

Supplementary Figure 13 (a & b) Immunolocalisation of CACANA1 in the lens, cornea, retina and optic nerve (CACANA1 IHC panel, 40x) in both non-XFS and XFS eyes show positive immunoreactivity in the anterior lens epithelium, corneal epithelial cells, stromal keratocytes and corneal endothelial cells. The optic nerve glia and vascular endothelial cells also show positive CACANA1 staining in both non-XFS and XFS eyes. In non-XFS retina, strong diffuse immunoreactivity is seen in the photoreceptor inner segments (IS), inner nuclear layer (INL) and outer nuclear layer (ONL) and nerve fiber layer (NFL) of non XFS globes in comparison to the XFS globes where focal and patchy immunostaining of the IS, ONL, INL and NFL is observed.

Panel c) CACANA1 positive immunoreactivity is seen in the anterior iris border, iris stromal cells, the iris dilator and sphincter pupillae as well as the pigmented iris epithelium in both XFS and Non-XFS irides (Iris panel: CACANA1 IHC panels). Exfoliated material does not show positive staining with CACANA1 (asterix, CACANA1IHC XFS2 panel) in contrast to its LOXL1 positive immunoreactivity (asterix, LOXL1 IHC XFS1 panel).

