CASE REPORT

Leri Disease – case report of uncommon disease

Choroba Leri’ego – opis przypadku

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Abstract

Introduction. Leri disease (synonyms: melorheostosis, candle bone disease, melting wax syndrome) is a rare disease which affect bones, first described in 1922 by Leri and Joanny. The disease can be asymptomatic or manifested by severe pain of the limbs. A radiological feature is the increasing density of the bone structure resembling wax flowing from a candle.

Case report. A 27-year-old Caucasian female presented with pain of her upper limb, a deformation of the limb and with a limitation of the active and passive range of motion of the joints. The x-rays showed irregular cortical sclerosis resembling wax that has dripped down the side of the candle. The analgetics were given to the patient. The patient has also underwent a surgical treatment.

Conclusions. The Leri disease is diagnosed based on the typical candle wax x-ray changes. There is no causative treatment. The treatment with bisphosphonates and physiotherapy can bring results. The surgical treatment does not help in relieving pain.

Key words: Leri disease, melorheostosis, candle wax syndrome

Streszczenie


Opis przypadku. 27-letnia pacjentka z silnymi dolegliwościami bólowymi kończyny górnej prawej, z deformacją i ograniczeniem ruchomości w stawach. Zdjęcia radiologiczne uwidoczniły zagęszczenie struktury kostnej oraz objaw kapiącej świecy. Pacjentka otrzymywała leczenie przeciwbólowe. Wykonano także zabieg resekcji nadmiaru kości.


Słowa kluczowe: choroba Leri'ego, meloreostoza, choroba kapiącej świecy
Introduction

Leri disease (synonyms: melorheostosis, candle bone disease, melting wax syndrome) is a rare disease which affect bones, first described in 1922 by Leri and Joanny [1]. It has sporadic characteristic. The changes concern both sexes. No hereditary features have been discovered. The disease can be asymptomatic or manifested by severe pain [1-3]. The changes are located in the bones of the limbs, usually one-sidedly. The most common part of the bone is diaphysis of the long bone of lower limb less often the axial skeleton [1-3]. A radiological feature is the increasing density of the bone structure resembling wax flowing from a candle [1-3].

The authors present a rare case of melorheostosis of the bones of the right upper limb.

Case report

A patient J.W., a 27-year-old Caucasian female, was admitted to the Orthopedic Department, with pain of her upper limb, a deformation of the limb and with a limitation of the active and passive range of motion of the joints, especially of the radiocarpal joint. The dorsal flexion of the joint was 50 degrees, and the volar flexion was about 60 degrees. The affected limb was thickened and slightly distorted.

The general history of the patient revealed melorheostosis. She did not give any history of an injury of her upper limb in the past. The patient has worked as a teacher.

The disease started when the patient was 15 years old with severe pain of the right upper limb without changes on the x-ray. During the diagnosing process she underwent CT scans of the upper limb, scintigraphy and genetic examinations. The diagnose was Leri disease. Two years after the first pain symptoms there were changes on x-ray. Firstly, it was the a deformation and sclerosis of the radius and the second and the third hand ray (Fig. 1). Three years later there were typical structures resembling wax flowing from the candle (Fig. 2, 3 and 4). The skin was normal. Laboratory findings for serum calcium, phosphorus, C-reactive protein and alkaline phosphatase were within normal limits.

According to the fact that it is a disease entity of an unknown etiology, there is no causative treatment. For this reason, the only treatment for the patient was analgesia. Ordinary painkillers did not help, so she was administered opioid medicines. She began to develop addiction to them, therefore the decision was made to remove the excess bones surgically.

The excess bone was surgically removed from the dorsal surface of the distal phalanx of the III finger, the second metacarpal bone and the radius bone. During the operation, an atypical location of the anatomical structures was found,
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sub periosteal bone formation. Other symptoms are the limitation of movement and deformity of the joints, and hyperpigmentation of the overlying skin or/and scleroderma. The signs and symptoms usually appear in childhood or adolescence [1-3]. Leri disease can be an independent disorder or can be a part of Buschke-Ollendorff syndrome, which is characterized by osteopoikilosis, connective tissue nevi and also can include melorheostosis [3,4].

Melorheostosis may be associated with tumors such as osteosarcoma or malignant fibrous histiocytoma [5,7]. Melorheostosis can affect one or multiple bones, or, which is the most common form, can be monomelic. The aetiology of this disease is still unknown. There are several hypotheses explaining the cause of the disease [8]. In 1979 Murray and McCreidie suggested that an embryonic nerve root infection leads to neural scarring and segmental bone sclerosis [9]. Kim J-E et al. (2000) showed that in melorheostosis in the regulation of osteoblasts, there is a lower involvement of adhesion proteins, especially transforming growth factor β induced gene product [10]. In 2004 Hellemans et al. [11] and in 2007 Mumm et al. [12] described a possible aetiology in mutation of the LEMD3 gene (12q12-12q14.3), a protein that interacts with BMP and TGF β signalling. Such hypotheses may confirm the genetic basis of this rare disease. However, further research on this matter should take place to solve the mystery of aetiology and help to determine the possible role of genetic therapy in treating this disorder.

Melorheostosis does not cause changes in standard laboratory tests or specific changes in a histopathological examination.

The treatment can be nonoperative and operative. Majority of the patients receive symptomatic treatment. The operative treatment includes the excision of hyperostotic bone, osteotomies, sympathectomy or even amputation.

There are reports of effective treatment with bisphosphonates. In Leri disease the bone pain is probably connected with osteoclastic bone resorption and the activation of pain receptors, increased intraosseous pressure and vascularity as a result of hyperostosis. Bisphosphonates inhibit osteoclastic bone resorption by a number of mechanisms on bone vascularity or cells such as osteoblasts and macrophages. Their effects on reducing bone pain and deceleration of bone lesions have been shown [6,13,14].

The Leri disease has a chronic, slow nature. However, there may be periodic exacerbations. After operative excisions of hyperostotic bone, recurrence can be expected [15].

Discussion

Leri disease is a chronic bone disease, which is very rare. It was first described by Leri in 1922 [1-3]. The frequency is 0,9/1 000 000. There are no hereditary features and both sexes can be affected. The first symptom is a pain which is connected with a sub periosteal bone formation. Other symptoms are the limitation of movement and deformity of the joints, and hyperpigmentation of the overlying skin or/and scleroderma. The signs and symptoms usually appear in childhood or adolescence [1-3]. Leri disease can be an independent disorder or can be a part of Buschke-Ollendorff syndrome, which is characterized by osteopoikilosis, connective tissue nevi and also can include melorheostosis [3,4].

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Conclusion

The Leri disease is diagnosed based on the typical candle wax x-ray changes. There is no causative treatment. The treatment with bisphosphonates and physiotherapy can bring results.
References


