

A Case of Geroderma Osteodysplastica Diagnosed by Recurrent Pathologic Fractures

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Abstract

Geroderma osteodysplastica (GO) is a rare autosomal recessive disorder. We report the case of a 33-year-old patient with GO, with generalized osteoporosis and frequent spontaneous fractures. We should consider this condition in the differential diagnosis of a patient with frequent spontaneous fractures.

Keywords: Geroderma osteodysplastica; Osteoporosis; Fracture; Bisphosphonate

Introduction

Geroderma osteodysplastica (GO) is a rare autosomal recessive connective tissue disorder first described by Bamatter et al (1950) in five members of a Swiss family [1]. Fewer than 35 patients have been reported to date [2]. It is characterized by an aged appearance with droopy eyelids, prominent forehead, and underdeveloped maxillary and malar bones. The skin is lax and wrinkled, especially on the extremities, and patients exhibit various musculoskeletal abnormalities including arm span greater than height, kyphoscoliosis, hyperextensible joints, osteoporosis, and hypotonia [3, 4]. GO is reported primarily in countries of the Middle East, where most patients have consanguineous parents [2].

In this report, we describe a patient with recurrent fractures and features of GO in the hope that it will provide essential information for endocrinologists and orthopedic surgeons to consider this condition in their differential diagnosis of spontaneous fractures.

Case Report

A 33-year-old female patient of healthy Saudi parents in a consanguineous marriage (parents were first cousins) was born by normal delivery after an uneventful pregnancy. The patient had multiple spontaneous fractures of the lower extremities between 3 and 12 years of age, which were treated conservatively, with subsequent limitation of her physical activity. Although she had multiple fractures, no diagnosis or treatment was provided until she was 14 years old, when she sustained a severe displaced fracture of the right femur that was fixed surgically. Her face appeared aged with a prominent forehead, low set posteriorly rotated ears, sagging cheeks, and short neck.

The patient's skin was lax, wrinkled mainly over the dorsum of the hands and feet, and she had an atrophic, band-like skin lesion on her forehead extending posteriorly to the scalp region. In addition, she had scoliosis with marked joint laxity. The initial dual-energy X-ray absorptiometry (DEXA) scan showed a severe decrease in bone density (total lumbar spine (L1-L4): 0.870 g/cm², Z score -3.5; total right femur: 0.566 g/cm², Z score -4.0).

The patient had the clinical criteria for GO, which prompted testing for *GORAB* mutations that turned out to be positive. She was treated with pamidronate (60 mg) twice for a period of 1 year. Three years later, in the absence of treatment, a repeat DEXA scan showed a bone mineral density (BMD) of 0.792 g/cm² for L1-L4 (Z score -4.1), but the total right femur was difficult to assess accurately. Two years later with intravenous (IV) bisphosphonate, bone density has improved (L1-L4: 0.842 g/cm², Z score -2.3) and was stable at the last clinical visit. The patient had been taking methotrexate as well for linear scleroderma of the scalp, with good clinical response.

Discussion

Our patient suffered from a disorder characterized by a prematurely aged face, lax skin mainly on the hand, and skeletal abnormalities including osteoporosis and increased susceptibility to fractures, particularly the vertebrae and lower extremities. In addition, a tight forehead appearance was due to linear scleroderma. GO is caused by mutations in the *GORAB* gene (1q24.2) [5], which was confirmed in our patient. This condition should be considered in the differential diagnosis of any patient with lax skin, especially if the acral areas are primarily

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involved [2].

Radiologic changes such as cone-shaped epiphyses and metaphyseal peg in addition to low BMD and skeletal abnormalities support the diagnosis of GO [6]. A histopathology finding of elastic fiber fragmentation in the affected skin can be also used to confirm the diagnosis [7].

Our patient sustained 10 spontaneous fractures, often involving the vertebrae, resulting in severe kyphosis. The proposed mechanism of fracture involves several aspects: inactivity, insufficient nutrition, and hyperlaxity of the joint, as well as a congenital disorder of collagen development [8]. It is important to test for low BMD in patients with congenitally lax skin in order to diagnose GO at an early stage.

Bisphosphonate (IV zoledronic acid) has been restarted, and the patient has been fracture-free for the last 2 years, with marked improvement of bone density. Significant improvement and stabilization of bone disease after bisphosphonate therapy have been observed in a previous study [9].

Conclusion

This case illustrates the need to consider GO in the differential diagnosis of any patient presenting with recurrent, spontaneous fractures with congenitally lax skin and abnormal facial features.

The devastating consequences of recurrent fractures illustrate the need for early evaluation, proper diagnosis, and early initiation of bisphosphonate therapy, with close follow-up of bone density.

Conflict of Interest

The authors have no conflict of interest.

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