

Idiopathic congenital lymphedema involving the upper limb: A case report

Pearl Mary Varughese

From Department of Pediatrics, Pondicherry Institute of Medical Sciences, Puducherry, India

Correspondence to: Dr. Pearl Mary Varughese, Department of Pediatrics, Pondicherry Institute of Medical Sciences, Puducherry-605 014, India. E-mail: pearlmaryvar@gmail.com

Received - 31 March 2018

Initial Review - 22 April 2018

Published Online - 24 May 2018

ABSTRACT

Congenital lymphedema is the rarest form of primary lymphedema. The best line of investigation is lymphoscintigram. The management of primary lymphedema is usually conservative with bed rest and skin care. The prognosis is usually good if there are no other complications. We report a case of 10-year-old boy presented with a complaint of swelling in left upper limb and scrotal edema soon after birth. Investigations were performed and it turned out to be a case of congenital lymphedema involving the upper limb. Although there are many cases of lower limb involvement, upper limb lymphedema has not been documented often, so we are presenting this case.

Key words: *Lymphedema, Lymphoscintigram, Lymphatics*

Lymphedema is defined as a regional accumulation of excessive interstitial protein-rich fluid caused primarily by an imbalance between the inflow and the removal of interstitial fluid and protein and secondarily by a malformation or malfunction of the lymphatic system [1]. Primary lymphedema is due to congenital absence, hypoplasia, or underdevelopment of the superficial lymphatic channel. Secondary lymphedema occurs when the lymphatics are damaged or blocked by any pathologic process such as infection, surgical excision, neoplasm, irradiation, or trauma [2]. Primary lymphedema exists in three different forms: (1) congenital lymphedema seen at birth or infancy, (2) lymphedema praecox seen during puberty, and (3) lymphedema tarda seen in the late second or third decades of life [3].

Congenital lymphedema is the rarest form of primary lymphedema, accounting for approximately 1:60,000 live births. Congenital lymphedema can be further classified into familial (hereditary) and idiopathic (non-hereditary) subgroups [4]. Milroy disease is an autosomal dominant inherited primary congenital lymphedema. There are several genetic syndromes including Noonan's syndrome, distichiasis-lymphedema syndrome, Aagenaes syndrome, yellow nail syndrome, and Turner's syndrome associated with lymphedema [5]. When lymphedema occurs without any known etiology, dysmorphic features, and no family history of lymphedema, the eponym idiopathic congenital lymphedema is utilized.

CASE REPORT

A 10-year-old boy was brought by his parents with a complaint of progressively increasing swelling in his upper left limb from birth and also scrotal swelling. Informed consent was obtained from the parents and assent from the patient for taking the photographs.

He was a first-order birth child to third-degree consanguineous marriage and was born as term baby by normal vaginal delivery with a birth weight of 2700 g in Calcutta. At birth, he had left-hand swelling with abdominal distension and decreased urine output. Investigations were performed and showed that he had some lymphatic duct obstruction with intestinal perforation and was advised laparoscopic abdominal surgery followed by a scrotal surgery. The abdominal surgery was post-operatively complicated by an increase in the scrotal and newly developed penile swelling.

When he was brought to our hospital in September 2017, he had non-pitting type edema in his left upper limb (Fig. 1). The dorsum of his right hand (Grade III lymphedema) and both the lower limbs had Grade I lymphedema and a positive Stemmer's sign (inability to pinch the skin of the dorsum of the foot). The penis and the scrotal region had Grade IV lymphedema, and bilateral testes were descended (Fig. 2). The circumferences in different regions on the right forearm and arm were 3–5 cm greater than corresponding regions on the left. He had no dysmorphic features. There was no localized overgrowth of bone and no varicose veins. His nails were normal, and no evidence of infection was present. There was no abdominal distension or organomegaly. Extensive skin lesions were also present on the back and the abdomen (Figs. 3 and 4). These lesions initially started as small macules and then became confluent lesions which later ruptured with white serous discharge and formed scaly, crust-like lesions, probably suspected to be lymphangiomas circum spectrum. The affected limbs and the abdomen had intact touch, pain, and temperature sensations. Systemic examination was normal.

