

Primary Upper Limb Lymphedema: Case Report of a Rare Pathology

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ABSTRACT

Introduction: Lymphedema is characterized by a defect in the lymphatic system that causes limb swelling. Impaired uptake and transport of lymphatic fluid through lymphatic vessels causes accumulation of protein-rich fluid in the interstitial spaces, which leads to swelling of the limb. Primary lymphedema often presents at birth. The rare cases that arise after age 35 years are described as lymphedema tarda. The great majority of patients with lymphedema have swelling of the lower limbs—upper limb lymphedema is a rare disorder.

Case Presentation: An 84-year-old woman presented with a 3-year history of unilateral swelling of the right upper limb. There were no constitutional symptoms and no evidence of lymphadenopathy or systemic disease. Blood tests, carcinoembryonic antigen test, computed tomography scans, and venous Doppler ultrasound were all normal. The diagnosis was primary upper limb lymphedema.

Discussion: The swelling that occurs in upper limb lymphedema is permanent and usually extends to the hand. About one-third of patients with this condition also present with lower limb lymphedema. Thorough investigations are warranted in cases of unilateral upper limb lymphedema to rule out occult malignancy and systemic disease.

INTRODUCTION

Primary upper limb lymphedema is a rare entity. Most reports of lymphedema affecting the extremities refer to disease of the lower limbs.¹ Cases are generally classified as primary or secondary. Primary lymphedema has been described mainly in the lower limbs.² The majority of cases of primary lymphedema are thought to be congenital.² Inheritance has been demonstrated in several cases, and a dominant gene has been identified in some families.^{3,4} Secondary lymphedema is more commonly recognized; typical examples include involvement of the upper limb following axillary dissection with or without radiotherapy for the treatment of breast cancer.⁵ Proximal or distal hypoplasia of lymphatics or disorderly hyperplasia has been described.⁶ Although primary lymphedema more commonly appears early in life, reviews of the literature have described later presentations of congenital lymphedema.⁷ Lymphedema occurring after age 35 years is sometimes referred to as lymphedema tarda.

Detailed clinical evaluation is necessary to exclude secondary causes of lymphedema. Lymphatic mapping with lymphoscintigraphy is frequently performed

to confirm the diagnosis but may not be necessary in the majority of cases once secondary causes of lymphedema are excluded. This report describes the case of an 84-year-old patient with isolated upper limb lymphedema.

CASE PRESENTATION

An 84-year-old woman presented to our clinic with a 3-year history of swelling of the right upper limb. There was no associated pain or constitutional signs. The patient's history was unremarkable, and she reported no similar family history. Apart from moderate swelling of the entire right upper limb (Figures 1 and 2), findings from physical examination were normal. She was in no apparent distress. Her mucous membranes were pink and moist, and her vital signs were normal.

Examination of the right upper limb revealed nontender, nonpitting edema. The



Figure 1. Photograph of dorsal aspect of both upper limbs showing swelling of the right side.



Figure 2. Photograph of the volar aspect of both upper limbs showing swelling of the right side.

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temperature of the limb was normal, and there was no evidence of localized cellulitis. All pulses were palpable. The superficial venous system was normal. There were no abnormal findings on examination of the breasts and axillae and no evidence of generalized lymphadenopathy. A provisional diagnosis of primary upper limb lymphedema was made. A full profile of blood tests was completed, including hematology, electrolytes, liver function, and carcinoembryonic antigen tests. Chest x-rays, contrast computed tomography scans of the chest and abdomen in both arterial and venous phases, and an upper limb venous Doppler ultrasound were also completed. All results were normal. Mammography findings were normal. Because the swelling was asymptomatic, the patient declined further investigation or treatment. See Figure 3 for a timeline of the case.

DISCUSSION

Lymphedema results from accumulation of protein-rich fluid in the interstitial spaces, which is caused by a large volume of lymph that exceeds lymphatic system transport capabilities. Normal drainage of lymph in the upper limbs occurs via thin-walled lymphatic channels, which drain to axillary nodes. Primary lymphedema may be caused by aplasia or hypoplasia of lymphatics or by valvular disease.

