

Milroy's Disease Associated with Scrotal Lymphangioma Circumscriptum

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INTRODUCTION

Milroy's Disease (MD) is characterized by peripheral edema of the lower extremities at birth or in early childhood which is due to complete aplasia of dermal lymphatics. Diagnosis of MD can be made easily by imaging the lymphatic channels with radionuclide lymphoscintigraphy or dynamic magnetic resonance lymphangiography. Lymphangioma circumscriptum (LC) is also a benign disorder of lymphatic vascular channels and considered to be a circumscribed developmental disease of lymphatic tissues in the dermis which is characterized by lymphatic cisterns in the subcutaneous tissue and communicate with dilated dermal lymphatics. LC is generally localized at any anatomic site of human body but more frequently affect the chest, axillary folds, shoulders, neck, buccal mucosa, proximal limbs and rarely hips, groins, and genital area. The pathognomonic appearance of LC is multiple translucent or hemorrhagic vesicular lesions with clear leakage. Association of MD with LC is very rare.⁽¹⁾

CASE REPORT

A 13-year-old boy presented with one year history of multiple vesicles with oozing serous fluid on his enlarged scrotum (Figure 1). On physical examination; mild lymphedema of the lower limbs were found. There were multiple, translucent and slightly pinkish papules on the skin of scrotum which were 2 mm to 4 mm in size. His parents emphasized that swelling of left lower limb has been present ever since birth, and at first, only dorsal aspect of his left foot was edematous then the edema has been progressed to upside of his left leg (Figure 2). Biopsy



Figure 1. Multiple vesicles with oozing serous fluid on scrotum.

of papular lesions exhibited papillated epidermal hyperplasia secondary to lymphedema and dilated characteristic of lymphangiectasia which was filled with eosinophilic proteinaceous substance [LC disease] (Figure 3). The patient was searched for etiology of edema of lower extremities. The hematological and biochemical parameters of the patient, urinalysis and urological examination were within normal limits. There was no abnormality in abdominal ultrasonography, and venous-arterial color Doppler ultrasonography of both lower limbs and testicles. But lymphoscintigraphy of lower limbs was confirmed the delaying and extremely impairment of lymphatic flow. So, this patient was diagnosed as MD in the light of his medical history and in the evidence of lymphoscintigraphy. At last, we diagnosed that this case was consistent with MD associated with LC and we treated the scrotal infection with appropriate antibiotic therapy. Scrotal edema was regressed and tenderness was vanished. Conservative treatment without surgical approach was preferred because of mild symptoms of the patient.

DISCUSSION

Lymphatic vascular insufficiency is a widespread problem in the adult population, but rare in the childhood. The lymphatic vessels mediate immune responses in inflammatory disease, whereas dysfunction of the lymphatic drainage leads to lymphedema and infection. Primary lymphedema is a hereditary condition arising from an abnormality of lymphatic de-



Figure 2. Edema has been progressed to upside of left leg.

velopment whereas secondary lymphedema is caused by an extrinsic process; e.g. surgery, radiotherapy, trauma or infection like tuberculosis, filariasis and etc. which damages a previously normal lymphatic system. The primary lymphedema is more common than secondary lymphedema, 97% and 3%, respectively in the pediatric age group. The pediatric primary lymphedema usually involves the lower limbs and genitalia, 91.7% and 4.3%, respectively. Boys are typically affected at birth, and girls most often present during adolescence.⁽²⁾ In one study; prevalence of congenital primary lymphedema under twenty-years of age was reported to be 1-15/100000.⁽³⁾ William Milroy was first published a case with hereditary lymphedema in the year 1892.⁽⁴⁾ MD is characterized by peripheral edema of lower extremities and mostly dorsal aspect of feet at birth or in early childhood which is due to the complete aplasia of dermal lymphatics. MD is an inherited autosomal dominant lymphedema caused by mutation in the gene for vascular endothelial growth factor receptor-3 [VEGFR-3, also known as Fms-related tyrosine kinase 4 (FLT4)]. VEGFR-3 is necessary for the development and functioning of the initial lymphatic system, but we could not have a chance for genetic molecular investigation neither the patient nor his family.^(5,6,7) MD is mostly a life-long disease but does not affect longevity. But MD is chronic condition with negative effects on physical, social and emotional level.⁽⁸⁾

