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MELORHEOSTOSIS

Report on 5 Cases with Follow-up

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Melorheostosis was described for the first time in 1922 by Léri & Joanny, and the first case in Denmark was reported by Bertelsen (1940).

The condition is presumably more common than indicated by the approx. 150 cases on record (Campbell 1968), but nevertheless rare enough for a report of 5 cases with follow-up to be of interest.

It is a peculiar and interesting fact that the disease has existed for centuries. Lester (1967) found radiological changes characteristic of melorheostosis in an Eskimo skeleton from Alaska dating from about 500 A. D.

The most characteristic change is hyperostosis affecting one or more bones. In typical cases the radiological changes are easy to recognize. They have been likened to melting wax dripping down the sides of a candle. The bony thickening may extend into the epiphyseal area of the bone as streaks, but the joints are seldom involved (Figure 1).

Biopsy specimens have been taken in about 20 cases (Morris et al. 1963), and the histological findings have been extremely uniform. In the sclerosed bone the Haversian system is of irregular arrangement with thickened, densely anastomosing trabeculae. As a rule there is cellular, fibrous tissue in the medullary canal and around proliferating bone. Fibrosis of subcutaneous tissue, perivascular obliteration of vessels, and atrophy of the skeletal muscles have also been observed by Morris et al. (1963).

SYMPTOMS, SIGNS AND COURSE

The most common symptoms and signs of melorheostosis are pain, paraesthesiae and sensory disturbances over the involved bones. The

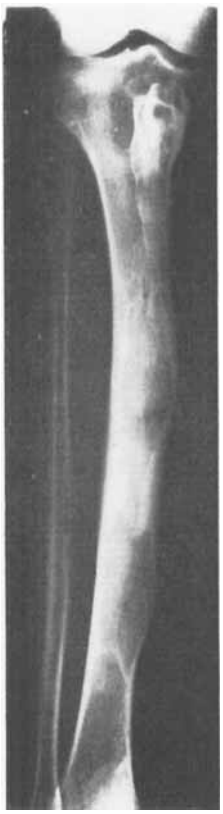

Figure 1. Case 3: Melorheostosis of the right tibia.

pain is of skeletal nature. It may be quite mild, but it is what usually makes the patient seek medical attention.

Bony deformity is sometimes present. Not infrequently the patients have noticed thickening or increased length of the affected bones right from childhood (Campbell et al. 1968). The limbs are most often affected, as a rule unilaterally, but any bone may be involved. Fairly mild muscular atrophy is common. Cutaneous changes are rare. Lymphoedema, haemangioma, and pigmentation of the skin have been described in a few cases.

The disease has been diagnosed at all ages, but mostly in the young age groups. In a few cases it has even been present at birth. Its aetiology and pathogenesis are unknown, but the findings made so far are most suggestive of a congenital origin, probably around the 4th foetal week (Campbell et al. 1968).

Among diseases of bone which may give rise to differential diag-

nostic problems, previous authors have emphasized Albers-Schonberg's osteopetrosis, osteopoikilosis, carcinomatous metastases with an osteoplastic tendency, chronic osteomyelitis, Paget's disease, Recklinghausen's disease, and syphilis.

Melorheostosis must be considered definitely benign. At any stage the changes may become stationary. If progression occurs, it is very slow. There have been no systematic studies to elucidate the long-term prognosis. Treatment is symptomatic.

CASE REPORTS

1. T. A. (Case rec. 2931/63). A female, aged 29. The only symptom was pain in the left ankle. Physical examination in 1963 disclosed no abnormality. X-rays revealed melorheostotic changes in the left tibia at the distal metaphysis, on the fibular and dorsal aspect of the bone.

At *follow-up* 5 years later the condition had improved. Now the patient rarely had pain in the left ankle. There was still no objective abnormality, and X-ray examination showed the changes to have remained unchanged since 1963. Routine radiography of the right foot and ankle region showed typical changes similar to those on the left. There have *not* been any symptoms or signs from the region of the right ankle.

