The patient is a 32-year-old female whose race she self-reported as African American. At age 13 years, she was diagnosed with glaucoma with elevated IOP, which was managed by bilateral trabeculectomy. Approximately 4 years later, significantly elevated IOP in both eyes was detected accompanied by severe cupping and open iridocorneal angle in each eye. She had no evidence of Haab striae, buphthalmos, or high iris insertion suggestive of PCG. Ultimately, she required bilateral glaucoma tube shunt implantation. Subtle lens dislocation superonasally in the right eye was observed at her initial visit at age 17 years, which slowly progressed over a 16-year period. At age 32 years, she developed lens dislocation in the left eye.

Given the constellation of taller stature than her relatives (180.4 cm), long fingers, and dislocated lens, a connective tissue disorder was suspected, especially Marfan syndrome, and the patient underwent cardiac and genetic evaluation. Serial echocardiograms failed to reveal any abnormalities, but hypermobility of the shoulders, digits, and knees was observed. A comprehensive connective tissue genetic testing panel including 92 genes (Invitae) revealed 2 heterozygous variants in the LTBP2 gene (c.709C>T p.Arg237\* and c.3776-1G>C splice acceptor), not previously reported in gnomAD (0.00%) or other publications. A complete family history was negative for similar findings. Cascade testing of the patient’s 2 children with normal ocular examination results at age 4 years and 10 years revealed 1 child carrying each variant, consistent with transheterozygous mutations responsible for the patient’s predominantly ocular and mild systemic phenotypic manifestations.