

AN UNUSUAL CASE OF PRIMARY LYMPHOEDEMA PRAECOX

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Primary lymphoedema praecox is a condition that predominantly affects women and involves swelling in one or more limbs which may extend to the trunk (Browse, 2003). The swelling usually occurs before the age of 35 years and most commonly presents during puberty. There does not have to be a family history of the condition, but a genetic link for two forms of primary lymphoedema has been identified; Milroy's disease and lymphoedema distichiasis (Browse, 2003).

The onset of oedema is gradual, occurring over a few weeks and, initially, the swelling can fluctuate and reduce a little, or completely, overnight. However, this initial stage does not usually last for more than three months. After this time, the swelling becomes permanent. Patients do not usually complain of pain during episodes of swelling, but instead describe feelings of heaviness or aching.

Anecdotal and clinical evidence suggests that with time, the lymphoedema usually progresses, with an increase in limb volume and accompanying skin changes and thickening of the subcutaneous tissues.

Primary lymphoedema praecox is usually diagnosed using thorough assessment and clinical examination, and, if further investigations are warranted, using lymphoscintigraphy. Investigation of the lymphatic system using lymphoscintigraphy can depict function of the lymphatics and is able to show pooling or backflow of lymph where there is poor or no function. In patients with primary lymphoedema praecox, lymphoscintigraphy often shows

distal lymphatic obliteration, with reduced lymphatics in the thigh, groin, and torso (Browse, 2003), though it is not clear why this occurs.

Primary lymphoedema praecox is not curable, but with conservative treatment can be managed.

This rare case article will now describe the investigation, differential diagnosis and treatment planning of patient X, a 40-year-old male with an unusual presentation of primary lymphoedema praecox.

Case study

At the age of 17 years, patient X first noticed intermittent and painful swelling of his left upper limb. Over the next 10 years, he also experienced swelling and pain in the right upper limb and both legs below the knee. Patient X finally sought advice from his GP, as the swelling to his left arm and hand had significantly increased and had become permanent. He was referred to a vascular surgeon who carried out a Doppler ultrasound scan of the left arm, to identify any underlying venous abnormality. The results showed no vascular abnormalities and the patient was discharged.

Approximately 10 years later, the patient once again contacted his GP, concerned about the swelling of his arm which had never resolved. He was again referred to a vascular surgeon, who felt the results indicated an oedema of lymphatic origin. At this point, patient X was referred to the lymphoedema clinic.

Assessment

Initial consultation consisted of thorough past medical history recording, and ascertaining the onset and progression of oedema. Any family history and previous investigations were also discussed. Clinical examination, including Stemmer's test, skin, subcutaneous tissue and limb-shape assessment, was performed. Limb volume measurements of both upper limbs were recorded (Table 1).

During the consultation, patient X pointed out that his right arm also had periods of swelling, although it was currently not swollen. This was documented and digital photographs were taken (Figures 1–3).

Diagnosis

A diagnosis of primary lymphoedema praecox was made, based on:

Table 1

Patient X's limb volume measurements in ml at initial assessment and first follow-up

	Left arm	Right arm	Right leg	Left leg
Total volume at initial assessment	2133	1998	6218	6542
Total volume at first follow-up	1940	2000	6117	6358

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- ▶▶ The patient's age at onset of oedema
- ▶▶ The exclusion of venous abnormality
- ▶▶ The nature of oedema to the right hand (thickened subcutaneous tissues, feel, texture, and duration of oedema).

Management plan

Patient X was given information on his condition and a treatment plan was initiated to manage the oedema in the left arm, and as a preventative measure in the other arm. The treatment plan consisted of:

- ▶▶ Skincare — to maintain the integrity and hydration of the skin and to reduce the risk of infection. Patient X was advised to wash, dry thoroughly and moisturise both arms daily. He was also advised to apply an antiseptic cream to any cuts, bites or scratches and to take other infection-reducing measures, e.g. wearing gloves while gardening. He was also told not to wear clothing or jewellery that was too restrictive or tight around the wrist.
- ▶▶ Exercise — to enhance the function of the lymphatics, patient X was advised to avoid gripping activities, such as racket sports, which can lead to a worsening of the condition, and instead maintain active exercises such as swimming.
- ▶▶ Manual Lymphatic Drainage (MLD) and Simple Lymphatic Drainage (SLD) (a simplified version of MLD) — these gentle skin-stroking techniques are designed to direct fluid away from swollen areas to enable it to drain more freely elsewhere. MLD was carried out at the clinic every day for one week, and then monthly for four months (Table 2). The patient was taught SLD techniques and was instructed to carry this out twice daily to move lymph fluid towards the groin.
- ▶▶ Compression hosiery — to provide a counter force for the muscles to work against and, in turn, increase lymphatic uptake. He was initially fitted with compression hosiery for his swollen arm only.

At his first follow-up appointment, patient X's left arm had reduced in volume (Table 1) but, during further discussions, he revealed that he had also suffered from

Table 2

Patient X's Manual Lymphatic Drainage plan (MLD)

What is Manual Lymphatic Drainage (MLD)?

MLD is a gentle skin-stretching technique which aims to manually move fluid away from congested areas towards functioning lymphatics. There is little clinical research regarding the efficacy of MLD, but it is anecdotally, strongly supported. MLD is a therapist-led, specialist skill requiring regular practice and updates.

Initial treatment plan

Sequence of Leduc method of MLD: the unaffected lymph nodes in the neck, axillae and left arm were treated.

Results: Softening of the subcutaneous tissues, reduction in pain and patient generally more comfortable. A reduction in limb volume was noted, though it is not clear which part of the treatment plan resulted in this.

Adapted treatment plan following lymphoscintigraphy results

Deep breathing, MLD sequence: neck, axillae, both arms, groin nodes and both legs. For the left arm and the lower limbs the technique was concentrated more distally.

Results: The left leg reduced in limb volume, again this cannot be clearly attributed to just MLD. The incidence of the occurrence of oedema in the right arm and right leg was also reduced with the patient not experiencing any swelling for one year after treatment. There are no studies relating to the effects of MLD preventatively for lymphoedema, but this would make an interesting area for further research.



Figure 1. Right arm.



Figure 2. Left arm.

intermittent swelling in both lower legs. Clinical examination revealed extremely mild swelling of the left leg and photos and measurements were recorded (Figure 4; Table 1). The treatment plan was revised so that skincare, preventative advice, exercise and SLD also included the lower limbs. However, compression hosiery for the right arm and both legs was not fitted, as the patient felt this would be too much to cope with.

Patient X's lymphoedema history was atypical of primary lymphoedema praecox because:

- ▶▶ It affected all four limbs
- ▶▶ The swelling completely resolved to all but the left upper limb for long periods of time, sometimes years
- ▶▶ When swollen, the limbs were often painful.

Patient X is a young, working gentleman with small children who understandably wanted as much information as possible about his diagnosis, concerning not only how it would affect the rest of his life, but also that of his children. He initially felt disappointment that the treatment methods offered were conservative and not curative, and felt that if there was any possibility of more information which might lead to different treatment options, then he was keen to undergo further investigation.

Lymphoscintigraphy

The patient, the GP and the lymphoedema clinic agreed that a referral needed to be made to a specialist centre for diagnostic lymphoscintigraphy. The patient was initially assessed and lymphoscintigraphy of all four limbs was arranged.

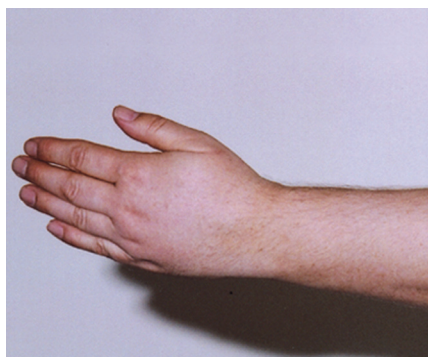


Figure 3: Close-up view of left hand.

