EUR J PHYS REHABIL MED 2008;44:89-91

Congenital lymphedema of the upper extremity.

A case report

Y.-J. SIM [1], J.-H. SEO [2]

[1]Department of Physical Medicine and Rehabilitation
Kosin University College of Medicine, Busan
Republic of Korea
[2]Department of Physical Medicine and Rehabilitation
Chonbuk National University Medical School, Jeanju
Republic of Korea

This study presents an unusual case of primary lymphedema of the upper extremity in a healthy 28-year-old woman. The onset of swelling of the left upper extremity was observed at birth, but was not accurately diagnosed until the patient visited our department. Diagnostic assessment included evaluating the patient's history and laboratory and radiological data, which were all normal except for the swollen upper extremity and the lymphoscintigraphy findings. The patient was diagnosed as suffering from primary lymphedema of her left arm. Complete decongestive therapy was done and her swelling mildly improved.

KEY WORD: Lymphedema - Upper extremity - Rehabilitation.

Congenital lymphedema is clinically evident at birth or within the first two years of life. Some patients with congenital lymphedema have a familial pattern of inheritance, known as Milroy's disease. Primary lymphedema almost exclusively affects the lower extremity (unilateral and bilateral) and it mostly involves females. However, the Authors encountered an unusual case of a 28-year-old woman who had congenital lymphedema of the left upper extremity.

Received on June 20, 2007.
Accepted for publication on December 10, 2007.

Address reprint requests to: J.-H. Seo, Associate Professor and Chair Person, Dept. of Physical Medicine & Rehabilitation, Chonbuk National University Medical School, San 2-20, Keumam-dong, Dukjin-ku, Chonju Chollabuk-do, Republic of Korea.
E-mail: vivaseo@chonbuk.ac.kr

Case report

A 28-year-old female patient was referred to the department with swelling of the left upper extremity (Figure 1A); this swelling started at birth, but had been inaccurately diagnosed. The swelling became more severe after puberty. In childhood, during a visit to a general physician, the patient underwent x-ray and examination, during which congenital lymphedema was not diagnosed; thus, she was not given any definite diagnosis or treatment for her symptoms. However, she later visited the department for evaluation and treatment.

The patient did not have a specific familial pattern of inheritance, a noteworthy past medical history, or any history of infection. Upon physical examination, there was non-pitting, non-erythematous tender edema of the hand, the forearm and the arm of the left upper extremity. Physical examination did not reveal any pathologic findings. The patient had no evidence of pathological reflexes, and displayed normal motor and sensory systems. The circumference of her upper extremity was measured 10 cm above the elbow, at the elbow, 10 cm below the elbow, the wrist and bilaterally at the metacarpophalangeal joint (Table I).

Laboratory findings, including a complete blood count, sedimentation rate, urinalysis findings, liver function tests (including the serum albumin concentration), renal tests, thyroid function tests and rheumatoid factors, were unremarkable.

Vascular computed tomography (CT) of the left upper extremity did not reveal any pathologic findings. A Tc-99m HS Albumin lymphoscintigraphy revealed the absence of flow at the left upper extremity and the left axillary lymph nodes (Figure 1B).

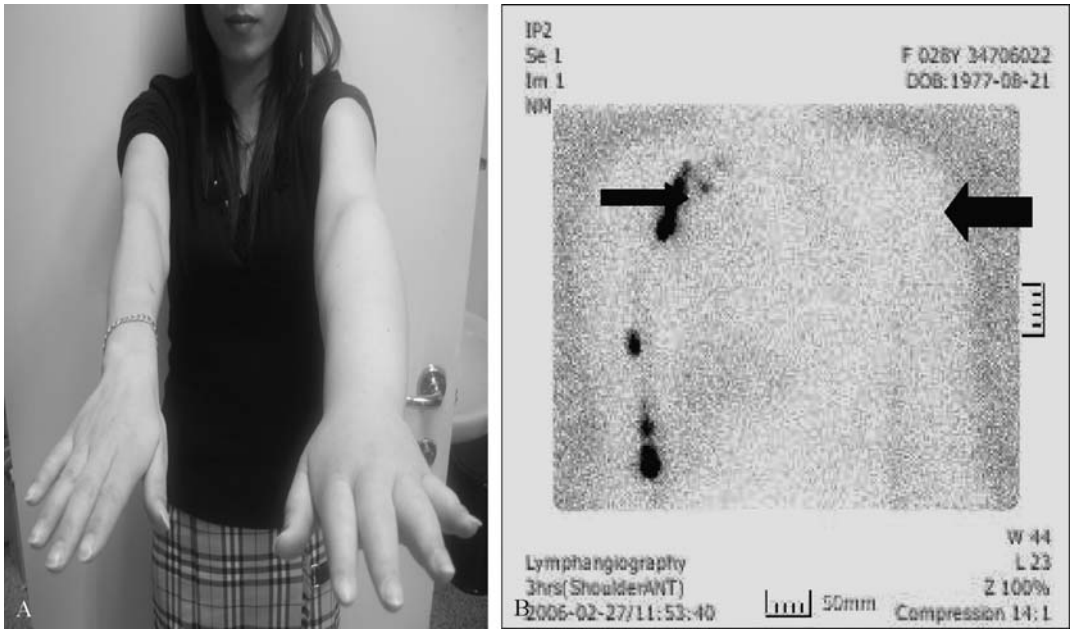

Figure 1.—A) Photograph of the patient's left upper extremity at the time of her initial visit to our department. Swelling can be observed on the left upper extremity. B) Upper extremity lymphoscintigraphy obtained three hours after injection of radionuclide reveals markedly delayed and diminished flow in the left upper extremity, with no lymphatic trunks and axillary lymph nodes imaged (thick arrow), compared with the uptake on the right side (thin arrow).

