A Story of Pancytopenia Revealing Stony Bones

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A 23 year old boy resident of Dharavi, Mumbai presented to our out-patient department in August 2015 with a two year history of symptomatic anaemia requiring monthly transfusions and occasional gum bleeds. His hemogram revealed pancytopenia – Hemoglobin - 6.3 g/dL, leukocyte count - 3300 cells/mm³ (Absolute Neutrophil count - 1500), platelet - 30,000 / mm³, corrected reticulocyte count < 1 %. Viral markers and autoimmune work-up was negative.

He did not have any lymphadenopathy or organomegaly on examination. His bone marrow was planned to rule out suspected aplastic anaemia. His bone marrow was not biopsable inspite of multiple attempts.

Retrospectively, his younger brother gave history of recurrent spontaneous fractures. Hence, X-rays were done (Figures 1 to 4) to find out the etiology of underlying hard “inaspirable/inbiopsable” bones. It revealed diffuse thickening of bones (Figures 1 to 4). Patient was counseled for genetic study and a transplant but he did not show intent due to financial issues. He was symptomatically treated with hematinics and erythropoietin injections.

Osteopetrosis (‘oste’ means bone and ‘petros’ means stone), also known as ‘marble bone disease’/‘Albers-Schönberg disease’, after the German radiologist reported this condition first in 1904. The estimated prevalence of osteopetrosis is 1 in 100,000–500,000 births.¹ It is a rare genetic disorder characterized by an increase of bone mass due to defective osteoclast function.²

Osteopetrosis comprises a heterogeneous group. Patients typically present with spontaneous fractures, anemia, and in the most severe forms hepatosplenomegaly and cranial nerve compression leading to deafness and blindness.² In addition, severe hypogammaglobulinemia and elevated alkaline phosphatase³ have been described. It has two major clinical forms: the autosomal dominant adult (benign) form is associated with milder symptoms often appearing in later childhood and adulthood whereas the autosomal recessive infantile (malignant) form has severe presentations appearing in very early childhood, if untreated, is typically fatal during infancy or early childhood. A rare autosomal recessive (intermediate) form is present during childhood with some signs and symptoms of malignant osteopetrosis. Diagnosis is mainly based on clinical and typical generalized increase in bone density.¹ X-rays reveal homogeneously increased density of bones with little differentiation between cortex and medulla in long bones and pelvis, X-ray vertebrae show sclerosis of vertebral endplates resulting in “sandwich vertebrae” appearance.¹ Hematopoietic stem cell transplantation (HSCT) is the only curative treatment. Interferon-gamma1b treatment has been tried in variants unresponsive to HSCT or as a bridging therapy.¹ Our case is unique as
he presented in the third decade with symptomatic pancytopenia without any bony symptoms/organomegaly thereby revealing its heterogenous nature.

To conclude, Osteopetrosis is heterogenous in presentation and should be ruled out by radiology in any patient with stony bones. HSCT is the only curative treatment.

References

