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Case Report

Anemia with massive splenomegaly an uncommon presentation of osteopetrosis in adult

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ABSTRACT

Osteopetrosis was first described by a Germen radiologist Albers - Schoenberg in 1904. It is characterized by excessive density of bones leading to typical "chalk bone" appearance on skiagram. There is a defect in the osteoclastic activity which leads to failure in remodeling of the developing bone. This excessive formation of bone with defective osteoclastic activity leads to mechanically weak bone. So, fractures on minor trauma are common presentation of the disease. Obliteration of marrow cavities leads to development of secondary anemia which is a feature of infantile form of osteopetrosis (malignant osteopetrosis), usually seen in infants and early childhood. Adult form of osteopetrosis known as benign osteopetrosis may remain asymptomatic and causes delay in the early diagnosis of it. Anemia and massive splenomegaly in adult is a very uncommon presentation of the osteopetrosis, as seen in this interesting case which we are discussing further in detail.

Keywords: Myelophthisic anemia, Marble bone, Osteopetrosis, Skeletal dysplasia, Splenomegaly

INTRODUCTION

Anemia with massive splenomegaly is not an uncommon clinical condition. The patients of Hematological disorders like hemolytic anemia, chronic myeloid leukemia, myelofibosis and other myeloproliferative disorders. In parasitic infections like malaria and kala-azar, anemia with massive splenomegaly is not an unusual feature. Patients of Osteopetrosis presenting as anemia and massive splenomegaly in adult is an uncommon presentation.

Osteopetrosis is characterized by excessive density of bones leading to similar radiological appearance of cortical and medullary compartments of bones, is a hallmark of "chalk bone" appearance on skeletal x-ray. There is a defect in osteoclastic activity which leads to failure in remodeling of developing bone. Obliteration of marrow cavities leads to development of secondary anemia (myelophthisic anemia).[1-8]. Type A, autosomal recessive, commonly seen in children is sever form of disease and death before puberty.[4] Type B of osteopetrosis is an autosomal dominant also known as adult type, it is less severe and usually possess a benign disease course and most of the time diagnosed incidentally in adult.[1]

CASE REPORT

A 31-year-old female, house wife, resident of Etawah Uttar Pradesh, presented with history of easy fatigability of long duration 10 years, abdominal discomfort in the

form of dragging sensation for 10 years, low grade fever on and off for last 1 year. There was no history of bleeding from any site, no history of dark colored urine. She did not give the history of any fracture/s in the past. She was giving history of blood transfusion (three unit of PRBC) over the last two years. There was no past history suggestive of tuberculosis and diabetes. She attained menarche at age of 14 year and menses were regular. She has been married for one year having obstetric history of Gravida 0 and Para 0. She was denying history of similar illness to any other member of the family.

Her built was small statured with height of 3 feet and 11 inches. Her weight was 34 kg which was low as per normal standard. Frontal bossing and severe pallor was detected on clinical examination. There was no icterus, cyanosis, significant lymphadenopathy, clubbing or edema. Her vital parameters like pulse, blood pressure and respiratory rate were normal, she was afebrile to touch.

On per abdominal examination, there was mild abdominal distention, no visible scar mark, pigmentation and dilated vessels. Liver was palpable two cm below the costal margins, it was firm, non-tender and having smooth surface and margins. Spleen was palpable crossing the umbilicus, firm and non- tender. There were no palpable abdominal nodes or free fluid on clinical examination and bowel sound were present. Skeletal system examination reveals short stature but proportionately equal upper and lower segment ratio and normal arm span. No abnormality of joints and ligament detected. Other systemic examinations including respiratory, cardiovascular and central nervous system were normal.

With the clinical diagnosis of anemia with hepatomegaly and massive splenomegaly and short stature, the patient was evaluated further. In view of chronic anemia requiring blood transfusion and signs of extra medullary hemopoesis like frontal bossing and hepato-splenomegaly the possibilities of hemolytic anemia was the likely diagnosis at this juncture with diffential diagnosis of other hematological disorders like chronic myeloid leukemia and other myeloproliferative diseases. In tropical countries like India in such patients presenting with anemia and massive splenomegaly likely diagnosis of chronic infections like kala-azar and malaria must be considered.

On investigation hemogram showing Hb 7.3 g/ dl, TLC - 5700 cmm, DLC % P56, L42, E02, MCV 81.7, MCH 24.7, MCHC 30.3, Plat- 1.14 lac, ESR- 65. Peripheral smear showing microcytic hypochromic red blood cells with anisocytosis, platelets and white blood cells were normal, no immature or abnormal cells were detected, malaria parasite not seen. Reticulocyte counts was 3%. Serum Lactate dehydrogenase (LDH) 230 IU, Hb electrophoresis was normal, reveals HbA 96.2%, HbA2 3.0%, HbF 0. 8%, serum Iron 87.0 mg/dl, TIBC 320 mcg/dl, S. ferritin 2.1 mg/dl.

