A 51-year-old female patient presented with complaints of swelling of the left hip and proximal thigh region with an inability to bear weight on the left leg for 3 days. There was no history of significant trauma.

She had a past history of a right proximal femur fracture 16 years ago. She was operated on, and the fracture was fixed with metallic implants. She developed an infection and nonunion at the fracture site that required revision surgery. She also had bilateral shaft humerus fractures 2 months ago, without any significant trauma.

She had a history of multiple blood transfusions due to low hemoglobin (Hb), since the age of 16 years (she was never investigated for chronic anemia). There was no history of chronic blood loss or postmenopausal bleeding. There was no history of diabetes mellitus, hypertension, thyroid dysfunction, or coronary artery disease. She had a family history of similar fractures in her brothers.

On examination, she had pallor with no cyanosis/icterus/clubbing/lymphadenopathy or pedal edema.

She had nontender globular swelling over the mid-shaft of the bilateral humerus. The left lower limb was in the attitude of external rotation, with abnormal mobility over the proximal femur shaft. There was a healed scar over the right hip and proximal thigh region with shortening of the leg. There was a chronic discharging sinus over the right proximal tibia at the site of metallic pin insertion for traction. There was no vision loss, deafness, or dental anomalies. The abdominal examination didn’t show any hepatosplenomegaly.

Blood investigations showed severe anemia along with thrombocytopenia. Peripheral smear showed dimorphic anemia with decreased platelets. (six units of packed red blood cells and one unit of single donor platelets were transfused). Other causes of low Hb and thrombocytopenia were ruled out.

Radiographs showed diffuse osteosclerosis and narrowed bone marrow canal of long bones. It also showed fractures of the bilateral shaft humerus with callus formation (Figs 1 and 2). A fracture of the left proximal femur was also visible on X-rays.

The differential diagnosis for osteosclerosis includes malignancy, leukemia, myelofibrosis, sickle cell anemia, Paget’s disease of bone, renal osteodystrophy, fluorosis, sclerotic dysplasias, hyperthyroidism, and hypoparathyroidism. All these causes were ruled out.
Adult Osteopetrosis Type-2

There are three types of osteopetrosis; (1) malignant (AR); (2) Intermediate (AR); (3) benign (AD). Malignant type or infantile osteopetrosis is mostly seen at an early age and is due to proton pump or chloride channel dysfunction due to TCIRG1 gene mutation. The intermediate type is due to carbonic anhydrase II dysfunction. The benign type is due to chloride channel dysfunction (CLCN 7 gene mutation). This benign type is commonly seen in adults and is of two types. Type 1 does not have an increased fracture risk. Type 2, also known as Albers-Schoenberg disease, presents with anemia, thrombocytopenia, pathological fractures, and premature osteoarthritis. These patients have normal general health, life span, mental function, and physique. This type is the most common form managed by orthopedic surgeons.

**References**