# A rare case of congenital lymphoedema in a 6-month-old infant

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Congenital lymphoedema is a rare disorder presenting in the newborn period and infancy. It is characterised by impaired lymphatic drainage due to lymphatic vessel abnormalities, which presents at birth or within two years thereafter. We report on a 6-month-old infant with congenital lymphoedema presenting with non-pitting unilateral lower-limb swelling and various cutaneous markings. This case underscores the importance of imaging techniques such as lymphangiography and the challenging aspects of management owing to limited interventional options.

**Keywords.** congenital lymphoedema; primary lymphoedema; lymphoscintigraphy

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Primary lymphoedema is a rare genetic disorder of lymphatic development, resulting in dysfunctional lymphangiogenesis and lymphatic vessel function.[1] The condition is considered congenital lymphoedema if present at birth or within 2 years postnatally.[1] The clinical onset varies, typically manifesting as painless swelling of the lower extremities, with most cases presenting in early childhood and predominantly affecting females.[1] Diagnostic challenges arise owing to its similarity to other causes of paediatric limb swelling, including cellulitis, trauma, and deep vein thrombosis.[2,3] Here we present a case of a 6-month-old infant with primary lymphoedema confirmed by lymphoscintigraphy. The importance of early specialised imaging is emphasised.

## Case

A 4-week-old infant weighing 4 100 g was admitted to the neonatal unit for investigation of a unilateral (left) swollen lower limb and bilateral lower limb cutaneous changes, present since birth. The baby was born at term, weighing 3 500 g, to a 32-year-old mother from her third pregnancy, with no known obstetric complications. The mother's previous two children were well, and there was no family history of a similar condition. Ethics approval for this case study was obtained from the University of Pretoria (ref. no. 328/2024).

## Clinical examination

Physical examination on admission revealed non-painful, firm, non-pitting oedema localised to the left leg. Measurements of the lower limbs demonstrated a significant discrepancy between the left and right legs: the mid-thigh circumference measured 19.5 cm (left) v. 17 cm (right), the mid-calf circumference measured 15.5 cm (left) v. 11.5 cm (right), and the mid-foot circumference measured 13.5 cm (left) v. 11 cm (right). Leg lengths were equal on clinical examination.

Fig. 1 (a and b) show the various cutaneous markings that were found on both lower limbs: an erythematous patch (blanching) over the dorsum of the right foot and extending onto the sole of the foot; right shin haemangioma (3 cm × 0.8 cm overall); and livedo reticularis extending from the right hip and thigh region to above the right ankle. On the left leg, there was a Mongolian spot on the knee.

The complete patient examination revealed additional findings of a nevus simplex over the mid-brow area, an umbilical hernia defect of 2 cm, as well as bilateral hypoplasia of the 4th and 5th toenails. No other limbs were affected.

## Management

The patient was managed conservatively. Physiotherapy was performed to enhance manual lymphatic drainage.

### Investigations

Lower-limb sonar and dopplers were normal. Lower-limb long-bone radiographs were normal with no limb length discrepancy. Lymphoscintigraphy findings at 6 weeks of age were as follows: left lower-limb findings equivocal for lymphatic dysfunction; repeat

Figure 1a Figure 1b

*Fig. 1. Cutaneous markings on lower limbs. (a) Erythematous patch over dorsum of the right foot and extending onto the sole of the foot. A right shin haemangioma (3 cm × 0.8 cm overall) can be seen. (b) Livedo reticularis extending from the right hip and thigh region to above the right ankle. On the left leg, there was a Mongolian spot on the knee.*

study recommended; normal right lower-limb lymphatic function (Fig. 2 (a and b)). At 6 months of age, lymphoscintigraphy showed impaired drainage in the left superficial lymphatic channels, with no lymphatic channels visualised in the left lower limb and normal right lower-limb lymphatic channels and drainage (Fig. 2 (c and d)).