Blood chemistry showing Serum bilirubin 0.8 mg/dl, SGOT 15 IU, SGPT 10 IU, alkaline phosphatase 144 IU S. Protein 7.0 gm%, S. Albumin 4.0 gm%, Globulin 3.0 gm%, Urea16 mg/dl, Creatinine. 0.4 mg/dl, Fasting blood sugar 87mg/dl, Na+ 136 mEq / L, K+ 3.4 mEq / L, serum calcium 6.9 mg/dl, phosphate 4.2 mg/dl, magnesium 1.23 mEq /L, PTH 116.9 pg/ml, Vitamin D (25 Hydroxy) 10.12 nmol/L, TSH 3.0 Miu/ml.

Negative for malaria antigen, rk 39 (antigen for kalaazar), IgM Typhoid, ANA (antinuclear antibody) and HIV. Blood culture was sterile. Routine and microscopic examination of urine was normal. ECG was normal. Bone marrow aspiration done, was dry tap and biopsy was deferring in view of fragile bones. Ultrasonography abdomen shows mild liver enlargement with normal echotexure, portal vein was normal, Spleen grossly enlarged, 23 cm. showing tiny echogenic focci suggestive of gamma - gandy bodies. No ascites or retroperitoneal lymphadenopathy, kidney, uterus and adnexa were normal. Upper GI endoscopy was normal.

Radiology shows increase density of bone in all skeleton with no distinction between cortical and medullary bone “chalk bone appearance” base of the skull shows marked radiopacity whereas the vault is generally less dense, x ray spine shows “Sandwich" vertebrae, alternating sclerotic and radiolucent transverse metaphyseal lines. Bone densitometry L.S. spine, femur & right radius T score of the L.S. spine, neck of both femurs and right radius are grossly increased.

Summarily, short stature and clinical features of secondary hematopoiesis in the form of frontal bossing and splenomegaly indicative of myelopathic anemia and distinct radiological feature suggesting skeletal dysplasia. Skeletal survey of her parents and siblings were carried out was normal.

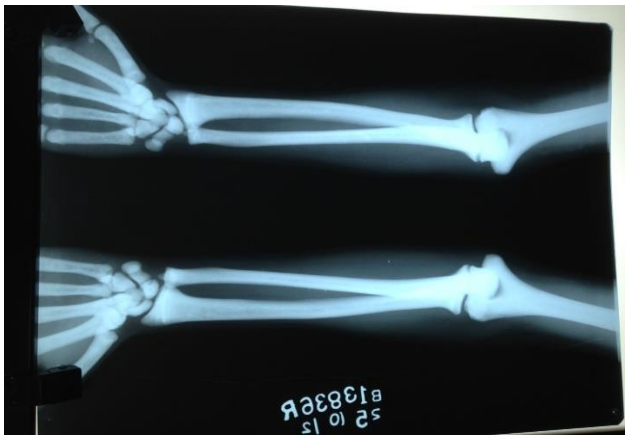

Figure 1: X-ray forearm ap view, sclerotic bone with normal interocious membrane.

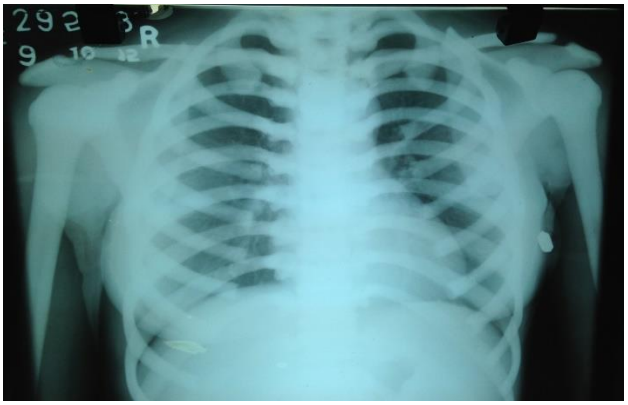

Figure 2: X-ray chest ap view, sclerotic.

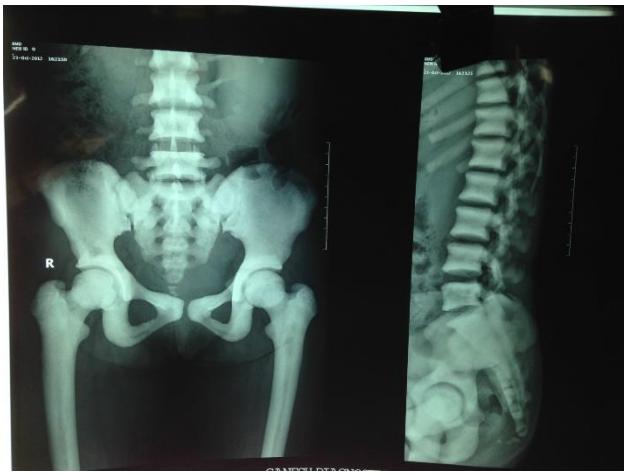

Figure 3: Dexa scan, dense pelvic bone and "rugger jersey vertebrae"

Primarily based on its clinical and radiographic features a diagnosis of osteopetrosis was made, with differential diagnosis of other skeletal dysplasia like Pyknodysostosis and cleido-cranial dysplasia. TRAP (Tartrate resistant Acid phosphatase) in serum and marrow and CK BB isoform in serum are usually increased due to destruction of defective osteoclasts, not done in our case because of non-availability at our institute.

