

## QUIZ SECTION

### Puffy Feet in an 11-month-old Infant: A Quiz

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An 11-month-old boy with puffy feet had first presented at birth after a normal pregnancy. The boy's father and 8-year-old sister had the same oedema of the foot unilaterally and the infant's paternal grandmother was also affected bilaterally. His parents were not related to each other. Physical examination of the patient was normal, apart from the pectus excavatus and pes planus of the feet. Dermatological examination showed a non-inflammatory oedema located on the dorsum of the feet, giving a puffy appearance, and congenital misalignment of the left foot big toe (Fig. 1). Laboratory investigations, including urine analysis, total protein and albumin levels in the blood, abdominal ultrasonography and echocardiography were normal. The patient was started on elastic bandage compression therapy.

*What is your diagnosis?* See next page for answer.



Fig. 1. Bilateral oedema of the feet and misalignment of the left foot big toe.

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**Puffy Feet in an 11-month-old Infant: Comment**

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**Diagnosis: Milroy disease**

Milroy disease (MD) (hereditary lymphoedema type I) may involve multiple limbs, the genitalia and even the face (1). MD is an autosomal dominant trait, also known as primary congenital lymphoedema, which was named after William Forsyth Milroy (1855–1942), who described lymphoedema of the lower extremities in six generations of an affected family (2). MD is considered as a benign disorder of mainly cosmetic importance. However, in certain cases it can cause cellulites, up-slanting toe nails, and papillomatosis. It may also necessitate life-long lympho-drainage by massage or compression therapy. Some patients demonstrate spontaneous improvement of the oedema over time (1). The oedema is usually limited to the feet, but can also involve the whole legs. Oedema of the legs can also be accompanied by chylothorax, chylous ascites and pericardial effusion (3, 4). The male to female ratio is 1:2.3 (5). The onset of the oedema is usually within the first 2 years, but it can also begin *in utero*.

A mutation of the VEGFR3 gene has been found to play a role in the aetiopathogenesis of MD. This gene is expressed in all endothelial cells during early embryogenesis, but in the course of development its expression is restricted specifically to the lymphatic endothelium (6). The mutation inactivates the VEGFR3 tyrosinase kinase mechanism specific to lymphatic vessels.

The diagnosis of lymphoedema is usually determined by a thorough history and physical examination. Lymphoscintigraphy and magnetic resonance imaging (MRI) have become helpful tools (1). In the differential diagnosis of infants, lymphoedema-distichiasis and hypotrichosis-lymphoedema-telangiectasia syndrome (6, 7) should also be kept in mind. Infants should be followed up with care in order to avoid infections, development of angiosarcoma (8) and podiatric problems.

Treatment of lymphoedema is difficult. Various combinations of extremity elevation with massage, exercise and compression therapy can be used. Simple elastic com-

pression garments have been shown to be effective, with a 30–40% reduction in oedema. Ultrasound therapy and localized hyperthermia have demonstrated improvement in certain patients with lymphoedema.

Diuretics do not improve the condition. Dietary manipulation has been proposed as a treatment. Flavonoids (Daflon<sup>®</sup>, Servier pharmaceutical company, Istanbul, Turkey) are believed to have a protective effect on the vascular endothelium and to improve the microcirculation (1).

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