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Vascular malformations of the lower limb with osseous involvement

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Vascular malformations are rare congenital lesions which often have associated skeletal changes. Over a period of ten years, 90 patients at our clinic had a vascular anomaly of the lower limb, examined by either CT or MRI. Of these, 18 (20%) had bony involvement. A questionnaire was sent to these patients (8 men, 10 women) to evaluate their age of presentation, initial symptoms and current complaints. Radiological imaging revealed 15 low- and three high-flow lesions. The mean age at presentation to a physician was six years of age. Pain was the most common complaint. Disparity in leg length of 2 cm or more was observed in ten patients. Of the 16 patients with muscle infiltration, 13 had four or more muscles involved. Treatment by resection alone would require radical surgery.

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Although most vascular malformations are treated in specialised centres, it is important for surgeons in general hospitals to be able to describe them correctly. Descriptions are often confusing with terms such as venous angioma, cavernous angioma or haemangioma being used.[1] According to the International Society for the Study of Vascular Anomalies, vascular anomalies are classified into either vascular tumours (mostly haemangiomas) or vascular malformations.[1-3]

Haemangiomas are common and distinguished by endothelial proliferation, characterised by a phase of rapid postnatal growth followed by slow involution. These lesions are rarely treated surgically unless for recalcitrant ulceration or bleeding or if they cause a functional deficit, such as dyspnoea, or obstruction of the upper eyelid.[2,3] Involution is nearly always complete by ten years of age.[1]

Vascular malformations have a different origin. They are rare congenital lesions caused by a defect during vascular embryogenesis. By definition they are always present at birth, but sometimes only become clinically evident later in life.[3] They can be divided into either high- or low-flow lesions. Any lesion with an arterial component is a high-flow lesion. Vascular malformations can be anatomically divided into either capillary, venous, lymphatic or arterial, or combinations of the above. Several syndromes have been described of which the Klippel-Trenaunay syndrome (KTS) and Parkes-Weber syndrome (PWS) are well known.[1] KTS is characterised by capillary malformations and venous anomalies with bony and soft-tissue hypertrophy of one or more limbs.[1] Often there is an associated lymphatic malformation. PWS has the same characteristics except that there are arteriovenous malformations (high-flow lesions). Vascular malformations usually grow proportionally with the child, but sudden progression can be seen secondary to trauma, thrombosis, sepsis, hormonal changes or surgical intervention.[3] The clinical manifestations of these lesions can vary considerably, ranging from small inconspicuous capillary malformations (port-wine stains), to large arteriovenous malformations causing overflow congestive heart failure. Most present at an early age. Late presentation is a feature of an arteriovenous malformation. Venous malformations (Fig. 1) are often visible early in life either as a small blue patch or a soft blue mass.[3]

Skeletal changes are commonly associated with vascular malformations while they are rarely seen in conjunction with haemangiomas.[4] There are only a few studies which have investigated the clinical symptoms and signs of vascular malformations with associated osseous involvement.[3-8] We have therefore reviewed the symptoms and the incidence of osseous involvement in patients with vascular malformations of the lower limb presenting at our clinic during the past ten years.

Patients and Methods

Between January 1990 and December 1999, 149 patients (65 men and 84 women) with vascular malformations of the

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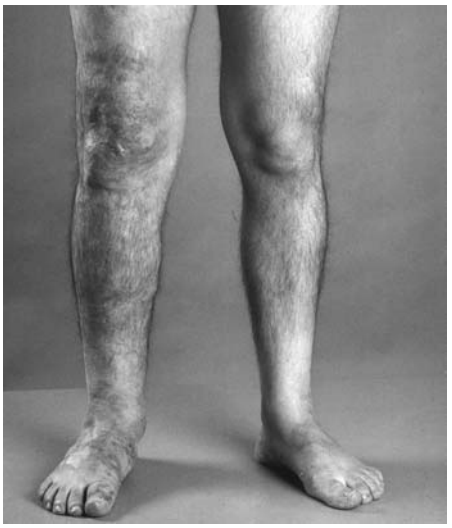

