

Osteopetrosis - Case Report

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Abstract

Osteopetrosis is a rare disease, whose patient's bone is extremely hard, so also called the "marble bone". The pathogen occurs due to injury on bone resorption. Has hereditary character and is usually diagnosed in the first decade of life, enabling a treatment through a bone marrow transplant. However, when diagnosed later, as in the case reported is not needed the transplant because it is benign and asymptomatic.

Keywords: *Osteopetrosis; Marble Bone; Bone Marrow Transplant; Hereditary*

Introduction

Osteopetrosis or marble bone is a disease of genetic inheritance, characterized by the reduction or complete functional loss of osteoclasts [1]. It was first described in 1904 by Albers Schonberg, which is why it is also called the Albers-Schonberg syndrome [1,2].

There are three main types of osteoporosis: autosomal dominant, autosomal intermediate recessive and autosomal recessive [2]. The autosomal dominant type is the mildest form [3]. While the autosomal recessive form or malignant childhood osteopetrosis is the most severe [1,3].

The autosomal dominant form is described as benign and late, which is present in adults. Autosomal recessive is malignant and congenital. Most patients fit the latter, who has a diagnosis in the first decade of life and has a poor prognosis [2].

It is a rare disease, reported by only a few hundred cases worldwide, affecting more males [1]. About 20% of cases result from inbreeding [1]. It has a higher incidence in Costa Rica, with 3.4 in every 100,000 live births, and in Chuvashia Republic of Russia, with 1 in 3,879 [4].

It is not yet well established, however, it is known that the disease is of a genetic character that affects several genes, some not yet identified [5]. These defects cause damage in bone resorption, either due to the total absence of osteoclasts, attributable to the absence of precursor cells, to a failure in the differentiation of the hematopoietic lineage or by an enzymatic defect in the metabolism of the osteoclast itself, making adequate bone resorption impossible [2,5]. In about two thirds of the patients, osteoclasts are normally formed, but unable to perform resorption effectively, due to mutations that affect the transport of Cl⁻ or H⁺ [1].

The osteoblastic function is normal, so there is excessive deposition of mineralized osteoid material in the bone marrow, leaving the bones more dense, sclerotic and radiopaque [1]. The increase in bone mass can cause phenotypic changes such as macrocephaly and modification of facial morphology, in addition to affecting other organs and tissues [6].

The diagnosis basically depends on the skeletal radiography. It usually appears with diffuse sclerosis affecting the skull, pelvis, spine and appendicular bones, and also a defect in the metaphyseal modeling of long bones, similar to the appearance of a funnel [6].

The treatment is carried out only through bone marrow transplantation, whose donor needs to have HLA compatible 100% [7]. Hematopoiesis is reestablished, improving bone lesions and preventing the disease from progressing. The radiological improvement occurs around the second month after the transplant and is completely resolved after approximately one year [3].

Case Report

LHLF patient, 65 years old, female, sought medical assistance at the Diagnostic Diamente Medical Specialty Center, in São Luís - MA, in 2017, for the evaluation of osteoporosis. She was asymptomatic. Physical exam not worthy of note. Hypertensive in use of Losartana, denies diabetes, alcoholism and smoking. She reports a diet low in calcium derivatives. Bone densitometry (BMD) was requested, whose spine result, in the L1-L4 region, was T-score 6.6; of the femur, neck region, T-score 7.2 and trochanteric region, T-score 14.4, totaling in the femur T-score 11.6. In addition to BMD, a radiographic examination was also requested, whose image showed diffuse sclerosis and great thickness of the cortical regions of the long bones, restricting the spinal canal (Figure 1-3).



Figure 1: Radiography of the right hemithorax and right upper limb showing increased thickness of the humeral cortex.



Figure 2: Radiography of the pelvis and femur showing sclerosis and cortical enlargement.



Figure 3: Radiography of the lower limbs with an increase in the cortical region and reduction of the spinal canal.

Due to the patient's age and the absence of symptoms, she was submitted only to conservative outpatient follow-up, without the need for pharmacological treatment. Currently, the patient is well and has no manifestations of the disease that justify adherence to medications or indication for bone marrow transplantation.

Discussion and Conclusion

Second, Borsato, *et al.* [3] osteopetrosis consists of an inherited condition in which the clinical manifestations can vary from asymptomatic to a fatal course of the disease. It also has two forms, an autosomal recessive, which is more severe and another autosomal dominant, characterized as benign and late.

The present report describes a patient in which the clinical picture is characterized in an autosomal dominant form, due to her age of 65 years and absence of symptoms, and the changes were observed only in bone densitometry and bone radiography.

As a rule, the disease is commonly diagnosed in childhood, according to Athar, *et al.* [2] so at this time in life it is possible to perform a more invasive and more successful treatment, such as bone marrow transplantation.

However, the case reported here does not match what is described in the literature, because, due to late diagnosis and the absence of clinical manifestations, invasive treatment with transplantation was not necessary, only conservative follow-up and monitoring of the disease was proposed.

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