

PRESENTATION OF CHILDHOOD LYMPHOEDEMA

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Childhood lymphoedema is a relatively rare condition, uncommon outside of specialist clinics, but which has a significant effect on the affected individual and the family. As a lifelong condition with, at present, no cure, management of the condition by dedicated lymphoedema therapists is of paramount importance. Increasingly, the underlying molecular genetic cause of some forms of childhood lymphoedema are being elucidated, which has led to a more precise diagnosis and may, in the future, lead to novel treatments. This paper describes the ways in which the condition may present, ways it can be investigated and the current forms of management.

Key words

Lymphoedema
Lymphatic
Milroy
Distichiasis

The main function of the lymphatic system is to return protein rich fluid, which has exited from the capillaries, back to the venous system (Damstra and Mortimer, 2008). Lymphoedema is a chronic, and often progressive swelling due to failure of the lymphatic system to drain this fluid from the interstitial spaces. This causes abnormal accumulation of tissue fluid in the interstitial spaces and can be due to a number of causes both primary and secondary (Rockson and Rivera, 2008). It usually affects the extremities as a result of abnormal regional lymph drainage, although visceral drainage can also be impaired. Lymphoedema in children, while not common, can cause considerable

diagnostic difficulty to clinicians and distress to parents. It is essential to obtain a rapid diagnosis and to implement correct treatment at the earliest opportunity. In the UK many physicians and surgeons will see less than 10 cases of lymphoedema in a year (Tiwari et al, 2006). It is therefore imperative that patients are referred at an early stage to a clinic with wide experience and expertise in diagnosis and treatment.

Phenotyping childhood lymphoedema

Primary lymphoedema

Primary lymphoedema is chronic oedema caused by a developmental abnormality of the lymphatic system (Mortimer, 1995) and is the most common type seen in the paediatric population. In primary lymphoedema, fluid accumulates due to either abnormal function or structure of the lymphatic system. In most cases, oedema will be present from birth but in some cases, although the lymphatic abnormality is presumed to be present congenitally, the swelling does not develop until some time later. It is thought that the lymphatic system normally functions at about 10% capacity and it is presumed that this functional redundancy allows homeostasis to be maintained for some time in this group of patients.

A population prevalence of 1.33 per 1000 for all ages has been reported but the authors acknowledge that this is probably an under estimation of the true burden of disease (Moffatt et al,

2003). A specific prevalence figure for primary lymphoedema in the paediatric population has been estimated at 1.15 per 100,000 population, but these numbers are based on those attending a single US clinic (Smeltzer et al, 1985). A female preponderance (M:F – 1:3) is documented, although this may represent ascertainment bias. Primary impairment of the lymphatic drainage system can occur as a non-syndromic mendelian condition or as part of a more complex syndromic disorder.

Well recognised forms of primary childhood lymphoedema

Over the past few years the underlying molecular defects in two types of lymphoedema have been well characterised; Milroy disease and lymphoedema distichiasis syndrome. This has given a greater understanding not only of the particular individual's disease, but also of the developmental pathways involved in the embryonic growth of the lymphatic system in the embryo. In the longer term, it is hoped that this new-found understanding will allow the development of novel medical treatments and perhaps one day the 'Holy Grail' of gene therapy to eradicate the disease in an individual.

Other forms of primary lymphoedema are also well recognised but the pathogenesis is less well understood. In the past there has been a tendency for all congenital lymphoedema to be

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incorrectly labelled as Milroy disease. The clinical and molecular findings in true Milroy disease are now clear, and the term should be reserved for the small group of individuals and families fitting the specific phenotype (Brice et al, 2005; Connell et al, 2009b). In addition to Milroy disease and lymphoedema distichiasis there are many other forms of primary lymphoedema; the phenotypic entities of primary lymphoedema vary in age of onset, site of the oedema, associated features, inheritance patterns and the underlying genetic cause. It is therefore important that when assessing a child presenting with lymphoedema that it is not only the lymphoedematous limb that is addressed. A careful examination of the child is essential. Other clinical features or aspects of the history may enable recognition of a specific diagnosis that has implications for management. A detailed family medical history should also be sought.

