

LERI'S DISEASE

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ABSTRACT

Melorheostosis or leri's disease is a rare bone disorder which is characterized by subperiosteal sclerosis of bones. The most common bone part is diaphysis of long bones of lower limb of one side with rare involvement of axial skeleton. Some times bones of the hands and feet may involved. Radiological pictures shows flowing hyperosteosis' resembling hardened wax which has dripped down the side of the candle. although patient presented with seizure and diagnosed as tubercular meningitis but we are reporting this case because of its rarity, in a young male.

KEY WORDS: Melorheostosis, Subperiosteal sclerosis, Candle dripping, flowing wax.**INTRODUCTION**

Melorheostosis is a relatively rare chronic sclerosing bone disorder also known as Leri's disease, candle bone disease, or melting wax syndrome. The disease was first described by Leri and Joanny^[1] in 1922. The disease affects men and women equally. Most common presentation is pain & most common bone part is diaphysis of long bones of lower limb of one side with rare involvement of axial skeleton. Radiological pictures shows flowing hyperosteosis' resembling hardened wax which has dripped down the side of candle.

CASE REPORT

A 35 years male presented to us in status epilepticus in emergency department. Patient has history of fever of high grade, headache and projectile vomiting from five days and became unconceous after seizure(GTCS type) from two days. Patient have history of right leg pain (dull aching) since 20 years of age & then develop mild swelling & limitation of knee & elbow joint movements which was gradually progressed so much that patient was unable to walk from 7 to 8 months. There was no relevant family history or trauma or any other musculoskeletal disorder.

On examination blood pressure of the patient was 100/80 mmHg, pulse rate 94 per minute, regular, pallor present but icterus, cyanosis, and, clubbing were absent & pedal oedema was present.

CNS- Patient was in postictal phase with GCS-E4V4M3. Signs of meningeal irritation were present. Deep tendon reflexes were diminished and planter reflex

were bilaterally equivocal, tone in right upper & lower limb increased due to flexion deformity at both knee and elbow joint and pupils were bilaterally semidilated and normally reacting to light. On respiratory system examination, bilaterally vesicular breath sound present. Per abdomen- soft, non tender, no organomegaly present.

There was no sign of inflammation in upper & lower limb. There was hard bony swelling over right hand & wrist joint and great toe & tibia along with hyperpigmentation of skin overlying deformed right hand and right lower limb (fig-1&2).

**Figure 1 Right leg showing multiple bony swellings**

Haematological examination Hb 12.3mg/dl, TLC 12000/cumm, N₈₅L₁₄E₀₁. Ceribrospinal fluid protein 244mg/dl, glucose 23mg/dl, N₀₅L₉₅, RBC 03. blood sugar 114mg/ dl, serum creatinine 1.2 mg/dl, blood urea 45mg/dl, Na 141 meq/l, K 4.5 meq/l, ca 9.9 meq/l, AST 45.0 U/L, ALT 55 U/L, urine 2-4 pus cells no albumin and sugar. Body mass index of the patient was 15.2

kg/mm². HIV ELISA, Hbs Ag, anti HCV were non reactive. On the basis of above investigation diagnosis of tubercular meningitis was made and treatment started accordingly along with antiepileptics.



Figure 2 Right hand showing multiple bony swelling

X-ray of right leg (Fig-3) showed dense irregular cortical hyperostosis, which looks like candle wax, extending along tibia of right leg resulting in deformity of bone. X-ray of right hand showed irregular cortical thickening of metacarpals and phalanges. X-ray picture of left upper and lower limb appear normal.



Figure 3 X ray both legs showing subperiosteal bony growths

The orthopedic and radiology experts were consulted and diagnosis of Leri disease with tubercular meningitis was made. Antitubercular treatment was given along with biphosphonate. Some operative treatment was advised by orthopedicians but patient refuses to take treatment.

DISCUSSION

Melorheostosis is a rare chronic bone disorder which was first described in 1922 by Leri and Joanny^[1]. Male and female are equally affected, and no hereditary features have been discovered. The onset of this rare diseases is insidious, and the first symptom is usually dull aching pain due to subperiosteal bone formation. Skin become rough, hard and in 17% of cases that may include hyperpigmentation. Melorheostosis mainly affects, the long bones of the upper and lower limb, and also short bones of hand and foot, but rarely the axial skeleton.^[2,3] Melorheostosis may present in a monostotic, polyostotic, or monomelic form. The monomelic form is most common.^[4]

The most accepted hypothesis was given by Murray and McCredie 1979.^[5] was that, embryonic infection of nerve root causes neural scarring and segmental bone sclerosis responsible for melorheostosis. One possible etiology of melorheostosis is a loss of function mutation in the LEMD3 gene (12q12–12q14.3), a protein involved in bone morphogenetic protein and tumor growth factor- β signaling.^[6]

It is associated with vascular malformations, soft tissue masses adjacent to the affected bone and scleroderma of the overlying skin.^[7] Routine laboratory findings usually are normal. Histological findings are usually nonspecific and often show dense bone formation, a mixture of mature and immature bone elements.^[7] Osteoclastic activity is not a prominent feature; however, osteoblastic activity along the margins of osteons is common.^[8] Treatment is mainly symptomatic. Most patient receives nonoperative treatment. Operative treatment consists of tendon lengthening, excision of hyperostotic bone, osteotomies, sympathectomy and amputation^[3]. Bisphosphonate are commonly use.^[3] Potential causes of the bone pain in melorheostosis include increased osteoclastic bone resorption and activation of pain receptors, raised intraosseous pressure and increased vascularity secondary to hyperostosis and soft tissue involvement around joints. Thus, bisphosphonate treatment via a number of mechanisms would be expected to reduce inflammatory bone pain and symptoms in melorheostosis. Bisphosphonates inhibit osteoclast mediated bone resorption by direct and indirect actions on osteoblasts and macrophages and bone vascularity. They have been shown to decrease bone pain, slow progression of bone lesion.^[9] The prognosis of a patient with melorheostosis is variable and depends on the anatomical location, extension into the soft tissues, and soft tissue changes. Melorheostosis does not shorten life span, however, morbidity may be considerable. The disease exhibits a slow, chronic course, with periods of exacerbation and arrest. Recurrence usually is expected after operative excision.^[10]

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