Fig. 1a

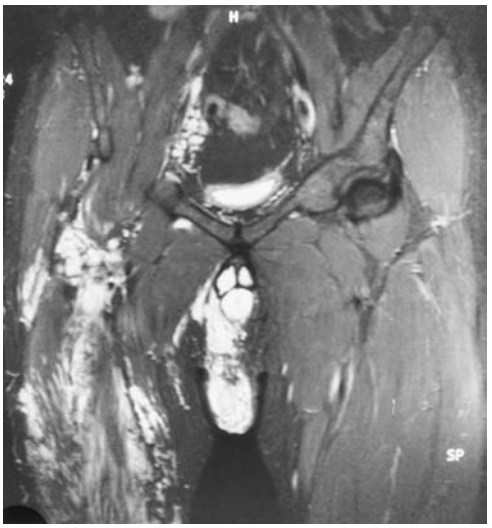

Fig. 1b

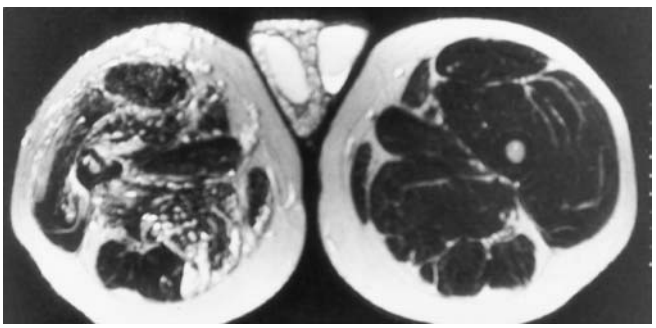

Fig. 1c

Figure 1a – Photograph showing diffuse involvement of a venous malformation in the entire right leg. Figure 1b – The MR coronal STIR image shows the vascular malformation as areas of high signal intensity seen in the right upper leg, extending towards the knee, in the scrotum and in the pelvis. There is asymmetry of vessels next to the bladder. Figure 1c – The MR axial T2-weighted image shows infiltration into subcutaneous fat and all muscles of the right leg. The femoral bone is deformed because of compression and infiltration of vessels. There is also infiltration of the scrotum.

Table I. Initial and current complaints of patients with a vascular malformation of the lower limb and bony involvement

Complaints	Number of patients
Initial	
a. Pain	6
b. Cosmetic reasons	3
c. Pain and cosmetic reasons	4
d. Leg length discrepancy	6
e. Bleeding/fluid discharge from skin	2
f. Other (non-healing of fracture)	1
Current	
a. Pain	9
b. Cosmetic reasons	8
c. Tiredness in the legs	7
d. Bleeding/ulcer	2
e. None	2
f. Other	1

lower limb were referred to our special vascular anomalies clinic. For diagnosis and/or treatment, 90 patients had the anomaly evaluated by either CT or MRI. Flow characteristics and muscle and bony involvement were retrospectively assessed in a similar way as reported previously.[9] Eighteen had bony involvement. A questionnaire was sent to these in order to evaluate their presenting symptoms. We wished to determine at what age they became aware of the vascular malformation, when a medical opinion was sought and what their initial symptoms were. The patient could choose from different categories as described in Table I. More than one answer was possible. The radiological involvement of the 18 patients was reviewed by a radiologist (MM) who had no clinical information.

Results

There were eight men and ten women. Imaging revealed 15 low- and three high-flow lesions. The different groups are summarised in Table II.

The mean age of patients at the time of the questionnaire was 32 years (10 to 61). Three patients failed to return their questionnaire, one of whom had died. It was possible to

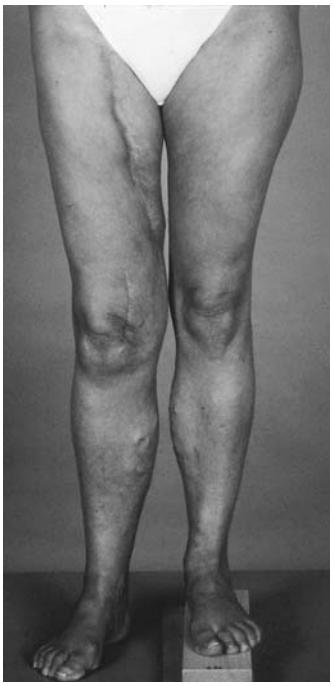

Fig. 2

Photograph showing hypertrophy of the right leg.

