Case Report

Primary Congenital Lymphedema Involving All Limbs and Genitalia

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Primary congenital lymphedema is the rarest form of primary lymphedema. Lymphedema of the extremities presents at birth and rarely involves the genitalia. There has never been a reported case in Thailand. The authors herby report a case of a 6-year-old boy who presented with progressive swelling of the lower legs since birth. The edema progressed into his scrotum and his arms. There was no history of lymphangitis or cellulites. Physical examination revealed a generalized non-pitting edema of all extremities, more on the right leg than the left leg. Swelling of the scrotum and penis was also detected. A diagnosis of primary congenital lymphedema was confirmed with lymphoscintigraphy.

Keywords: *Primary congenital lymphedema, Scrotal edema, Lymphoscintigraphy*

J Med Assoc Thai 2005; 88 (12): 1958-61
Full text. e-Journal: *http://www.medassocthai.org/journal*

Lymphedema is defined as a swelling of a part of the body caused primarily by an imbalance between the inflow and the removal of interstitial fluid and protein, and secondary by a malformation or malfunction of the lymphatic system[1]. It was classified into primary and secondary forms[2].

Primary lymphedema is defined as lymphedema that presents at birth or appears in childhood. Primary lymphedema is more common in childhood[2]. The secondary lymphedema is caused by obstruction of lymphatic flow following infection, surgical excision, neoplasm, irradiation, or trauma.

To the authors' knowledge, this is the first case report of primary congenital lymphedema involving all limbs and genitalia in Thailand.

Case Report

A 6-year-old boy presented with swelling of his lower limbs since birth, more on the right foot. At the age of one, he began to have edema of his forearms. The swelling gradually progressed proximally. During the clinical examination, the edema was found on both thighs and scrotum. He also had chronic dermatitis of the arms and legs.

He did not complain of any pain in his legs after walking long distances or standing for a long period of time. He had no history of lymphangitis or cellulites.

The patient was a single child. He was delivered by caesarean section, because of a malalignment of the fetus. His birth weight was 3,900 grams. He had a normal growth and development. There was no history of consanguineous marriage and no known family history of limb swelling.

His physical examination revealed a generalized non-pitting edema of all extremities, the right leg more than the left. There was no localized overgrowth of bone and no varicose veins. His nails were normal. Multiple localized discrete hyperpigmented macules and excoriation marks over the arms and legs, were noticed. He also had swelling of the scrotum and penis (Fig.1). There was no evidence of infection.

The diagnosis of primary lymphedema was confirmed by lymphatic scintigraphy. The findings revealed no ascent of tracer into the lymphatic vessels of the right lower extremity and both arms (Fig. 2). On follow-up the swelling has not progressed for one year.

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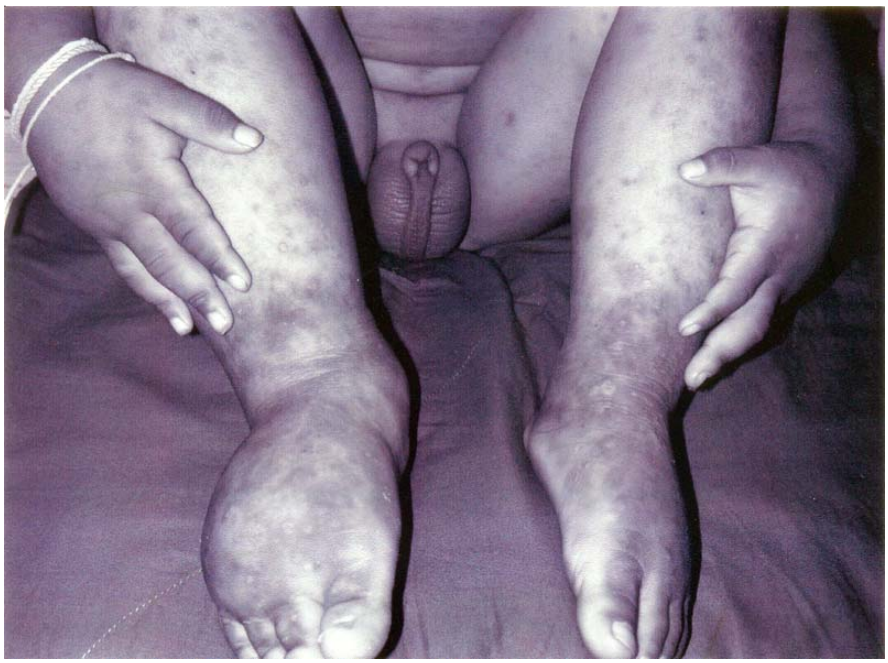

