COL4A1 MUTATIONS IN TWO INFANTS WITH CONGENITAL CATARACTS AND PORENCEPHALY: AN OPHTHALMOLOGIC PERSPECTIVE

To the Editor: The recent short report by Nau and colleagues[1] suggests that there is a phenotype–genotype correlation between posterior lenticonus or posterior cortical cataract and heterozygous *COL4A1* mutation. The authors reference in their discussion a study in which I participated that suggested posterior lenticonus cataract can also be caused by biallelic *FYCO1* mutations.[2] This suggestion was based on three children from two Saudi families who had bilateral posterior lenticonus cataract and were homozygous for one of two *FYCO1* mutations (NM_024513: c.2505del;p.Ala836ProfsX80 or c.T449C;p.lle150Thr). Recently I was referred a fourth child (a third Saudi family) who was found to be homozygous for *FYCO1* mutations (c.2505del;p.Ala836ProfsX80). Lens opacities were noted since birth but had been visually insignificant. At four-year-old she had bilateral posterior lenticonus cataracts; in addition, she had an anterior polar lens opacity in the right eye (Figure 1). This additional case strongly supports biallelic *FYCO1* mutations as a cause for posterior lenticonus cataract specifically. Recognition and documentation of such phenotype–genotype correlations by clinicians is essential in order to enable accurate interpretation of molecular genetic testing results for our patients with specific phenotypes.

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References

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2. Khan AO, Aldahmesh MA, Alkuraya FS. Phenotypes of recessive pediatric cataract in a cohort of children with identified homozygous gene mutations. Trans Am Ophthalmol Soc 2015;113:T7.

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REPLY

We thank Dr. Arif O. Khan for his interest in our study.[1] We reported 2 cases of posterior lenticonus/cortical cataracts associated with *COL4A1* mutation. Other conditions are also associated with posterior lenticonus cataracts, including *FYCO1* mutations. We thank Dr. Khan for providing an additional report of biallelic *FYCO1* mutations as a cause for posterior lenticonus cataract.[2] We agree that recognition of these phenotype–genotype correlations will enable clinicians to accurately diagnose and counsel affected patients and their families.

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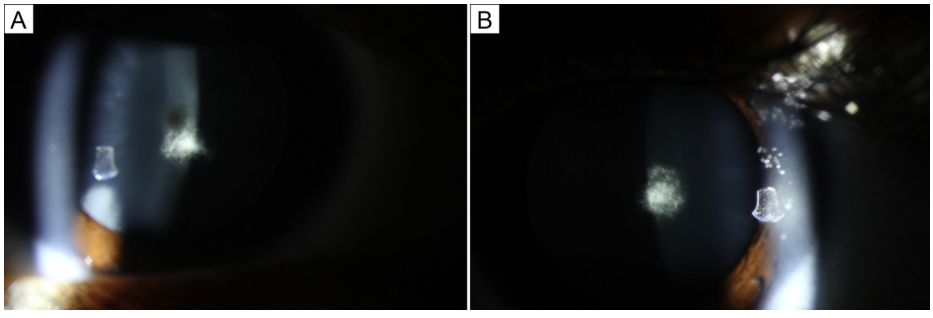

FIG 1. A, Slit-lamp photograph of the right eye showing posterior lenticonus cataract. The dark opacity seen overlapping the cataract superiorly is an anterior polar lens opacity. B, Slit-lamp photograph of the left eye showing posterior lenticonus cataract.