DISCUSSION

Osteopetrosis commonly known as Marble Bone Disease was first described by a Germen radiologist Albers - Schoenberg in 1904. Disease is characterized by excessive density of bones "chalk bone" appearance there is a defect in osteoclastic activity which leads to failure in remodeling of developing bone. There is an excessive bone formation which is mechanically weak so the fractures are common. Delayed eruption of teeth and osteomyelitis is a common complication of tooth extraction.[3] Commonly two types were described. Type A infantile type, malignant and progressive, inherited as autosomal recessive occur early in life with severe bone fragility, death usually occurs before puberty.[2,5,6,9] Adult type B, a benign autosomal dominant less severe as our case possess a benign disease course, there may be repeated fracture following minor trauma which were not seen in this case. Anemia and splenomegaly is not commonly reported in adult, obliteration of marrow cavities leads to development of secondary anemia (myelophthisic anemia).[1,8] Radiology features of increase density of bone in all skeleton with no distinction between cortical and medullary bone, base of the skull shows marked radiopacity whereas the vault is generally less dense and sandwich (rugger jersey) vertebrae appearance has been described 50% of cases in Costa Rica.[7]

Pyknodostosis an autosomal recessive disease, there is systemic osteosclerosis. Bones are therefore abnormally dense, brittle and easily fracture. Characteristic phenotypical appearance with proportionate dwarfism and dysmorphic facies the maxilla may be under developed with high narrow arched palate. There may be acro-osteolysis (aplastic terminal phalanges with loss of ungual tufts), sparing of the medullary cavity within the long bones is characteristic of the disorder, resulting in normal hematopoietic function differentiate it from osteopetrosis. Cleidocranial Dysostosis is also presents similarly to pyknodysostosis, with persistent open fontanels and cranial sutures. It always involves the clavicle; partial or complete absence of clavicles allows the shoulders to be brought forwards until they meet in midline.[10]

CONCLUSION

Osteopetrosis is a rare condition that is diagnosed primarily on its clinical and radiographic features, bone biopsy is not essential for the diagnosis. Adult type is a mild form of disease, repeated fracture is common, myelophthisic anemia is uncommon presentation in adults. Usually require no specific treatment, patient has to be kept in hypocalcemic state with supplement of calcitriol to enhance osteoclastic activity and blood transfusion may be required. If hypocalcemia is symptomatic than calcium supplement is needed. The importance of recognition of these features in the diagnosis and prevention of future complications in adult type is stressed in this case report.

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REFERENCES

1. Tripathi AK, Tandon V, Kumar A, Sarkari M, Chaturvedi R. A rare cause of Leucoerythroblastic anemia. India J Hematol Blood Transf. 1998;16:62-4.
2. Bodamer OA, Braverman RM, Craigen WJ. Multiple fractures in a 3-month-old infant with severe infantile osteopetrosis. J Pediatr Child Health. 2001;37:520–2.

3. Bénichou OD, Laredo JD, de Vernejoul MC. Type II AD osteopetrosis; clinical and radiological manifestations in 42 patients. Bone. 2000;26:87–93.
4. Chen CJ, Lee MY, Hsu ML, Lien SH, Cheng SN. Malignant infantile osteopetrosis initially presenting with neonatal hypocalcemia: case report. Ann Hematol. 2003;82(1):64-7.
5. Mahdi AH. Osteopetrosis in Saudi children: A report of 10 cases. Ann Trop Pediatric. 1988;8:112-5.
6. Stark Z, Savarirayan R. Osteopetrosis. Orphanet J Rare Dis. 2009;4:5.
7. Loria-Cortes R, Quesada-Calvo E, Cordero-Chaverri C. Osteopetrosis in children: A report of 26 cases. J Pediatr. 1977;91:43-7.
8. Kumar P, Shashi Kala P, Chandrashekhar HR, Basavargin Band AS. Osteopetrosis presenting as leuko-erythroblastic anemia: A case report. Indian J Haematol Blood Transf.1998;116:260-1.
9. Sharma A, Kumar S. Osteopetrosistarda: A case report. J Indian Med Assoc.1999;97:22-4.
10. Hunt NP, Cunningham SJ, Adnan N, Harris M. The dental, craniofacial, and biochemical features of pyknodysostosis: a report of three new cases. J Oral Maxillofac Surg. 1998;56:497-504.

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