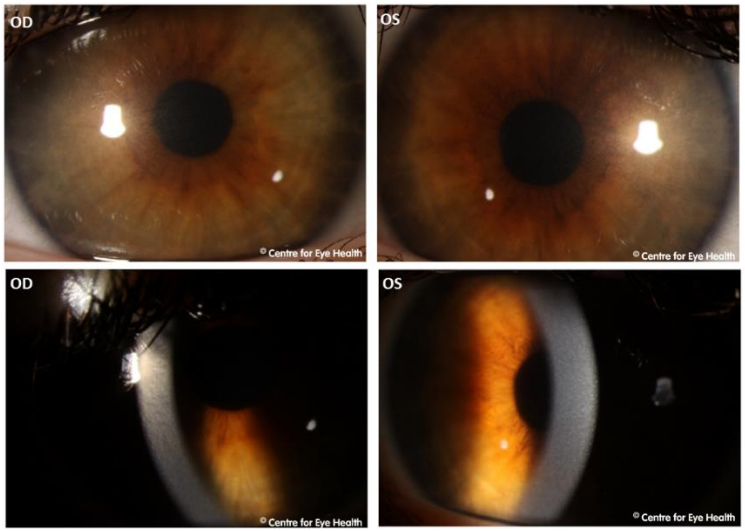
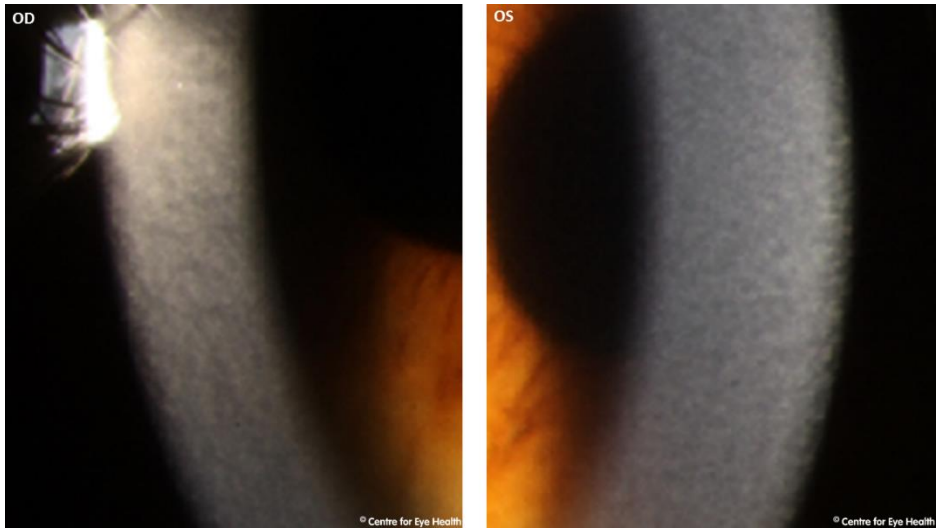
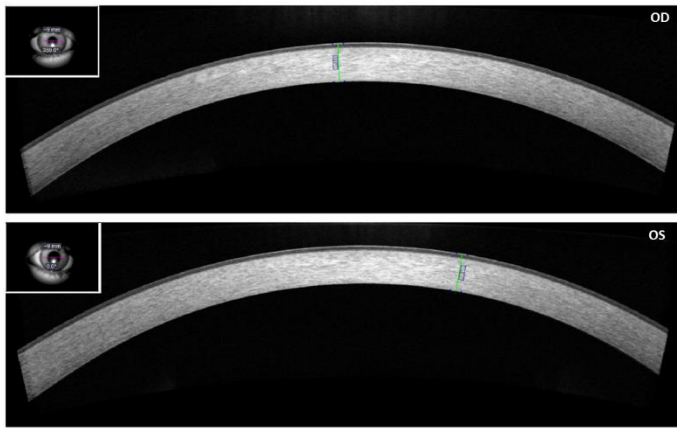
CFEH facebook Cases

CFEH Facebook Case #57

A 7 year old Caucasian male was referred for investigation of an unusual corneal appearance. VA was 6/9.5 OU. His general health is reported to be good, he takes no medications and does not have any allergies. There was no staining of the cornea with fluorescein dye and central corneal thickness was 511µm in the right and 508µm in the left eye. What is the likely diagnosis?







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ANSWER

The findings are most consistent with congenital stromal corneal dystrophy, which has previously been referred to as “congenital hereditary stromal dystrophy” This is an autosomal dominant dystrophy that is either non-progressive or slowly progressive. Typically, diffuse bilateral corneal clouding is noted with flake-like stromal opacities as seen in the corneal photos. The corneal stroma is affected throughout and anterior corneal surface is unaffected. As is typical, in this case the anterior OCT shows an increased stromal reflectivity in both eyes. Usually, pachymetry will show a thickened cornea, however in this case the cornea is actually at the thin end of normal.

Examination of immediate family members (mother and 2 brothers) revealed a similar corneal appearance in each: