A rare case of thigh asymmetry in an infant

A 6-month-old baby boy presented with asymmetry of thighs since birth. On clinical examination, anteromedial aspect of the left thigh was bulky with pale yellow discolouration of the skin and no tenderness. The child was referred for high-resolution ultrasonography. On ultrasonography, there was evidence of a well-defined isoechoic soft tissue lesion measuring 1.5x6.0cm in the subcutaneous plane of the anteromedial aspect of the upper two-thirds of the left thigh with multiple cystic spaces (Fig. 1A). The lesion was noted to extend into the intermuscular fascial planes with no evidence of deeper muscle invasion (Fig. 1B). There was associated thickening of the skin and subcutaneous fat with increased echogenicity. The lesion was non-compressible on application of pressure by the ultrasonography probe. Colour Doppler ultrasonography revealed absent vascularity within the cystic spaces of the lesion (Fig. 1C).

What is your diagnosis?

- A. Rhabdomyosarcoma
- B. Cavernous haemangioma
- C. Venous malformation
- D. Arteriovenous malformation
- E. Mixed lymphatic malformation

Findings and diagnosis. A diagnosis of lymphatic malformation of the thigh was made based on the findings of high-resolution ultrasonography. Lymphatic malformations tend to be infiltrative, permeate across fat planes, and manifest with associated diffuse soft tissue thickening and surrounding lymphoedema. The child was referred to the department of paediatric surgery for further management where an open surgical excision under general anaesthesia was performed. Surgical debulking of the soft tissue lesion was performed, which demonstrated intermuscular extension via the fascial planes on the subcutaneous plane of the lesion. However, there was no deeper intramuscular invasion. The intraoperative findings correlated very well with the preoperative findings on ultrasonography. Intraoperatively, the mass lesion consisted of a plethora of lymphatic vessels, some >1cm in diameter and some <1cm in diameter. Histopathology of the resected specimen demonstrated multiple macrocystic and microcystic

Fig. 1. (A) High-resolution ultrasonography image demonstrating an isoechoic soft tissue lesion with multiple macrocystic (e.g. area indicated by a square) and microcystic (3 arrowheads) spaces in the anteromedial aspect of the left thigh. Note the interspersed soft tissue component (2 stars). (B) Far field high-resolution ultrasonography image demonstrating the extent of the soft tissue lesion in the subcutaneous plane with extension into intermuscular fat planes (arrow). (C) Doppler ultrasonography image demonstrating absent vascularity within the cystic spaces (in rectangle box) consistent with features of lymphatic malformation.

Answer: E

Fig. 2. Histopathology image demonstrating multiple macrocystic and microcystic lymphatic channels in the background of loose connective tissue stroma consistent with features of mixed lymphatic malformation. Note the focal disorganisation of smooth muscle in the walls of larger channels (haematoxylin and eosin staining, x200 magnification). (Colour figure available online.)

lymphatic channels in the background of loose connective tissue stroma, consistent with features of mixed lymphatic malformation (Fig. 2). Postoperative course was uneventful and the child was discharged home after 1 week.

Discussion. Lymphatic malformations are developmental anomalies of the lymphatic system. They are benign soft tissue lesions and congenital in origin. They are also often slow growing and frequently located in proximal extremities—in this case, the trunk and axilla.¹ Lymphatic malformations are classified as macrocystic, microcystic or mixed, based on their imaging appearances.² Lymphatic malformation involving soft tissues can be confidently diagnosed based on high-resolution ultrasonography and colour Doppler findings, which include isoechoic to hypoechoic soft tissue masses showing multiple cystic spaces and absent vascularity. Differential diagnosis of soft tissue masses involving the extremities in the infantile age group are vascular malformations such as haemangioma (the most common); benign tumours such as lipoma; malignant tumours of the smooth muscle such as rhabdomyosarcoma; and lymphatic malformation. Lymphatic malformations do not show spontaneous resolution after birth, and first-line treatment includes sclerotherapy, with debulking surgery reserved only as second-line treatment.³

Although cystic lymphangiomas of congenital origin involving extremities have been previously described,⁴ this case report deserves special attention due to the lymphangiomas' mixed appearance on ultrasonography. The formations were composed of large cysts interconnected with clustered smaller lymphatic cysts,

which were large sized at presentation across the upper two-thirds of the thigh, a rare site for an infant. Mixed lymphatic malformation of the extremity is rare, which makes this case unique. Moreover, the lesion described is confined only to the extremity, with no abdominal extension. Literature review reveals varied presentation of the entity such as juxta-articular lymphangioma of the extremity,⁵ retroperitoneal and genital lymphangioma,⁶ and even acquired cases of progressive lymphangioma.⁷ Previously published case reports documented abdominal lymphatic malformations extending to the extremities, mostly in autopsy specimens of preterm infants after termination of pregnancy.⁸ This case illustrates an uncommon yet important cause for unilateral extremity swelling of congenital origin. Although there were complaints of asymmetry of thighs since birth in this case, the child had a delayed presentation at 6 months when the mother felt an obvious mass in the left thigh. However, prompt diagnosis of the entity on ultrasonography and timely management resulted in a favourable outcome. Since fetal lymphatic malformations carry high risk of aneuploidy and coexisting fetal malformations, prenatal diagnosis of the entity on ultrasonography has a prognostic significance.