Primary lymphedema has been classified into 3 types on the basis of age at onset. Lymphedema presenting at birth is called congenital lymphedema (Milroy disease); from birth to age 35 years, lymphedema praecox (Meige disease); and after age 35 years, lymphedema tarda.^{8,9} Primary lymphedema has also been associated with a number of genetic syndromes and cutaneous disorders,^{10,11} including Turner syndrome, Klinefelter syndrome, and neurofibromatosis type 1. Lymphedema has been found in multiple sites in the body, including the lower limbs, trunk, genitalia, head, and neck—even involvement of internal organs has been observed.¹² However, primary lymphedema has mainly been described as affecting the lower limbs.

Secondary lymphedema, which is most common in the lower limbs, results from lymphatic obstruction caused by a pathologic process such as malignancy, burn, radiation, trauma, or surgical resection.

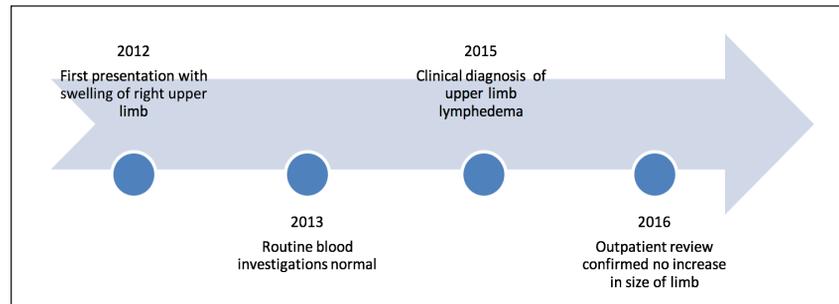


Figure 3. Case timeline.

Lymphatic filariasis, a mosquito-borne infection caused by the parasite *Wuchereria bancrofti*, mainly involves the lower limbs and is the most common form of secondary lymphedema worldwide.¹³

Reports of primary lymphedema involving the upper limbs are rare. Most cases of upper limb lymphedema in the literature resulted from surgical lymph node resection for cancer, malignant infiltration, radiation, or trauma.^{14,15}

In women, deep vein thrombosis is associated with malignancy and is a common cause of unilateral limb swelling. Because of this association, malignancy should be considered in all cases of unilateral limb swelling.¹⁶ In a large review of 650 patients with primary and secondary lymphedema, 65 patients were found to have a malignant lesion.¹⁷ Ovarian carcinoma and uterine carcinoma were the most common malignancies.^{10,17}

Vignes et al,¹⁸ in a review of 60 patients with primary upper limb lymphedema, found similar incidence in males and females, with a mean age at onset of 38.5 years (range 3–82 years). The hand was affected in all patients, the forearm in 55%, and the upper arm in 23%.¹⁸ Twenty-one patients (35%) had associated lower limb lymphedema. Patients were followed for a median period of 103 months, and lymphedema was stable in 95%.¹⁸

Lymphoscintigraphy has now superseded lymphangiography as the choice means of confirming the diagnosis of lymphedema.¹⁹ Computed tomography and magnetic resonance imaging are very helpful in outlining nodal architecture and have the added benefit of being able to rule out systemic disease. In our case, a contrast computed tomography scan was performed and excluded the presence of systemic disease.

Numerous methods for treating lymphedema have been reported in the literature.²⁰ Many of these methods are supportive rather than offering a definitive cure, and the results have been generally unpredictable. Conservative treatments have been tried, with variable results, and include elevation, intermittent pneumatic compression, gradient compression stockings, pumps, physiotherapy, and massage.^{21,22} Special techniques referred to as “complete decongestive physiotherapy” have also had promising results.^{23,24} This technique has gained popularity in Europe and includes multilayer compression bandaging, massage, and long-term use of compression garments with graded exercises.²⁵ Consistently good results have been reported when this technique is administered by skilled practitioners.²⁶

Primary lymphedema of the upper limb is a rare phenomenon. The majority of cases are unilateral, and swelling is usually stable and involves the hand. About one-third of patients with primary lymphedema present with involvement of both upper and lower limbs. Thorough investigation is warranted in cases of unilateral upper limb lymphedema to rule out occult malignancy and systemic disease. ❖

Disclosure Statement

The author(s) have no conflicts of interest to disclose.

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