

# An adolescent with recurrent ankle swelling

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A 14-year-old girl was admitted to our institute with a history of intermittent bilateral ankle swelling, and moderate but progressively worsening pain which has lasted for 2 years.

The patient's history was unremarkable. She did not take medications and was not involved in any sports activity. She reported no fever, gastrointestinal symptoms, fatigue, weight loss, travels abroad or previous infections. She reported moderate pain at night, associated with a sense of heaviness, tightness and general discomfort, and with no response to ibuprofen.

Physical examination was remarkable only for bilateral ankle non-pitting oedema, more evident on the left leg, with a thickened skinfold at the base of the second toe, and without redness, swelling or skin warming.

The patient had been previously examined, and her foot and ankle X-rays, ultrasound (US) and MRI were all negative. Blood tests (white cell count, C reactive protein, erythrocyte sedimentation rate, albumin, anti-nuclear antibodies, creatinine, transaminase, creatine kinase, lactate dehydrogenase, thyroid function and glucose) and urinalysis were in the normal range. Her ocular assessment and echocardiogram were also normal.

## QUESTION 1

Based on the clinical picture and laboratory tests, what is the most likely diagnosis?

- A. Deep venous thrombosis.
- B. Osteochondritis.
- C. Lymphoedema.
- D. Juvenile idiopathic arthritis.

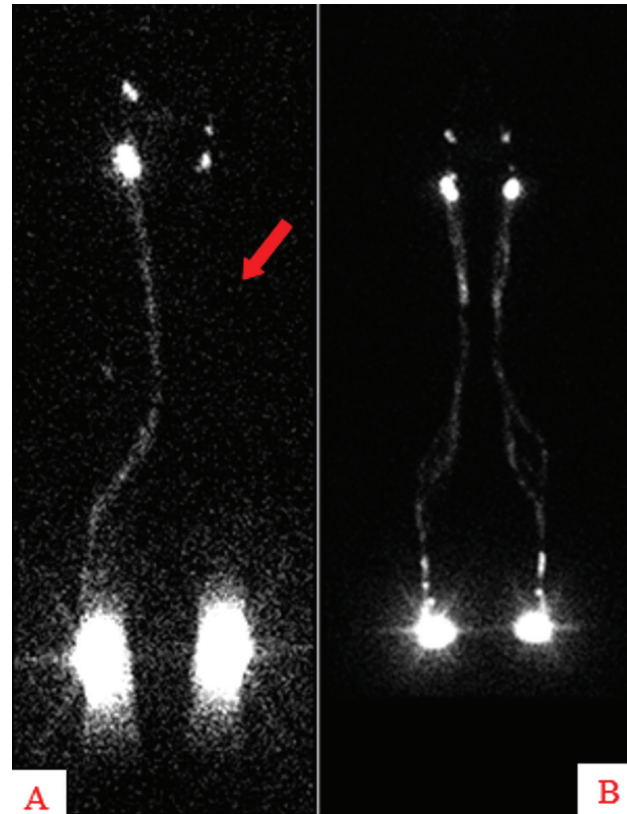
## QUESTION 2

Based on what you see in [figure 1](#), what is the underlying cause?

- A. Recurrent bacterial lymphangitis.
- B. Primary lymphoedema.
- C. Tumour.
- D. Filariasis.

## QUESTION 3

Which is the best diagnostic test to confirm the diagnosis?



**Figure 1** Lymphoscintigraphy of the lower extremities showing insufficient deep lymphatic circulation in the left leg (red arrow, A) replaced by superficial drainage (B).

- A. US scan.
- B. MRI.
- C. Lymphoscintigraphy.
- D. Reassurance and clinical follow-up.

## QUESTION 4

What is the mainstay of management of this condition?

- A. Wait and see.
- B. Antibiotic course.
- C. Supportive therapy (ie, physical activity, elevation of extremities, pneumatic compression).
- D. Surgical intervention.

*Answers can be found on page 345.*

### ANSWER TO QUESTION 1: C

Lymphoedema refers to the low-output failure of the lymphovascular system characterised by an increased collection of protein-rich fluid in the tissues, leading to swelling and pain. In the present case, the long-standing history of oedema is unlikely to be related to deep venous thrombosis, and a standard blood test can rule out myxoedema and nephrotic syndrome as underlying conditions. Typical values of inflammatory markers, autoantibodies and US findings can help to exclude juvenile idiopathic arthritis. While differential diagnosis can be challenging, specific clinical signs, such as non-pitting oedema and Stemmer sign (inability to pinch the base of the second finger or toe), can be useful in the diagnosis.<sup>1</sup>

### ANSWER TO QUESTION 2: B

Lymphoedema can be either primary or secondary, and caused by multiple conditions, such as damage to the lymphatic vessels or lymph nodes, leading to impaired lymphatic outflow (secondary lymphoedema), or congenital malformation of the lymphatic system (primary lymphoedema).

Secondary causes of lymphoedema include cancer, infections, lymphadenectomy, obesity, inflammatory disorders, hereditary syndrome (ie, Turner syndrome, Klippel-Trenaunay-Weber syndrome), radiation therapy, hypothyroidism, thromboembolism and trauma.<sup>2</sup> Primary lymphoedema is more frequently detected in the lower limbs and has an increased incidence in females. It is categorised according to the age of onset as congenital, praecox and tarda. In this patient, the absence of any local signs of inflammation did not suggest recurrent lymphangitis or filariasis, while standard laboratory and imaging tests ruled out a tumour. A Doppler US of the leg and an abdominal CT or US helped to exclude secondary causes in monolateral cases, such as deep venous thrombosis and pelvic pathology.<sup>3</sup> Negative test results, including CT and MRI, confirmed the diagnosis of primary lymphoedema. Although it also occurs in newborns, the onset of primary lymphoedema is much more frequent in puberty, possibly related to the loss of a previous relative balance in lymphatic drainage due to increased sex and growth hormones, together with pubertal growth spurt.<sup>4</sup>

### ANSWER TO QUESTION 3: C

In lymphoscintigraphy, a small amount of radioactive tracer, usually radiolabelled albumin, is injected into the web space between the first and second digits of the affected limb, and using a gamma camera it is possible to see the passage of the tracer through the


lymphatic system. The lymphatic oedema refers to the image of the radioactive tracer outside the lymphatic vessels.<sup>5</sup> While ultrasonography and MRI can also be useful in staging the lymphoedema,<sup>6</sup> lymphangiography is considered the gold standard not only for diagnosis but also for preoperative assessment of lymphatic function.<sup>7</sup> Particularly in this patient, the lack of deep lymphatic circulation was evident, partially replaced by superficial drainage (figure 1). A clinical diagnosis based on history and physical evaluation after exclusion of secondary causes is acceptable. Reassurance and clinical follow-up may also be a reasonable option.

### ANSWER TO QUESTION 4: C

While primary lymphoedema has a benign course and a favourable prognosis,<sup>7</sup> the impact on a patient's self-image and daily life can be bothersome.<sup>8</sup> Treatment consists of decongestive therapy, use of elastic garments or pneumatic compression devices, manual lymph drainage, and physical activity. Surgical intervention is an option in case of medical failure or when mobility has been compromised, but the efficacy of this approach seems limited and is debated in the literature.<sup>9</sup>

### PATIENT OUTCOME

Supportive therapy improved the oedema and limb function, resolving the pain and allowing thoroughly normal ambulation after 2 weeks.

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