

# Congenital Lymphatic and Venous Abnormality of Leg

DWIJESH KUMAR PANDA



## ABSTRACT

Primary lymphoedema arises from congenital disorders of Veins and Lymphatic Vessels. The diagnosis of congenital oedema leg is confirmed by the absence of Venous and Lymphatic Vessels and by Vascular Doppler and Lymphoscintigraphy. Congenital lymphoedema of leg is defined as lymphoedema that is present at birth. It may be due to defect of the venous and lymphatic systems which results in excessive fluid load at the tissue level. The important evaluations that aid in the diagnosis of lymphoedema are detection of circulating adult filarial antigen, lymphoscintigraphy to identify patient lymphatic vessels and duplex ultrasound of leg to detect patency and competency of venous system. Physiotherapy and compression stocking can control swelling and prevent development of irreversible skin changes. Surgical intervention may help by creating alternate pathways. In this case report, a 35-year-old man reported for treatment of his bilateral oedema leg which was present since childhood. The patient consulted the surgeons and physicians of different super-specialist hospitals of the state. He was treated with antibiotics and analgesics for the last five years without any effect. After that, patient was presented to the author, where the diagnosis of patient was made as Congenital Lymphoedema based on Vascular Doppler and Lymphoscintigraphy.

**Keywords:** Filariasis, Lymphangitis, Lymphoedema, Lymphoscintigraphy, Vascular doppler

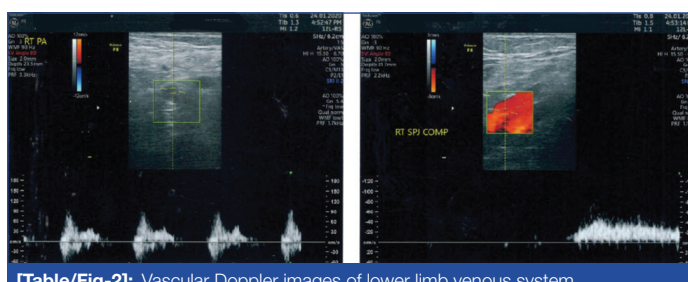
## CASE REPORT

A 35-year-old male reported to Sanaria Clinic and Research center for treatment of his bilateral oedema leg since childhood [Table/Fig-1]. The patient consent was obtained for the publication of information and images. Positive family history of patient was not available but there was a history of fever or pain in the body. Patient was treated previously in other hospitals by administering different antibiotics and anti-inflammatories. The consulting physician advised the patient fourth generation Cephalosporins along with analgesics although the CRP level (0.2 mg/dL) was found to be normal (Reference range below 0.6 mg/dL). The patient was non-diabetic and non-hypertensive. However, the effect on swelling was not satisfactory. After that, patient was presented to the author. On examination both the legs were swollen, soft, non-tender and without induration. No pigmentation, blister or ulcer was found on the skin. All haematological and biochemical parameters were within the reference range. The diagnosis of filaria was ruled out by the negative result (positive more than 128 antigens Units AU) of OG<sub>4</sub>C<sub>3</sub>. Duplex Ultrasound determined incompetent patency of venous system. Ultrasound of abdomen and pelvis revealed fatty liver without any other abnormality. The differential diagnosis of bilateral passive oedema is hypothyroid state, Congestive Heart Failure (CHF), renal disease, portal cirrhosis and gross anemia. All these conditions were ruled out by estimation of serum TSH, serum urea and creatinine, liver function tests and blood haemoglobin, the results being within reference range. The 2D-Echo (Echocardiography) was done for CHF. The report showed normal echo study with good LV functioning.

The Vascular Doppler study of the right side venous system revealed incompetent perforator near the ankle, delayed obliteration of Saphenofemoral, Saphenopopliteal junction, with mild dilated superficial veins. The venous Doppler study of both lower limbs presented in [Table/Fig-2] showed no evidence of deep



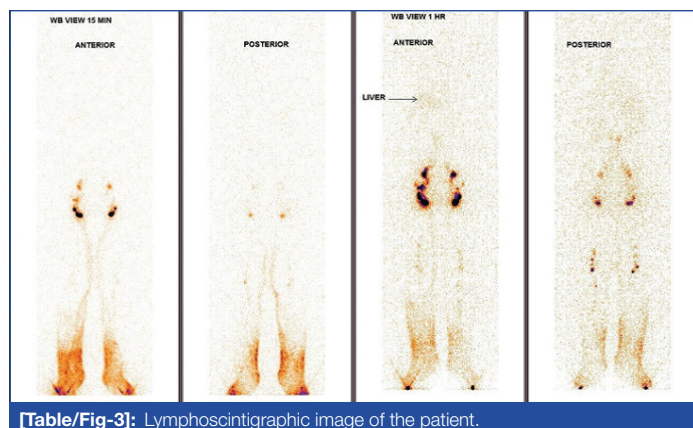
[Table/Fig-1]: Clinical picture of oedema of bilateral lower limb.



[Table/Fig-2]: Vascular Doppler images of lower limb venous system.

vein thrombosis/reflux, mild superficial varicosity in the right side of the medial calf along with diffuse subcutaneous soft tissue swelling in both the side of calves.

