

Macrocystic lymphatic malformation in adolescence. A case report and literature review

Malformación linfática macroquística en la adolescencia. Presentación de caso y revisión de la literatura

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Summary
Cystic lymphatic malformations are rare benign vascular anomalies affecting various anatomical surfaces. The cervicofacial region is the most commonly affected. Their diagnosis is usually prenatal through ultrasound, and their peak incidence occurs around 2 years of age, when the growth of cervical masses is highly suggestive of lymphatic malformations. Late presentations are exceptional and are usually associated with chromosomal abnormalities and/or genetic syndromes. The treatment of these abnormalities depends on the patient's condition, the characteristics of the lesion, and the availability of personnel to perform invasive or percutaneous procedures, with surgical excision and sclerotherapy being the two most commonly used modalities in these cases. An unusual case of microcystic lymphatic malformation in a 13-year-old patient is presented, along with its diagnostic and therapeutic approach, as well as the findings from a brief updated literature review.

Resumen
Las malformaciones linfáticas quísticas son anomalías vasculares benignas poco comunes que afectan diversas superficies anatómicas, aunque la región cervicofacial es la más frecuente. Su diagnóstico suele ser prenatal mediante ecografía y su pico de incidencia máxima se encuentra cerca de los 2 años, edad en la que el crecimiento de masas cervicales es muy sugestivo de malformaciones linfáticas. Las presentaciones tardías son excepcionales y suelen asociarse a cromosomopatías y/o síndromes genéticos. El tratamiento de estas anormalidades depende del estado del paciente y de las características de la lesión, así como de la disponibilidad de personal para realizar procedimientos invasivos o percutáneos, como la extracción de la pieza quirúrgica y la escleroterapia, que son las dos modalidades más usadas en estos casos. Se presenta un caso inusual de malformación linfática microquística en un paciente de 13 años de edad, su aproximación diagnóstica y terapéutica, así como los hallazgos encontrados en una breve revisión de literatura actualizada.

Introduction
Cystic lesions of the lymphoid system are vascular malformations composed of dilated lymphatic vessels that are usually congenital and benign (1). Most of these masses are evident at birth (50%-65%) and up to 80%-90% present before the age of 2 years (1, 2). In the latter cases, lesions may be evident with slow, progressive growth or sudden, rapid growth following infection or hemorrhage at the site (1, 2). Its incidence is not well established; however, it is known to have a prevalence of approximately 1 in 2,000 to 4,000 live births and to cause one miscarriage for every 750 spontaneous abortions (1, 3).

According to the ISSVA classification for simple vascular malformations, three morphological types of cystic malformations are recognized: microcystic (< 1 cm), macrocystic (> 1 cm) and mixed lesions. The most frequent presentation is macrocystic, with 1-3 lesions predominantly in the cervicofacial area, which represents 20-25% of cervical lymphatic tumors (3).

Its etiology is still not well known; however, the main theories correlate anomalies in vasculogenesis that compromise the connection between the lymphatic and venous systems, abnormal sprouting of lymphoid tissue and isolated lymphoid remnants that retain embryonic growth potential. They typically occur at the end of the sixth week of gestation (4, 5). Recent research has studied the genetic basis involved in the development of these lesions, describing genetic variations in specific cells and associations with genetic syndromes, such as

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Turner and Klinefelter syndromes, and chromosomopathies, such as trisomies 21, 18 and 13 (1, 3).

Case report

A 13-year-old male patient with no relevant history was admitted to the emergency department for eight days with right cervical pain and the presence of a progressively growing mass associated with respiratory distress. Neck ultrasound was performed with findings of an oval lesion of 2.3 × 4.0 × 4.4 cm, expansive, heterogeneous, well-defined contours, with posterior enhancement in levels III, IV and V of the right neck and predominantly solid (Figure 1). No flow was evident in the Doppler study. Due to the characteristics, an abscessed adenopathy or infiltrative type lesion was suspected.

No relevant findings were found in the hemogram and active phase reactants were negative. Computed axial tomography (CT) of the neck reported a 50 × 30 mm mass with areas of liquid and solid density, septate, multilobulated, with displacement of the sternocleidomastoid muscle and the carotid artery, as well as decreased caliber of the jugular vein (Figure 2). The above findings were suggestive of a lymphatic anomaly, which was corroborated by magnetic resonance imaging (MRI). The MRI showed a cystic image with a liquid-liquid level in its interior in sequences with T1 and T2 information measuring 6.6 × 5.6 × 3.8 cm, without enhancement after the administration of contrast medium (Figure 3). Extension was observed occupying levels IIA, III, IV and VB of the right hemicollar. There was no evidence of airway involvement. The lesion was compatible with a low-flow cystic venolymphatic malformation.