Table II. The different vascular groups according to gender in the 18 patients with vascular malformations of the leg with bony involvement

	Number of patients
Low-flow lesions	
Total	15
Total venous malformations	11
F	6
M	5
Total KTS	4
F	2
M	2
High-flow lesions	
Total	3
Total PWS	2
F	1
M	1
Total arteriovenous malformations	1
F	1

Table III. Leg-length discrepancy in ten patients with vascular malformations and bony involvement

	Hypertrophy	Hypotrophy
Total number of patients	8	2
Type of vascular malformation		
KTS	4	
PWS	2	
Arteriovenous	1	
Venous	1	
Amount of disparity (cm)		
>5	2	
>2 to <5	3	2
2	3	
Mean age (years) at which disparity became visible to family	3.5	2

retrieve all the necessary information from the patient's medical file. The mean age at which the family became aware of the vascular anomaly was one year. Patients with high-flow lesions became aware of their lesion at a mean age of six years, while the low-flow lesions were evident from a mean age of one year. The age at which medical help was sought was six years (0 to 36). The mean age of presentation at a medical clinic was the same for the low- and high-flow lesions. Due to pain in her leg one patient presented at her general practitioner at the age of three years. No diagnosis was made and she presented again at the age of 17 years with a femoral fracture after minor trauma. Cutaneous capillary lesions and subcutaneous varicose veins had been visible for many years.

Ten patients had a leg-length discrepancy; eight had hypertrophy (Figs 2 and 3) of the affected leg while two patients had hypotrophy (Table III). Three of the eight patients with hypertrophy presented initially with pain and not because of the hypertrophy. These three included one with a high-flow anomaly, one with KTS and one with venous malformation. The two patients with hypotrophy both had venous malformations. The hypotrophy was 2.5 and 4 cm and became visible at a mean age of two years (Table III).

The different areas of bony involvement are shown in Figure 4. Six had intraosseous extension of the malformation while 12 had reactive bony changes. The latter included cortical thickening or depression. Intraosseous extensions were seen in all three high-flow lesions (Fig. 5), two venous malformations and one case of KTS. Seven patients had intra-articular extension diagnosed by MRI. Two had involvement of the calcaneus (one high- and one low-flow lesion) and had no muscle involvement. Of the 16 patients with muscle infiltration, 13 had four or more associated muscles involved. One patient had only one involved muscle while two had three involved muscles.

Discussion

In our study of patients with vascular malformations of the lower limb, 20% had bony involvement. We are aware that this is a select group since all had lesions for which MRI was indicated. This is not a true reflection of vascular malformations of the lower limb in general. The size of the group is too small for statistical analysis. We have shown that patients with vascular malformations and associated bony involvement often present at a late stage. All, however, had cutaneous lesions. Pain is the most common presenting symptom. Leg-length discrepancy of 2 cm or more was detected in more than 50% of the patients. Patients with bony involvement often have associated multiple muscular