Milroy disease

Milroy disease is congenital onset lymphoedema which classically affects the lower half of the legs only (Milroy, 1892). It can, although rarely, affect the entire of the lower limbs but is not reported to affect the arms. Males and females are affected equally. In males, hydroceles are common, affecting up to 30% of those carrying the altered gene. Other characteristic clinical findings include upslanting 'ski-jump' toenails (Figure 1) and prominent large calibre veins in the legs (Figure 2), most commonly the great (long) saphenous veins (Brice et al 2005).

The causative gene was first located on chromosome 5q in 1998 (Ferrell et al, 1998) and subsequently numerous causative mutations in the *VEGFR3* gene have been described (Evans et al, 2003; Ghalamkarpour et al, 2006; Ferrell and Finegold, 2008). In those individuals not conforming to the Milroy phenotype, molecular testing of *VEGFR3* is not warranted (Connell et al, 2009b).

Inheritance of Milroy disease is autosomal dominant with a penetrance of approximately 85%. This gives up to a 50% recurrence risk for siblings if a parent carries the gene change. *De novo* mutations have been reported so a family history is not mandatory (Ghalamkarpour

et al, 2006; Connell et al, 2009b). For this reason, it is always worthwhile testing the parents of an affected child before calculating recurrence risks.

Lymphoedema distichiasis

Lymphoedema distichiasis syndrome is a rare, dominantly inherited condition for which the underlying genetic cause was identified by Fang et al (2000). Almost all individuals with lymphoedema distichiasis syndrome have mutations in *FOXC2* (Sholto-Douglas-Vernon et al, 2005). This condition is the association of primary, pubertal, or post-pubertal, onset lymphoedema with aberrant eyelashes arising from the Meibomian glands in the eyelids (*distichiasis* – from Greek for 'two rows') (Figure 3). Other associations include; cleft palate, congenital heart disease, varicose veins and ptosis (Brice et al, 2002). Half of affected individuals will have clinically evident varicose veins from an early age, while 100% have venous abnormalities when assessed by ultrasound scanning (Mellor et al, 2007). A lymphoscintigram showing lymph reflux (Figure 10) is suggestive of a diagnosis of lymphoedema distichiasis. If distichiasis is not present in the patient or a family member, the chance of finding a mutation in the causative gene, *FOXC2*, is extremely unlikely. As distichiasis can be difficult to see, slit lamp examination by an ophthalmologist is advised wherever possible.

Due to the increased risk of both cleft palate and congenital heart disease, additional pre-natal scans may be recommended for a fetus at risk of inheriting the condition.

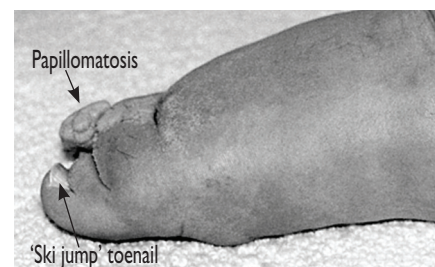


Figure 1: Adult patient with Milroy disease showing pedal oedema, 'ski jump' nails and papillomatosis due to congenital lymphatic abnormalities.



Figure 2. Prominent veins, commonly seen in Milroy disease.

