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Posterior staphyloma in oculocutaneous albinism: another possible cause of reduced visual acuity

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Abstract

Posterior staphyloma is typically associated with myopic degeneration and has not been recognized as a cause of reduced visual acuity in albinism. We report 3 cases of posterior staphyloma, each with oculocutaneous albinism (OCA) defined by phenotype and genotype. Two cases are biological sisters with OCA type 2; one was myopic and the other was hyperopic. The third case involves a man with OCA associated with Hermansky-Pudlak syndrome (HPS-5). Staphyloma may be another cause of reduced visual acuity in albinism, particularly with increasing age. It may occur in association with myopia or hyperopia.

Case 1

A 9-month-old girl with nystagmus was referred to the Albinism Clinic at the University of Minnesota for ophthalmic evaluation. Her biological mother was African American, and her maternal grandmother had albinism. Visual acuity was estimated at 20/270 binocularly and 20/360 monocularly, with the acuity card procedure at 38 cm (Teller Acuity Cards, Stereo Optical Co. Inc, Chicago, IL). Physical examination showed blond hair, milky-white skin, and blue irides. Slit-lamp examination was normal, with the exception of grade 3-4 iris transillumination.¹ Cycloplegic refraction was plano +3.50 × 90 in the right eye and −0.75 + 2.75 × 90 in the left eye. Dilated examination of the fundus of each eye showed a sharp, pink optic nerve, a grade 2 macula,¹ foveal hypoplasia, and absence of melanin pigment in the retinal pigment epithelium (RPE).

At 4.5 years of age, best-corrected visual acuity was 20/200 in the right eye, 20/250 in the left eye, and 20/150 + binocularly using the HOTV chart. Cycloplegic refraction was −7.50 + 1.00 × 90 in the right eye and −6.25 + 2.50 × 90 in the left eye. A posterior macular staphyloma was noted in each eye.

At age 10, her best-corrected visual acuity was 20/100 in the right eye, 13/100 in the left eye, and 20/70 binocularly. Cycloplegic refraction was $-12.75 + 3.50 \times 95$ in the right eye and $-12.00 + 4.00 \times 81$ in the left eye. Axial length measured 26.88 mm in the right eye and 25.32 mm in the left eye (2500 A-scan, Sonomed, Lake Success, NY). Genetic testing confirmed the clinical diagnosis of type 2 oculocutaneous albinism (OCA2), with a previously described mutation, 615_632 del 18bp (in frame deletion), in exon 6. Case 1 also carries a common variant in exon 9: 913C>T(Arg305Try).

Case 2

A 3-month-old girl, the younger sister of case 1, was referred to our clinic for evaluation of nystagmus. Visual acuity was estimated at 20/470 in the right eye, 20/710 in the left eye, and 20/380 binocularly at 38 cm (Teller Acuity Cards). Physical examination showed dark blond hair and eyelashes, white skin, and blue/gray irides with golden brown pigment centrally. Slit-lamp biomicroscopy showed grade 2 iris transillumination.¹ Dilated examination of the fundi was significant for a grade 3 macula,¹ foveal hypoplasia, and absent melanin pigment. Cycloplegic refraction was $+9.50 + 1.00 \times 90$ right eye and $+8.50 + 1.50 \times 90$ left eye.

At 6 years of age, her best-corrected visual acuity was 20/60 in the right eye, 20/80 in the left eye, and 20/60 binocularly. Cycloplegic refraction was $+4.25 + 2.50 \times 85$ in the right eye and $+4.75 + 3.00 \times 91$ in the left eye. Dilated fundus examination and B-scan ultrasonography showed a posterior staphyloma in each macula (Figure 1). Axial length measured 21.23 mm in the right eye and 21.24 mm in the left eye (Aviso-Quantel Medical, Bozeman, MT). Genetic testing confirmed the same OCA2 genotype as seen her sister (case 1), 615_632del 18bp (in frame deletion) in exon 6 and a common variant in exon 9: 913C>T(Arg305Try).

Case 3

A 91-year-old man with a clinical diagnosis of Hermansky-Pudlak syndrome (HPS) was seen at our clinic for a 10- to 20-year gradual decrease in vision. At age 78, visual acuity was 20/400 in the right eye and counting fingers in the left eye at 2 inches. At age 88, he was only able to count fingers in the right eye and perceive hand motion in the left eye. At age 91, visual acuity was light perception in each eye. He had horizontal nystagmus, grade 3 iris transillumination,¹ and bilateral brunescient nuclear sclerosis. Dilated examination of the fundus showed a pink, tilted optic nerve without significant cupping and large atrophic posterior staphyloma in the macula of both eyes. Cycloplegic refraction was $-13.00 + 1.00 \times 180$ in the right eye and $-13.75 + 0.50 \times 180$ in the left eye. Axial lengths measured 29.21 mm in the right eye and 30.68 mm in the left eye (Accutome Inc, Malvern, PA). Molecular testing showed two homozygous frameshift mutations, described as c.1081 delC causing HPS5. This individual has been previously reported as the oldest reported survivor with HPS.² Following cataract surgery in the right eye at age 94, visual acuity was hand motion at 2 feet in the right eye and light perception in the left eye.

Discussion

OCA is an autosomal recessive condition that affects melanin production in the skin, hair, and eyes. It may rarely be associated with a bleeding disorder due to HPS. All individuals with OCA have some degree of foveal hypoplasia, contributing to the frequent but variable reduction in visual acuity. Our 3 cases suggest that yet another cause of reduced visual acuity is the development of a posterior staphyloma, with RPE atrophy over time.

The first 2 cases presented here have OCA2 with mutations in the *P* gene on chromosome 15q11-q13.³ Visual acuity typically ranges from 20/80 to 20/400.⁴ The third patient has HPS, characterized by abnormal biogenesis of lysosomes, platelet-dense granules, and melanosomes.⁵ We are not aware of other reports of posterior staphyloma in albinism.

Posterior staphyloma is a localized ectasia of the posterior globe which develops due to the abnormal composition of scleral fibrils that are smaller, more immature and have greater elasticity, occurring most often in older adults with myopia.⁶ Children with high myopia rarely have posterior staphylomas, and they may occur unilaterally.⁷ The staphylomas in our patients were confined to the macula (type 2),⁸ and would be described as narrow in character.⁶ Over time, RPE atrophy in the area of the sclera ectasia can cause reduced visual acuity. Other changes can include choroidal neovascularization, and/or development of a macular hole, retinoschisis, or retinal detachment. The reduced visual acuity in case 3 was felt to be due to RPE atrophy. If adults with albinism develop reduced visual acuity, the differential diagnosis should include posterior staphyloma.

Literature Search

PubMed was searched (1958-2015) in June 2015 using the following terms: *albinism* and *staphyloma* and *posterior staphyloma*. The reference sections of relevant articles were also reviewed. Untranslated foreign articles were excluded.

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Fig 1.
Case 2. A, Fundus photograph of posterior staphyloma in left macula at age 6. B, B-scan ultrasonography of staphyloma (arrows) at age 8.