Puffy feet in a female neonate

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Accepted 26 July 2016

DESCRIPTION

A female neonate was evaluated due to limb oedema. Her father and three older siblings were healthy. Her mother had a subclinical autoimmune hypothyroidism. Gestation was uneventful, except for an abnormal maternal serum screening for aneuploidy during the first trimester. Amniocentesis revealed a normal female karyotype. A normal vaginal delivery occurred at 39 weeks. Physical examination showed bilateral dorsum foot oedema (figures 1 and 2). Cerebral ultrasound was normal and echocardiography revealed a patent foramen ovale. She was discharged at 48 hours of life and had regular follow-up consultations in neonatology, physical medicine and rehabilitation, paediatric cardiology and genetics. Later, the family learned about some paternal relatives with swollen legs and feet.

Congenital primary lymphoedema can be caused by several genetic diseases (Trisomy 13, 18 and 21,

Figure 1 Bilateral lymphoedema of the lower limbs at first physical examination of the neonate.

Figure 2 Bilateral lymphoedema of the lower limbs at first physical examination of the neonate.

Figure 3 The appearance of the feet at 18 months.

Turner syndrome, Noonan syndrome, lymphoedema-distichiasis or yellow nail syndrome). As she had an isolated lymphoedema, normal karyotype and a positive family history, a Milroy's disease was considered. This diagnosis was confirmed by the identification of a mutation in *FTL4/VEGFR3* gene.

Milroy's disease is a developmental disorder of the lymphatic system which leads to disabling and disfiguring swelling of the extremities. It is a rare condition with an estimated frequency of 1:6000.^{1 2} This disease shows an autosomal dominant pattern of inheritance with penetrance of 80–90%.² It is considered a benign disorder which, nonetheless, can be associated with cellulitis, upslanting toenails and papillomatosis.³

Currently, our patient is 18 months old and has a normal growth and development. She presents mild-to-moderate asymmetric bilateral lymphoedema of the lower limbs (figure 3).

Learning points

- ▶ When evaluating a neonate with oedema of the lower limbs genetic causes should always be considered. If the infant has no other associated malformations, Milroy's disease is also a possibility.^{1 3}
- ▶ Milroy's disease is an autosomal dominant form of primary lymphoedema characterised by a congenital onset and non-progressive lower limb involvement.¹
- ▶ Treatment for Milroy's disease is conservative based on physical therapy, simple elastic compression with garments and exercise.^{1 2}

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To cite: Espírito Santo R, Moldovan O, Costa P, et al. *BMJ Case Rep* Published online: [please include Day Month Year] doi:10.1136/bcr-2016-215872

Acknowledgements Mariana Soeiro e Sá.

Competing interests None declared.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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