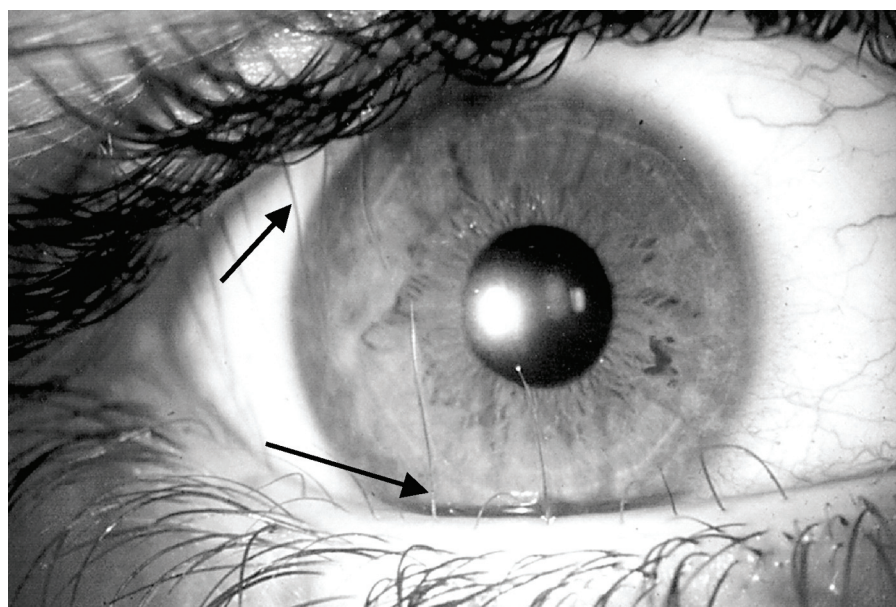


Figure 3. Distichiasis on upper and lower lids. Note presence of more than one row of normal lashes but anterior to, and clearly separate from the abnormal lashes (arrowed) growing from the Meibomian gland.

Syndromic lymphoedema

Lymphoedema can be seen as part of a syndromic diagnosis. Assessment of a child with primary lymphoedema requires obtaining a full personal and family history along with clinical examination, as other clinical signs may point towards a specific diagnosis. Turner, Noonan, Prader-Willi and CHARGE (Coloboma, heart defect, atresia choanae, retarded growth and development, genital hypoplasia, ear anomalies/deafness) are all well recognised conditions in which lymphoedema is known to form part of the phenotype.

Primary lymphoedema can also be seen in rarer syndromes including microcephaly-lymphoedema-chorioretinopathy, hypotrichosis-telangiectasia-lymphoedema and Hennekam syndrome. In patients with lymphoedema and other congenital anomalies and/or dysmorphic features, a review by a geneticist should be sought. Karyotype analysis is appropriate in all dysmorphic patients with primary lymphoedema as chromosome abnormalities have been identified in a proportion of these patients (Radhakrishnan and Rockson, 2008). Other molecular investigations should be targeted at confirmation of a specific syndromic diagnosis. Accurate recognition of a syndromic diagnosis has implications for prognosis, management and genetic counselling for the patient and family in terms of risks to other family members having affected offspring.

Lymphoedema in association with vascular malformations/overgrowth

Lymphoedema can be seen in association with vascular malformations or as part of an overgrowth syndrome such as Proteus or Klippel-Trenaunay syndromes. This group of patients tend to have a low recurrence risk, given the sporadic, mosaic nature of these conditions. The phenotyping and management of these conditions is not straightforward and may require specialist input (Brouillard and Vikkula, 2007). Management of overgrowth may require orthopaedic input.

Segmental lymphoedema

Primary lymphoedema affecting one or more body segments (segment = upper

limb[s], lower limb[s], face, genitalia or conjunctiva) is a recognised phenotype in the paediatric population. Facial, genitalia and conjunctival lymphoedema tend to be seen in association with lymphoedema of one or more limbs. Unisegmental lymphoedema (affecting one segment only) usually affects a limb (Figure 4). This group of patients do not have systemic involvement of their lymphoedema and the family history is usually negative, suggestive of a sporadic form of primary lymphoedema, with a low recurrence risk (Connell et al, 2009b).

Generalised lymphatic dysplasia (types I and II)

Generalised lymphatic dysplasia is a form of primary lymphoedema in which systemic involvement is a feature. Recognising systemic lymphatic problems has important implications for management. The lymphoedema in these patients can be: multisegmental (type I), affecting upper and lower limb(s), face, conjunctiva and genitalia in a segmental, asymmetrical pattern, with any combination of body parts being affected; or generalised, affecting the whole body (type II) as seen in Figure 5 (Connell et al, 2009a). These patients can present *in utero* with signs of oedema or hydrops. A full family and antenatal history should be obtained as, particularly in type II generalised lymphatic dysplasia, in which mendelian inheritance patterns are recognised, a positive family history has an impact on the risk of having another affected child.