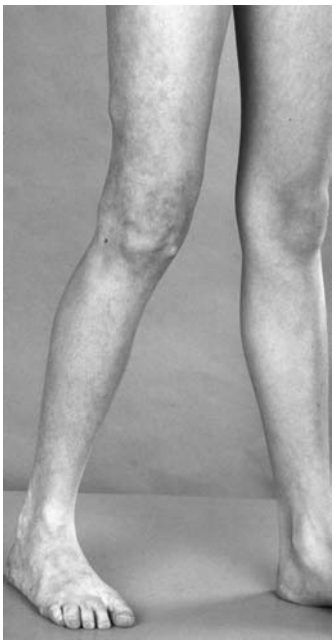

Fig. 3a

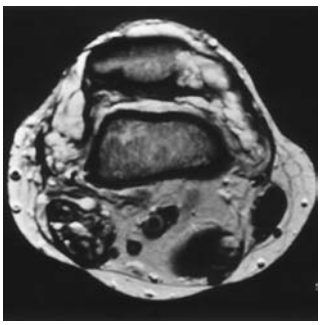

Fig. 3b

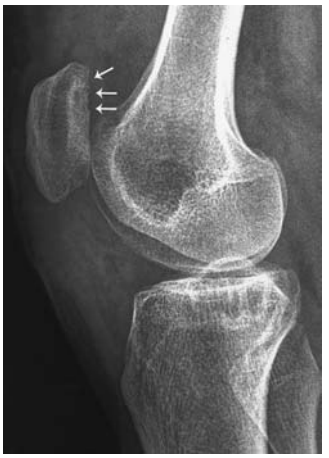

Fig. 3c

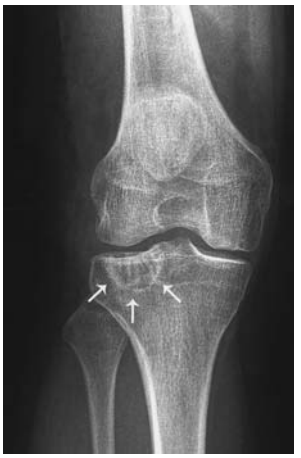

Fig. 3d

Figure 3a – Photograph showing lateral deviation of the right lower leg due to hypertrophy. There is vascular malformation from the thigh to the foot. Figure 3b – The MR axial T2-weighted sequence at the level of the knee showing vascular malformation seen as dilated vessels infiltrating subcutaneous fat, muscles, knee, patella and patellar retinaculum. Figures 3c and 3d – Radiographs showing c) the soft-tissue involvement and destruction on the underside of the patella and d) involvement of the proximal tibia.

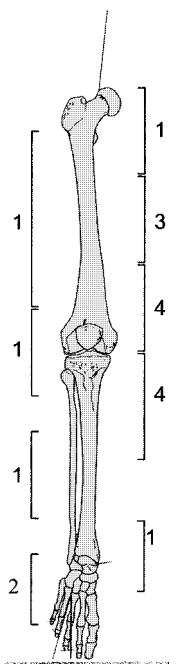

Fig. 4

Diagram showing the number of patients with the areas of bony involvement.

involvement. For surgery to be curative a severely mutilating procedure would have to be undertaken.

The mean age at which these lesions were noticed was one year, but the patients presented later for medical attention. If there is no cutaneous lesion, the asymptomatic intraosseous lesion is often discovered by coincidence when imaging studies are undertaken for other reasons.[7]

Pain is mainly related to episodes of thrombosis or haematoma, while chronic venous hypertension and muscle involvement causes tiredness of the leg. Of the six patients with intraosseous lesions, none initially complained of pain, while ten of the 12 with reactive bony changes did. At review, three of the six patients with intraosseous lesions complained of pain. Of our 18 patients, 14 specifically complained of intermittent episodes of pain in the knee. Specific symptoms from the knee were anticipated in the seven patients with intra-articular extension and MRI confirmed ligamentous involvement in the other seven patients. Although haemarthrosis was not a presenting symptom it is clear that repetitive haemarthroses would have disastrous consequences. Episodes of effusion and haemarthrosis in the knee may cause a flexion contracture, muscular atrophy, equinus deformity of the foot and progressive ankylosis and early osteoarthritis of the knee.[2,3] In a study by Enjolras et al,[5] 15 of 17 patients with pure venous malformations of the limbs had a chronic localised intravascular coagulation disorder. This coagulopathy caused episodes of thrombosis or bleeding, leading to a haemarthrosis. This condition became worse after surgical intervention. If surgery is considered, treatment with low-molecular-weight heparin is advised to minimise thromboembolic complications.

Discrepancy in leg length was a presenting symptom in six patients, while at the time of the questionnaire ten patients (55%) had a leg-length discrepancy of 2 cm or more. This became evident in the hypertrophy group at a mean age of 3.5 years but was seen at two years in the group