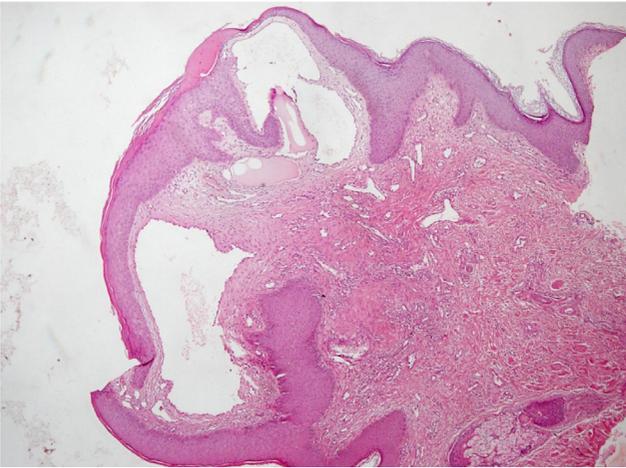


Figure 3. Pathologic examination demonstrates papillated epidermal hyperplasia and lymphangiectasia which has been filled with eosinophilic proteinaceous substance.

Lymphangiomas are rare and benign proliferations of the lymphatic system. Circumscriptum form (or capillary form), cavernous form, and cystic form are the three types of congenital lymphangiomas. LC may be acquired due to injury of lymphatics after inflammations, trauma, infection etc. LC is most common type of lymphangiomas involving skin and subcutaneous tissue. LC is caused by an abnormality of the dermal lymphatics, and lymphedema of skin occurs as a result of lymphostasis. The dilated cutaneous lymphatics are associated with large muscular-coated lymphatic channels deep within the subcutaneous tissue without general lymphatic communication. LC commonly appears at upper part of the body, but rarely in the hips, groins and genital area. LC is characterized by persistent clusters of thin-walled translucent vesicles. These vesicles are varying size, though commonly 2 mm to 4 mm diameter, bright, pinkish hue and usually asymptomatic. But scrotal LC lesions can be bleed into the cysts spontaneously and this hemorrhage causes acute painful swelling of scrotum and mimics acute scrotum of childhood. Genital LC sometimes presents as verrucous papules that mimicking warts especially in adult patients, although very rare in childhood. The diagnosis of LC is usually made by means of biopsy. Hemangioma, melanoma, lymphangiectasia, lymphangiosarcoma, maculopapular herpetic rash, and carcinoma telangiecteticum all should be thought in the differential diagnosis of LC. Treatment of patients with MD and LC is primarily directed against the prevention of

infection. Elevation of extremities and elastic bandages application diminish the lymphedema of MD. But there is no definitive medication or prevention of MD. LC disease is primarily treated with adequate surgical excision of affected region. The CO₂ laser, electrocautery, cryotherapy and sclerosants can also be effective in LC treatment.⁽⁹⁾

Here in, we presented a MD associated with LC disease as the second child case in current medical literature.⁽¹⁰⁾ MD should be kept in mind when LC disease was diagnosed in child's genital area associated with lymphedema of lower limbs. This case is important for dermatologists, pediatricians, cardiovascular surgeons and urologists because of a thorough diagnosis of the lymphedema and for treating the complications of MD and LC. Differentiating the mimicking diseases and avoiding the mistreatment as a consequence of misdiagnosis of the lymphedema will be possible in the child patient with the light of this rare case report.

CONFLICT OF INTEREST

None declared.

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