Secondary lymphoedema

Secondary lymphoedema is the commonest form of oedema affecting adults. In developed countries this is most often following surgery for cancer during which lymph nodes are either removed or damaged. Further insults can occur to the lymphatic system with radiotherapy treatment, leading to fibrosis of lymphatics in the treated field. Other causes of secondary lymphoedema include injury and infection. In those parts of the world in which filariasis is endemic, this is the leading cause of lymphoedema (Szuba and Rockson, 1998). Lymphoedema due to filarial infection is rarely seen in the UK and Europe but should always be considered when assessing a patient who has lived



Figure 4. Unisegmental oedema in a child.



Figure 5. Widespread lymphatic dysplasia with four limb oedema and ascites.

or travelled in areas where filariasis is prevalent.

Secondary lymphatic impairment as a side-effect of drug treatment is not uncommon in adults but rarely seen in children. Treatment with calcium channel blocking drugs and sirolimus, an anti rejection medication, have been implicated in the development or exacerbation of oedema (Sahney, 2006; Desai et al, 2009;).

Diagnosis of lymphoedema

The diagnosis of lymphoedema is predominantly a clinical one. It is important at the outset to distinguish lymphoedema from other conditions



Figure 6. Lipoedema, note relative sparing of the feet.

with overlapping phenotypes. This is necessary as the treatments, long-term outcomes and reproductive risks may vary according to the diagnosis. Incorrect diagnoses can be dangerous and delay the institution of specific treatments. An incorrect diagnosis can also be expensive in that unhelpful investigations and treatments may be undertaken.

Differential diagnoses to be considered when faced with a patient with oedema include; cardiac failure, venous reflux, venous obstruction, e.g. thrombosis, drug-induced oedema, low albumin levels, recurrence of carcinoma, lipoedema and any circumstances of increased capillary filtration overwhelming lymph drainage (Levick and Mortimer, 2004).

Lymphoedema can be seen in association with or be misdiagnosed as hemi-hypertrophy or segmental overgrowth. This is an important diagnosis to make in the paediatric population due to the increased risk of Wilms tumour, and the need for renal screening in this group (Scott et al, 2006). The co-existence of vascular malformations and overgrowth should point towards differential diagnoses including Klippel-Trenaunay syndrome and Proteus syndrome.

It should always be considered that primary lymphoedema can co-exist with, or be exposed by, any of the secondary causes. The finding of a secondary cause does not rule out an underlying primary 'weakness' of the lymph system. Lymph drainage may be adequate until an inherent weakness is unmasked by a secondary event. This suggests that

there is a range of lymphatic function and establishing an accurate history will facilitate an accurate diagnosis.

Lipoedema (Figure 6) deserves special mention as it is often misdiagnosed as morbid obesity or lymphoedema and therefore needs to be included in the list of differential diagnoses. Lipoedema, first described by Allen and Hines (1940), describes a condition in which excess fat is preferentially stored in the legs and buttocks, and is often erroneously diagnosed as lymphoedema. Lipoedema almost exclusively affects females and presents at, or after puberty. It is not often seen in paediatric clinics but should be considered when seeing a teenage girl with recent onset of lower limb enlargement. In addition to symmetrical lower limb enlargement, there is often a history of easy bruising and tenderness in the affected limbs. Lymphoedema can complicate lipoedema, particularly when it is longstanding, and then lymphoscintigraphy may become abnormal (Bilancini et al, 1995). A recent report using MR lymphangiography suggests covert lymphatic vessel dilatation may exist in lipoedema before lymphoedema development (Lohrmann et al, 2009).

Management is difficult but essentially is similar to that prescribed for lymphoedema patients. Liposuction is sometimes recommended, although large scale, long-term follow up is not

yet available (Schmeller and Meier-Vollrath, 2006).

