CASE REPORT

Massive localised lymphoedema: a rare vascular malformation

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SUMMARY

Lymphatic malformations are a subset of congenital vascular malformations, and are caused by a defect in lymphatic development during embryogenesis. When lymphatic mesoderm development is prematurely arrested, it retains its proliferative potential. Stimulus in the future can cause the lesion to proliferate locally without coordination or regulation, resulting in the rare condition known as massive localised lymphoedema (MLL). We present a case report of MLL, a rare and ill-defined soft tissue mass reported in the morbidly obese, with reference to the existing literature.

BACKGROUND

Massive localised lymphoedema (MLL) is a recently described clinical entity characterised by lymphoedema secondary to focally deranged lymphatic drainage in morbidly obese patients. These are difficult to characterise on imaging and create a significant challenge to clinicians and pathologists. With the obesity epidemic, the incidence of MLL is likely to increase and clinicians should be aware of the correct approach towards management of this pathology.

CASE PRESENTATION

The patient presented with a mass on the upper medial aspect of her left thigh, approximately 30 cm×15 cm in size, which had been present for 12 months, but had increased in size rapidly within the preceding 3 months. It was associated with discomfort on mobilising, frequent skin infections and made personal hygiene difficult, significantly affecting the patient's quality of life. She was morbidly obese with a body mass index of 64 kg/m². She was born in Kuwait, and had a medical history of non-insulin dependent diabetes. On examination the mass was soft and deeply tethered, with indurated and dimpled overlying skin, non-pitting oedema and a slight serous discharge.

INVESTIGATIONS

Contrast enhanced CT characterised the mass as a large non-enhancing focal swelling of soft tissue density arising from the posterior-medial aspect of the thigh. The mass was superficial to deep fascia. Incisional biopsy had been performed 1 year previously, and showed lymphatic tissue only with no evidence of malignancy.

DIFFERENTIAL DIAGNOSIS

Soft tissue sarcoma.

TREATMENT

The patient elected for open surgical excision under general anaesthetic. Intraoperatively it was noted that the mass consisted of a plethora of lymphatic vessels, some greater than 1 cm in diameter. The mass was marginally excised and primary closure was achieved. Suction drains were left in situ for a week and then removed. Continuing serous discharge was treated with local infusions of doxycycline. The patient was discharged home, but required a short readmission for treatment of a lower respiratory tract infection.

The excised specimen weighed 1857 g. The surface of the skin was indurated with a peau d'orange appearance (figure 1). On sectioning, the tissue was significantly oedematous and leaking fluid. The cut surface showed a lace-like network of fibrous septae between fatty lobules (figure 2). Microscopy showed skin with hyperplastic epidermis overlying fatty lobules separated by thick fibrous septae (figure 3A). In addition, plasma cells and mast cells were noted in the stroma together with predominantly perivascular lymphocytes. There was associated lymphangectasia and pseudocyst formation (figure 3B). The presence of myofibroblasts and floret like giant cells were noted in the fibrous septae (figure 3C).

The macroscopic and microscopic features are consistent with MLL.

OUTCOME AND FOLLOW-UP

Well at 1 year follow-up with no evidence of recurrence.

DISCUSSION

Lymphatic malformations form part of a spectrum of congenital vascular malformations that are classified using the Hamburg Classification System.¹ They are first categorised according to predominant vascular component, and then subclassified according to the temporal defect in embryological development stage (truncular or extratruncular), and whether they are localised or diffuse.

The classification is of clinical benefit, allowing the clinician to predict 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.

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Figure 1 (A) Minimal intensity projection image from thick coronal slices of a left leg CT angiogram, lesion shown with arrows. (B) Axial image from a contrast enhanced CT through the femurs demonstrating a large soft tissue mass arising from the posteromedial aspect of the left thigh distally, lesion shown with arrows. (C) Three dimensional reconstruction from a CT angiogram centred on the left knee demonstrating the anatomic relations of the soft tissue mass.

MLL may therefore be classified as a rare extratruncular localised lymphatic malformation, with premalignant potential. First described in the literature as an anomaly of the morbidly obese by Farshid and Weiss (1998),² it has a propensity for occurrence in the fourth decade of life, and anatomically in the thigh and anterior abdominal wall, although it has also been described in the upper arm and the external genitalia.^{3–8} Brewer *et al*⁷

reviewed 41 cases in the literature, and proposed a female predominance of 1.6 to 1, an average patient weight of 191 kg and a 58% majority of cases in the thigh. There is only one reported recurrence, with a lack of follow-up data. Perhaps more important to note is the significant risk of sarcomatous transformation, with the presence of angiosarcoma in nine reported cases of MLL, of whom five died within 11 months of diagnosis of this aggressive disease.⁹

Historically, cases present after an identifiable stimulus, commonly extreme weight loss, or after surgery or trauma to the area. The natural history is one of quiescence followed by rapid progression of uncontrolled growth.

The diagnosis of MLL remains challenging for the pathologist owing to the histological resemblance of this entity to well-differentiated liposarcoma. Therefore, misdiagnosis is not uncommon.³ The essential morphological features are considered to be preservation of architecture of the normal subcutaneous fat, absence of atypical lipocytes and the paucity of the cytological atypia in the fibrous bands in between the fat lobules.^{10 11}

Suspicion of MLL can be reasonably raised by an accurate history and examination, and is confirmed by biopsy, imaging (ultrasound/CT/MRI) and multidisciplinary discussion. This is important given its similarity to well differentiated liposarcoma and its potential for malignant transformation.

Management

Despite often existing as a diffusely infiltrating independent lesion the majority of lesions are relatively benign and seldom become a life-threatening or limb-threatening condition. Patient-tailored management depends on the anatomical site, related neurovascular structures, ease of surgical access and anticipated effects on quality of life.

Surgical excision can be performed where required for functional or cosmetic reasons, but in areas where radical surgery with clear margins would cause excess morbidity a debulking procedure is seen to be preferential. Recurrence has shown to be reduced when pre-operative embolo-sclerotherapy is performed.¹² An intravascular sclerosant is injected under radiological guidance to the abnormal vasculature in an effort to cause scar tissue to form and close off feeding vessels. The

Figure 2 (A) Skin surface showing the characteristic peau d'orange appearance. (B) Cut surface showing prominent fibrous septae exhibiting lace-like configuration.

Figure 3 (A and B) Low power views of H&E stained sections (magnification $\times 2.5$) show fatty lobules separated by broad fibrous septa and pseudocyst formation in photographs (A) and (B) respectively. (C) A high power view of H&E stained sections (magnification $\times 20$ and $\times 40$ inset) shows myofibroblasts, which display large nuclei and scattered floret giant cells (arrows).

resulting reduction in vascular supply by the action of the sclerosant can also reduce operative bleeding.

Summary

MLL is a recently described clinical entity characterised by lymphoedema secondary to focally deranged lymphatic drainage in morbidly obese patients. These are difficult to characterise on imaging and create a significant challenge to clinicians and pathologists. With the obesity epidemic, the incidence of MLL is likely to increase and clinicians should be aware of the correct approach towards management of this pathology.

Learning points

- Massive localised lymphoedema (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. Surgical excision is the preferred modality of treatment.

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