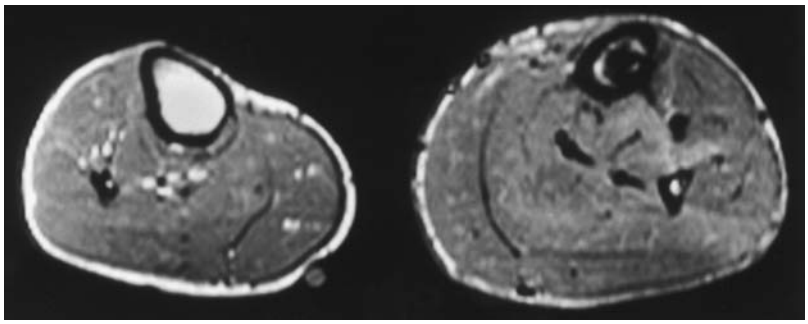

Fig. 5a

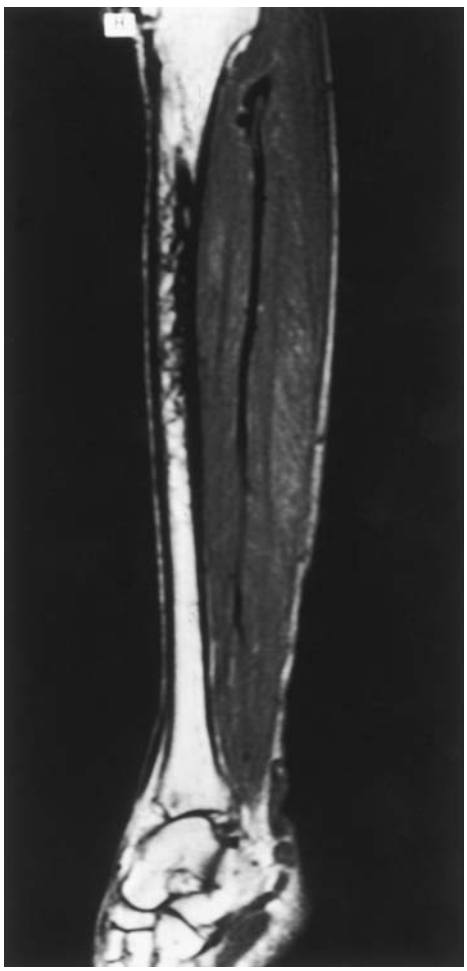

Fig. 5b

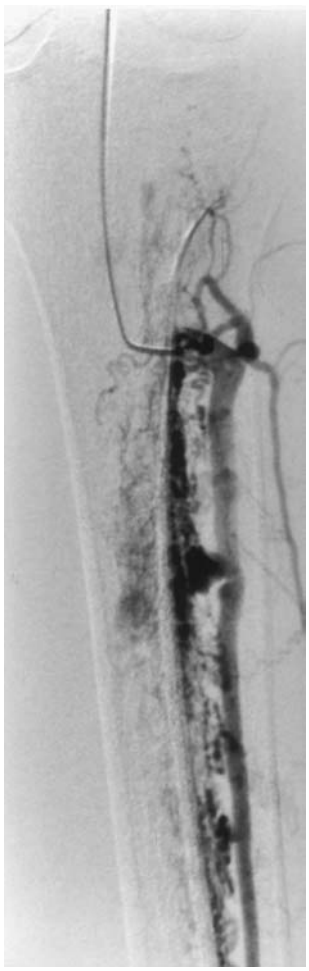

Fig. 5c

Figure 5a – MR axial proton-density-weighted image of both lower legs. There is a marked hypertrophy of the left leg, with signal voids, representing a high-flow malformation in the muscles and in the tibia. Figure 5b – MR sagittal T1-weighted image showing the bony involvement of the tibia. Figure 5c – Angiography of the same lesion, infiltrating muscles and bone.

with hypotrophy. The resultant secondary scoliosis is rarely noted before the child starts to walk.[1] All four KTS and the three high-flow lesions had limb overgrowth while one venous malformation had hypertrophy of 2 cm. Hypotrophy was seen with two other venous malformations. This confirms that venous malformations involve bones which are usually normal or hypoplastic.[3-5,10] The mild leg-length discrepancy seen in 70% of patients with KTS does not seem to progress after the age of ten years.[11]