Fig. 1 A generalized non-pitting edema of all extremities, the right leg is more involved than the left. Multiple localized discrete hyperpigmented macules of insect bite reaction are also noticed. The patients also suffer from swollen scrotum and penis

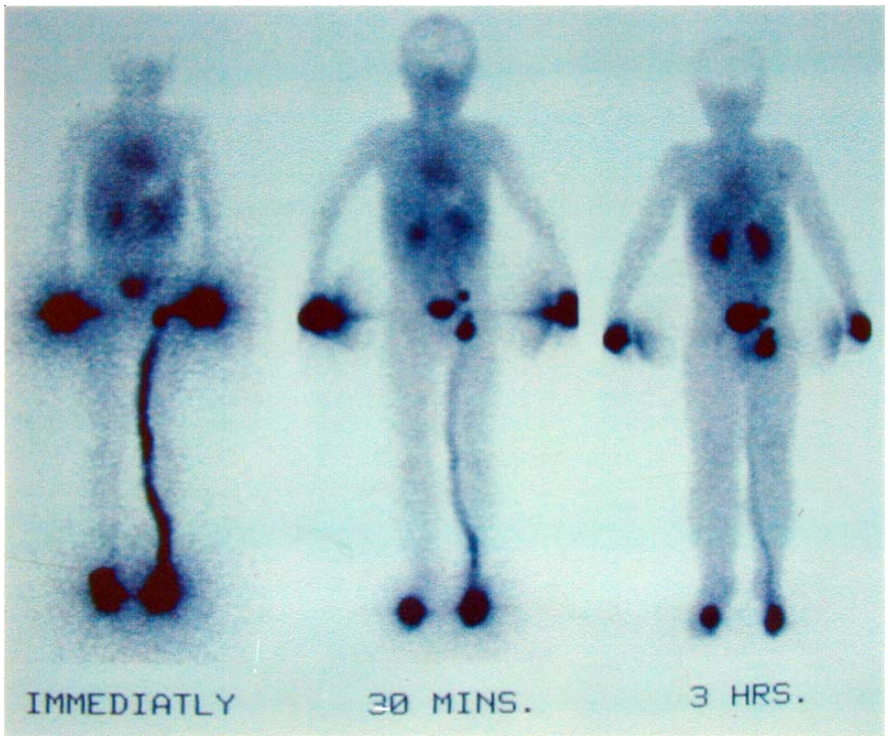

Fig. 2 Lymphangioscintigraphic findings show no ascent of tracer into the lymphatic vessels of the right lower extremity and both arms

Discussion

Lymphedema can either be primary or secondary. In childhood, primary lymphedema is more common[2]. Congenital lymphedema is found in patients with edema present at birth or shortly afterwards. Primary lymphedema is estimated to occur in about 1.15 in 100,000 persons who are less than 20 years old with a sex ratio of about one male to three females. It is the rarest form of primary lymphedema, accounting for approximately 1:60,000 live births[1]. Generally it involves more than one limb, but rarely the genitalia[3,4]. Inherited primary congenital lymphedema gene has been mapped to the 5q35.3 region[5] with code for vascular endothelial growth factor receptor 3 (VEGFR-3)[6]. The rare associated anomalies include microcephaly, distichiasis, extradural cysts, intestinal lymphangiectasia, protein-losing enteropathy, cardiac anomalies and gonadal dysgenesis[1,2]. Lymphedema has also been associated with several genetic syndromes[7-10] including Noonan's syndrome, distichiasis-lymphedema syndrome, Aagenaes' syndrome, yellow nail syndrome, and Turner's syndrome. Here, the patient had swelling of the lower limbs, without any dysmorphic feature and no family history of lymphangioma.

The diagnosis of congenital primary lymphedema is made from a carefully taken clinical history and findings in the physical examination[1]. Since 2002, the condition could be detected prenatally via fetal ultrasonography. Radionuclide lymphoscintigraphy is a useful technique for differentiating lymphedema from other causes of swelling, and it may sometimes be useful for delineating collateral lymphatics, the level of obstruction, and the presence of lymphoceles or abnormal collections of lymphatic vessels[12], can be used to follow up the progress of treatment in primary lymphedema. Three dimensional magnetic resonance imaging (MRI) is known to provide more anatomic information about lymphatic malformation and dysfunction than lymphoscintigraphy[13].

