INTRODUCTION – Melorheostosis is a rare but well publicised disease. About 320 cases were reported up to 1994. The incidence is estimated 0.9 cases per million.

CASE REPORT – We report a 40 year-old man diagnosed with severe melorheostosis and unusual radiographic appearances. No dripping candle wax cortical thickening was present. All the long bones of the upper extremities showed extensive periosteal and endosteal hyperostosis with extension of the changes into the hands and shoulder girdle. Osteosclerosis was present at both sides of the right sacro-iliac joint. There was also hypoplasia of the scapulae, distal ulnae, carpal bones and the left thumb. Distinctive features of the clinical history were that the disease was of prenatal origin. The lower extremities were normal and in spite of extensive bone and soft tissue changes in the upper extremities this patient never experienced pain – a prominent feature of adult melorheostosis.

CONCLUSION – Melorheostosis is a rare disorder, diagnosis of which can be made from clinical presentation and radiographs. In case of atypical melorheostosis, the lower extremities may be found to be normal on radiographs. The presentation may be painless.

Melorheostosis
– Ritka megjelenési forma

BEVEZETÉS – A melorheostosis ritka, de jól dokumentált betegség, 1994-ig 320 esetről számoltak be. Az előfordulási gyakoriság 0,9 eset/egymillió.


KÖVETKEZTETÉS – A melorheostosis ritka betegség, amely a klinikai tünetek és a röntgenvizsgálat alapján diagnosztizálható. Atípusos melorheostosis esetén az alsó végtagok normálisak lehetnek a röntgenfelvételeken, és a betegség nem jár fájdalommal.

Melorheostosis
– Uncommon appearances

Anna Kutkowska-Kazmierczak, Ewa Obersztyn, Kazimierz Kozlowski

melorheostosis, hyperostosis, hypoplasia
Melorheostosis is a rare but well publicised disease\textsuperscript{1–3}. About 320 cases were reported up to 1994. The incidence is estimated 0.9 cases per million\textsuperscript{28}.

The purpose of this paper is to present an adult man with melorheostosis and unusual clinical course, and not yet reported radiographic findings.

**Case Report**

This patient was born at 40 weeks to a 20 year-old, second gravida after an uneventful pregnancy and delivery. The unrelated parents were of Polish decent and were of good health. The father was aged 24 years. The family history was unremarkable.

Birth weight 3600 g, length 52 cm. Immediately after the birth, deformities of the hands with thickening of the skin at the extension surface of the palms were noted. In the following years not only the deformities of the hands progressed but deformities of the forearms developed. At the age of seven years he underwent tenotomy of the extensors and capsulotomy of the 3rd–5th metacarpo-phalangeal joints of the left hand. At that time the diagnosis of melorheostosis was made at the Orthopaedic Pediatric Clinic in Lublin, Poland.

The patient presented at the Department of Medical Genetics at the age of 40 years for consultation regarding his wife’s pregnancy. Later a normal son was born. At the time of examination his height was 166 cm, weight 56 kg, head circumference 56 cm. His face was normal. The chest was narrow. There was shortening of the deformed upper extremities. Right arm –1.6 SD, forearm –4.8 SD; left arm –3.5 SD, forearm –5.4 SD.

Deformities of the hands made measurements impossible. The lower extremities were normal.

His mental development was normal (Fig 1. a, b).

Routine blood and urine examinations were normal.

Radiographic examination documented extensive periosteal and endosteal hyperostosis in all the long tubular bones of the upper extremities extending into the hands and shoulder girdles. There was hypoplasia of the distal end of the ulnae, hypoplastic/dysplastic changes in the carpal bones.

![Image of the patient](image-url)
Anna Kutkowska-Kazmierczak: Melorheostosis
and hypoplastic 1-st right metacarpal and proximal phalanx. The scapulae were small (Fig 2. a–f).

Osteosclerosis was present in the right sacroiliac joint (Fig 3.).

Discussion

The clinical and radiographic features of melorheostosis in childhood differ from those of adults\(^2, 25, 26, 30\). In children soft tissue contractures with inequality in limb length are usual presenting features. Skin manifestations are described as scleroderma or scleroderma like changes and soft tissue fibrotic contractures. Pain, rare in childhood, is an almost constant feature in adults. Hyperostosis is seen in children as endosteal linear areas of increased density. Classical irregular, linear periosteal hyperostosis along the axis of the long bones resembling flowing candle was is rarely seen in the first decade of life\(^30\). Often only one side of the long bone is involved while the other appears normal. Hyperostosis continuous into the hand or foot. In the forearm and the leg one bone is more severely affected. In the rare instances when the disease progresses extensive peri- and endosteal hyperostosis occurs. Extension into the axial skeleton and soft tissue is frequently seen in more advanced cases. Campbell et al stress that in melorheostosis, contracture and deformity (especially of the hands and feet) precede the X-ray signs in infants, and that faint linear hyperostosis in childhood is difficult to diagnose\(^5\). They also say that the early contractures and abnormalities of mesodermal origin cause greater disability than the disease of bone. This case fits these criteria very well. The plain film appearances of melorheostosis are diagnostic. CT and MR are helpful in documenting the details of architecture and extension of the disorder\(^13, 14, 16\). The histology is nonspecific.