2. L. T. J. (Case rec. 8143/64). A male, aged 16, who had previously been in good health. The patient complained of pain in the lower part of the right thigh, especially when bending the knee. No objective changes of the limbs. X-ray examination of the femora showed melorheostosis of both shafts medially and dorsally.

At *follow-up* 4 years later the condition had considerably improved. There was only intermittent pain in the lower part of the right thigh. No treatment had been required. X-ray examination showed melorheostosis of *both femora* medially and dorsally, of the same appearance as previously.

3. L. S. H. (Case rec. 7389/60). A male, aged 64, previously in good health. The patient presented himself for the first time 8 years ago because of severe pain on the posterior aspect of the right lower limb, mild paraesthesiae, but no sensory disturbances or pareses. There was slight limitation of movement in the right hip, especially on internal rotation and abduction. No symptoms from the right knee. X-ray examination revealed severe changes of the right femur, involving the entire shaft centrally, medially, and dorsally, but most marked in the lower part. There were also severe changes of the right tibia, involving the entire shaft, from the proximal condyle to the distal part. No changes of the left lower limb. The patient received symptomatic treatment wih a favourable effect.

At *follow-up* 8 years later the patient had been symptom-free during the past 7 years. Physical examination did not show any abnormality, and on X-ray examination the appearances were unchanged.

4. E. N. J. (Case rec. 5801/66). A female, aged 46. During the past 10 years she had had periodical pain in the region of the left shoulder, left arm, and fingers. Always free mobility of the joints of the left arm. No paraesthesiae or pareses.

Radiography revealed melorheostosis at the transition between the metaphysis and shaft proximally in the left humerus, on the ulnar and dorsal aspect. The patient received symptomatic treatment, and the pain yielded.

At *follow-up* 2 years later the patient was still symptom free, and X-rays showed unchanged appearances.

5. U. I. S. (Case rec. 5930/56). A female, aged 43, previously in good health except for the present disease. For about 5 years she had been having increasing pain in the right knee. No complaints from the left knee. X-ray examination revealed melorheostosis distally, medially, and dorsally on the femoral shaft on *both* sides, but more marked on the right.

At *follow-up* 17 years later the condition had appreciably improved. There was still pain in the right knee, but not as severe as previously. The patient had not received any treatment in the meantime. Physical examination did not reveal any changes of the right lower limb. X-rays of the left and right femora showed the appearances to be unchanged.

At the time of follow-up all 5 patients were subjected, *inter alia*, to the following laboratory tests: ESR, Hb, serum creatinine, alkaline phosphatases, serum calcium, and serum phosphorus. The results were normal in all cases.

DISCUSSION

The material comprises 5 patients whose disease was diagnosed during the period 1935–1967. Two were males and three females, and at the time of the diagnosis they ranged in age from 16 to 64. The age at onset of the disease cannot be stated, as the symptoms need not appear until long after the disease process has started. The follow-up examinations were performed in 1968.

In our cases the diagnosis was based upon the radiographic findings, but it is usually vague, uncharacteristic pain at the site of the affected bones which makes the patient consult a doctor. However, the bone pain may be absent. For instance, 3 of our patients (Cases 1, 2, and 5) had symmetrical melorheostotic changes in the contralateral limb, but only symptoms from one.

The symptoms have subsided during the follow-up period, from 1–10 years, in all the patients.

We have not observed radiological progression of the osseous changes.

There is a tendency for the osseous changes to be localized at the site of the metaphysis and dorsally in the long bones.

The soft-tissue changes described in the literature were not present in any of our patients, presumably because all the cases were mild. On

the other hand, Campbell et al. (1968), among their 14 cases, had 5 fairly severe ones—showing massive osseous changes with secondary articular contractures and deformities.

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SUMMARY

The authors report 5 cases of melorheostosis with follow-up. Unlike most previously reported cases, the present ones improved. There was no instance of contractures or deformities. The diagnosis was based upon the radiographic findings in all cases. The findings are briefly discussed in relation to those reported by previous authors.

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