Given the imminent risk of airway obstruction, the patient underwent ultrasound-guided collection drainage with fluoroscopic venography and sclerotherapy. During the procedure, guided by Doppler ultrasound, a mixed lymphatic malformation was identified in the right hemicollar. Subsequently, percutaneous puncture was performed with catheter # 16 and drainage of approximately 50 mL of serohematic material, followed by administration of bleomycin 45 IU. Neck venography with contrast medium performed to identify communication of the lymphatic malformation with the venous or arterial system was negative (Figure 4). Histopathology study confirmed a macrocystic lymphatic malformation.

Diagnosis

The anamnesis and clinical findings on physical examination, such as large, soft, translucent, non-compressible masses, which usually appear from birth or early childhood, mainly in the neck and axillae, usually suggest the presence of a lymphatic lesion in up to 41% of cases, especially in typical cases, which appear before the age of two years. When the age of presentation is older than two years, other lesions of similar appearance such as lipomas, dermoid cysts, branchial cysts, thyroglossal duct cysts, teratomas, salivary gland or thyroid tumors

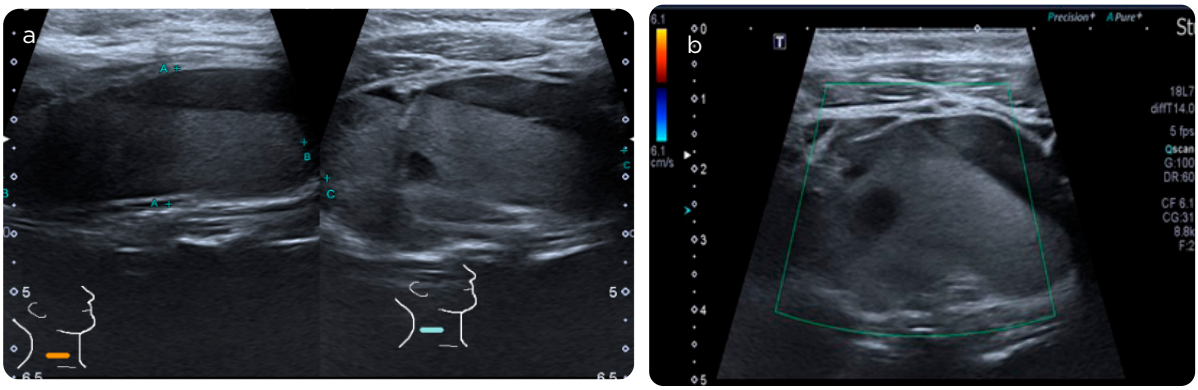


Figure 1. Cervical ultrasound. a). B-mode ultrasound: oval expansive lesion with liquid-liquid level, with well-defined contours in the right hemicollar at levels III, IV and V. b) Color Doppler ultrasound of the lesion, which does not show vascularization.



Figure 2. Neck CT with contrast medium. a) Coronal. Low density lesion, homogeneous, displacing the sternocleidomastoid muscle and adjacent vascular structures. b) Axial. Compressive effect of the lesion on the jugular vein and right carotid artery. Considerable reduction of the jugular vein caliber. There is no evidence of contrast medium enhancement within the lesion.

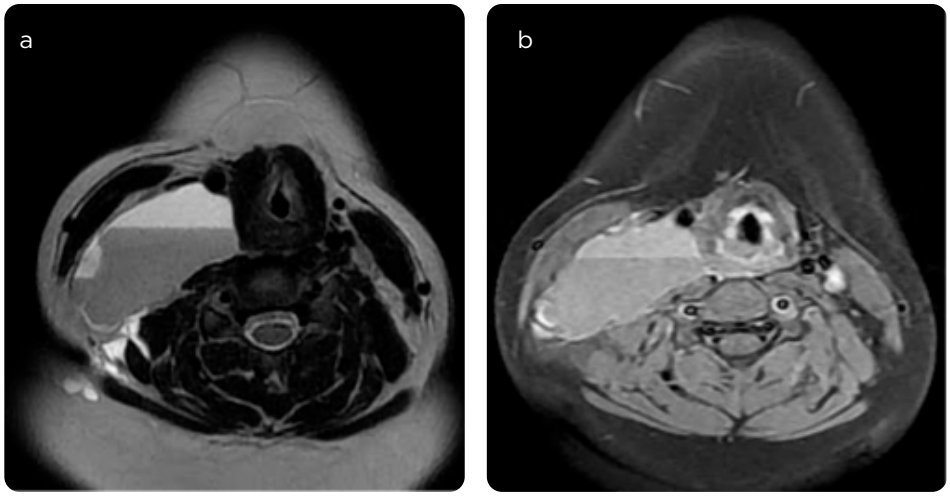


Figure 3. a) Axial MRI with T2 information. Cystic lesion with liquid-liquid level inside, measuring 6.6 × 5.6 × 3.8 cm extending through levels IIA, III, IV and VB of the right hemicollar. b). Axial MRI with T1 information after contrast medium injection. There is no enhancement of the contrast medium within the lesion.

