Case Report

Acute life-threatening presentation of unknown lymphatic malformation

Abstract

Lymphatic malformations (LMs) are rare congenital vascular malformations. Lymphatic malformations include a wide variety of diseases, such as lymphangiomatosis, macro or microcystic lymphatic malformation, and lymphangiectasia. Lymphatic malformations are often first seen at birth, although intrauterine diagnosis is not uncommon. Depending on their size and location, they can be asymptomatic or threaten life by compression of vital structures. We report 2 children admitted to the emergency department with an acute life-threatening presentation of undiagnosed LM. They both required immediate cardiopulmonary resuscitation. Because acute presentations of LM are rare, proper diagnosis can be difficult. Emergency physicians must be aware that sudden enlargement of an unknown thoracic LM can result in airway compression, cardiac tamponade, or rapidly progressive pleural or pericardial effusion. Prompt diagnosis is essential to avoid further morbidity or mortality. The treatment of extensive forms is challenging, requiring a multidisciplinary approach. The prognosis can be poor.

Lymphatic malformations (LMs) consist of rare and complex congenital disorders of the development of the lymphatic system. Several forms of LMs are described depending on the timing of the arrest in lymphangiogenesis, including micro and macrocystic lymphatic malformation, lymphangiectasia, lymphangiomatosis, and lymphatic dysplasias [1,2]. Although most of LMs are diagnosed in utero or during the first 2 years of age, it can be diagnosed as late as in adulthood. Diagnosis requires recognition of the clinical features, including bone and spleen involvement, analysis of fluid effusion, and imaging studies. The evolution is usually slow, but rapid deterioration can occur after local infection or bleeding into the malformation or accumulation of chyle in a serosal cavity.

A previously healthy 22-month-old boy presented in the emergency department with rapidly progressive respiratory distress associated with a 24-hour history of rhinitis, without fever. On admission, he presented respiratory insufficiency with obnubilation, exhaustion, and hypoxemia despite full flow face mask oxygen. On auscultation, air entry was very poor. There was no evidence of neck swelling. The child required urgent endotracheal intubation, but airway stabilization was complicated by a severe upper airways deviation. In the following hours, a suprasternal and left laterocervical swelling appeared. A magnetic resonance imaging (Fig. 1) revealed a macrocystic lymphatic malformation involving the neck and the upper mediastinum. The child underwent a complete surgical resection with favorable outcome, except for vocal cord paralysis.

A 16-month-old boy was admitted to the emergency department for cardiorespiratory collapse. This child had been previously observed for several episodes of respiratory distress for which the diagnosis of asthma was suggested. Previous investigations, including a chest x-ray, were normal. On admission, the child was in shock; the heart rate was 200 beats per minute, respiratory rate was 40 beats per minute, blood pressure was 75/35 mm Hg, and oxygen saturation as measured by pulse oximetry was 93% on full face mask oxygen. The auscultation was characterized by diffuse and bilateral crepitations. An enlarged spleen was palpable at abdominal examination. Chest x-ray revealed severe

Fig. 1 Macrocytic lymphatic malformation of the neck and upper mediastinum with upper airways compression.
cardiomegaly and diffuse interstitial edema. The cardiac echography confirmed massive pericardial effusion and tamponade. Pericardiocentesis was urgently performed, and 300 mL of chylous fluid were evacuated (15 300 cells/μL, 91% of mature lymphocytes—triglycerides 3.2 mmol/L). Over the next 24 hours, the child developed pleural effusions requiring bilateral chest tube insertion. Magnetic resonance imaging revealed disseminated lymphangiomatosis of the neck, mediastinum, lungs, spleen, mesentery, and thoracic vertebra (Fig. 2). The child’s further evolution worsened. Despite treatment with total parenteral nutrition, steroids, vincristine, and finally, interferon α-2b, massive pleural and pericardial drainage was necessary. Fifty liters of chyle was drained over the next 3 months (child’s weight, 9 kg). The child underwent pleuro-pericardio-peritoneal window. He eventually improved and could be discharged home after a 10-month hospitalization on interferon α-2b and nasogastric feeding (adapted diet with medium-chain triglycerides).

Cystic lymphatic malformations and lymphangiomatosis are embryologic remnants of lymphatic tissue sequestered during embryologic development. They are usually diagnosed during pregnancy or at birth but can occasionally be discovered later during childhood. Acquired lymphocele may develop after lymphatic trauma (eg, surgery or chronic infection). Diagnosis is based on imaging (ultrasound, computed tomographic scan, or magnetic resonance imaging). Accurate diagnosis is essential to assure proper management. The evolution of a cystic lymphatic malformation is usually progressive, with a propensity to grow. Treatment involves surgical excision and/or sclerotherapy [1,2]. Lymphangiomatosis is a rare congenital malformation of the lymphatic system characterized by cystic lymphatic malformation, either microcystic or macrocystic, involving multiple organs. Interstitial lung disease, pleural and/or pericardial effusion, spleen involvement, and lytic bone infiltrations are characteristic, but lesions can occur in any tissue containing lymphatic vessels [3]. Diagnosis is based on imaging and, when possible, biopsy. The prognosis of these disseminated forms is poor, especially those involving the mesentery. Treatment is supportive, including total parenteral nutrition and drainage of the fluid collections. Modulation of epithelial growth factor expression by steroids, vincristine, or interferon has been proposed [4].

Most cases are characterized by a slow chronic evolution. Nevertheless, emergency physicians must be aware that a fulminant new presentation or an acute deterioration of a stable LM may occur, often in relation with a viral infection, a trauma, or an accumulation of chylous fluid. Because of predominant neck and chest localizations, LM may cause life-threatening cardiopulmonary compromise. Management of such disease in the emergency department can be difficult and requires expertise and a multidisciplinary approach.

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Fig. 2  Diffuse lymphangiomatosis involving subcutaneous tissues, lungs, pleura, pericardium, spleen, mesentery, and thoracic vertebra.
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**References**


