



Multiple long bone fractures in a man with pycnodysostosis; a case report

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Abstract

Pycnodysostosis is a rare genetic disease that is diagnosed in childhood however in very rare conditions, maybe diagnosed in adulthood. It is presented by dental malformation, short stature and short extremities, multiple fractures with low-pressure forces, and some other manifestations. In this case report, we present a patient with typical pycnodysostosis manifestations including a hard palatine groove, poor hygiene oral situation, dental malformation, and several fractures with a low threshold pressure.

Introduction

Pycnodysostosis is a rare autosomal recessive disorder that affects 1 to 1.7 people out of every million (1). Loss-of-function mutations of the cathepsin K (CTSK) gene cause pycnodysostosis. CTSK is responsible for collagen type 1 degradation at low pH. However, collagen degradation does not occur and there are several cytoplasmic vacuoles filled with undegraded collagen in these cells (2). Patients typically have short stature or dysmorphic features in face and extremities. Pycnodysostosis is a form of osteosclerosis, and osteopetrosis, associated with decreased bone resumption and bone fragility. The disorder is usually suggestive of atypical facial features. However, osteosclerosis and acroosteolysis of the distal phalanges indicate a more definitive diagnosis (3). It is important to recognize the clinical manifestations of the disorder, but it is difficult sometimes (4). In pycnodysostosis, subtrochanteric and proximal femoral fractures are common. Due to the hard-but-brittle bone, surgical treatment for the patient with a fracture is a real challenge for the orthopedic surgeon (5,6). In this case report, we aimed to present a 51-year-old man with some dysmorphic features and multiple fractures.

Case Presentation

A 51-year-old man that worked in a printing house, had lower height and weight than his

Key point

In this report, we describe a patient with pycnodysostosis with some manifestations including short stature, multiple fractures as a rare disease.

peers since he was a child. There was an uneventful maternal and neonatal history and he had no similar history in his family. The first time when he was 15 years old, he experienced a left tibia fracture due to low threshold trauma. Two years later, his left tibia bone fractured again. The patient mentioned numerous fractures in his whole life such as femoral fracture leading to trivial trauma (patient hit his femur by himself then his femur fractured) or hip fracture about nine years ago due to car-car accident but it was not very vigorous. He had a history about several times hospitalization because of several fractures in an orthopedic ward. Last time, three days ago, he had a fracture in right femur and left ulna due to falling on the same level place. In general appearance we saw some abnormalities in his face, teeth, and extremities. In physical examination he had the following findings; a groove in hard palate (Figure 1), nail deformities, short extremities (Figure 2), frontal and occipital bossing, caries and tooth loss, poor dental hygiene, beaky nose and obtuse mandibular angle. All laboratory tests were normal, such as complete blood count, serum calcium,

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Figure 1. A groove in hard palate in pycnodysostosis.



Figure 2. Short extremities in pycnodysostosis.

phosphate, alkaline phosphatase, 25-hydroxy vitamin D, and parathormone and also cortisol values. A generalized increase in bone density and some fractures was found in radiographic examination (Figure 3). These clinical and radiological features with normal laboratory study, supported the diagnosis of pycnodysostosis. The patient underwent orthopedic surgery to treat the fractures. Due to the small size of the medullary canal, we decided to internally fix the fracture with a 95-degree angled blade plate. We placed the patient in lateral decubitus position, then opened the fracture and refreshed and opened the medullary canal with a 4mm drill bit. Then we insert the guide pin for starting point of the blade. During insertion of the chisel, we found excessive retroversion of the femur neck, while the chisel penetrates and breaks the anterior femoral neck. Then, we decided to use an intramedullary rush nail to repair and fix the fracture.

Discussion

Pycnodysostosis is a rare hereditary disorder characterized by short stature and bone fragility (7). In the present report our patient had several fractures in several bones with low-pressure. In a case report by Puri et al, a 34-year-old woman with specific feature such as short stature, particularly limbs, short broad hands, frontal and occipital bossing, and normal laboratory data was reported. Our patient, had a hard palatine groove, dystrophic nails and normal laboratory findings. Usually the age of pycnodysostosis's detection is variable ranging from nine months to fifty years. One of the points of this case report was that patients with pycnodysostosis may have higher age but remained undiagnosed(8). It is estimated that the universal frequency of pycnodysostosis is 1 to 1.7 per



Figure 3. Fractures and increase in bone density in pycnodysostosis.

million with equal gender distribution (9). Dhameliya et al in a study showed a 25-year-old female who had a typical manifestation of pycnodysostosis; however they emphasized that it is a very rare disease (10).