Boyd et al[4] indicated that skeletal changes are commonly associated with vascular malformations while they are seen in less than 1% of patients with haemangiomas. MRI is, at present, the most effective way of demonstrating involvement of the surrounding structures and defining the type of

flow.[9,12] Despite some reports indicating that vascular malformations with associated bony involvement are rare, our findings seem to indicate the opposite.[13] Of our group of patients with vascular malformations of the lower limb, 20% (18 of 90) had bony involvement. Most bony vascular malformations described in the medical literature are in the craniofacial bones and the bodies of vertebrae.[6-8] In a study of 108 patients, Wenger and Wold[6] found 84 (77%) bony vascular malformations in either the craniofacial bones or the spine. The characteristic radiological appearance of vascular malformations of the skull and spine is often a well-circumscribed zone of rarefaction which may have a honeycomb appearance. It may also have a polka-dot appearance in the spine and a sunray appearance in the skull.[7] Involvement of long bones may lack these features and present a diagnostic challenge.[6,8] There have been only a few reports of intraosseous vascular malformations affecting long bones.[6-8,13-21] In a study by Boyd et al[4] of 158 patients with vascular malformations in the limbs, 50 (31%) had bony changes. Most had hypertrophy or hypotrophy. The extent of bony involvement was not stated and the patients were not divided into those affecting the upper and lower limbs. Intraosseous and lytic changes were characteristic of high-flow lesions. The distinctive thickened irregular trabecular pattern with well-defined lesions having a lattice-like trabecular pattern on plain radiographs, and the presence of a high signal intensity on MRI are the most helpful diagnostic imaging features for making the diagnosis of an intraosseous vascular malformation in long bones.[6] It is important to remember that the periosteal reaction may mimic osteosarcoma and chondrosarcoma.[12] Phleboliths are characteristic of venous malformations and not of all vascular malformations. Massive intraosseous involvement results in decreased bone density and an increased risk of fractures.

Vascular malformations with associated bony involvement often involve the diaphysis of long bones.[13-21] As shown in Figure 5, we could not confirm this since the distal half of the femur and the proximal half of the tibia were most often involved. In many patients without hypertrophy the distal femur or proximal tibia were affected indicating that the theory that increased vascularity in this growth-plate area results in hypertrophy seems to be unlikely.[4]

While a full discussion of treatment of these malformations falls outside the scope of this study, it should be emphasised that each case should be treated on its own merits. The management of all types of vascular malformation requires a multidisciplinary approach and treatment should, if possible, be conservative.[22] Options for treatment include symptomatic treatment with compression stockings and analgesia, surgical resection and/or intralesional transarterial embolisation and amputation.[23] Often patients with bony involvement also have muscular involvement making treatment more complicated. Surgical treatment is often associated with profuse bleeding, incomplete resection and local recurrence.[7,23] The difference in treatment between a high- and low-flow lesion is crucial.[23-27] Arteriovenous malformations can be excised and/or embolised.[22,23,27] Embolisation alone as a method of treatment for intraosseous arteriovenous malformations of the limbs is associated with a high incidence of recurrence.[27] Definitive solutions have been achieved in patients treated by a combination of surgery and interventional radiological techniques. The management in the growing child with leg-length inequality requires careful assessment, sequential leg-length evaluations and careful formulation of a treatment plan. Treatment should be by a multidisciplinary team since certain 'simple' procedures such as the ligation of varicose veins in KTS can have disastrous consequences.[23,25] Treatment should be conservative and intervention only initiated when symptoms develop, except when prophylaxis against progression is feasible or when complications arise.[23] In general, elastic support is supplied for the venous hypertension. Orthopaedic procedures should be prophylactic to control bony overgrowth. Matassi[10] has suggested that the vascular malformation should be treated before the bony involvement. Patients with high-flow lesions and hypertrophy (often PWS) who require correction of excess leg length, are often made worse by epiphysiodesis.[10] If the symptoms are severe and the surrounding soft tissue allows resection, some authors recommend *en-bloc* resection of the vascular malformation, including the affected bone.[12,15,20] Reconstruction consists of skin expansion or local flaps in the simple cases, and free flaps in more complicated ones. If the malformation is cosmetically and functionally acceptable, a conservative approach is often advised.[22] In our study, 70% of the patients had four or more muscles affected, indicating that resection would result in a mutilating intervention. Here analgesia and compression stockings are the mainstay of treatment.

Because of the rarity of vascular malformations, experience in diagnosis and treatment is limited. This may lead to misdiagnosis and a poor outcome. Vascular malformations are generally treated in specialised centres by a multidisciplinary team. They are probably more commonly associated with bony involvement than has been realised hitherto.

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