### Differential diagnoses

In this case, there are a few considerations for the diagnosis, including Klippel-Trenaunay-Weber syndrome, Milroy disease, Milroy-like lymphoedema, and Meige's disease. Klippel-Trenaunay-Weber syndrome includes limb overgrowth, cutaneous angiomas, venous disease, and lymphoedema.[1] Milroy disease and Milroy-like lymphoedema both have isolated lymphoedema, but it is usually bilateral.[1] Meige's disease also has isolated lymphoedema of the lower extremity.[1]

### Outcome and follow-up

A follow-up examination 4 months after presentation demonstrated a minor enlargement of the haemangioma, the right foot marking was less obvious, and the livedo reticularis had resolved. There was persistent, unilateral non-pitting oedema of the left leg with the following measurements: left mid-thigh circumference 24.5 cm v. 20.8 cm on the right; left mid-calf circumference 18 cm v. 14.5 cm on the right; and the circumferential measurement of the left foot being 16 cm v. 13 cm on the right. Although all measurements of the left limb had increased since presentation, it may be secondary to normal postnatal growth and not necessarily limb overgrowth. Given that the leg lengths remained equal, we think that limb overgrowth is unlikely; however, further follow-up is required. There were no neurological deficits at follow-up.

## Discussion

Lymphoedema can be categorised as either primary or secondary. Primary lymphoedema is a much rarer form and is due to abnormal lymphatic vessel development (aplasia, hypoplasia, dilatation) or malfunction of the lymphatic drainage capacity, with secondary lymphoedema due to lymphatic injury.[1] The reported prevalence of primary lymphoedema is 1.15/100 000 individuals younger than 20 years old.[2] Only one previous case report of congenital lymphoedema has been published in South Africa, and described a case of Milroy's Disease[4] – a form of congenital lymphoedema not associated with cutaneous markings as is found in the case of our patient.

The diagnosis of primary lymphoedema is challenging. The condition usually presents in early childhood as limb swelling and enlargement, for which a wide differential diagnosis exists. Primary lymphoedema was historically sub-classified as either congenital (present at birth or within 2 years after birth), lymphoedema praecox (during puberty or shortly after), and lymphoedema tarda (present after 35 years of age).[1,4] Recent insights into the condition give understanding to the fact that it is highly heterogeneous in its aetiology and clinical presentation. This has subsequently led to this historical classification of the condition being replaced by a more recent and appropriate classification by Connell *et al.*,[5] which considers the sites affected and the presence of associated features, such as vascular malformations, limb overgrowth, as well as systemic lymphatic involvement and family history of lymphoedema. The advent of isotopic lymphoscintigraphy lends itself to assisting the clinician in making a correct diagnosis for this challenging condition.

Isotopic lymphoscintigraphy was the primary mode of diagnostic imaging for the present case, as it has a sensitivity of 96% and a specificity of 100% in the case of lymphatic vessel abnormalities.[3] It is a minimally invasive investigation that involves injecting a particular tracer protein (in our case Technetium-99m) in the distal extremities and observing how it is absorbed along the lymphatic drainage pathway.[3] The tracer protein has a high molecular weight that is detected on the gamma camera as it follows the lymph vessel pathway from the most distal point to either the axillary or inguinal lymphnodes.[3] This investigation is a superlative diagnostic measure, as the radiotracers have the ability to selectively bind to the specific tissues under investigation, thus illuminating even the most minor defects. Repeat studies allow for a comparative assessment to be made irrespective of how the lymphatic channels develop from infancy to 6 months, ensuring the opportunity to assess the presence of impairment in the development of the lymphatic system.

