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CLINICAL REPORT

Congenital Elephantiasis-like Lymphangiomatosis of a Lower Limb

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The case of a newborn girl with a rare, giant, congenital, tissue lymphangioma giving rise to elephantiasis of the right lower limb is presented. The different imaging methods, especially magnetic resonance imaging, showed no extension of the lesions into the deep structures. At the age of 2 years, the child underwent a roentgenographic skeletal survey, which revealed osteolytic lesions in the femurs and the right tibia. There was no clinical evidence of systemic involvement. The place of this affection among the different lymphatic malformations was discussed and the diagnosis of elephantiasis-like lymphangiomatosis of the limb, an extremely rare disorder, has been retained. Early surgical reduction was performed, followed by application of a pressure dressing. Five years later the result remains satisfactory, but the excision of a persistent fluid-filled pouch around the knee will probably be necessary in the future. ***Key words: lymphangioma; lymphatic malformations.***

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Lymphangiomas are considered as hamartomatous malformations composed of dilated lymph channels of various sizes, lined by normal lymphatic endothelium. There are multiple sites of development, including the skin. The classification of lymphangiomas tends to be confusing. At the present time, 2 types of lymphatic malformations are distinguished: cystic lymphangiomas and tissue lymphangiomas, this second type including localized and diffuse forms (1). We report a case of a giant tissue lymphangioma, giving rise to elephantiasis of a lower limb in a newborn girl. The malformation was associated with asymptomatic osteolytic lesions of the femurs and the right tibia, emphasizing that cutaneous lymphatic malformations may be an expression of lymphangiomatosis, which is a more widespread disease. Classically, this entity has a very poor prognosis because of extensive visceral involvement but localized forms have been reported recently, characterized by a predominant or exclusive involvement of the soft tissues of a limb, with or without bone lesions (2). This case confirms the necessity to individualize these localized forms of good prognosis and we report on the first congenital elephantiasis-like lymphangiomatosis of a limb.

CASE REPORT

A newborn girl was referred to our hospital after birth with a suspected diagnosis of cystic lymphangioma of the right lower limb. The malformation, which presented as a multiloculated mass, was detected *in utero* by fetal echography at 32 weeks' gestation.

Physical examination showed an unusual malformation of the right lower limb, extending from the proximal part of the thigh to the upper two-thirds of the leg, sparing the foot. The malformation consisted of a severe sponge-like swelling of the thigh associated with a monstrous redundancy of the skin on the distal part of the thigh and the upper leg. The many loose folds gave the appearance of scrotal skin (Fig. 1*a*). On palpation, there was the sensation of underlying fluid-containing pouches. On standing-erect examination, the fluid filled the redundant skin, which immediately changed into a large, elongated bag hanging on the leg.

Chylomicrons were detected in the fluid aspirated with a syringe, confirming the lymphatic nature of the malformation. Echo-Doppler of the affected limb was normal. A full skeletal survey seemed to be normal and a chest X-ray showed no abnormalities. An MRI scan showed high signal intensity in the subcutaneous tissues of the entire right lower limb, with the exception of the foot, on T2-weighted images (Fig. 2). The underlying muscles were spared and there was no extension of the lesions into the pelvis and retroperitoneum. Isotopic lymphangiography was performed via the lymph vessels on the dorsa of the feet. This revealed a blockage of lymphatic drainage at the right knee level. The lymphatic system within the left limb and from the right foot to the knee was normal. Two biopsy specimens were taken from the redundant skin and the proximal part of the thigh. Histologic examination revealed numerous dilated and interconnecting lymphatic vessels extending through the middle and deep dermis, lined by a thin layer of endothelial cells. The

