**Case Report**

**Unusual presentation of osteopetrosis in a young female with anemia and hepatosplenomegaly**

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**ABSTRACT**

Osteopetrosis (or Albers-Schonberg disease) is a rare, hereditary bone disorder that is characterised by increased bone density caused by osteoclast dysfunction. Bones become sclerotic and thick, but their abnormal structure results in them being both weak and brittle. Excessive bone density can lead to associated problems which may be life threatening. Nerves in the skull may be compressed and cause vision loss, hearing loss, or paralysis of facial muscles. Crowding of the bone marrow can lead to low levels of cells and platelets needed to fight infection, carry oxygen to the body’s cells, or control bleeding. Clinicians managing patients with osteopetrosis face a difficult task of treating the complications. Although advances in molecular genetics have led to an improved understanding of the cellular basis of the disease, clinical management has still remained unchanged.

The characteristic radiological findings are usually sufficient for diagnosis of the disease. This article presents a case of autosomal dominant osteopetrosis type II in a young female. Most of the cases present clinically with fracture because of the weakened bones. Fractures are often transverse with multiple areas of callus formation and normal healing. In this case there was no evidence of any fracture in the bones and patient presented with symptoms of anemia. In addition, the literature is reviewed with a comparative discussion of the difficulties associated with osteopetrosis.

***Key words****:* osteopetrosis, bone dysplasia, autosomal dominant