Clinical features of lymphoedema

Features typically seen in lymphoedema patients, whether primary or secondary, include; cutaneous and subcutaneous thickening, pitting oedema which does not improve significantly on elevation, positive Kaposi-Stemmer sign (inability to pinch a fold of skin at the base of the second toe (Figure 7; Stemmer, 1976) and skin changes including fibrosis, hyperkeratosis, papillomatosis (Figure 1) and lymphangiectasia. Papillomatosis is the occurrence of the out-pouching of pockets of skin between the fibrosed areas of tissue. Lymphangiectasia are 'blister-like' lesions on the surface of the skin that represent engorged lymphatic vessels. Leakage of lymph from these vesicles is termed lymphorrhoea. Many of the skin changes are dependent on the severity of oedema, efficacy of treatment and, in particular, duration of swelling, so are not often evident in the paediatric lymphoedema population.

Systemic involvement can be a feature of a more widespread lymphatic problem. It includes problems such as pleural and pericardial effusions, ascites, chylous effusions, and pulmonary and intestinal lymphangiectasia. The latter can cause malabsorption from the gut. This is most often a clinical diagnosis as invasive tests for this condition are rarely diagnostic. A history of symptoms



Figure 7. Demonstration of positive Stemmer sign, the inability to pick up a fold of skin at the base of the second toe, indicating thickening of the skin. This is useful in differentiating lymphoedema from other forms of oedema. Also known as Kaposi-Stemmer sign.

of diarrhoea/steatorrhoea and failure to thrive should be specifically asked when assessing paediatric patients. Respiratory and cardiac complications can arise from pleural or pericardial effusions caused by abnormal thoracic lymphatics and may require treatment.

Complications

Complications associated with lymphoedema are common and often dependent on the degree of oedema present. More severe oedema is likely to be associated with a greater number and severity of complications.

Psychological morbidity can be significant and is one of the major reasons given for presenting for medical attention. Lymphoedema can be very disfiguring and the perception of disfigurement contributes to the psychological morbidity. It can also be physically disabling and for this reason can impact on quality of life. In the paediatric population this can be particularly evident during school years with teasing by peers a common problem.

Cellulitis and lymphangitis are common complications of lymphoedema. Impaired immune function due to disturbed cellular trafficking from abnormal lymph drainage encourages infection. An initial local infection can quickly become septicaemia with systemic symptoms, requiring intravenous antibiotics and hospitalisation. Recurrent cellulitis not only leads to acute illness, but causes further damage to an already compromised lymphatic system, predisposing to further infective episodes. It is imperative that this cycle of events is arrested to prevent rapid deterioration in function of the lymphatics.

A consensus document outlining agreed treatment and prophylaxis for adults has been published and is available online at: www.lymphoedema.org/lsn. While this was developed for the adult population it can be adapted for use in children.

Tumours

In the adult lymphoedema population there is reportedly an increase in tumours developing in the lymphoedema-affected

areas. Lymphangiosarcoma is the most commonly associated malignancy. This is not reported in the paediatric population but information regarding the risks of these cancers should form part of the long-term health advice given to patients by medical staff and lymphoedema therapists.

Investigations

For many years the lymphatic system has been difficult to visualise both *in vitro* and *in vivo*. *In vitro* analysis has in the past been hampered by a lack of lymphatic-specific markers which is slowly being remedied with an increasing number now being available for laboratory use. These new markers can help to differentiate between lymphatics and other elements of the vascular system, allowing for the first time the ability to understand the embryological development of the lymph system. Lymphatic vessel markers of interest in humans include; LYVE1, VEGFR3, PROX1, Podoplanin, Neuropilin-2, and FOXC2 (Baluk and McDonald, 2008).

Functional imaging of the lymphatic system has improved with advances in technology and knowledge of molecular markers.

In vivo investigations are not always necessary to make a diagnosis but can at times add to the overall understanding of the condition in an individual and can help to classify them into a diagnostic group. As more is understood about the underlying molecular abnormalities in lymphoedema, the ability to classify patients correctly will become increasingly important if specific therapies are developed.