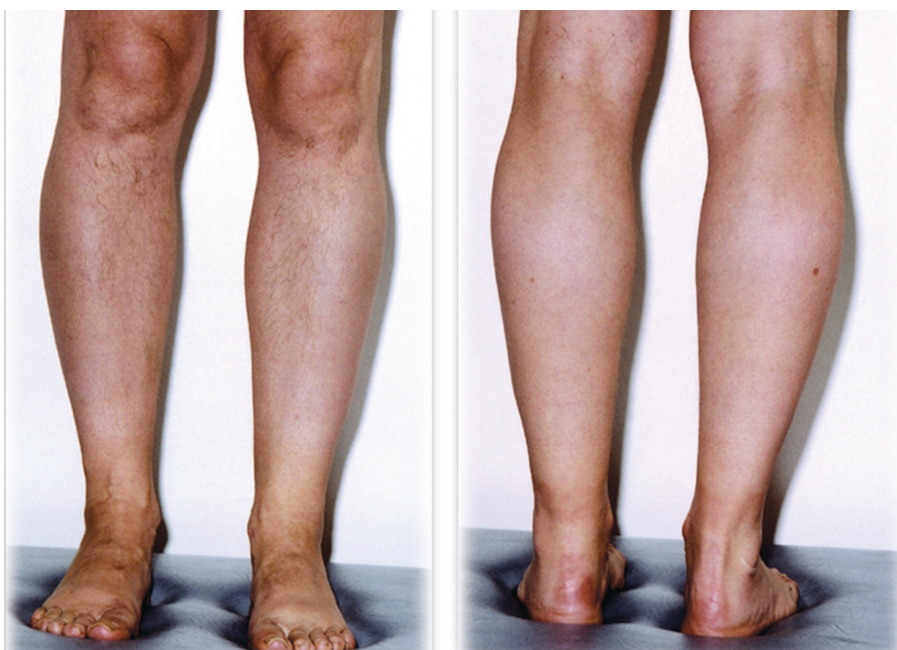


Figure 4. Legs, front and back at first assessment.

Lymphoscintigraphy involves a small amount of radioactive macromolecule being injected between the fingers, or toes, before the limb is imaged by a gamma camera to show function of the lymphatic system. It shows the superficial lymphatic system and the inguinal and parailiac nodes.

Active exercise of the limbs takes place between images to enhance lymphatic uptake. A diagnosis of lymphoedema is usually confirmed if there is an uptake of less than 0.3% of the initial injected dose in the groin at 30 minutes, and little more at an hour (Browse, 2003). Lymphoscintigraphy is minimally invasive but, in patient X's case, the radiation exposure would be increased with all four limbs being assessed at one time.

Results

The results of patient X's lymphoscintigraphy showed distal hypoplasia of both legs and the right arm, and proximal and distal hypoplasia of the left arm. Distal hypoplasia is a reduced number of lymphatics in the lower limb and fewer than five entering the lymph glands (Browse, 2003). The proximal lymphatics are normal and mild oedema is usually visible at the extremities, e.g. ankles. In cases of proximal and distal hypoplasia, the lymphatics are reduced throughout the limb, the swelling may be moderate and affect the whole limb and there will

be a probable underlying genetic cause. In both conditions, there may be associated lymphatic valvular incompetence.

As lymphoscintigraphy cannot depict valvular incompetence or the number of reduced lymphatics, patient X may benefit from undergoing a 3D MRI scan to give a deeper understanding of the lymphatics and their appearance, or, indeed, further lymphoscintigraphy to focus specifically on the deep lymphatic system.

Liu et al (2005) stated that in order to provide optimal clinical management, patients with lymphoedema should undergo proper imaging. The more that is known regarding the underlying pathophysiology of a patient with lymphoedema, the more confident the approach to treatment will be. The results of patient X's lymphoscintigraphy influenced his treatment plan in three main ways.

