BRIEF HISTORY: A six-year-old boy with severe visual impairment (as a result of optic canal stenosis) was referred to the pediatric clinic of Imam Khomeini Hospital, affiliated to Tehran University of Medical Sciences (TUMS). Chest X-ray (PA view) revealed an abnormal increase in bone density. Other radiographic imaging helped to secure the diagnosis of “osteopetrosis”. These findings are typical bone-within-bone appearance in hand bones, increased bone density in vertebral column and hyperdensities in pelvic bones. Osteopetrosis (marble bone disease) is shown by dense and brittle bones which are susceptible to spontaneous fractures. Bone-within-bone is a dense bone filling the medullary cavity of other bones. Figure 1 (metacarpals, phalanges, and ulna) and Figure 2 (hip bones) show the bone-within-bone appearance. In addition, bilateral deformity in head of both femurs (Figure 2) and increased density of bone in the margins of vertebral bodies (Figure 3) is seen. We referred the patient to an expert neurosurgeon for decompressing the optic nerve.

Osteopetrosis is a rare inherited disease whereby the bones harden and get more density, in contrary to osteoporosis that gives rise to a decrease in bone compaction. The pathophysiology of this disease is due to osteoclast dysfunction, i.e. osteoclasts fail to resorb bone while osteoclast mediated bone formation persists leading to excessive bone formation [1]. Osteopetrosis can manifest through a spectrum of symptoms, from nearly asymptomatic phenotype to disabling limb deformities. Because of the excessive bone formation, bone marrow space narrows and hematopoiesis retards leading to anemia. As a compensatory mechanism, extramedullary hematopoiesis forms, culminating in hepatosplenomegaly [1]. Anemia is not the only sign of bone overgrowth. Several nerve fibers that pass through cranial bony canals and foramen could be trapped so they may lose neural function. Based on this mechanism,
compressed optic, auditory, and facial nerves it can result in impairment of eyesight, hearing loss, and facial paralysis, respectively. Although bone formation is enhanced in osteopetrotic patients, their bones are fragile and they are prone to the fractures [2]. Treatment of osteopetrosis is mainly symptomatic. Fractures are common in these patients and should be managed by an orthopedic surgeon [3]. Continuous activity of osteoblasts may reduce blood calcium which requires vitamin D and calcium supplementation. It also can be a therapeutic option for hypocalcemic seizures. Transfusion of blood factors is one of the therapeutic options for treatment of bone marrow failure and anemia, although bone marrow transplantation may be applied if necessary [4]. In case nerve fibers are under pressure, surgical decompression can spare cognate sensory function [5]. The novel treatment options currently being investigated as therapeutic options such as gene therapy may one day become the absolute cure for patients suffering from osteopetrosis [6]

Figure 3: Increased bone density of the vertebral body margins is evident as shown by arrows in this lateral vertebral column X-ray graph.

REFRENCES