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Swelling of Extremities in a Toddler: Primary Lymphedema?

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Abstract

Background: Primary lymphedema is a rare disorder, affecting mainly pediatric patients. It results from maldevelopment of the lymphatic vasculature. Its differential diagnosis is wide and includes secondary lymphedema (the most frequent), and several syndromes.

Methods: We report the case of an 8 months old girl who presents primary Lymphedema, and we review the literature published on the topic.

Conclusions: Diagnosis of primary lymphedema might be difficult since several syndromes might present swelling. We have recently diagnosed a case of primary Lymphedema. Since it is a pretty rare disorder we find it interesting for the scientific community, to learn about it. Besides, our two new contributions are the possibility of performing nuclear medicine before magnetic resonance or computed tomography, avoiding sedation and risk associated, and the accurate diagnosis made by an adult-specialized Nuclear Medicine Service, avoiding the inconvenience of going to a Pediatric Centre which might be far in distance.

Keywords: Lymphedema; Lymphoscintigraphy; Toddlers; Milroy disease

Introduction

Lymphedema is a chronic and progressive swelling of tissue due to inadequate lymphatic function. It can be classified as primary (due to anomalous development) or secondary (due to injury to lymph nodes or vessels). Genitalia and lower extremities are the most frequent locations affected by this pathology. Complications include infection, functional disability, chronic cutaneous changes and psychosocial morbidity [1-5].

Primary lymphedema is thought to happen only on 10% of the patients with lymphedema [2,6] and it is mainly a pediatric disorder. It is rare, affecting 1.2 per 100,000 persons younger than 20 years [3,4,7]. This report is based on a case of primary lymphedema of an 8 month old girl.

Clinical Report

A woman, 8 months old, presented edema located on the back of her left foot associated with dismorphic appearance. There is no family history of lymphedema. On her history antecedent of a surgical intervention was found, when she was 4 days old, because of an ovarian cyst on her right ovary (51 mm diameter). It was detected prenatally by ultrasound. Surgery had no incidence, and the ultrasound post surgery was normal. Neurodevelopment was accorded to her age. On the physical examination indurated edema on her feet back, more prominent on her left side, antimongoloid palpebral fissure (as her father), and mild hyperthelorism were found. No extra row of eyelashes, no yellow nails and no genitalia edema was found. Her weight and height were within the normal percentiles.

Caryotype resulted 46 XX, dismissing Turner Syndrome. On the hemogram there was a mild microcytic hypocromic anemia (Hemoglobin 10.4 g/dl, MCV 99.4 CHM 33.8) and light hypoproteinemia (total proteins 5.7 g/dl). Urinalysis showed no proteinuria and no microalbuminuria that could explain edema. Echocardiography was performed with no abnormal result. Transfontanelar ultrasound was also normal. Because of the antecedent of surgical intervention, secondary lymphedema was the main diagnosis and an abdominal MRI was performed: Uterus of normal morphology and location for her age, left ovary of 11×15 mm with several follicles inside, biggest of 8 mm. Right ovary is not recognizable, there is no free liquid no tumors on the pelvis. No lymphadenopathy of significant size. During the weeks the tests were being performed, edema appeared on her left hand back. Primary lymphedema was then suspected and a Tecnecium 99 lymphoscintigraphy of her low extremities was performed. It was informed as lack of radiocoloid lymphatic migration, consistent with primary lymphedema. Milroe disease genetic screening was negative (gene FLT4, mutation in the vascular endothelial growth factor receptor 3-VEGFR3), although 3 polymorphisms (with no clinical relevance according to the literature published) were found in the patient: rs446003 in homozygosis, rs55657009 in heterozygosis, rs2934600 in homozygosis.

With the diagnosis of primary lymphedema treatment was started, with education of the parents to moisture and massage the affected extremities. The patient was referred to vascular surgery. So far she hasn´t developed any cellulitis, nor functional disability. She is doing well with rehabilitation.

Discussion

Primary lymphedema is a rare disorder in pediatrics. The progression and morbidity is not influenced by the age of onset [1]. It affects males and females equally. Clinically the edema slowly enlarges over time because of the accumulation of subcutaneous lymph, which stimulates adipose deposition and fibrosis [2]. It is a diagnosis made by exclusion of familial and syndromic congenital lymphedema [7], and it is made by history and physical examination in 90% of patients

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[6,7]. It can be confirmed by lymphoscintigraphy which is 92% percent sensitive and 100% specific [2]. Some authors recommend the performance of a CT or MRI as the first test [4]. However, since pediatric patients, especially toddlers may need some kind of sedation for the performance of CT or MRI with the risks associated to it, we believe lymphoscintigraphy should be the test to be done first. In our case, lymphoscintigraphy was made and interpreted by an adult-specialized nuclear medicine facultative. Accessibility to a Pediatric Nuclear Medicine Service is difficult, since there are not many pediatric hospitals with that kind of service. According to our experience the lack of a Pediatric Nuclear medicine service shouldn´t mean a problem to proceed and make the most suitable test in order to achieve the right diagnosis. Since the technique of lymphoscintigraphy is reasonable to be made on a child and general medicine nuclear facultative are trained to read and interpret the results, diagnosis of primary lymphedema becomes easier, and there is no need to refer the patient to a third level hospital with the expenses associated to such efforts.

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S.No.	Name of methods/ sample	Distance travel by solute	Distance travel by solvent	RF value
01	Expeller	10 cm	10.7 cm	0.93 cm
02	Soxhlet	9.2 cm	10.7 cm	0. 85 cm
03	Utrasonicator	5 cm	10.7 cm	0.46 cm
04	Crude jatropha oil	9.7 cm	10.7 cm	0.90 cm
05	Jatropha biodiesel	9.2 cm	10.7 cm	0.85 cm
06	Crude karanja oil	9.7 cm	10.7 cm	0.90 cm
07	Karanja biodisel	10.2 cm	10.7 cm	0.95 cm

Table 3: RF value of different oil sample using TLC.