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Multimodality treatment of a massive cervicothoracic lymphatic-venous malformation in a 13-year-old boy

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ABSTRACT

We report the multi-modality treatment of a massive cervicothoracic lymphatic-venous malformation (LVM) in a 13-year-old boy that was present at birth as a small cervical mass. At four years of age, sclerotherapy significantly reduced the size of the malformation but over subsequent years the mass increased dramatically in size and resulted in a consumptive coagulopathy and limited left shoulder function. Treatment with sildenafil was without effect. The consumptive coagulopathy improved with subcutaneous enoxaparin. A multi-disciplinary team excised part of the mass when he was 11 years old. After surgery, gradual enlargement of the mass and rise in d-dimer prompted treatment with sirolimus. Two sessions of sclerotherapy arrested enlargement of the mass. Multi-modality treatment resulted in a marked reduction of the LVM in the child. However, enlargement of the mass and further treatment is anticipated as a cure has not yet been found.

1. Introduction

Lymphatic-venous malformations (LVMs) are congenital lesions that occur due to poor communication between the venous and lymphatic circulation and abnormal proliferation of lymphatic vessels during fetal development [1]. These malformations have an incidence of 1 in 6000 births and most commonly present in children under the age of two years [2,5,6]. Lymphatic-venous malformations are associated with several syndromes [2,11] and sporadic mutations. The pathogenesis of these lesions is still not well-understood [16]. Up to 95% of LVMs occur in the neck and the axilla [6]. This report describes an extensive cervicothoracic LVM in an adolescent boy and the multi-modality therapeutic approach used to reduce the mass.

2. Case report

2.1. Presentation

A 9-year-old boy presented to the University of New Mexico Hospital with an extensive cervicothoracic mass. He had mild pain, limited movement of the left shoulder, and occasional cutaneous bleeding and drainage of clear fluid. The mass would increase in size during infections. The mass involved the left side of his face and filled with fluid when he was recumbent (Fig. 1). At initial presentation, he had no complaints of shortness of breath, dyspnea, or dysphagia. On examination, he had massive fluctuant mass with areas, which would compress and then slowly refill (Figs. 3–5). The overlying skin had hemosiderin deposits and small blebs, but no erythema or induration. His cardiopulmonary function was normal but there was tracheal compression from the intrathoracic malformation but no obstructive sleep apnea on polysomnography.

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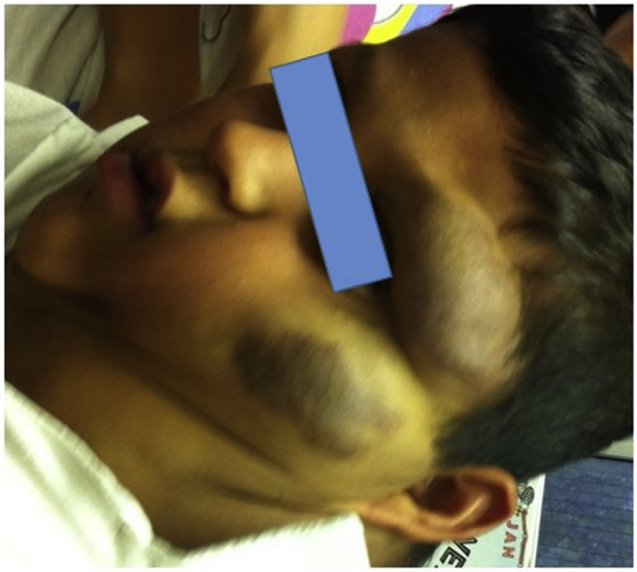

Fig. 1. Prior to treatment, the mass extended along the left side of his face and filled with fluid when he was recumbent.

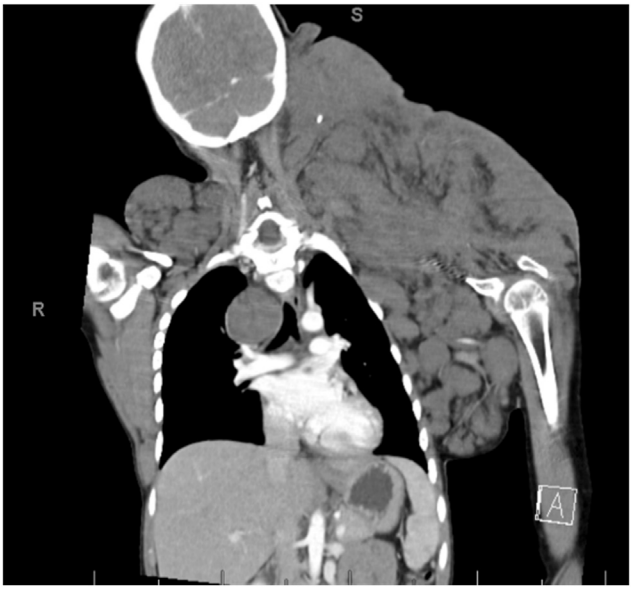

Fig. 2. The massive VLM infiltrated the neck, chest wall, left axilla, mediastinum and compressed the trachea.

2.2. Investigation

Initial laboratory results revealed that the patient had a consumptive coagulopathy due to a venous component of the LVM with a platelet count of 91 (N: 170–380), prothrombin time (PT) of 18.9 (N 11–13 s), partial thromboplastin time (aPTT) of 39 (N 20–30 s), and fibrinogen of <60 mg/dL (N 170–470 mg/dL). In addition, his hemoglobin decreased from 12 to 9 (N: 10–13) and his hematocrit decreased from 37% to 28% (N: 30–40%) in the first four months following presentation. His d-dimer value was also elevated to >20 (N < 0.5). His liver function tests and electrolytes were all within normal limits. Radiologic imaging demonstrated a massive lymphatic-venous malformation involving the neck, chest wall, left upper extremity, left axilla, mediastinum, and compression of the trachea (Fig. 2).

2.3. Treatment

Sildenafil was administered for one year but had no effect. Coagulation studies normalized with enoxaparin. A multidisciplinary team consisting of pediatric surgeons, pediatric otolaryngologists, pediatric radiologists, pediatric anesthesiologists, pediatric oncologists, pediatric dermatologists and pediatric cardiothoracic surgeons participated in planning and performance of a partial excision of the mass from the left face and neck when he was 11 years old. The area of the specimen removed was 21.5 × 11 cm. Additional resection of the tissue was not possible due to extensive intra-operative blood loss. He recovered in the intensive care unit for 9 days for support through massive post-operative facial and neck edema. He was treated for cellulitis and discharged 36 days after surgery.

Pathologic analysis of the mass described abundant malformed channels of fibrotic smooth muscle with vessels extending throughout the lesion. The mass was histologically consistent with predominately venous proliferation in addition to a lymphatic malformation.

Several months after surgery, the mass gradually increased in size and his d-dimer level increased. Sirolimus stabilized the lesion size and improved the coagulation profile. However, sirolimus was stopped after two months when cellulitis recurred. Subsequently, two sessions of sclerotherapy using a mixture of sodium tetradecyl sulfate, lipiodol, air and gelfoam arrested enlargement of the mass.

2.4. Outcome and follow-up

Partial excision, treatment with sirolimus and two sessions of sclerotherapy achieved over 50% reduction in the size of the mass (Figs. 3–5). Recumbent facial swelling diminished significantly and his coagulopathy normalized.

3. Discussion

Massive complex multi-spatial LVMs are fortunately uncommon but morbidity and mortality are high. Masses involving the head and neck, or mediastinum may obstruct the airway or esophagus, impair speech or normal bone growth and result in significant cosmetic deformity. Bleeding and infection are relatively common. A local consumptive coagulopathy associated with vascular malformations (Kassabach-Merritt Syndrome) can result in severe hemorrhage and death [1–5].

No single modality of treatment is universally effective and multi-modal therapy is often necessary.

Surgical excision is no longer considered first-line therapy. Complete excision is often challenging due to growth around vital structures [2,9] and bleeding due to coagulopathy and large vascular channels. The benefit of surgery should be carefully calculated in each case because post-operative mortality is 2–6% and recurrence rate is 27% [2]. In our patient, a surgical approach was chosen when medication failed to produce significant improvement.

Sclerotherapy is commonly used as primary therapy or to shrink LVMs before surgery. Results with sclerotherapy is generally better and carries less risks than surgery [17]. The most commonly used sclerosants are bleomycin, tetracycline, sodium tetradecyl sulfate, inactivated OK-432, and 100% ethanol [1,2]. These irritants induce an inflammatory reaction and fibrosis in the cystic walls of LVM resulting in elimination or size reduction [2,5,10].