Another report of eight children with this rare disease demonstrated that some patients with this syndrome have defective growth hormone (GH) secretion since GH therapy improves their linear growth(11).

Some disorders, such as osteopetrosis and cleidocranial dysostosis, can be confused with pycnodysostosis since they show certain symptoms that are clinically and radiographically close to each other. Osteopetrosis is characterized by a generalized elevated bone density. The malignant types are recessive, with extreme aplastic anemia caused by medullary canal obliteration and early death. Accordingly, a normally average height and normal texture characterize cleidocranial dysostosis (12).

Some reports demonstrated the prevalence of manifestation of pycnodysostosis including obtuse mandibular angle in 87.09%, occipital and frontal bossing in 70.96%, groove in hard palate in 48.38% and impacted teeth in 48.57% of cases. These findings were seen in our patient however, there are some other features such as open bite, midfacial hypoplasia detected in teeth. Additionally, in children, open fontanelles may be detected (13,14).

The most common type of bone fracture in Pycnodysostosis is a transverse fracture of long bone midshafts. In our patient, fractures were transverse in femur and other bones mostly. Moreover, our patient had several fractures with trivial trauma that was similar to a patient that was reported by Pangrazio et al (4). It seems that these patients are at high risk for bone fracture. Therefore particular attention to self-care for prevention from fractures is necessary.

Atypical facial features usually indicate the disorder, but acroosteolysis and osteosclerosis of the distal phalanges provide a more definitive diagnosis. Molecular genetic testing can reveal the presence of a CTSK mutation in patients with clinical manifestations of pycnodysostosis (3). Recognizing the clinical manifestations of the disease is fundamental at this point, however identifying its dysmorphic features can be difficult. In the absence

of acroosteolysis, misdiagnosis is possible. The absence of acroosteolysis can be deceiving, since osteopetrosis is frequently diagnosed erroneously in such cases, a finding that is mentioned by Panrazio et al (15).

Although pycnodysostosis is not a life-threatening situation, frequent fractures, craniosynostosis, respiratory-sleep problems, and dental problems, as well as their treatments, can cause considerable agony to patients. Treatment with growth hormone in pycnodysostosis has recently contributed to a significant improvement in final height but it is a routine in children, not in adults. Patients should be monitored for complications and treated accordingly by neurosurgery, orthodontics, respiratory medicine, sleep medicine, and rehabilitation, despite the fact that no specific treatment for the disorder has been described. Oral hygiene and regular dental care are essential for avoiding oral complications. Increased bone density can cause post-extraction osteomyelitis, therefore risk factors should be closely considered when preparing tooth extraction and other surgeries (15-17). Fractures are treated in a variety of ways, from conservative to operative, with open reduction and plate fixing to closed reduction and exterior fixation or intramedullary nails. Strong intramedullary nails are recommended. However, the insertion of these nails is accompanied by several problems (18).

Conclusion

In patients with several fractures who are referred to the hospital and have one or some maxillofacial deformities, pycnodysostosis should be one of the top differential diagnoses because this rare disease may be not considered by physicians.

Authors' contribution

SH, MZ and MHAB were the principal investigators of the study. MAS, SH and MZ were included in preparing the concept and design MZ and MHAB revisited the manuscript and critically evaluated the intellectual contents. All authors participated in preparing the final draft of the manuscript, revised the manuscript and critically evaluated the intellectual contents. All authors have read and approved the content of the manuscript and confirmed the accuracy or integrity of any part of the work.

Conflicts of interest

All authors have no conflict of interest disclosure to disclose.

Ethical issues

This case report was conducted in accord with the World Medical Association Declaration of Helsinki. Written informed consent was obtained from the patient for publication of this report. Besides, ethical issues (including plagiarism, data fabrication, double publication) have been completely observed by the authors.

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