MELORHEOSTOSIS ASSOCIATED WITH ARTERIOVENOUS ANEURYSM OF THE LEFT ARM AND TRUNK

Report of a Case with Long Follow-up

J. H. PATRICK, WETHERBY, ENGLAND

Formerly House Surgeon, Professorial Surgical Unit, St Thomas's Hospital, London

Melorheostosis is a rare cause of pain and stiffness in a limb and has an unknown etiology. It can present at any age and the diagnosis is made by radiography. The appearances are of “candle greasing” down one side of one or several of the bones of one half of the body. The prognosis is uncertain, and treatment symptomatic. Melorheostosis was first described by Léri and Joanny in 1922. Morris, Samilson and Corley (1963) described a specimen as consisting histologically of “sclerotic bone, formed basically by Haversian systems irregularly arranged with dense thick anastomosing trabeculae. In some areas immature and adult bone are interlaced with conspicuous osteoid seams. Cellular fibrous tissue within the marrow spaces and about the proliferative bone is usually present. Sometimes islands of cartilage with evidence of enchondral bone formation are seen in addition to the intramembranous bone formation within the cellular fibrous tissue.” In a survey of the world literature these authors found 131 case reports (sixty-three male and sixty-eight female), in seven of which were associated vascular abnormalities.

The patient now to be described—previously reported to the Royal Society of Medicine in 1951 by Murray—came to St Thomas’s Hospital in 1966 because of new symptoms arising from the arteriovenous aneurysms.

CASE REPORT

The patient was a fit fifty-nine-year-old clerk. In December 1966 he was admitted complaining of severe pain in the left amputated arm stump and of a watery discharge from it for two weeks. He had also noticed vibration in the left side of his neck and increased swelling of the amputation stump in the last ten years (Figs. 1 and 2).

![Figure 1](image1.png)  
![Figure 2](image2.png)

**FIG. 1**—The left arm stump, with congenital arteriovenous aneurysms. **FIG. 2**—The left scapular region, showing skin malformation from multiple haemangiomas.
He had been told that as an infant he crawled awkwardly and he remembered an abnormal left arm from his earliest days. It was always larger than the right arm, and painful. There were "strawberry" angiomata and dilated veins over it and he remembered its being splinted to relieve pain. When eight years of age (in 1915) he had had the left subclavian artery ligated above the clavicle, and at the age of twelve (1919) he underwent supracondylar amputation of the left arm because of pain and excessive size of the arm. The stump healed well but was always large. Soon after operation he noticed a pulsating lump in the neck. There was no history at this time of leg abnormality. Careful enquiries about congenital abnormalities in the immediate family were negative.

The patient remained well until 1930 when he presented with urinary tract symptoms and was found to have tuberculosis of the left kidney. This was treated by nephrectomy (Sir Max Page). The amputation stump was noted to be large but symptomless. In subsequent years he remembers swelling and stiffness of the left ankle and local skin sloughing over the lower leg. Local treatment sufficed and it was not until 1950 that he began to complain of pain in the left ankle and leg on exercise. Radiographs then showed the typical appearance of melorheostosis in the left leg and foot, the left sacro-iliac region, the fourth cervical and ninth thoracic vertebrae, the left eighth and ninth ribs, and the superior and inferior angles of the left scapula (Murray 1951). The arteriovenous aneurysms of the left arm stump and trunk were symptomless.

For the next sixteen years there was a gradual increase of stiffness of the left leg, and four months before his admission he began to notice dyspnoea on exertion.

On examination the amputation stump was swollen, hot and cystic, and its lower end was weeping serous fluid. There was a pulsatile mass in the left supraclavicular region with a palpable thrill over it. The circumference of the left upper arm was 42 centimetres and of the right 22 centimetres. Over the left scapula and chest wall there were multiple haemangiomas which appeared to fill from the neck but probably communicated with the intercostal and scapular vessels. The left leg was discoloured and the skin ulcerated, the tibia was thickened and bowed and the ankle joint was ankylosed (Fig. 3). He was not in cardiac failure and his other systems were normal. Radiographs of the left leg showed changes typical of melorheostosis (Fig. 4). An arch aortograph showed dilation of the stump of the left subclavian artery at its origin, with many collateral vessels around the site of the previous ligation and malformed vessels in the stump (Fig. 5).

Treatment and progress—It was thought that amputation of the arm stump would be a formidable procedure; so preliminary ligation of the left subclavian artery at the aortic root was undertaken through an anterior third rib thoracotomy. The haemangioma then collapsed.
Pulsation in the stump was noted the same evening and became worse in the next three weeks although the pain and serous discharge did not recur.

Three and a half weeks after the subclavian ligation left forequarter amputation was performed. The multiple haemangiomata on the back were found to be very large and fed by posterior and lateral branches of the intercostal vessels (Fig. 6). Not all of the abnormal skin on the back could be removed but recovery this time was excellent.

**DISCUSSION**

It is generally agreed that melorheostosis usually affects only one side of the body, as in this case. Only one case has been reported (Elkeles 1954) in which there was bilateral involvement of the long bones and pelvis. It is interesting that in the case reported here all the abnormalities—bony and vascular—were on the left side. The pain in the left arm during childhood is suggestive of involvement of the bones of the forearm, for pain is usually the only presenting symptom of melorheostosis (Léri and Joanny 1922; Boggon 1938; McGuinness, Watson, Lindsell and Inglis 1953; Morton 1957; Hall 1961).

Both McGuinness et al. and Morris et al. explored the possible causes of melorheostosis, but the varied number of associated conditions reported, and their own histological and biochemical investigations, allowed no definite conclusions. It is tempting to postulate that unilateral bony and vascular abnormalities are both due to some congenital defect. In ten of the 131 recorded cases the diagnosis of melorheostosis was made very soon after birth, when deformity or stiffness of joints prompted radiographic investigations (Morris et al. 1963). However, the disease is usually noted in early adult life when stiffness, deformity or other abnormality such as lymphoedema (Franklin and Matheson 1942), neurofibromatosis (McCarroll 1950) or scleroderma (Dillehunt and Chuinard 1936; Thompson, Allen, Andrews and Gillwald 1951) is being investigated. Review of the radiographs of the limbs affected in the case reported here shows little change with the passage of sixteen years. This suggests that melorheostosis is a benign condition in that its progression is slow and nearly symptomless. In no case reported has it been incriminated in the cause of death, and its natural history seems only to be one of extreme chronicity.
SUMMARY

A long-term follow-up of a patient with melorheostosis and arteriovenous aneurysms confined to the left side of the body is reported. The patient was earlier investigated and shown at the Royal Society of Medicine by Murray in 1951.

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REFERENCES