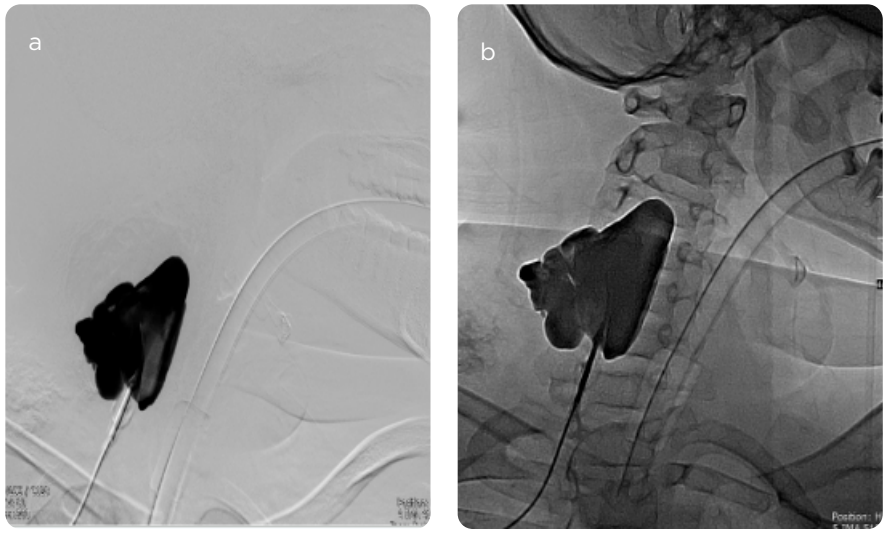


Figure 4. Fluoroscopic venography study with direct injection of contrast medium. There is no evidence of communication of the malformation with the venous or arterial system.

and neurofibromas should be ruled out (2, 5). In the case presented, one of the initial differential diagnoses was lymphoproliferative lesion, which can also manifest as a solid cervical lesion; however, the normal behavior of the cell lines indicated another etiology.

Imaging evaluation is necessary in the presence of a cervical mass. Although the diagnosis and extension can be evaluated by ultrasonography (US), CT or MRI, the first line tool is the US of the neck, which is particularly useful to differentiate a cystic mass from a solid tumor. Likewise, US has the advantage of intrauterine detection of cystic hygroma (3, 4), while radiographs -although they are not routine in the evaluation of vascular malformations- are more useful in the evaluation of the respiratory tract; in addition, they allow the detection of phleboliths and calcifications (3, 5).

CT, like US, is useful for detecting the solid or cystic nature of the lesion and, in addition, allows delineation of the relevant anatomical structures. Plain radiography is of limited value, even for assessing the degree of bone involvement and calcifications; in contrast, CT is markedly more sensitive in this respect. However, MRI provides greater anatomical and soft tissue differentiation, making it the most useful modality for assessing the extent of the lesion (3). To differentiate the lymphatic lesion from other masses with a fluid component, needle aspiration or biopsy is required.

The definitive diagnosis of these lesions is histopathological, confirming that the cystic malformation has conjunctive stroma separated from lymphoid tissue and smooth muscle, as well as endothelial lining covering the formation (6).

As for treatment and prognosis, intervention depends largely on the size of the lesion and the risk of affecting adjacent nerves, the stability of the patient and the availability of trained personnel. In microcystic lesions, drainage of lymphatic fluid together with sclerotherapy may be sufficient (7, 3). Percutaneous sclerosis has been described with different agents, such as bleomycin, ethanol or OK-432, and surgery is often used as an adjunctive therapy (7). Macrocysts may require surgical removal, which should be performed by gentle dissection to avoid severing nerves and tearing the walls of the lesion (7). The evolution of the patient in the case documented here was satisfactory, with no recurrence of the lesion in the following five months after the intervention.

Discussion

Cystic lymphatic malformation is an anomaly seen almost exclusively in children under 2 years of age; it is very rare in older children and adults. Although congenital masses and the appearance of cervical lesions in the first years of life are usually benign, the differential study of each of them must be exhaustive to rule out malignant etiologies, especially in solid masses. The most frequent causes of cervical lesions and differential diagnoses of lymphatic malformations in children include thyroglossal duct cysts, gill anomalies and dermoid cysts (2, 5).

According to the literature, malformations with late onset - after 2 years of age - are often associated with a sudden increase in size secondary to subclinical infection, which is common during childhood and adolescence (6). Different variants of vascular malformations have been described: capillary, lymphatic, venous and arteriovenous, among which lymphatic malformations, in turn, can be differentiated into common (cystic), generalized lymphatic anomalies, Gorham-Stout disease, canal-like malformations, acquired progressive lymphatic anomalies and primary lymphedema, according to the ISSVA 2018 classification (7, 8).

As evidenced, finding a congenital lymphatic malformation debuting in adolescence is not improbable; however, its low incidence should be a reason to exclude other benign and malignant pathologies that may present in a similar manner. Similarly, in the presence of cervical masses with progressive growth and respiratory symptomatology, imminent risk of airway involvement should be suspected, so early imaging studies should be the mainstay of diagnosis and treatment of these patients. In case of suspected and diagnosed cystic lymphatic malformation, sclerotherapy has proven to be an effective treatment.

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