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Case Report

Idiopathic Congenital lymphedema in A 9-Year-Old Girl; Diagnosis, and Management in A Limited Resource Setting: A Case Report

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Abstract

Congenital idiopathic lymphoedema is a comparatively uncommon disorder characterized by abnormal lymphatic development resulting in chronic lymphatic dysfunction and subsequent localized swelling (lymphedema). The condition's etiology remains largely unknown, adding to the complexity of diagnosis and management. In resource-limited settings, where access to specialized medical facilities and comprehensive healthcare resources is limited, these challenges become particularly daunting. This case report presents the experiences encountered while diagnosing and managing a 9-year-old girl with congenital lymphedema of the left lower limb in such a setting.

Keywords: Congenital; Lymphedema; Resource-limited settings

Introduction

Lymphedema of the limbs is the consequence of a dysfunction of the lymphatic system responsible for a stasis of the lymph and incidentally an increase in volume of the affected limb [1]. Congenital lymphedema is the rarest form of primary lymphedema, accounting for approximately 1:60,000 live births. Congenital lymphedema can be further classified into familial (hereditary) and idiopathic (non-hereditary) subgroups [2]. When lymphedema occurs without any known etiology, dysmorphic features, and no family history of lymphedema, the eponym idiopathic congenital lymphedema is utilized [3].

Patients with congenital idiopathic lymphoedema face additional complications related to infectious manifestations. These infections can exacerbate the patient's symptoms, leading to impaired mobility, chronic pain, and recurrent hospitalizations. This case report aims to shed light on diagnosing and managing idiopathic congenital lymphedema and its infectious complications in a 9-year-old girl. This case is reported according to the SCARE guideline [4].

Case Presentation

A 9 years old female patient presented with swelling of the right lower limb since birth. She had also a fever and a ruptured blister on the swollen limb.

Initially, at birth, the swelling was confined to the foot and then it was gradually progressing with involvement of her right leg and thigh which was grossly edematous. She also had chronic dermatitis of the legs. She had moderate pain during walking and standing. She is the fourth child of non-consanguineous parents. There was no known family history of limb swelling. She was delivered normally at 40 weeks gestation to a 24 years mother whose first prenatal visit was in the second trimester. The mother took all supplements; there were no complications at delivery. APGARs are unknown but mom says that the baby did breathe spontaneously at birth and doesn't require any respiratory support or phototherapy. Her birth weight was 3,000 gm. She had normal growth and development, and she Immunizations are up to date.

Her physical examination revealed non-pitting edema of the right foot, right leg, and right thigh (Figure 1). The dorsum of her right foot and the leg had a Grade III lymphedema with skin fibrosis and the right thigh had Grade I lymphedema. We noticed also a positive Stemmer's sign. The circumferences in different regions on the left foot and leg were 2 cm to 10 cm greater than the corresponding regions on the right. She had no dysmorphic features. There was no localized overgrowth of bone and no varicose veins. Her nails were normal. Multiple localized excoriations and disrupt blisters with serosanguineous discharge were noticed on the concerning limb. There was also a 1 cm inguinal lymphadenopathy that was mobile and tender.

Renal and liver function tests revealed normal findings. FBC shows a discrete lymphocytosis. The ESR was 50 mm/1h (ref value: <12 mm/1 h) and the Quantitative CRP was 1193.000 ng/mL (ref value: 0.000 ng/mL-700.000 ng/mL). Doppler ultrasonography of affected limbs, as well as pelvic and abdominal ultrasonography to exclude deep vein thrombosis or any obstruction, was found normal.

