Melorheostosis

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SUMMARY

This case study is a report on the rare mesodermal disorder typically characterized by abnormalities of the skeleton and soft tissues. We present a 56 years old woman who was diagnosed with melorheostosis affecting the left arm.

Chronic pain odema and cosmetic deformities were her presenting problems.

Melorheostosis is a bony dysplasia with characteristic X-ray appearance resembling wax dripping down one side of the candle. Soft tissue calcification and even ossification may rarely be seen. In some rare and complicated cases corrective surgery or amputation may be done in very painful and ischemic limbs (1).

Until very recently the aetiology of melorheostosis was unknown but now it has been established that melorheostosis is due to a loss-of-function mutation in LEMD 3 gene (also called MAN 1), which encodes an inner nuclear membrane protein (2).

This is the first reported case in this region. The purpose of this case report is to describe the presentation and course of the disease. A comprehensive review of literature describing etiology, clinical aspects, diagnosis and treatment is included. Patients symptoms vary considerable in melorheostosis and consequently their treatment should be individualized.

INTRODUCTION

Melorheostosis, also known as LERI'S disease or flowing periosteal hyperostosis is a rare benign disorder affecting the skeleton and adjacent soft tissue, which was first described by Leri and Joany in 1922 (2) as hyperostose en coutee "Melorheostosis" is derived from the Greek: Melos; Limb; rhein; flow; osteon; bone; referring to the radiographic appearance that resembles wax flowing down one side of the candle (3-6).

The estimated minimum incidence of the disorder is 0.9 per million persons (7,8).

The illness affects soft tissue and bone resulting in contractures, deformities and limitation of movement, joint ankylosis and limb shortening (4).

One bone (monostotic) or many bones (polystotic) may be affected (9).

HISTORY

She was a 56 year old female of Asian decent who had a long standing right upper limb pains and swelling. This started after a fall in childhood. No treatment was offered then, may be there was no fracture noted or any significant swelling to warrant medical care. The problem has been progressively increasing since then. In the last two years she has had associated body hotness on and off recurring every three to four months. The most recent episode was 2 weeks ago where she was admitted with a fever of 39 degrees centigrade and treated with antibiotics. Most of the other times, the fevers would resolve on their own, without treatment. She has however been on analgesics for pain relief. There is no history of pus discharge from any spot in the upper limb.

PHYSICAL EXAMINATION

The patient was stable and freely mobile. The vital signs were within normal, afebrile. Right upper limb:

- Generalised swelling of the whole limb extending to the right breast.
- Edematous right upper limb.
- Patchy erythematous lesions.
- Hard protrusions in the fingers.
- No areas of tenderness noted
- Patchy areas of increased warmth, especially in the erythematous

She had reduced range of motion of interphalengeal and metacarphalangeal (MCP) joints. The wrist, elbow and shoulder all had good range of motion. The neurovascular function was normal.

The left upper limb was normal. No significant findings were noted on general systemic examination.

DISCUSSION

Since the original description in 1922 (2) more than 400 cases have been reported worldwide but none is from this region. The disease affects soft tissues (10) and bone leading to deformities, bony ankylosis more in lower limbs and upper limbs and rarely in the skull, ribs, hands and spine (4). There is equal sex distribution (4).

Various names have been used for melorheostosis such as Leri-Joanny syndrome (11), osteosis eburnicans monomelica (12) and rhizomonomelor heostosis (13).

The age of the first clinical manifestation varies from the 3 years to 61 years (12), our patient presented symptomatic at age 56 years. She had progressive bone pain of varying intensity. She has been treated by non-steroidal anti-inflamatory drugs and analgesics over the years with fair relief of pain.

Other medications that have been tried are Nifedipine (14) and diphosphonates (15) for pain reduction. She has over the years progressively gotten more pains, stiffness, swelling of the whole arm with marked limitation of shoulder movements. This is the usual course of the disease which is insidious and slowly progressive with periodic exercerbations with eventual soft tissue contracture, fibrosis and even ossification (16,17).

The patient also has arm shortening as compared with the right arm and also noted tendon and marked oedema probably due to malformation of blood and lymphatic vessels or soft tissue fibromatosis (18,19). The left arm was grossly swollen and odemation from the hand to the shoulder and had hyperpigmented skin patches which has been reported in literature (20).