Lymphoscintigraphy is currently the 'gold standard' for investigating the lymph system. It has the advantage of being relatively non-invasive and, other than mild discomfort as the radioactive tracer is injected, is not unpleasant for the patient. Lymphoscintigraphy is a radionuclide-based method of investigating lymphatics. An injection of technetium-99 is placed subcutaneously into the web spaces of the limb and gamma camera imaging is undertaken at various time points to visualise the

uptake of the tracer (Wheatley et al, 1996) (Figures 8–10). Various protocols are used, but generally a comparison is made with known normal control values. A low uptake is generally indicative of a lymphatic abnormality while an unusually high uptake of tracer can be suggestive of increased filtration, e.g. venous disease. Further evaluation of the images can, sometimes, give more information as to the underlying cause of the abnormality (Mortimer, personal communication). The sensitivity and specificity of this type of scanning is not 100%. A normal scan does not rule out a lymphatic abnormality but an abnormal scan indicates an underlying lymphatic abnormality. Lymphoscintigraphy in children is not often necessary to make a diagnosis and can be performed using anaesthetic gel to avoid injection discomfort. Scans can give added information in cases where there appears to be widespread lymphatic abnormalities (Bellini et al, 2008).

Magnetic resonance imaging (MRI) has rarely been used for diagnosis of lymphoedema but can be used to differentiate from hypertrophy and may identify an obstruction to lymph drainage. Clinical examination can sometimes suggest tissue hypertrophy and, in these cases, MRI is the definitive test to differentiate between the two conditions, for example, fat hypertrophy in lipoedema or muscle enlargement in Klippel-Trenaunay syndrome. In the paediatric population MRI scanning can be problematic as the patient is required to remain still for a considerable period of time during the scanning process and they may need a general anaesthetic or sedation.

Newer MRI techniques, such as MR lymphangiography, are currently being developed in an effort to improve imaging of lymphatic vessels and lymphatic function.

Ultrasound scanning of the lymphatic system is rarely used, as visualisation of the lymph vessels is difficult. The use of ultrasound scanning is usually restricted to ruling out secondary causes of oedema such as tumours, venous disease, and cardiac failure (echocardiography).



Figure 8. Normal lymphoscintigram, note significant tracer collection in groin and evidence of major lymph vessels in legs.

Blood tests can be used to rule out some secondary causes of oedema such as low albumin and filariasis.

Genetic tests

The *VEGFR3* and *FOCX2* genes can be sequenced to look for mutations that are pathogenic for Milroy disease and lymphoedema distichiasis, respectively. In well-phenotyped patients, there is a high likelihood of finding a mutation (Bell et al, 2001; Connell et al, 2009b). Identification of a mutation means an unequivocal diagnosis and molecular diagnosis can then be offered to other family members. Prenatal diagnosis would be feasible, although rarely requested. In the future, if therapies specific to the underlying molecular abnormality are developed, this form of testing will become more important.

Genetic counselling

While identification of the causative genetic fault is unusual in lymphoedema conditions, genetic counselling can be extremely useful in providing better quantification of the recurrence risks

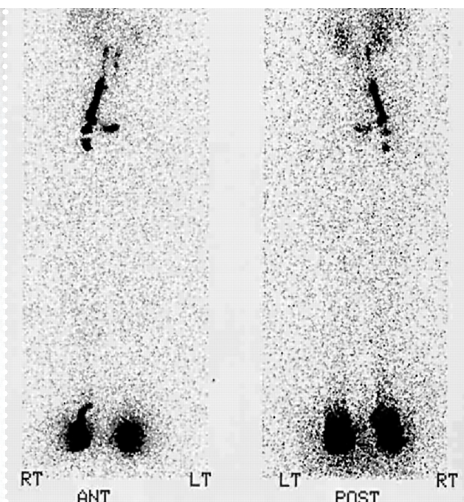


Figure 9. Lymphoscintigram scan as seen in Milroy disease patient with one leg affected. Note uptake of trace to groin on right side but no uptake on left side.

and pre-natal diagnosis/pre-implantation genetic diagnosis.

