A Rare Patient with Osteopetrosis Presenting in Whole-Body Bone Super Scan

Yan-Li Xue, Chen-Tian Shen, Zhong-Ling Qiu and Quan-Yong Luo*

Department of Nuclear Medicine, Shanghai Sixth People’s Hospital, Shanghai Jiao Tong University, 600 Yishan Rd., Shanghai 200233, China

Abstract: Osteopetrosis is a hereditary group of bony dysplasia caused by diminished osteoclast activity. It is a rare bone disease with an incidence of 1/500000, therefore, doctors may experience only few cases, or only a single case within their lifetime. We hereby reported a case of osteopetrosis who presenting with a super-scan found by 99mTc-MDP whole-body bone scan.

Keywords: Osteopetrosis, 99mTc-MDP, whole-body bone scan, super-scan.

A 11-year-old girl was referred for 99mTc-MDP whole-body bone scan with unexplained skeletal pains and markedly elevated serum alkaline phosphatase activity. Approximately 3 hours after intravenous injection of 555 MBq (15mCi) of 99mTc labeled methylene diphosphonate (99mTc-MDP), anterior and posterior views of the whole-body were obtained using a dual-head gamma camera with low-energy general purpose collimators. Intense radioactive uptake was noted at the epiphyses and metaphysis of the proximal humeri, distal femora, proximal and distal metaphysis of tibiofibula, and the spine (Figure 1). The patient was suspected for high risk of osteopetrosis, then a X-ray examination was performed in order to confirm the diagnosis. The X-ray images showed notably increased density of the bone, thickening cortical bone, narrowing even occluded of the bone marrow cavities of lower limbs, and marked “sandwich sign” of the vertebral bodies (Figure 2). The X-ray manifestations were the typical characteristics of osteopetrosis. The final diagnosis of osteopetrosis was confirmed by bone biopsy.

Osteopetrosis is a hereditary group of bony dysplasia caused by diminished osteoclast activity [1,2], resulting in defective bone remodeling and increased bone density, which was known as marble bone disease, congenital osteosclerosis, osteosclerosis generalized fragilitas, or chalky bone, etc. Because it was first described by Albers-Schonberg in 1904, it is also called Albers-Schonberg disease. Osteopetrosis is a rare bone disease with an incidence of 1/500000 [3], therefore, doctors may experience only few cases, or only a single case within their lifetime.

Osteopetrosis is generally divided into three types: severe infantile malignant autosomal recessive, intermediate mild autosomal recessive, and benign autosomal dominant [4,5]. The prognosis of the first two types is very poor and is characterized by an early onset, usually within the first decade of life, and early death. The third type is characterized by a later onset and a longer life span [4]. Generally, osteopetrosis
lacks typical clinical features and the radiological findings are well-known, including increasing bone sclerosis with eventual marrow cavity obliteration, long bone metaphyseal remodeling deformities, obliteration of the skull diploic cavity, and sclerotic vertebral bodies often with a “sandwich vertebra” appearance [6,7].

The interesting and unusual feature of this case is the $^{99m}$Tc-MDP whole-body bone scan resembled a typical super-scan, including symmetric intense radioactive uptake at the epiphyses and metaphysis of long bones and the spine [8-10] and nonvisualization of both kidneys. The intense radioactive uptake at the metaphyseal regions of the long bones and spine may be due to the defective osteoclasts in these regions resulting continual osteoblastic activity [11,12]. The super-scan image may be related to the rapid and enhanced uptake of $^{99m}$Tc-MDP by the abnormally thickened cortical bone, resulting in reduced radioactivity excretion by the kidneys [10]. It is a typical characteristic of metabolic bone disease.

Infantile osteopetrosis warrants treatment because of the adverse outcome associated with the disease. Vitamin D (calcitriol) appears to help by stimulating dormant osteoclasts, thus stimulating bone resorption. Treatment with gamma interferon has produced long-term benefits. Erythropoietin can be used to correct anemia. Corticosteroids have also been used to treat anemia, as well as to stimulate bone resorption. Adult osteopetrosis requires no treatment unless complications arise which do require treatment. No specific medical treatment exists for the adult type.

In conclusion, whenever a super-scan detected by $^{99m}$Tc-MDP whole-body bone scan in a teenager, we should be aware of the possibility of osteopetrosis, although osteopetrosis is a rare bone disease. The patterns of $^{99m}$Tc-MDP uptake as in this case can significantly enhance the interpretation of whole-body bone scan and radiograph in osteopetrosis.

REFERENCES


Figure 2: (A) Radiographs of lower limbs demonstrated bone density increased notably, cortical bone thickening and bone marrow narrowing even occlude. (B) Chest and abdomen X-ray images showed “sandwich sign” in the vertebral bodies.
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