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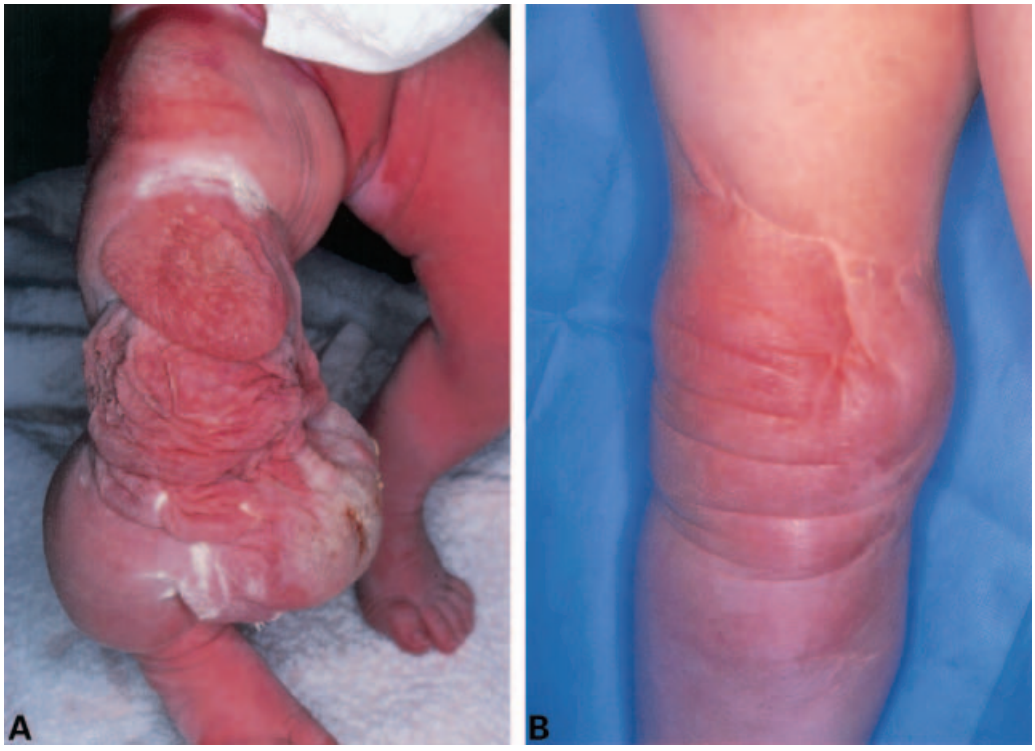

Fig. 1. (a) At birth: enlargement of the right lower limb and redundancy of the skin, sparing the foot; multiple loose folds giving the appearance of scrotal skin. *(b)* Feature 5 years after surgery (at 5 years of age): a fluid-filled pouch persists around the knee.

lymphatic spaces dissected between normal persistent dermal structures and collagen bundles (Fig. 3).

These features were consistent with the diagnosis of congenital diffuse lymphangioma of the limb with a clinical aspect of elephantiasis. Through excoriations of the redundant skin, leakage of lymphatic fluid was continuous and induced hypoproteinemia. A surgical reduction was performed at 3 months of age associated with a remodeling excision of most of the excess skin.

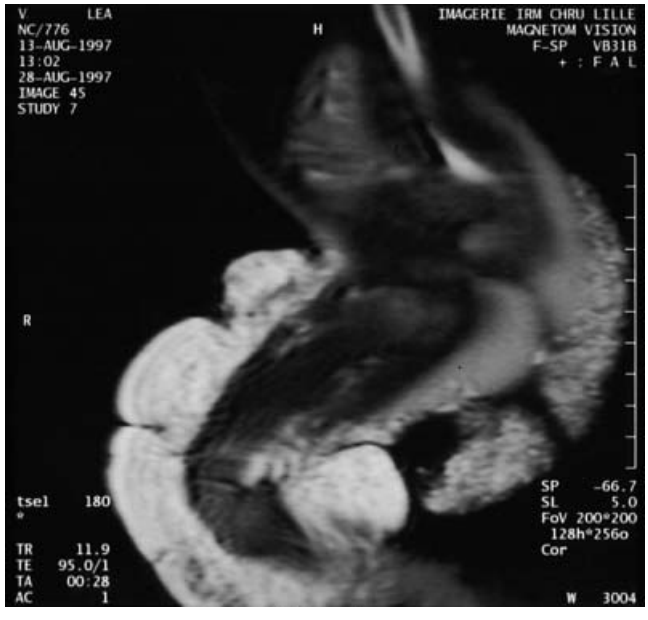

Fig. 2. Sagittal MR T2-weighted image: high signal intensity in the subcutaneous tissues without involvement of the deep structures.

