### Citation

### Published Version
doi:10.1097/GOX.0000000000001223

### Accessed
April 11, 2017 7:43:31 PM EDT

### Citable Link
http://nrs.harvard.edu/urn-3:HUL.InstRepos:32072216

### Terms of Use
This article was downloaded from Harvard University's DASH repository, and is made available under the terms and conditions applicable to Other Posted Material, as set forth at http://nrs.harvard.edu/urn-3:HUL.InstRepos:dash.current.terms-of-use#LAA

(Article begins on next page)
Lymphedema is the progressive swelling of tissue due to inadequate lymphatic function; the lymphatic system either develops abnormally (primary) or there is an injury to the lymph nodes and vessels (secondary). Primary lymphedema is rare, affecting 1 in 100,000 persons, and 92% of cases involve the lower extremities. Primary lymphedema is a progressive, noncurable disease. Over time, the extremities enlarge because of subcutaneous fibroadipose deposition. We present an unusual patient who, without treatment, had resolution of primary lymphedema clinically and confirmed by lymphoscintigraphy.

CASE PRESENTATION
A 5-year-old male twin conceived via in vitro fertilization presented at 3 months of age with right foot swelling. There was no family history of lymphedema. On physical examination, the right foot had pitting edema and a positive Stemmer’s sign (inability to pinch the skin of the dorsum of the foot). The left foot was normal. The child was thought to have infant-onset primary lymphedema, and a lymphoscintigram was performed to confirm the diagnosis (Fig. 1). The study showed abnormal lymphatic function in the right lower extremity confirming lymphedema.
Because the child was an infant, compression treatment was not prescribed, and the patient was re-evaluated at 12 months of age. At that time, his mother noted that the right foot was improved, which was corroborated on physical examination. Repeat lower extremity lymphoscintigraphy showed normal lymphatic function in the right leg (Fig. 2). The findings confirmed the clinical impression that his lymphedema had resolved.

**DISCUSSION**

Primary lymphedema is a rare, progressive disease of the lymphatic vasculature for which there is no known cure.1 Over time, the extremities enlarge because of fibroadipose deposition.2 We present the first patient with primary lymphedema in our experience of 303 to have spontaneous resolution of the condition. In our literature search, we could only find one other potential case of primary lymphedema that improved; a Pakistani family with a high degree of consanguinity from which 25 subjects, across 5 generations, exhibited congenital-onset, progressive lymphedema confined to the lower limbs, which resolves by 40 to 45 years of age.3 However, these patients did not have lymphoscintigraphy confirming either the presence of lymphedema or its improvement.3

The mechanism for our patient’s disease resolution is unclear. He did not receive compression treatment because of his young age, and thus, the lack of compression may have enabled an environment for his lymphatic function to improve. Lymph flow is based on load and clearance. Perhaps the child had normal lymphatic channels, but his lymph production in early infancy overwhelmed his lymphatic system causing an abnormal lymphoscintigram result. As the child grew older, his lymph production may have decreased resulting in a normal repeated study. Alternatively, the patient may have had consistent load but developed increased lymphatic vessel function by lymphangiogenesis or another mechanism. The observation that primary lymphedema has the rare ability to improve during infancy has implications for patient care and provides a potential clue into the pathogenesis of the disease.

_Arin K. Greene MD, MMSc_  
Department of Plastic and Oral Surgery  
Boston Children’s Hospital  
300 Longwood Ave.  
Boston, MA 02115  
E-mail: arin.greene@childrens.harvard.edu

**REFERENCES**