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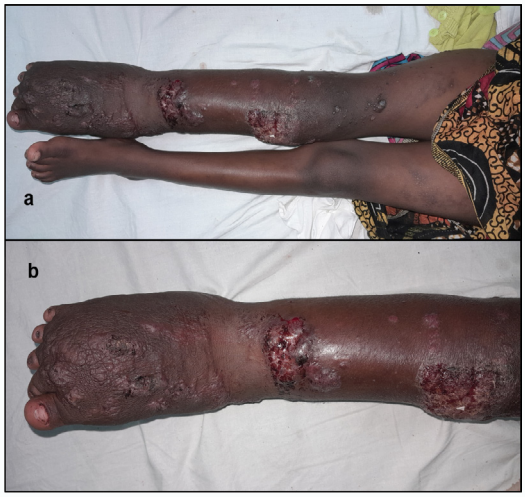

Figure 1: Lymphoedema of the right lower limb. (a) comparative view with the contralateral limb. (b) View of the affected lower limb.

We were unable to do the genetic tests or the lymphoscintigraphy because of their unavailability in our center. Based on clinical and anamnestic elements, we retained the diagnosis of sepsis secondary to cellulitis complicating primary congenital lymphedema. The patient was admitted to the pediatrics surgery ward and commenced on IV ceftriaxone (500 mg BD for 5 days), IV metronidazole (250 mg TDS for 5 days), and IV paracetamol (300 mg TDS for 3 days); limb elevation, manual foot massage along with skincare and daily dressing.

On day 3 post-admission, the fever had completely subsided and the patient's general condition had improved. On day 5 after completing her antibiotic treatment, the patient was discharged. The mother was advised to avoid prolonged standing of the child and use compressing stockings. The lymphedema did not show any progression in the next three months of follow-up.

Discussion

Idiopathic primary lymphedema is a rare disorder causing persistent swelling in an extremity due to impaired lymphatic drainage. The current concept regarding 'primary' lymphoedema accepts its definition as a clinical manifestation of a lymphatic malformation developed during the later stage of lymphangiogenesis [5,6].

Our patient was female and 10 years old at diagnosis. In 1985, Smelzer et al. [2] noted an annual French incidence of 1.15 cases per million inhabitants aged under 20 with a clear female predominance and Baulieu et al. [7] a sex ratio of 1, with an average age at diagnosis of 9 years; 1/4 of cases before 2 years.

Studies have shown cases of idiopathic primary congenital lymphedema without any family history [8,9]. Our patient also had no family history of lymphedema.

The majority of patients with lymphedema can be diagnosed by a thorough history, physical examination, and ultrasound. Lymphatic permeability can be assessed by isotope lymphoscintigraphy, this test is generally considered the gold standard for diagnosing lymphedema because the procedure is minimally invasive, easy to perform, and safe for the lymphatic endothelium [10,11].

Infectious complications are frequently described in advanced forms ranging from erysipelas, pleuropneumonia, digestive infections, and myocarditis. Lymphangiosarcoma can also occur in rare cases [7,12]. Our patient with lymphedema had lesions of cellulitis and sepsis.

Treatment of primary lymphedema is conservative and generally successful in most patients. Avoidance of prolonged standing and elevation of the affected extremity combined with skin care is sufficient for mild, uncomplicated cases of lymphedema. Maneuvers that improve lymphatic drainage, such as compression, special exercises, and manual lymphatic drainage, are also recommended [11]. If medical treatment fails, surgical interventions are indicated as a last option: these include bypass procedures or debulking operation plus or minus skin grafting, to improve lymph flow and to eliminate lymphedema, respectively [2,13].

Conclusion

Idiopathic congenital lymphedema, a condition rarely seen in children, poses physical and psychological challenges to patients. Its diagnosis is usually determined by a thorough clinical evaluation, including a detailed family history and the results of a physical examination. Physical examination plays a crucial role in distinguishing lymphedema from other potential causes of extremity swelling. In cases where the diagnosis remains uncertain, isotope lymphangioscintigraphy may be performed to confirm the presence or absence of lymphatic dysfunction. The approach to the management of primary lymphedema generally involves conservative measures in most cases. However, if conservative methods prove ineffective, surgery may be considered as an alternative option.

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