

Corneal Fleck dystrophy in a Spanish family

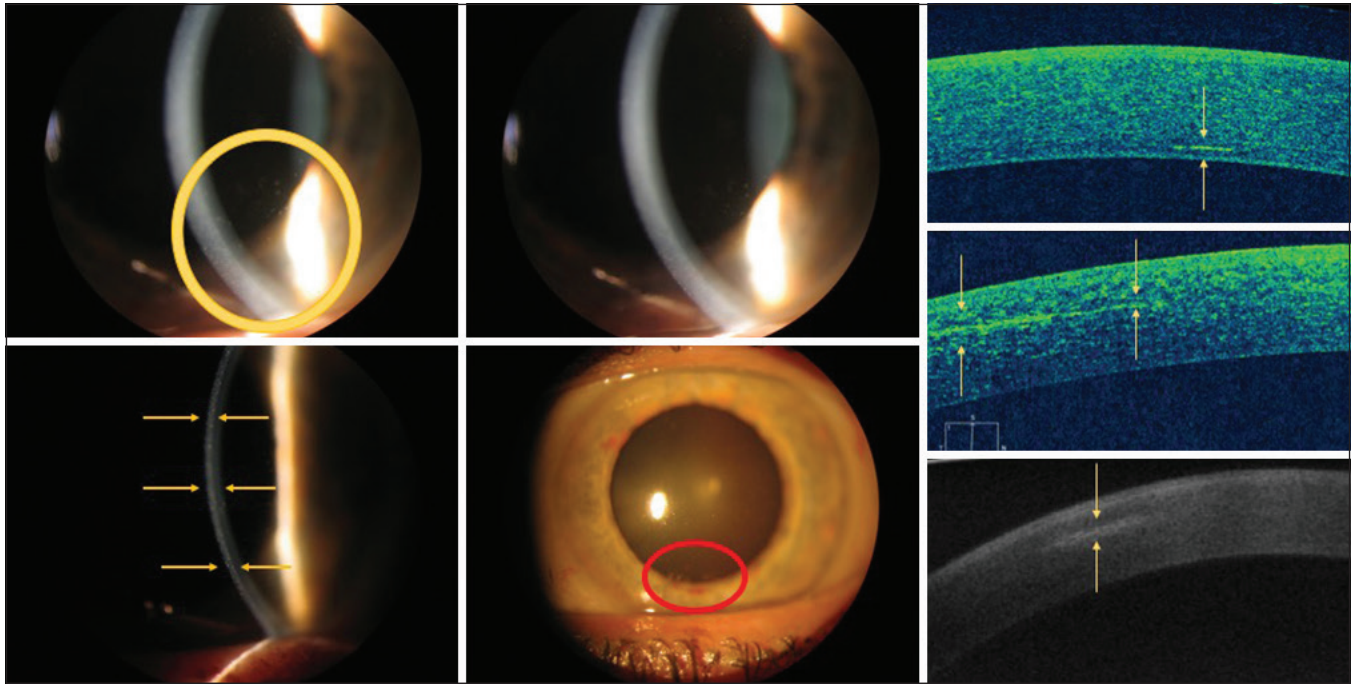


Figure 1: Composition of slit-lamp and anterior segment OCT showing the stromal flecks. Look at the circles and arrows, pointing to the flecks

Corneal fleck dystrophy is an unusual, asymptomatic, autosomal dominant condition.^[1,2] In the pathogenesis, it is hypothesized that a hydrolytic enzyme deficiency of keratocytes is involved, leading to accumulation of mucopolysaccharides and intracellular lipids.^[2]

We present an asymptomatic family, mother and daughter, who showed small, bilateral, translucent flecks, scattered throughout the whole corneal stroma without a defined pattern. Due to the suspicion of a mottled dystrophy, they were referred to the Genetics department and they were finally diagnosed with the PIKFYVE gene in heterozygosis, confirming the diagnosis [Fig. 1].

To our knowledge, this is the first Spanish family with corneal fleck dystrophy reported in the literature.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

*Carlos Rocha-de-Lossada,
Carmen Alba-Linero, Carlos Hernando Ayala,
Marina Rodriguez-Calvo-de-Mora*

Regional University Hospital of Malaga,
Plaza del Hospital Civil s/n, Málaga, Spain

Correspondence to: Dr. Carlos Rocha-de-Lossada,
Regional University Hospital of Malaga,
Plaza del Hospital Civil s/n, Málaga, Spain.
E-mail: Carlosrochadelossada5@gmail.com

References

1. Streeten BW, Falls HF. Hereditary fleck dystrophy of the cornea. *Am J Ophthalmol* 1961;51:275-8.
2. Assi A, Ebenezer N, Ficker L. Corneal fleck dystrophy in an English family. *Br J Ophthalmol* 1999;83:1407-8.

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10.4103/ijo.IJO_1702_19

Cite this article as: Rocha-de-Lossada C, Alba-Linero C, Hernando Ayala C, Rodriguez-Calvo-de-Mora M. Corneal Fleck dystrophy in a Spanish family. *Indian J Ophthalmol* 2020;68:918.