Massive localised lymphoedema: a rare vascular malformation

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▲ Introduction

Lymphatic malformations are a subset of congenital vascular malformations, and are caused by a defect in lymphatic development during embryogenesis. Future stimulus can cause the lesion to proliferate, resulting in the rare condition known as Massive Localised Lymphoedema (MLL). It is characterised by lymphoedema secondary to focally deranged lymphatic drainage in morbidly obese patients. It is difficult to characterise and likely to increase in incidence.

Case presentation

The patient presented with a mass on the upper medial aspect of her thigh, 30 cm x 15 cm, present for 12 months but increasing in size. She was obese (body mass index 64kg/m^2) and diabetic. The mass was soft, deeply tethered, with oedema and serous discharge. Contrast enhanced CT showed a focal swelling of soft tissue density arising from the thigh, superficial to deep fascia (Figure 1). Incisional biopsy showed lymphatic tissue only.

The 1857 g mass was marginally excised under general anaesthetic. Large lymphatic vessels were noted intra-operatively. Histologically the lesion was consistent with MLL.

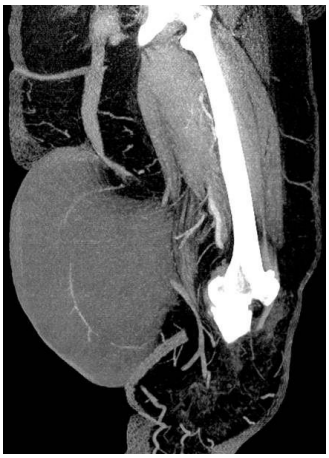

Fig. 1: Left leg CT angiogram.

Discussion

Lymphatic malformations form part of a spectrum of congenital vascular malformations that are classified using the Hamburg Classification System (4). The classification is of clinical benefit, allowing prediction of clinical course, response to treatment, and risk of recurrence.

Extratruncular lesions are embryonic tissue remnants of mesoderm that retain the potential to proliferate in response to hormonal or traumatic stimulus.

Truncular lesions arise as a result of developmental arrest during embryonic vascular trunk formation, and no longer have the ability to proliferate.

Massive localised lymphoedema is a rare extratruncular localised lymphatic malformation, with pre-malignant potential. It occurs around the 4th decade of life, in the thigh and abdominal wall, and also in the arm and external genitalia (1, 2, 5-8). There is a female predominance of 1.6:1, an average patient weight of 191 kg and a 58% occur in the thigh (2). There is a significant risk of sarcomatous transformation, with angiosarcoma in 9 reported cases of MLL, 5 of whom died within 11 months of diagnosis (9). Cases present commonly after extreme weight loss or after local surgery/trauma. The natural history is one of quiescence followed by rapid uncontrolled growth.

The diagnosis of MLL remains challenging for the pathologist due to the histological resemblance of this entity to well-differentiated liposarcoma (WDL), and misdiagnosis is not uncommon (6). The essential morphological features are preservation of normal subcutaneous fat architecture, absence of atypical lipocytes and paucity of cytological atypia in the fibrous bands between the fat lobules (3, 10).

Suspicion of MLL can be reasonably raised by accurate history and examination, and confirmed by biopsy, imaging and multidisciplinary discussion. Patient management will depend on the site, related structures, ease of surgical access and anticipated effect on quality of life.

Learning Points

MLL is a rare but significant congenital vascular malformation. Morbid obesity predisposes to clinical manifestation of MLL. It is part of a spectrum of malformations

that can coexist – and should be investigated as such. It has a significant risk of malignant transformation, and surgical excision is the preferred modality of treatment.

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