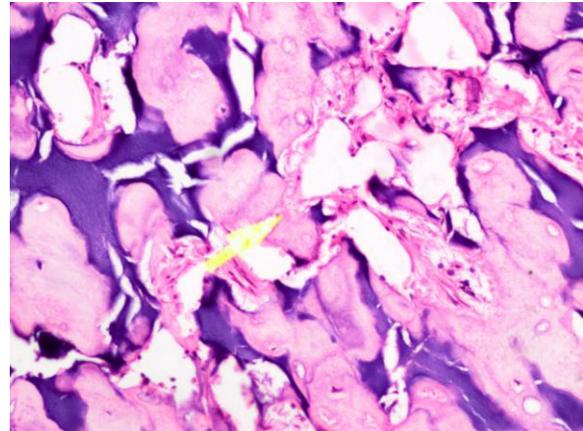


Figure 2: The bonemarrow biopsy showing thick sclerotic trabeculae. Note the compromised marrow spaces. (Hematoxylin & Eosin; x200).

The trephine biopsy from the iliac crest showed thickened, disorganized bony trabeculae with regular cement lines and reduced marrow spaces. There was variable fibrosis of marrow spaces and severe effacement of hematopoietic spaces by the fibrous tissue. Erythroid and myeloid series showed all stages of maturation. Megakaryocytes were adequate with few hypolobated forms and focal clustering. Significantly, a focus of calcific material in the cartilaginous matrix was noted in the trabeculae (figure 2). No osteoclast activity was seen. Thus, a diagnosis of osteopetrosis was rendered.

The vertebral radiogram showed dense sclerotic bony shadows and end-plate thickening. The typical "ruger-jersey" appearance of the vertebrae was not seen. These features were suggestive of osteopetrosis.

Based on the clinical, hematological and radiological findings, a final diagnosis of osteopetrosis was rendered.

DISCUSSION

Osteopetrosis (OP) is a disease where the etiology is not known. [1] It is also called as “marble bone” disease or the Albers Schonberg disease. It is a genetically determined heterogeneous disorder classified either as autosomal recessive and dominant type. The recessive type, also called as congenital / infantile OP; manifests in infancy and portends a bad prognosis due to a severe bone marrow failure. [2] The autosomal variety, also known as benign variety, is detected in older children, adolescents and adults and has a normal bone marrow function. [2,3] The basic pathophysiological mechanism in all three types of OP is however the same, i.e. failure of the osteoclasts to reabsorb bone, leading to a thickened, sclerotic bone. The clinical features are manifested due to a poor mechanical and tensile property of the bone.

The age of presentation may range from 2 to 4 years as described by earlier published literature.³ Although a presentation in infancy is also described.² The present case involved a 4 month old infant. Anemia and thrombocytopenia were seen in our patient and has also been described in other case reports as well. [1-4] They manifest due to bone marrow failure resulting from decreased volume of the medullary cavity. [3,5] The same explains the organomegaly due to extramedullary hematopoiesis. Bone marrow aspirate studied in this case was a dry tap which has been reported by Mahdi et al and Saluja et al. [6,7] This can be explained on the basis of fibrotic changes. Subsequent bone marrow biopsy revealed the histopathological features consistent with osteopetrosis. The degree of disorganization of bony trabeculae and effacement of hematopoietic spaces by

fibrous tissue suggested a possibility of osteopetrosis. But these histopathological features are not specific to osteopetrosis. The small focus of calcific material in the cartilaginous matrix, noted in the trabeculae is regarded as the most specific feature.⁷ Hence the bone marrow biopsy is quintessential for the final diagnosis of osteoporosis. The degree of disorganization of bony trabeculae and fibrosis has also been used as distinguishing criteria to classify cases of osteopetrosis as either autosomal dominant OP or autosomal recessive OP. [8]

Histologically, the other prominent features characteristic of osteopetrosis include the thickening of the trabeculae due to an increased deposition of lamellar bone and osteoclasts. [9]

CONCLUSION

Osteopetrosis is a rare cause of anaemia due to effacement of hematopoietic tissue by fibrous tissue. A leukoerythroblastic picture subsequent to such a marrow pathology is rarely, if at all, implicated in the presentation of this disease. The bone marrow biopsy and radiological features are instrumental in the diagnosis of this rare disease. This case highlights osteopetrosis as a rare cause of anemia.

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