Based on the size of the lymphatic lumen, lymphatic malformations (previously termed lymphangiomas) can be classified into microcystic lesions (previously termed lymphangioma circumscriptum), macrocystic lesions (previously termed cystic hygromas or cavernous lymphangiomas), and a combined form as described in this case. The mean diameters of cystic spaces are >1cm in macrocystic lymphatic malformation and <1cm in microcystic lymphatic malformation, and a combination of these in mixed lymphatic malformation. On ultrasonography, lymphatic malformations present as soft, easily compressible masses that increase in size in dependent positions or when venous pressures increase (through crying or the Valsalva manoeuvre). Lymphatic malformations appear as multilocular cystic masses with internal septations of varying thickness.

The cystic contents in lymphatic malformations are usually anechoic, and appear hyperechoic if debris, high lipid concentration, infection or haemorrhage is noted within. In lymphatic malformations, the cystic spaces are noted in the skin and subcutaneous fat. The cystic spaces are usually round or ovoid, rather than tubular as seen with venous malformations. Lymphatic malformations are non-compressible unlike venous malformations. On colour Doppler ultrasonography, no flow is demonstrated within the cystic spaces, but

blood flow is demonstrable in the septae. Ultrasonography including colour Doppler and spectral Doppler tracings can evaluate vascularity and determine types of vessels present in congenital vascular malformations. Soft tissue venous malformations (haemangiomas) are compressible with the typical multicystic and/or partially solid heterogeneous echotexture, and can be hypoechoic, hyperechoic, or isoechoic with respect to surrounding structures.⁹ Although phleboliths are classic of venous malformation and noted on plain radiographs, they are rarely detected on ultrasonography. On colour Doppler ultrasonography, monophasic waveforms with low flow are noted in venous malformation, while continuous high flow is noted in arteriovenous malformation, and no flow is noted in large hypoechoic cysts in lymphatic malformation. The presence of pulsatile triphasic flow of nearby arteries in a venous malformation should not be confused with arteriovenous malformation. Rhabdomyosarcoma is the most common soft tissue tumour of extremities in children, and presents as a heterogeneous, well-defined and irregular mass of low to medium echogenicity on ultrasonography.¹⁰

Lymphatic malformations are congenital lesions, which are present since birth. Although there were complaints of asymmetry of thighs since birth in this case, the child had a delayed presentation at 6 months when the mother felt an obvious mass in the left thigh. However, prompt diagnosis of the entity on ultrasonography and timely management resulted in a favourable outcome. Lymphatic malformation needs to be included in the differential diagnosis of soft tissue masses involving the extremities, especially when the lesion is congenital and present since birth.

REFERENCES

1. Lal A, Gupta P, Singhal M, et al. Abdominal lymphatic malformation: Spectrum of imaging findings. *Indian J Radiol Imaging* 2016; 26:423-8.
2. Wassef M, Blei F, Adams D, et al. Vascular Anomalies classification: recommendations from the International Society for the Study of Vascular Anomalies. *Pediatrics* 2015;136:e203-14.
3. Okazaki T, Iwatani S, Yanai T, et al. Treatment of lymphatic malformation in children: our experience of 128 cases. *J Pediatr Surg* 2007;42:386-9.
4. Thakur SK. Unilocular cystic lymphangioma of thigh—an extremely rare clinical entity. *Indian J Surg* 2010;72:417-8.
5. Jain D, Selhi HS, Yamin M. Lymphangioma presenting as juxta-articular swelling in children: a case series. *APSP J Case Rep* 2013;4:30.
6. Olabanji J, Oladele A, Famurewa O, et al. Retroperitoneal and genital lymphangioma therapeutic challenges in a developing country. *Libyan J Med* 2009;4:44-5.
7. Larkin SC, Wentworth AB, Lehman JS, et al. A case of extensive acquired progressive lymphangioma. *Pediatr Dermatol* 2018;35:486-9.
8. Durukan H, Gözükar İ, Cevikoglu M, et al. Isolated fetal lymphatic malformation of the thigh: prenatal diagnosis and follow-up. *Autops Case Rep* 2017;7:49-53.
9. Chan YC, Giam YC. Guidelines of care for cutaneous haemangiomas. *Ann Acad Med Singap* 2005;34:117-23.
10. Aung LL, Soe TA, Chang KT, et al. Singapore Rhabdomyosarcoma (RMS) Experience: Shall We Change Our Practice? *Ann Acad Med Singap* 2014;43:86-95.

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