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Double Concentric Hyperautofluorescent Ring in EYS-Associated Retinitis Pigmentosa

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A 37-year-old Caucasian female with consanguineous parents and a brother with nonsyndromic retinitis pigmentosa (nsRP) presented with longstanding nyctalopia. The best-corrected visual acuity was 20/20 in both eyes. Anterior segment examination and fundoscopy were unremarkable. Blue light fundus autofluorescence (FAF) revealed a bilateral and symmetrical double concentric hyperautofluorescent ring (Fig. 1). On spectral domain optical coherence tomography, the retinal structure was preserved inside the inner ring, whereas outer retinal atrophy was observed outside its limits. Genetic testing identified the c.5928-2A > G p.? pathogenic variant in homozygosity in EYS gene.

Whereas a perifoveal/perimacular ring of hyperautofluorescence is a common FAF finding across several RP genotypes,[1] the presence of a double concentric hyperautofluorescent ring was deemed pathognomonic of NR2E3-associated autosomal-dominant RP.[2] We describe for the first time the occurrence of this peculiar FAF phenotype in EYS-associated nsRP, highlighting that FAF alone does not seem to be a reliable method of distinguishing between RP genotypes.

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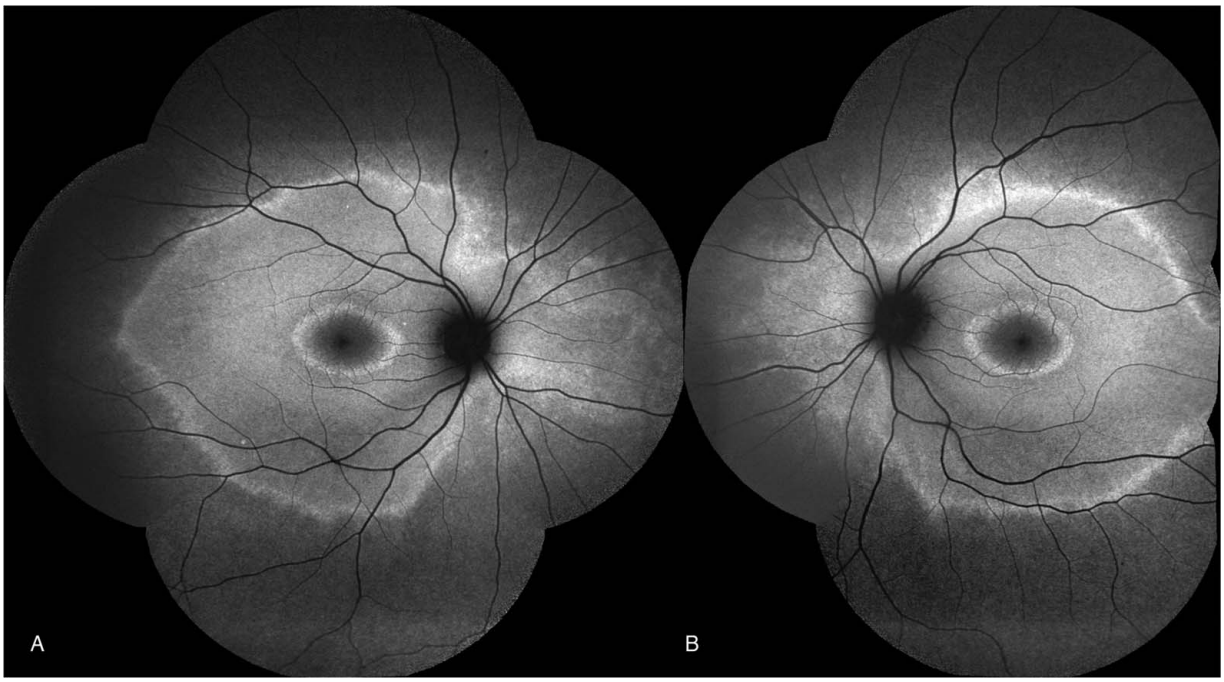

FIGURE 1. Double concentric hyperautofluorescent ring in EYS-associated retinitis pigmentosa. Right (A) and left (B) FAF highlights 2 hyperautofluorescent rings, an inner perifoveal ring and an outer ring, located along the vascular arcades and demarcating a diffuse hyperautofluorescent annular surface area. FAF indicates fundus autofluorescence.

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