Abstract:
Melorheostosis is a rare sclerosing skeletal dysplasia affecting bones of the appendicular skeleton. Radiography shows the characteristic dribbling candle wax appearance and streaky endosteal bone formation. Soft tissue calcification and even ossification may rarely be seen. Radiological appearance forms the basis of the diagnosis, as clinical and histological findings are non-specific. This is the first reported case of melorheostosis in this part of the country among the literature known to us. We present a case report of a 26 year old woman, who was diagnosed with melorheostosis affecting the left hand. The purpose of this case report is to describe the presentation of the disease and the principles of treatment, because the patients symptoms vary considerably in melorheostosis and consequently their treatment should be individualized.

Keyword: melorheostosis, dysplasia, dribbling candle wax

INTRODUCTION:
Melorheostosis, also known as LERI’s disease or flowing periosteal hyperostosis is a rare dysplastic disorder affecting the bones of the appendicular skeleton and adjacent soft tissue. It was first described by Leri and Joany in 1922. The word 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. The estimated incidence of the disorder is 0.9 per million persons. The disease affects bone and soft tissue resulting in contractures, deformities, ankylosis of the joint and limitation of movement. One bone (monostotic) or many bones (polyostotic) may be affected.

CASE REPORT:
A 26 year old female presented with a history of pain and swelling in her middle finger of the left hand for the past three years. She had persistent dull aching pain which was relieved temporarily by taking medications and she got accustomed to her ailment for the past three years.
She does not have similar complaints in any of the other regions of the body. On physical examination, she had a diffuse swelling over the proximal and middle phalangeal region of the middle finger of left hand, which was more prominent on the ulnar aspect. The skin over the swelling was stretched and shiny. The phalanges of the middle finger was not warm and found to be tender and thickened on palpation. The range of movements of the third metacarpophalangeal joint and the interphalangeal joints of the middle finger were painful and restricted. The wrist, elbow and shoulder joints all had good range of motion. No significant findings were noted on general systemic examination.

**Fig: clinical picture**

Laboratory studies including complete blood hemogram, urine analysis, serum calcium, phosphorus, alkaline phosphatase and serum proteins were all within normal limits. X-ray of the left hand showed diffuse sclerosis of all the three phalanges of the middle finger and the radial aspect of the third metacarpal bone. Both the cortex and medulla were found to be involved and the characteristic flowing candle wax appearance was also seen in this patient. A skeletal survey of the whole body of the patient was done to find similar lesions in other bones, but was found to be normal.

**Fig : x ray left hand Anteroposterior and Oblique views**

The CT scan of the left hand and wrist was taken which showed findings identical to those seen on the radiographs. In addition, the CT scan also revealed the involvement of the capitate bone, just proximal to the affected third metacarpal. Hence this patient had involvement of the entire ray of the middle finger of left hand.
An excision biopsy was done, which was taken from the ulnar aspect of the proximal phalanx of the middle finger. Histological examination of the biopsy specimen showed fragments of dense bone with fibrotic marrow, which is consistent with the diagnosis of melorheostosis. No evidence of active inflammation or neoplasm was noted.

Surgical management of this patient may need entire excision of the middle finger ray of the left hand, which will be a debilitating procedure and may cause significant physical handicapping and psychological trauma to the patient. Hence the patient was planned to be treated conservatively. She was started with bisphosphonates (etidronate 1200 mg/day) and non steroidal anti inflammatory drugs. Subsequently she was encouraged to do active mobilization exercises of the fingers of the left hand. The patient responded very well to the conservative management. She experienced a significant pain relief and her range of motion of the fingers of the left hand improved to a great extent. The patient will be followed up regularly, since isolated cases of malignancy have been reported in association with melorheostasis including osteosarcoma. (7)

DISCUSSION:
Melorheostosis is a rare developmental sclerosing mesenchymal disease of bone mineral density due to a loss of function mutation in LEMD3 gene. (1) LEMD3 (also known as MAN1) encodes for an integral protein of inner nuclear membrane. The developmental error is at the site of both intramembranous and endochondral bone formation, predominantly the former. There is a definite tendency for monomelic distribution i.e., involvement of one limb, but single bone or multiple bones may be affected.
Melorheostosis was described by Leri and Joanny as: hyperostose en coulee”, i.e., flowing hyperostosis resembling dripping candle wax. The age of first clinical presentation varies widely from 3 years to 60 years and there is no sex predilection. (11)
The course of the disease is insidious, with a slow, chronic progression of symptoms and periodic exacerbation. Soft tissue involvement is evidenced by restricted motion and stiffness. Serum calcium, phosphorus and alkaline phosphatase levels are normal. Cortical hyperostosis is readily seen on x rays, extending along the length of one side of bone resembling “flowing candle wax”. Soft tissue calcification or ossification may be seen.

Scintigraphy reveals abnormal increased tracer uptake in the bone and soft tissue lesions. It may be instrumental in confirming the diagnosis in equivocal cases. CT and MRI are not needed in vast majority of case, especially in children.

In most of the cases of melorheostosis, biopsy is not needed for diagnosis. If performed, it is not diagnostic. Proper diagnosis is almost always evident on plain radiographs.

Treatment of melorheostosis depends upon the exact symptoms and findings exhibited by a given patient and the problem varies depending upon the type and the number of bones involved. Most of the patients respond well to the conservative management with drugs like bisphosphonates, nifedipine and non steroidal anti-inflammatory drugs. Surgical excision or resection may be done to increase range of motion in joints of patients with bone enlargement or soft tissue calcification. Other reported treatments include: osteotomies and bone lengthening for patients with limb shortening or angular deformities, excision of hyperostosis and fibrous tissues, fasciotomies, capsulotomies, arthrodesis, sympathectomies and in extremely rare cases, amputation is indicated in very painful limbs with contractures and ischaemia. (3)

Surgeries for the sole purpose of relieving pain (non-mechanical) is rarely effective, also contracture releases are more effective in adults than in children. Soft tissue releases alone in skeletally immature patients have a 100% "failure" rate in the literature. This statistics does not necessarily mean that they should not be done, but families need to be advised that the procedure may need to be repeated in the future.

CONCLUSION:
Neither con-servative nor surgical methods of treatment appear to be very effective in the management of melorheostosis. Hence, treatment of melorheostosis is suggested to be individualized based on the patient’s lifestyle, pain level, progression of the disease, and age. The authors suggest that the diagnosis of melorheostosis should be kept in mind in cases of hyperostosis of bones.

REFERENCES:


