Lymphoedema-distichiasis syndrome

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Accepted 16 December 2015

DESCRIPTION

A 29-year-old woman with a history of renal cysts, hypertension and lymphoedema-distichiasis syndrome, was referred to ophthalmology, with bilateral blurred vision, hyperaemia and ocular pain, developed over months.

The patient had no positive family history for lymphoedema-distichiasis or other diseases.

Clinical examination revealed stunted height (144 cm), neck webbing (figure 1), bilateral and asymmetric lymphoedema (figure 2), bilateral distichiasis (figure 3) and keratitis (figure 4). Other ocular manifestations of lymphoedema-distichiasis, such as ptosis and strabismus, were excluded through ophthalmological examination.

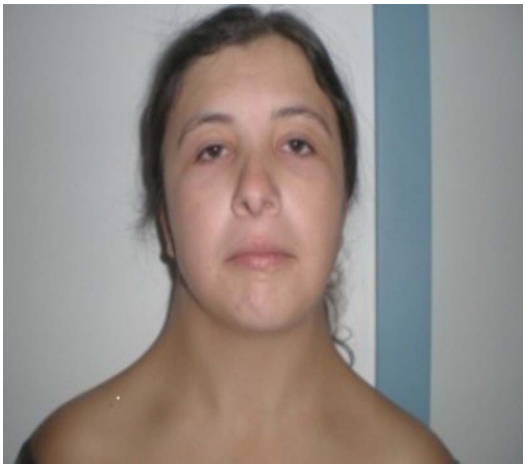

Figure 1 Patient's neck webbing.

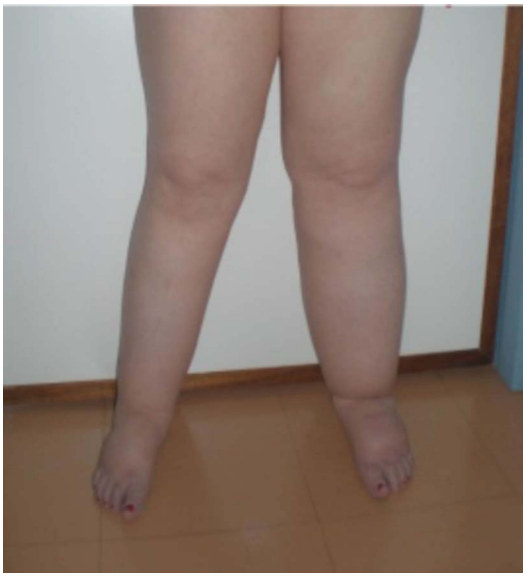

Figure 2 Bilateral and asymmetrical lymphoedema, which the patient presented since puberty, with progressive worsening and episodes of cellulitis.

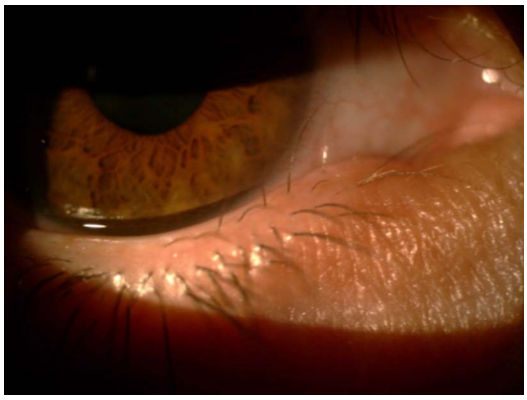

Figure 3 Inferior distichiasis.

Spinal MRI revealed multiple arachnoidal extradural cysts in the thoracolumbar and sacral spine (figure 5, arrows).

The patient's symptoms regressed with bilateral electrolysis of the abnormal follicles after unsuccessful attempts at epilation and follicle removal using an argon laser.

Lymphoedema-distichiasis syndrome is a rare condition, associated with diminished quality of life, being linked with chronic keratitis, conjunctivitis and photophobia in 75% of cases. Distichiasis, which may be present at birth, is observed in 94% of affected individuals.

The FOXC 2 gene is the only gene in which mutations are known to cause lymphoedema-distichiasis syndrome. Its protein has a role in a variety of developmental processes, such as formation of veins, lungs, eyes, kidneys, urinary tract, cardiovascular system and lymphatic vessels. Any pathogenic variant of this gene could lead to varicose veins, absence of lymphatic valves, lymphoedema, and cardiovascular and kidney malformations.

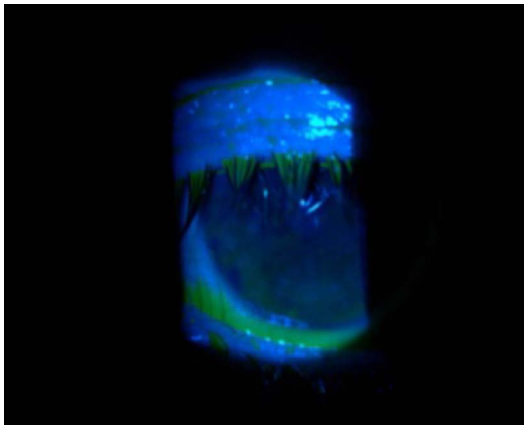

Figure 4 Discrete inferior keratitis.

To cite: Marques NS, Miranda A, Barros S, *et al*. *BMJ Case Rep* Published online: [*please include* Day Month Year] doi:10.1136/bcr-2015-213651

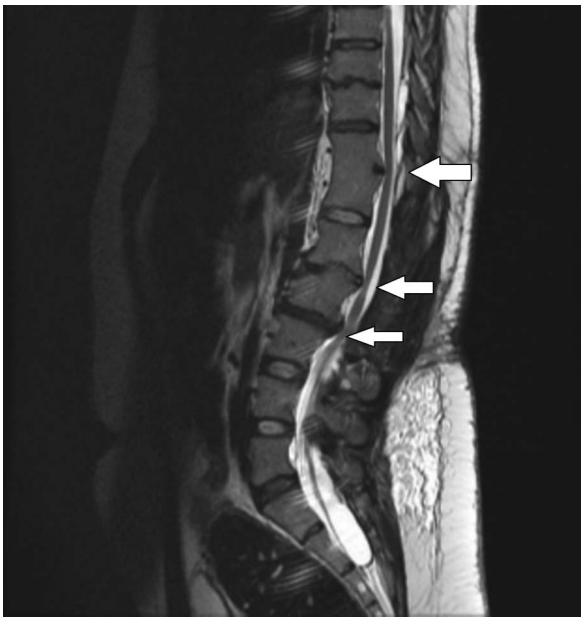

Figure 5 MRI, sagittal plane, of the thoracolumbar spine with spinal extradural arachnoid cysts (white arrows).

Our patient did not have a family history for this syndrome. For this reason, lymphoedema-distichiasis syndrome in this case was a probable phenotypic manifestation of a de novo mutation in the FOXC 2 gene.

Learning points

- The diagnosis of lymphoedema-distichiasis syndrome is made clinically based on the presence of primary lymphoedema and distichiasis. This syndrome can also present with ptosis, strabismus, renal and cardiac abnormalities, spinal extradural cysts,[1] neck webbing and varicose veins.[2]
- Lymphoedema-distichiasis syndrome is inherited in an autosomal dominant manner. Approximately 75% of affected individuals have an affected parent; about 25% have de novo pathogenic variants.[3]
- Conservative management of symptomatic distichiasis is with lubrication or epilation, or more definitive management with cryotherapy, electrolysis or lid splitting. Recurrence is possible even with more definitive treatment.

Competing interests None declared.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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