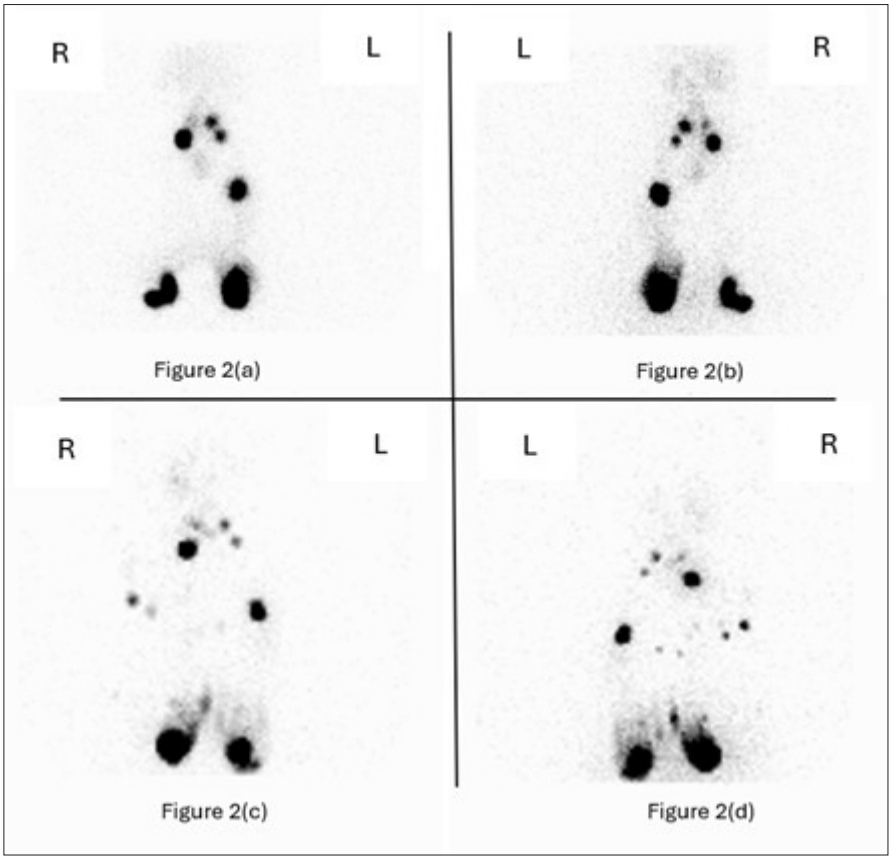


*Fig. 2. (a and b) Lymphoscintigraphy findings in lower limbs at 6-weeks-old showing visualisation of the deep popliteal lymphnodes and less intensely visualised inguinal lymphnodes on the left, which may have been secondary to deep tracer injection.*
*(c and d) Lymphoscintigraphy findings in lower limbs at 6-months-old showing impaired drainage in left lower limb lymphatic channels, with the first lymphnodes on the left seen in the popliteal fossa in keeping with deep rerouting. Additionally, dermal backflow is noted on the left.*

Treatment of primary lymphoedema is limited to physiotherapy and skin care to prevent local complications. This includes exercise and elevation of limbs if affected, massage of the affected area to enhance manual lymphatic drainage, compression therapy with elastic garments/stockings over the affected area, pneumatic compression devices, and optimal skincare (regular moisturising and protective clothing).[1,3] There are no drug treatments available, and surgical intervention is ineffective.[6]

This 6-month-old infant presented with unilateral limb swelling as well as cutaneous lower limb markings. Using the revised classification of Connell *et al.*,[5] as well as the confirmatory lymphoscintigraphy, we can confirm primary lymphoedema with combined vascular malformation/lymphangioma/mixed angioma. The infant was managed conservatively, and follow-up demonstrated persistent lower limb swelling but no skin complications.

## Teaching points

Congenital lymphoedema is a rare cause of localised limb swelling in newborns

- The most recent aetiological approach for congenital lymphoedema employs three main considerations: extent of swelling; cutaneous manifestations; and family history.
- The gold standard of diagnosis involves lymphoscintigraphy.
- The mainstay of treatment is conservative and employs techniques that assist with manual lymph drainage.

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