Management of childhood lymphoedema

Management of lymphoedema is lifelong and multidisciplinary. Parents should be encouraged to take an active role in the day-to-day provision of massage and application of garments as required. They will also take on a major part of the health education which is an important part of lymphoedema care, and essential to reduce complications. From the outset, the concept of long-term management, rather than cure, must be emphasised.

Lymphoedema therapist

All children with a diagnosis of lymphoedema should be referred to a trained lymphoedema therapist. A list of trained therapists can be accessed through the website of the British Lymphology Society (UK) (www.lymphoedema.org/bls). Currently in the UK, therapy provision for primary paediatric lymphoedema is patchy and those affected may need to travel considerable distances to access services.

Best practice for management will focus on the needs of each individual patient. This will include:

- ▶▶ Swelling reduction and improvement of shape
- ▶▶ Treatment and prevention of infection
- ▶▶ Treating skin problems such as

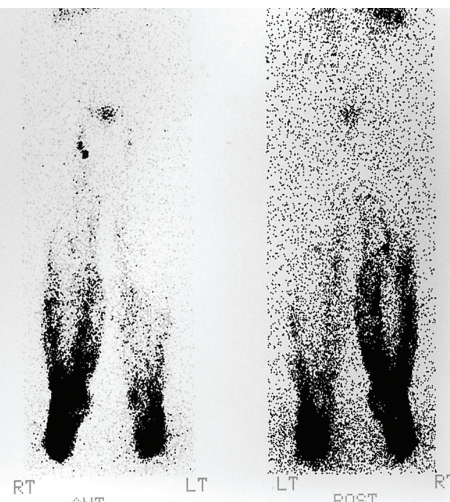


Figure 10. Lymphoscintigram showing characteristic 'dermal backflow' in lymphoedema distichiasis.

papillomatosis, lymphorrhoea and discouraging tissue fibrosis

- ▶▶ Pain and psychosocial management.

Therapy assessment will also include setting the benchmarks against which improvement can be judged, e.g. limb volume measurement, mobility and functional assessments. A treatment plan will depend on the site and severity of the lymphoedema.

Physical therapy

Physical methods of treating lymphoedema have been practised for many years. Therapy essentially aims to control lymph formation and to improve lymph drainage through existing lymphatics and collateral routes by enhancing what lymphatic function is present. It should be appreciated that lymph flow still exists in lymphoedema, otherwise swelling would be a relentlessly progressive process.

Physical treatment can, in the majority of cases, improve quality of life considerably and reduce the incidence of complications such as cellulitis and papillomatosis. Key to management is assisting patients and parents to understand their condition and to become partners in their care. Doing no harm and allowing any natural improvement through further development of the lymphatic system are important. With an enhanced understanding of their condition and

the need for treatment a high level of motivation and compliance can be generated (Mortimer and Todd, 2007).

Main elements of physical therapy

Care of the skin and prevention of infection

Skin changes are not only unsightly, but can lead to problems such as infection, fibrosis and lymphorrhoea. Regular application of an emollient is important for hydrating any hardened skin, thus making it more supple and discouraging hyperkeratosis. Tinea pedis is almost invariably because of the closely apposed swollen toes (not helped by elastic hosiery). Modern antifungal creams unfortunately macerate skin further and therefore it is suggested that Terbinafine cream is applied for two weeks followed by an alcohol wipe, assuming the skin is not broken. For deep cracks and crevices which bacteria may readily colonise, regular cleaning is necessary followed by an antiseptic soak, e.g. potassium permanganate. Prevention of infection, particularly lymphangitis/cellulitis, is crucial to the control of lymphoedema. Care of the skin, good hygiene, control of tinea pedis and good antisepsis following abrasions and minor wounds are important to reduce the risk of cellulitis.