The lymphoscintigraphy of the lower limb suggests severe obstruction of bilateral superficial lymphatic of lower limbs. No deep lymphatic obstruction on either side was noted. The whole-body scan showed an increased number of ilioinguinal lymph nodes along with visualisation of the liver, indicates that there is no deep lymphatic system obstruction. The Lymphoscintigraphic image is presented in [Table/Fig-3]. So the final diagnosis was made as congenital lymphoedema. The patient was treated in conservative line involving skin care, weight management, physiotherapy and elastic compression stocking. These procedures could not address the underlying cause of lymphoedema. This could only control the swelling and prevent development of skin changes.



[Table/Fig-3]: Lymphoscintigraphic image of the patient.

## DISCUSSION

The lymphatic system resembles the venous blood system and travel parallel to each other. Primary lymphoedema arises from congenital disorders. The prevalence of primary lymphoedema is estimated at 1.15 in 100,000, for people under the age of 20 years [1]. Lymphoedema, without an inciting factor, termed primary lymphoedema, is generally due to a congenital or inherited condition associated with the pathologic development of the lymphatic vessels. Primary lymphoedema often results from genetic or inherited conditions that impair lymphatic vessel development or function [2].

Lymphoedema is a clinical condition in which there is an accumulation of interstitial fluid. The pathologic collection of fluid may be either a congenital improper development of the lymphatic system (primary) or an acquired condition (secondary) [3]. Venous malformations are slow-flow vascular malformations that occur due to abnormalities in the development of veins. When the venous malformation is more extensive, there may be more widespread swelling of the affected leg and foot. In this case the Stemmer sign was absent. Tenting of the skin over the toe was not possible due to fibrosis. In later stages of lymphoedema of toe, skin thickens, deposition of fat occurs, leading to wart like changes [4]. Most venous malformations do not cause any life-threatening problems. Compression garments provide external support to the affected leg. This reduces the venous swelling in the venous malformation, thus causing the reduction of pain. Regularly wearing a compression garment can be very useful [5]. Limb hypertrophy is often noted at birth and is usually secondary to soft tissue overgrowth, although the presence of venous malformations and lymphoedema may be the contributing factors [6]. The C-reactive protein level measured was within biological reference range. So no antibiotic was administered to the patient. It is an indicator of grade of inflammation [7]. The level of circulating adult filarial antigen of *Wuchereria bancrofti* in serum ( $OG_4C_3$ ) was below biological reference range ( $<128$  AU). This test can be performed at any time. Night blood examination is not required. It may be positive even in amicrofilaremic

individuals [8]. This is a monoclonal antibody-based Enzyme Linked Immunosorbent Assay (ELISA) which gives a quantitative result and detects antigens released by adult filarial worms and microfilaria present in tissue or body fluids [9]. Diagnosis of primary lymphoedema could be made by duplex ultrasound. The patency, competency and reflux of the venous system were assessed correctly [10]. Patients with severe lymphoedema may benefit from Intermittent Pneumatic Compression (IPC). Surgical intervention is effective with appropriate indications. Physiologic procedures (Lympho-venous anastomosis, lymph node transfer) improve lymphatic drainage by creating alternate pathways [11]. It is concluded that unfortunately the compliance of lower extremity lymphoedema is difficult.

Hypothyroidism is a condition of low secretion of Thyroxine Hormone. An elevated TSH level indicates that the thyroid gland is not producing enough thyroid hormone, which results in oedema of legs [9]. Lymphoedema due to infection of *W. bancrofti* is diagnosed by the detection of circulating adult filarial antigen in serum ( $OG_4C_3$  test). It is 98% sensitive and greater than 95% specific for the detection of the parasite's antigen [10], (reference range below 128 antigen units). The typical secondary causes of bilateral oedema legs are lymphatic obstruction (filariasis), chronic venous insufficiency, congestive heart failure, hypothyroidism, renal, and liver failure. All these conditions were ruled out by the investigations cited above. The vascular Doppler study revealed incompetent perforator near the ankle, obliteration of Saphenofemoral, and Saphenopopliteal junction [9].

The lymphoscintigraphy study shows severe obstruction of the bilateral superficial lymphatic system of lower limbs. This indicates that the oedema of the lower extremity of the patient is primary. The C-reactive protein level directly reflects the activity of inflamed tissue. The value was within the reference range. As a result of which there was no need for prescribing antibiotics and anti-inflammatory drugs in the case of primary atresia of lymph-venous vessels. The treatment, in this case, was graded elastic compression stocking. It is contraindicated in peripheral obstructive arterial disease. The Ankle-Brachial Pressure Index (ABPI) must be more than one [11].

## CONCLUSION(S)

Primary oedema of legs should be diagnosed before administration of antibiotics and other medications. Usage of elastic compression stocking can provide relief to the patient. Many pathological conditions may cause bilateral pedal oedema which should be ruled out by appropriate investigations.

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**PARTICULARS OF CONTRIBUTORS:**

1. Lymphologist, Pathologist and Microbiologist, Department of Lymphology, Sanaria Clinic and Research Center, Bhubaneswar, Odisha, India.

**NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:**

Dr. Dwijesh Kumar Panda,  
M5/12, Acharya Vihar, Bhubaneswar, Odisha, India.  
E-mail: doctordwijesh@gmail.com

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