He had a normal growth and development. There was no history of weight loss or recurrent respiratory tract infections. He had no history of lymphangitis or cellulitis and had no complaint of any



Figure 1: Lymphoedema involving only left upper limb, lower limbs are normal



Figure 2: Scrotal swelling

pain during walking and standing. There was no family history of similar complaints. There was no history of trauma, spontaneous bleeding, or any hemangiomas or any vascular lesions increasing in size, which ruled out vascular malformations.

Ultrasound of the abdomen, scrotum, and the upper limb revealed lymph collection with a minimal abdominal fluid collection and multiple hydroceles, but otherwise normal internal organs. Complete blood count, serum albumin (3.3 g/dl), and coagulation profile were within normal limits. Electrolyte sedimentation rate was 42 mm/h. Biochemical tests were done to exclude hepatic or renal etiologies and urinalysis to exclude proteinuria - all were within normal limits. Dermatologists opined that the skin lesions were due to the lymph extravasation causing the white discharge and the extensive lesions. Lymphoscintigram was done to determine the level of lymphatic hypoplasia or agenesis. Pediatric surgeon opined that reconstructive surgery can be done if the parents request for the same. Plastic surgery opinion was obtained, and lymph nodovenous shunt followed by debulking surgery with skin grafting was advised.

He was treated conservatively with the skin care protocol and the elevation of affected limbs. On follow-up, the swelling did not progress further in next 6 months. The prognosis was explained to



Figure 3: Extravasation of lymphatic fluid on the abdomen



Figure 4: Extravasation of lymphatic fluid on the back

the parents. They were not willing for any surgeries and they have been lost to follow-up after that.

DISCUSSION

Lymphatic vessels play a central role in maintaining interstitial fluid balance. Angiogenesis and lymphangiogenesis are tightly regulated by growth factors, intercellular, and cell-extracellular matrix signaling mechanisms. Lymphedema is characterized by a chronic disabling swelling of the extremities caused by an increase in the interstitial protein-rich fluid, which subsequently results in insufficient lymphatic transport and drainage [1].

Idiopathic primary lymphedema is a rare disorder causing persistent swelling in an extremity due to impaired lymphatic drainage. There have been studies showing cases of idiopathic primary congenital lymphedema without any significant family history [6,7]. Our patient also had no family history of lymphedema and no dysmorphic features, so we categorized this patient as idiopathic primary congenital lymphedema. There are many lymphedema cases reported in the literature for lower limb involvement; however, it may also affect the upper limb, face, or genitalia [8]. In this patient, the distal part of the extremity is affected initially, with proximal extension occurring later.

Differentially, we thought parasitic infestation, infection, deep vein thrombosis, secondary to tumors, nodal resection, and trauma. The lack of any congenital heart defects and facial anomalies ruled out Noonan syndrome in our patient. Our patient also did not meet any of the cardinal features (hypohidrosis and hypotrichosis) of the (hypohidrosis and hypotrichosis) of the rare syndrome osteoporosis-lymphedema - anhydrotic ectodermal dysplasia with immunodeficiency or Aagenaes syndrome associated with lymphedema and neonatal cholestasis. In our case, filariasis was excluded through peripheral smear studies for microfilariae. Doppler ultrasonography of affected limbs and abdominal ultrasonography were done, and no evidence of deep vein thrombosis and obstructive lesions was found.

The natural history of primary lymphedema classically has been stated to have a slow but constant progression from a mild swelling of an ankle to a swollen extremity. The swelling enlarges at a slower rate than the growth of the body. A firm, non-pitting edema, fibrokeratotic skin, verrucous growths, squaring of the toes, and a tendency toward recurrent attacks of cellulitis and lymphangitis are its common manifestations [9]. The swelling normally remains unchanged in 60% of the patients for several years. In most patients, a static point is reached after several years of increased swelling, and irrespective of treatment measures, the swelling remains stable [4,10]. In the presented case, the swelling was on a plateau stage for 1 year.

Isotopic lymphangioscintigrams are generally considered the gold standard for the diagnosis of lymphedema as it is simple, easy, and harmless and has good reliability [11]. Lymphangioscintigrams in patients with primary lymphedema display absent or delayed tracer transport, lack or paucity of lymphatic channels, retrograde diffusion (backflow), and poorly visualized or absent regional lymph nodes. Computed tomography scan imaging has been shown to be highly sensitive (97%) and specific (100%). Although more costly, magnetic resonance imaging offers greater detail of lymphatic architecture and confers no radiation exposure [12].