Exercise

Exercise and movement are crucial to lymph drainage (Roddie, 1990) and should be encouraged. Dynamic muscle contractions encourage both passive and active phases of lymph drainage. Overexertion and excessive static exercise increase blood flow, which therefore tends to increase oedema.

External compression (hosiery, bandaging or pneumatic compression)

External compression complements the exercise programme. Such compression is not intended to 'squeeze' oedema but to act as a counterforce to muscle activity and so generate higher tissue pressures during contractions. This provides the most powerful stimulus to lymph drainage. Compression also limits capillary filtration by opposing capillary pressure. Without exercise, compression is much less effective.

Hosiery (below-knee or full-length stockings, half or full tights, sleeves) usually

requires high compression and double hoses may occasionally be required. Most garments last no more than six months. Two garments (or pairs) should be provided; one to wear and one for the wash. The correct technique for application, removal and care of garments are crucial for a successful outcome.

Multilayer bandaging is rarely required in the paediatric lymphoedema population. It can be used for limb reduction, but also has the advantage of restoring limb shape so that subsequent use of compression garments (hosiery) is more effective at controlling swelling (Badger, 2000). The digits are also bandaged to control swelling of fingers and toes. The strategic positioning of rubber pads 'irons out' pockets of swelling and deep skin folds. Multilayer bandaging is a skill which takes time to learn and should not be undertaken without appropriate training.

Pneumatic compression therapy (intermittent/sequential pneumatic compression) should not be used in preference to exercise and compression, but can be useful when exercise is difficult. An inflatable boot, legging or sleeve is connected to a motor-driven pump and lymph is displaced towards the root of the limb. If hosiery is not fitted immediately following compression therapy, the swelling readily recurs. Pneumatic compression softens the tissues and reduces limb volume during treatment, but it is doubtful that any long-term benefit is gained over hosiery and exercise alone.

Massage (manual lymphatic drainage therapy, MLD)

Massage is an important component of treatment, particularly for midline lymphoedema where there are seldom effective alternatives. The problem is that there are different forms of massage, used mostly for increasing blood flow and therefore of vigorous nature. In lymphoedema patients tissue movement needs to be gentle, as a stimulus to lymph flow without increasing blood flow (Mortimer et al, 2000). Indeed, in the absence of lymphatic contractility, massaging of filled lymphatics by muscle exercise or surface massage may be the major

Key points

- ▶▶ Always take a comprehensive family history. Lymphoedema can occur as part of several well described syndromes and it is vital to recognise these as the natural history and recurrence risks are well described.
- ▶▶ If in doubt about the diagnosis or best treatment, seek advice from your local lymphoedema clinic, genetics department or the Lymphoedema Support Network (LSN).
- ▶▶ Methods for visualising the lymphatics are not readily available and most diagnoses rely on clinical examination and medical history.
- ▶▶ Management by a lymphoedema therapist with experience of paediatric lymphoedema is vital.

stimulus to lymph flow. The practice that encourages such flow, referred to as manual lymphatic drainage therapy (MLD), is directed at normally draining lymph node regions; this 'siphons' or 'milks' lymph away from congested lymphoedematous areas, so complementing attempts to improve drainage 'upstream' by exercise and external compression. Continuous MLD delivered by a therapist is expensive and few healthcare providers will fund. Once a patient or parent has been taught the techniques, it is possible to do a simplified version of MLD at home. This is sometimes called simple lymphatic drainage.

Monitoring treatment

Monitoring of efficacy of treatment in children differs somewhat from adults; volumetric measurement is usually unsuitable as a parameter because of natural growth in length and weight. Monitoring of height and weight, and the physical and psychological development

of the child is important. Surgical management of lymphatic abnormalities in children is rarely required. Occasionally, surgery may be indicated for visceral involvement or ingrown toenails. In males with genital oedema, circumcision is sometimes indicated (Damstra and Mortimer, 2008).

It is important to integrate therapies into the child's daily life. Physical activities should be positively encouraged and the child must be allowed to do recreational activities as normally as possible (Damstra and Mortimer, 2008). JL

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