Frey Schmidt who reviewed 23 patients with melorheostosis stresses the variability of the radiographic appearances of the disease\(^8\). Most of his patients must have been adults. The mean age was 34 years and the age range 7–70 years. He recognises the classic “candle wax appearance”, osteoma-like appearance, myositis ossificans-like pattern, osteopathia striata-like pattern and a mixed pattern.

There is little doubt that the disease in our patient had a prenatal origin, as deformed hands and abnormal skin over the back of the palms were noted at birth. We do not have previous radiographic documentation of our patient. Extensive hyperostosis and endostosis involving symmetrically, uniformly the whole length of all the long bones of the upper extremities, is unusual indeed. Hypoplastic scapulae, distal ulnae and small carpal bones, have to the best of our knowledge, not been described in melorheostosis although they are illustrated but not commented upon in some reports\(^4, 12, 13, 25\). Hypoplasia of the distal ulna and first metacarpal bone are well illustrated in the papers of Butkus et al\(^4\) and Radlo and Michno\(^25\). We believe that hypoplasia of bone is a precursor of more severe form of the disease. Hypoplasia of the
carpal bones is associated with heavy spotting. The large sclerotic area present at both sides of the right sacro-iliac joint documents a rare involvement of the sacrum. The lower extremities showed no abnormality.

**DIFFERENTIAL DIAGNOSIS**

Differential diagnosis includes disorders presenting with endosteal and/or periosteal thickening such as trauma, osteoma, various types of osteosarcoma, fibrous dysplasia and myositis ossificans when soft tissue ossification is present.

Scleroderma, scleroderma-like changes, subcutaneous fibrosis, anomalous pigmentation and vascular abnormalities corresponding in location to the skeletal involvement are important clinical diagnostic clues. In any instance of increase in bone density in adults Paget’s disease should be considered. The axial skeleton is predominantly involved and subarticular areas are most severely affected. In the third, bone forming stage, new bone is laid down in a mosaic pattern. The demarcation between the cortex and medullary cavities becomes obliterated. Some osteoclastic activity – from previous phases – is also present. This Paget’s pattern of changes is quite different from that of our patient.

The etiology and pathogenesis of melorheostosis is unknown. The two major theories are the mosaicism theory and the sclerotomal theory. The first one regards melorheostosis as an early post-zygotic mutation of the mesenchyme resulting in asymmetrical involvement of the skeletal structures with concomitant vascular and hamarthromatous changes in the overlying soft tissues. Murray and McCredie proposed that melorheostosis may be the result of a segmental sensory nerve lesion, to account for its sclerotomal distribution.

They speculated that melorheostosis represents some form of post-natal peripheral neuropathy, affecting segmental spinal sensory nerves with scarring of the bone. Hyperostosis represents nerve root irritation and resultant bone overgrowth, whereas segmental skeletal aplasia and hypoplasia in dysmelia reflects repression of the trophic effect of sensori-neural tissue in the embryo. It is possible that disregulation of the corresponding myotome innervation early in life results first in skeletal hypoplasia and secondly, in melorheostosis. We believe that documentation of hypoplastic bone changes in melorheostosis indicates a more severe form of the disease.

**SUMMARY**

In summary, this patient with melorheostosis combines several rare and/or unusual clinical and radiographic features. The disease had prenatal origin as it was detected already at birth. The patient never experienced pain – a cardinal sign in adult melorheostosis. All the long bones of the upper extremities showed extensive, uniform periosteal and endosteal hyperostosis. The sacrum was involved. The lower extremities were normal. There was hypoplasia of the scapulae, distal ulnae, carpal bones and first left metacarpal.

**Acknowledgement**

We thank dr. Jane McCredie of Sydney for reviewing the paper.

**Fig 3. Radiograph of the right side of the pelvic girdle. There is a sclerotic mass at both sides of the sacro-iliac joint**
References


HÍREK

Elismerés


dr. Lombay Béla