- ▶▶ First, as the lymphoscintigraphy showed normal proximal lymphatics in both legs and the right arm, patient X could be safely fitted with below-knee compression garments and a glove, rather than wearing full limb-length garments which were needed for the left arm (a glove and a sleeve), thus improving his quality of life
- ▶▶ Second, manual lymphatic drainage (MLD) (Table 2) could be focused

on particular areas of hypoplasia (distally) and less where there was good function (proximally). Bourgeois (1997) suggests that when carrying out lymphoscintigraphy for upper limbs it would be helpful to have views of the thorax, as these can show eventual drainage pathways, axilla to axilla, or to internal mammary nodes. This information would be particularly helpful when prescribing a plan for MLD. In patient X's case, this procedure was not carried out as it was not standard protocol for treatment

- ▶▶ As there was no evidence to suggest any reduced function of the deep lymphatic channels in the trunk, deep breathing was introduced to patient X's regimen, in an attempt to maximise drainage. Some therapists recommend the use of breathing exercises to help clear the deep central lymphatics in an attempt to increase lymphatic uptake from the extremities. Although there is a lack of evidence relating to the effectiveness of deep breathing, it is not harmful and it was incorporated in the case of patient X (Table 2).

Földi et al (1985) indicate the use of lymphoscintigraphy after treatment to evaluate its effectiveness, and this view is supported by Boris et al (1994). Despite the financial implications, possible increased risks and inconvenience to the patient, this could be an excellent outcome measure for research into the effectiveness of lymphoedema management. Weiss et al (2003) utilised lymphoscintigraphy as a method for evaluating changes following lymphatic microsurgery, and found lymphoscintigraphy to be a reproducible method to demonstrate functional ability of the lymphatics for a follow-up study. Miranda et al (2001) used lymphoscintigraphy to evaluate the effects of sequential intermittent pneumatic compression on leg lymphoedema, the study again found lymphoscintigraphy to be a valid outcome measure.

It would certainly be interesting to carry out repeat investigations for patient X to assess any changes in function. Unfortunately, due to lymphoscintigraphy not being carried out at the hospital

providing the treatment, this would involve further inconvenience for the patient in travelling and time off work, and may involve a long waiting list. Such an approach would also have financial implications.

Discussion

Patient X was diagnosed as having primary lymphoedema praecox by using only basic diagnostic techniques, including thorough assessment, examination and the recording of measurements. As he was atypical in his presentation, and in line with the patient's wishes, further investigation was requested.

Lymphoscintigraphy is a low risk, relatively inexpensive, easy to perform and moderately accurate system for diagnosing functional failure of the lymphatic system.

The lymphoscintigraphs he underwent did not provide a different diagnosis, but a deeper understanding of the function of his lymphatic system that allowed the patient's treatment plan to be refined. The identification of distal hypoplasia in all four limbs and proximal hypoplasia in the right upper limb influenced the choice of hosiery and the sequence and concentration of hand movements for MLD.

Patient X did feel that the lymphoscintigraphy results had perhaps raised more questions than answers, and this may be a problem for some patients if they go looking for a definite diagnosis. The fact that it was highlighted that there may be a possible genetic link also raised concerns about his children and whether, indeed, this problem could have been passed on to them. The ability of the patient to deal with the outcome of any investigations must be considered before referral for investigation is made.

At the present time, patient X is doing well. He has been able to completely stop wearing compression hosiery on his right leg and right arm (these areas have not had an incidence of swelling for 12 months). He wears his stocking on the left leg every day to work, but removes it at all other times. Although there remains mild oedema in this limb, it is not painful and does not cause patient X any problems. His left arm continues to require strong compression hosiery throughout the day, but he no longer wears full-limb hosiery.

He has been fitted with a full-fingered glove which he tolerates well and which maintains the oedema. The patient states that this limb is no longer painful and the swelling does not cause him any problems. He continues with skincare and exercise to all four limbs and has reduced the SLD to once daily. He visits the lymphoedema clinic every six months for repeated limb volume measurement and assessment of his compression garments.

Conclusions

Both the lymphoedema clinic and the patient agreed that the implementation of treatment based upon the function of an individual's lymphatic system is beneficial. Not only does it allow for less time to be spent on areas not requiring extensive treatment, but it empowers the patient with knowledge of the true underlying cause of oedema. Although lymphoscintigraphy is not necessary for every patient, and will not necessarily change their diagnosis, it can have very real benefits for the patient and the healthcare professionals involved in their care. JL

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