There was an interesting improvement with a significant reduction in the volume of the limb. Only a little leakage of lymphatic fluid persisted during the 4 months after surgery, and then stopped. Hypoproteinemia was rapidly corrected by giving additional nutriments. There were no infectious events and walking ability developed without any problems. At the age of 2 years, the child's roentgenographic skeletal survey revealed osteolytic lesions in the distal part of the right femur and in the middle part of the right tibia and left femur, which were totally asymptomatic. There was no clinical evidence of systemic involvement. At the present time, 5 years after surgery, the bone lesions

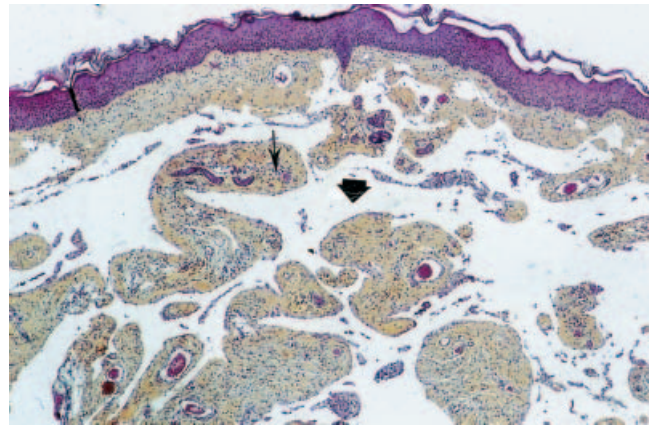

Fig. 3. Histopathologic features: large lobules of interconnecting dilated lymphatic channels extending through the dermis (*arrowhead*) dissecting between normal persistent tissue (*arrow*) (H&E, original magnification $\times 40$).

have tended to regress and there is no difference in the length of the lower limbs. A pressure dressing made to measure is permanently carried. On standing-erect examination, a pouch of moderate volume can be seen around the knee, which becomes completely empty when the limb is elevated, filling a part of the thigh, which swells and feels sponge-like on palpation, attesting to a large communication (Fig. 1*b*). The morphological and functional results remain satisfactory but the excision of this persistent fluid-filled pouch will probably be necessary in the future.