The natural history of primary lymphedema classically has been stated to have a slow but constant progression from a mild swelling of an ankle to a swollen extremity. The swelling enlarges at a slower rate than the growth of the body. Firm, non-pitting edema, fibrokeratotic skin, verrucous growths, squaring of the toes, and a tendency toward recurrent attacks of cellulites and lymphangitis are its common manifestations. The normal course of primary lymphedema is not necessarily progressive. The swelling normally remains unchanged in 57% of the patients for up to 27 years. In most patients, a static point is reached after several years of increased swelling and, irrespective of treatment measures, the swelling remains stable[1]. In the presented case, the swelling was on a plateau stage for one year.

For the treatment of primary lymphedema, expectant management by compression and elevation can maintain the status of the swelling; it reduces the incidence of infection, and stops the development of verrucous growths and hyperkeratotic skin[1]. Compression is so far the therapy of choice in a mild to moderate primary lymphedema in children[14,15]. It is associated with a long term maintenance of reduced limb circumference, especially in secondary lymphedema. Jobst-type of compression with high-pressure (up to 50 to 60 mm Hg) stockings is expensive and has poor compliance because of the discomfort when applied[1]. Ace-type elastic bandage is inexpensive, and it is more comfortable but it lacks sufficient pressure to decrease the edema significantly[1]. Pneumatic pumping is a new method of compression. The provided actions are sequential, intermittent, pneumatic compression of the limbs which must be used together with stockings. It is effective and decreased swelling after several hours of use but they are costly and immobile[14,15].

Elevation is routinely and universally recommended[1]. Surgery is still the most controversial treatment. The excision procedures relatively produce good results in reducing the size of the swollen extremities but often leave scarring[16,17]. Improved results can be expected with operations performed during the early stages of lymphedema. Over the past 25 years of follow-up, there was a significant improvement in 83%, 85% of the patients were able to discontinue the use of conservative measures. There was an 87% reduction in the incidence of cellulites[16]. The latest physiological operation uses microsurgery to create lymphaticovenous anastomoses in an attempt to bypass blocks in the lymphatic system. Currently, this is the therapy of choice in patients who are not sufficiently responsive to conservative treatment[17].

In conclusion the authors describe here a case of primary and progressive congenital lymphedema that was brought to a stable course after the age of 6 years. Conservative treatment with nighttime elevation is recommended during the stable condition. Surgical treatment should be performed in patients whose conservative measures have failed.

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ภาวะบวมจากน้ำเหลืองคั่งแต่กำเนิดที่แขนขาและอวัยวะเพศโดยไม่พบร่วมกับความผิดปกติอื่น

ศิริวรรณ วนานุกูล, ศานตี จิตติถาวร

ภาวะบวมจากน้ำเหลืองคั่งแต่กำเนิดที่ไม่พบร่วมกับความผิดปกติอื่น เป็นภาวะที่พบได้น้อยมากในกลุ่มภาวะบวมจากน้ำเหลืองคั่งซึ่งมักพบความผิดปกติหรือจัดเป็นกลุ่มความผิดปกติทางโครโมโซม ภาวะน้ำเหลืองคั่งแต่กำเนิดที่ไม่พบร่วมกับความผิดปกติอื่นนี้จะพบการบวมที่แขนขาตั้งแต่เกิดแต่มักไม่เป็นที่บริเวณอวัยวะเพศ

เนื่องจากยังไม่มีรายงานผู้ป่วยที่มีภาวะบวมจากน้ำเหลืองคั่งแต่กำเนิดในประเทศไทย จึงได้รายงานผู้ป่วยเด็กชายไทย 1 ราย อายุ 6 ปี ที่มีการบวมจากน้ำเหลืองคั่งตั้งแต่แรกเกิดที่แขนขา และเป็นมากขึ้นจนบวมที่อวัยวะเพศด้วย ผู้ป่วยไม่เคยมีการอักเสบของผิวหนังชั้นลึกหรือท่อน้ำเหลืองอักเสบมาก่อน ตรวจร่างกายพบการบวมแบบกดไม่บุ๋มที่แขนและขาโดยบวมมากที่ขาข้างขวาและมีการบวมที่อวัยวะเพศชายด้วย ยืนยันการวินิจฉัยโดยทำการตรวจต่อมน้ำเหลืองวิธี Lymphoscintigraphy