The abovementioned diagnostic assessment led to the diagnosis of primary lymphedema, and medication with *vitis vinifera* (Entelon,)[1] and complete decongestive therapy (CDT) were prescribed. Such therapy consists of skin care treatments, manual lymph drainage, a compression bandage and daily exercise for eight weeks. This treatment was done by a physical therapist for the first two weeks and then by the patient herself for six weeks.

During the treatment with CDT, the circumference of the left upper extremity was checked with measuring tape. The patient showed reduced circumferences (see Table I), and the skin and subcutaneous tissue of the left upper extremity became smoother and softer.

Discussion

Lymphedema is a very common and serious disease affecting about three million people in the USA. It occurs if the transport capacity of the lymphatic system has fallen below the normal volume of the lymphatic load, causing abnormal water and protein accumulation, mainly in the subcutaneous tissues. However, as reported in one study, the protein concentrations in the lymphedema fluid from the edematous arm are significantly lower than in the fluid from the non-swollen arm.[2] Lymphedema may be present in the extremities, trunk, abdomen, head and neck, external genitalia and inner organs; according to the etiology, it is divided into two groups: 1) primary lymphedema, when no definite etiology can be found, and 2) secondary lymphedema, when the etiology is definite. Common causes for secondary lymphedema include surgery and radiation, trauma, infection, malignant tumors, immobility and chronic venous insufficiencies, whereas primary lymphedema represents a developmental abnormality of the lymphatic system, either congenital or hereditary. Although developmental abnormalities are present at birth, lymphedema may also develop later in life. Congenital lymphedema is clinically evident in the newborn. Primary lymphedema is often classified according to the age of the patient at the onset of swelling. If primary lymphedema presents after birth but before the age of 35, it is called *lymphedema praecox*, whereas *lymphedema tarda* is relatively rare and develops after the age of 35.[3]

Primary lymphedema has only been reported in the medical literature with reference to those cases involving swellings of the lower extremities[4-6] or the

TABLE I.—*Circumference of the upper extremities before and after treatment.*

Laterality sites	Before treatment		After treatment	
	Affected site	Unaffected site	Affected site	Unaffected site
MCP* joint	23	18.2	23	18.2
Wrist	20	15	19	15
Below 10 cm to elbow	29.5	21	28	21
Elbow	29.5	21.4	28	21.5
Above 10 cm to elbow	29.2	22.8	27.8	22.9

*MCP: metacarpophalangeal.

genitalia. Primary lymphedema of the upper extremity has rarely been reported in the medical literature. Most of the lymphedema patients described in medical articles with upper extremity swelling are cases of secondary lymphedema caused by cancer, radiation therapy or traumatic lesion.[7, 8]

It is not known why primary lymphedema is more common in the lower extremity than the upper. Genetic or congenital factors, as well as the gravity effect, may help explain the more common involvement of the lower extremity.

Lymphedema has recently gained interest for physicians as it causes pain and discomfort as well as a serious worsening in quality of life. There are only a few experts who can diagnose and properly treat this disease. Until recently, and especially in Korea, patients affected by lymphedema were not given any information about reasons and treatments for the swelling of their extremities. Treatments for lymphedema include CDT, pneumatic compression pump, some kinds of medications and surgery. Of these, CDT is considered the best therapy for the vast majority of patients suffering from primary and secondary lymphedema;[3] in the present study, the Authors used CDT and medication. Standard medications include diuretics and benzopyrones, together with *vitis vinifera*. Most experts agree that the use of diuretics in managing uncomplicated lymphedema is ineffective and may lead to worsening of symptoms. Benzopyrones include flavonoids, and have been shown to stimulate macrophage activity and promote the breakdown of proteins in the lymph fluid.

Conclusions

Early diagnosis and treatment are important for lymphedema, as these can effectively prevent the progression of the disease. However, if diagnosis is delayed, as in the case reported here, resulting fibrosis of the lymphatic system can prevent effective treatment. Even though the Authors tried to reduce the lymphedema, the effect of treatment was not complete. Early diagnosis and treatment are recommended, especially for patients who show swelling of the unilateral upper extremity.

References

1. Doutremepuich JD, Barbier A, Laecheretz B. Effect of Endotelon (procyanidolic oligomers) on experimental acute lymphedema of the rat hindlimb. Lymphology 1991;24:135-9.
2. Bates DO, Levick JR, Mortimer PS. Change in macromolecular composition of interstitial fluid from swollen arms after breast cancer treatment, its implications. Clin Sci (Lond) 1993:85:737-46.
3. Zuther JE. Pathology. In: Zuther JE. Lymphdema management. The comprehensive guide for practitioners. New York: Thieme; 2005. p. 46-73.
4. Prpic I, Hartl-Prpic V, Montani G, Vladovic T. Lymphangiographic studies of primary lymphedema. Plast Reconstr Surg 1969;44:383-6.
5. Tilley AR, Douglas LG. Staged treatment of lymphedema praecox. Can Med Assoc J 1974;110:309-12.
6. Wolfe JHN, Kinmoth JB. The prognosis of primary lymphedema of the lower limbs. Arch Surg 1981;116:1157-60.
7. Weissleder H, Weissleder R. Lymphedema: evaluation of qualitative and quantitative lymphoscintigraphy in 238 patients. Radiology1988;167:729-35.
8. Gloviczki P, Fisher J, Hollier LH, Pairolero PC, Schirger A, Wahne HW. Microsurgical lymphovenous anastomosis for treatment of lymphedema: critical review. J Vas Surg 1988;7:647-52.