The management of primary lymphedema is usually conservative and successful for most patients. Decongestive lymphatic therapy (DLT) is now well established as the treatment of choice for lymphedema regardless of the underlying etiology (primary or secondary) or its clinical stage. DLT consists of movement exercises, manual lymphatic drainage, and compression (bandaging, garments, and intermittent pneumatic compression) therapy in addition to basic skin care and education for risk reduction. A particular emphasis on skin care is important to reduce the increased risk of cellulitis and lymphangitis. When medical management fails, surgical intervention is indicated as the last option [13].

There are numerous potential surgical procedures, such as resection approach, debulking, buried dermal flaps, and microsurgical techniques such as the creation of anastomoses between lymphatic vessels and veins, between lymph nodes and veins, and between distal and proximal lymphatics. However, complications are common

and reported success rate of surgery is only 30% [4]. Conservative management, as well as surgical consultation, was taken in our patient and no further deterioration occurred for next 12 months.

CONCLUSION

Idiopathic congenital lymphedema is a rare disease in children but exhibits psychological and physical burden. Diagnosis of congenital lymphedema is done through clinical history including family history and findings in the physical examination. Primary lymphedema of the upper limb is a rare phenomenon, and a thorough investigation is warranted in cases of unilateral upper limb lymphedema to rule out occult malignancy and systemic disease. In cases where the diagnosis is unclear, lymphangioscintigraphy should be performed to confirm the presence or absence of lymphatic dysfunction.

REFERENCES

1. Wang Y, Oliver G. Current views on the function of the lymphatic vasculature in health and disease. *Genes Dev* 2010;24:2115-26.
2. Mortimer PS, Rockson SG. New developments in clinical aspects of lymphatic disease. *J Clin Invest* 2014;124:915-21.
3. Connell F, Brice G, Jeffery S, Keeley V, Mortimer P, Mansour S. A new classification system for primary lymphatic dysplasias based on phenotype. *Clin Genet* 2010;77:438-52.
4. Smeltzer DM, Stickler GB, Schirger A. Primary lymphedema in children and adolescents: A follow-up study and review. *Pediatrics* 1985;76:206-18.
5. Northup KA, Witte MH, Witte CL. Syndromic classification of hereditary lymphedema. *Lymphology* 2003;36:162-89.
6. Wananukul S, Jittitaworn S. Primary congenital lymphedema involving all limbs and genitalia. *J Med Assoc Thai Chotmaihet Thangphaet* 2005;88:1958-61.
7. Sim YJ, Seo JH. Congenital lymphedema of the upper extremity. A case report. *Eur J Phys Rehabil Med* 2008;44:89-91.
8. Schirger A, Harrison EG, Janes JM. Idiopathic lymphedema. Review of 131 cases. *JAMA* 1962;182:14-22.
9. Wu JJ, Wagner AM. Verruciform xanthoma in association with milroy disease and leaky capillary syndrome. *Pediatr Dermatol* 2003;20:44-7.
10. Wolfe JH, Kinmonth JB. The prognosis of primary lymphedema of the lower limbs. *Arch Surg Chic Ill* 1960 1981;116:1157-60.
11. Szuba A, Shin WS, Strauss HW, Rockson S. The third circulation: Radionuclide lymphoscintigraphy in the evaluation of lymphedema. *J Nucl Med Off Publ Soc Nucl Med* 2003;44:43-57.
12. Witte CL, Witte MH, Unger EC, Williams WH, Bernas MJ, McNeill GC, *et al.* Advances in imaging of lymph flow disorders. *Radiogr Rev Publ Radiol Soc N Am Inc* 2000;20:1697-719.
13. Lee B, Andrade M, Bergan J, Boccardo F, Campisi C, Damstra R, *et al.* Diagnosis and treatment of primary lymphedema. Consensus document of the International Union of Phlebology (IUP)-2009. *Int Angiol J Int Union Angiol* 2010;29:454-70.

Funding: None; Conflict of Interest: None Stated.

How to cite this article: Varughese PM. Idiopathic congenital lymphedema involving the upper limb: A case report. *Indian J Child Health.* 2018; 5(4):312-314.

Doi: 10.32677/IJCH.2018.v05.i04.021