DISCUSSION

Taking the results of biochemical, histologic and imaging investigations into consideration, there was no doubt about the lymphatic nature of the malformation in this newborn girl. Congenital malformations of the lymphatic system constitute a spectrum of disorders that may manifest with a variety of clinical presentations that imply very different therapeutic problems and prognoses. The diagnosis of lymphedema was not considered in this case since this affection is usually bilateral, does not spare the dorsa of the feet and there are no fluid-filled pouches on palpation and no sponge-like swelling (3). Clinical and histologic features were consistent with lymphangioma. The term "lymphangioma", usually used when lymphatic vessels are dilated to tumor-like proportions, applies to 2 types of vascular malformations (1). The first type, cystic lymphangioma, also referred to as macrocystic lymphatic malformation (4), is usually a benign condition that clinically presents as a well-defined tumefaction with a normal overlying skin. In our patient, this diagnosis was suspected on fetal echography but the appearance of the malformation at birth was inconsistent with this hypothesis. The second type of malformation is tissue lymphangioma, a deeply infiltrating lesion, characterized by a cutaneous and subcutaneous or mucous thickening, not well limited. This entity includes the previously termed lymphangioma circumscriptum, also referred to as localized microcystic lymphatic malformation (4), and a diffuse form, previously referred to as deep cavernous lymphangioma, which is less frequent. There is no clear distinction between lymphangioma circumscriptum and diffuse lymphangioma and the difference depends solely on the extent of the malformation. These tissue lymphangiomas may manifest with fluid-filled vesicles, usually grouped into structures resembling frogspawn. However, malformations situated in the subcutaneous fat may present as localized swelling with normal overlying skin. In our patient, the diagnosis of diffuse lymphangioma was a strong hypothesis but the clinical aspect of sponge-like swelling is not reported in this entity. Clinical and histologic features were consistent with lymphangiomatosis. Classically, lymphangiomatosis is a very rare systemic disorder characterized by a widespread abnormality of the lymphatic system, with both bone and extensive visceral involvement, especially chylothorax and chylous ascites, associated with a poor prognosis (5, 6). The condition becomes apparent after the first year of life and is rarely present at birth. Recently, another type of lymphangiomatosis has been described. Gomez et al. (2) have reported 6 cases of patients with almost exclusive involvement of soft tissues of the extremities, with or without bone involvement, associated with a slow clinical progression and a good prognosis. This condition, termed lymphangiomatosis of the limbs, has rarely been reported in the literature and seems to be relatively underdiagnosed. It is present earlier in life, and usually affects a lower limb but can involve an upper limb. Clinically, lymphangiomatosis of the limbs is characterized by a sponge-like swelling of the affected limb, the fluid contained in the large pouches can be moved about within the swollen area. Overlying skin changes such as hyperplasia, pigmentation and vesicle formation may appear secondarily. Lytic bone lesions, which are always asymptomatic, are inconstant. They may appear after a few months, remain stable throughout life or even regress. They may be located in bones neighboring the affected soft tissues or in distant bones, as in our report. Occasionally, the malformation involves the underlying fascias and muscles. Histopathologically, the lesions of lymphangiomatosis consist of large lobules of interconnecting dilated lymphatic channels involving the dermis and subcutaneous fat, dissecting between collagen bundles and around pre-existing dermal and subcutaneous structures. Lymphangiography shows dilated lymphatic spaces, sometimes with connections to normal lymphatics. In our observation, the association of a diffuse tissue lymphangioma giving a sponge-like swelling, histologic examination and findings of lymphangiography were consistent with the diagnosis of lymphangiomatosis of the limb. The bone abnormalities were absent at birth and appeared secondarily, emphasizing the importance of renewing roentgenographic skeletal survey to reinforce the diagnosis. To our knowledge, the clinical aspect of elephantiasis has never been reported in this entity. Furthermore, these malformations are rarely present at birth. We have found only one case clinically similar to our observation, also congenital and associated with multiple bone lesions in the tibia but the extension of the lesions into the pelvis and retroperitoneum raises the diagnosis of generalized lymphangiomatosis (7).

REFERENCES

1. Riche MC. Lymphangiomas. Rev Prat (Paris) 1992; 42: 2041 – 2043.
2. Gomez CS, Calonje E, Ferrar DW, Browse NL, Fletcher CD. Lymphangiomatosis of the limbs. Clinicopathologic analysis of a series with a good prognosis. Am J Surg Pathol 1995; 19: 125 – 133.

3. Lorette G, Vaillant L, Baulieu F. Lymphoedema of children. Ann Dermatol Venereol 2001; 128: 674 – 676.
4. Mulliken JB, Young A. Vascular bithmarks, hemangiomas and vascular malformations. Philadelphia: WB Saunders, 1988.
5. Requena L, Sangueza OP. Cutaneous vascular anomalies. Part I. Hamartomas, malformations, and dilation of preexisting vessels. J Am Acad Dermatol 1997; 37: 523 – 549.
6. Dutheil P, Leraillez J, Guillemette J, Wallach D. Generalized lymphangiomatosis with chylothorax and skin lymphangiomas in a neonate. Pediatr Dermatol 1998; 15: 296 – 298.
7. Castillo M, Dominguez R. Congenital lymphangiectatic elephantiasis. Magn Reson Imaging 1992; 10: 321 – 324.
8. Wells RG, Ruskin JA, Sty JR. Lymphoscintigraphy. Lower extremity lymphangioma. Clin Nucl Med 1986; 11: 523.
9. Breugem CC, Maas M, Reekers JA, van der Horst CM. Use of magnetic resonance imaging for the evaluation of vascular malformations of the lower extremity. Plast Reconstr Surg 2001; 108: 870 – 877.
10. Katz VL, Watson WJ, Thorp JM Jr, Chescheir NC, Miller RC. Prenatal sonographic findings of massive lower extremity lymphangioma. Am J Perinatol 1992; 9: 127 – 129.