IMAGING

The X-ray appearance are highly distinctive with extraosseous sclerosis appearing on the outside of the diaphysis of the humeral shaft giving the classical "molten wax" appearance (Figure 1). Differential radiological diagnosis are osteochondroma, myositis ossification, calcified haematoma, fluorosis, osteomyelitis and

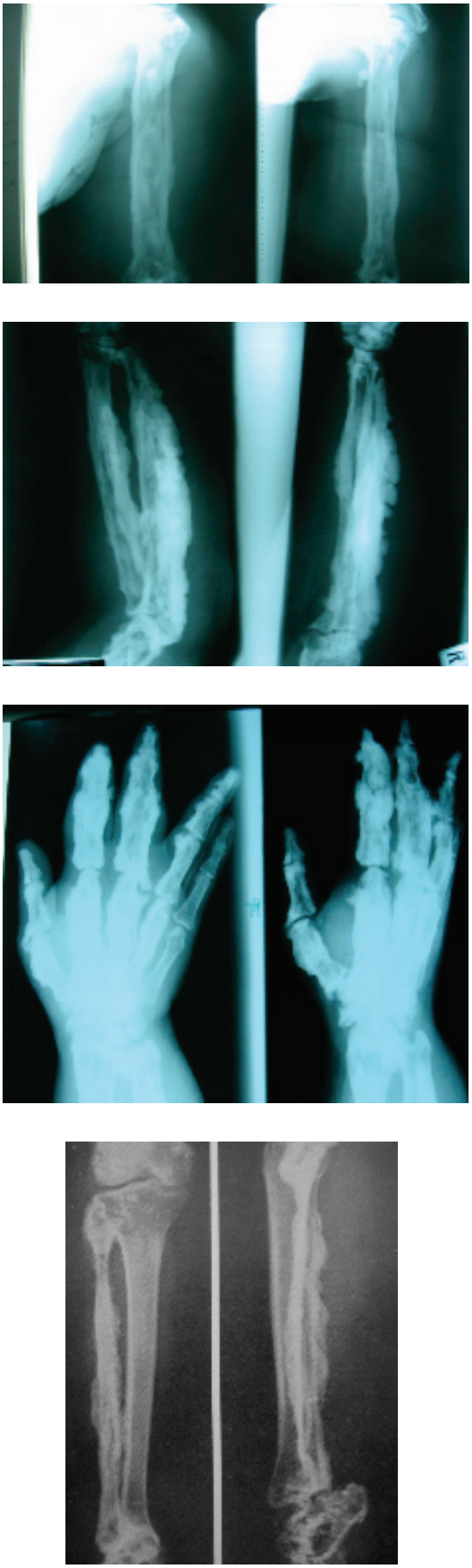

Figure 1: *Candle wax appearance in melorheostosis*

periosteal osteosarcoma (21). These were excluded in this patient clinically by lack of supportive clinical symptoms and presence of normal blood count (WBC)) and C. Reative protein (CRP) and normal erythrocyte sedimentation rate (ESR). Serum calcium, phosphorus and alkaline phosphate were normal.

The value of scintigraphy as a diagnostic tool for melorheostosis is not clear since large uptake or slight uptake in the sclerotic bone have been reported (21,22) while other reports (23) advocate its use as a confirmatory tool in equivocal cases. Computerised tomography (CT scans) will show the lesions and clear demarcation between normal and abnormal bone while magnetic resonance (MRI) shows the bone and soft tissue lesions as areas of low signal (24,25).The last two investigations were not done due to financial constraints and may not have added any new information.

Pain control in this patient has been medications only. Surgical modality such as soft tissue release, shoulder capsulotomy, sympathectomy will be considered if pains get worse and as a last resort in case of unbearable pains disarticulation at the shoulder can be considered.The patient should be followed up regularly since isolated cases of malignancy have been reported in association with melorheostosis including osteosarcoma (7).

Patients with melorheostosis may have associated cutaneous and soft tissue lesions such as vascular malformations, neurofibromatosis, haemangiomas, arterial aneurysms, linear scleroderma, tuberous sclerosis and focal subcutaneous fibrosis. Out of these one, the last entity of focal subcutaneous fibrosis was noted in this patient. However, biopsy was not done since the X-rays were definitely diagnostic of melorheostosis. Histological analysis shows dense bone without distinctive cellular abnormality but microscopic appearance of the lesions are not identical in all cases. The histology appearance is determined by the time of biopsy in relation to the course of the disease (5,8).

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