a single guanine to adenosine substitution resulting in a Gln213Lys amino acid change. The exact effect of the amino acid substitution, from glutamic acid (which has a negative charge at physiologic pH) to lysine (which has a positive charge), is unclear. However, a similar missense mutation in hemoglobin (specifically a glutamic acid to valine substitution) retards ionic cross-linking and results in altered tertiary protein structure to yield, most famously, the “sickling” of erythrocytes characteristic of sickle cell anemia.

Dysfunction of bestrophin likely indirectly impairs apical fluid transport. This then indirectly impairs RPE phagocytosis of photoreceptor outer segments, lysosomal function, and regulation of subretinal fluid, yielding the characteristic vitelliform lesions and retinoschisis, characteristic of BVMD. Similarly, the phenotypic severity of the siblings we describe, particularly serous retinal detachments and retinoschisis, suggests the mutation they harbor grossly affects chloride transport and Ca+2 signaling, both thought to underlie RPE ionic transport and fluid homeostasis.

In summary, we herein present 2 siblings with BVMD, both exhibiting a previously unreported missense mutation in BEST1 as well as the novel findings of retinoschisis and a full-thickness macular hole.

Ruwan A. Silva, MD
Audina M. Berrocal, MD
Byron L. Lam, MD
Thomas A. Albini, MD

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Author Affiliations: Department of Ophthalmology, Bascom Palmer Eye Institute, Miller School of Medicine, University of Miami, Miami, Florida.

Correspondence: Dr Albini, Department of Ophthalmology, Bascom Palmer Eye Institute, 900 17th St NW, Miami, FL 33136 (talbini@med.miami.edu).

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Congenital Bilateral Aplasia of Medial Recti in a Family

A 50-year-old male patient from a nonconsanguineous marriage (case 1) was seen with outward deviation of both eyes since birth. He and his 2 sons (case 2, 24 years old, and case 3, 21 years old) had similar concerns of outward deviation of eyes. There was no history of any orthoptic treatment like occlusion, prism, or convergence exercises. There was no other medical or surgical ailment in either the father or the sons. Pedigree analysis revealed no similar ailment in any family member other than the father and his 2 sons; the only other issue was mild hypoplasia of the patient showed thin MR muscle on both sides (Figure 3B). Postoperative recovery was uneventful. His deviations measured 25 prism dioptries exotropia at distance and he had limited adduction (−1) after 4 weeks (Figure 1B). Subsequent follow-up after 3 months and 6 months did not show any further change.

Cases 2 and 3. Both his sons had similar large exodeviations. Magnetic resonance imaging in both cases revealed thin MR on both sides while intraoperatively, only an empty muscle sheath was seen. They were managed similarly with good postoperative results. Follow-up of both cases was satisfactory with minimal (<20–prism dioptrie exotropia) deviation.

Comment. Agenesis or hypoplasia of the extraocular muscles have been grouped as congenital cranial dysinnervation disorders with absent muscle development or abnormal innervation of the target muscle. Magnetic resonance imaging may show affected muscles and cranial nerves to be normal, hypoplastic, or absent. There is only 1 reported case of unilateral agenesis of MR muscle by Girard and Neely in 1958 and another case of bilateral agenesis of MR muscle by Houtman et al in 2009. However, familial occurrence of this anomaly has never been reported, to the best of our knowledge.

Our case appears to be a very rare case of autosomal dominant aplasia of MR seen in a male patient and his 2 sons. In all 3 cases, MR was not absent on the magnetic resonance image, which only showed mild hypoplasia; however, the intraoperative findings showed only a thin sheath of muscle capsule. As pointed out by Demer et al, the orbital and global fibers have differences in...
fiber types, fiber sizes, electromyography characteristics, vascular content, and metabolic activity. It is speculated that these cases had a lack of development of global fibers but complete development of orbital fibers. More research is needed into this differential development of the extraocular fibers in the future.

Congenital strabismus in humans can result from mutations in a number of genes, including PHOX2A, SALL4, HOXA1, ROB03, and KIF21A, that are essential to the normal development of brainstem motor neurons or axons. Because of the nonspecific nature of the disorder, genetic analysis was not possible in our case.

The aim of this case report was to highlight the possible separate global and orbital development of MR and the management of relatively larger deviations and to be ready to alter the surgical plan based on intraoperative findings. The management of these types of cases is very challenging and involves supramaximal resections and muscle transpositions in cases of muscle palsy. The management by partial vertical rectus transposition with Foster augmented sutures and lateral rectus recession had reasonable postoperative surgical success in all our cases as well and avoided an anterior ischemic complication.

Anirudh Singh, MS
Anudeepa Sharma, MBBS
Pradeep Sharma, MD, FAMS

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Author Affiliations: Strabismus and Neuro-ophthalmology Division, R. P. Centre for Ophthalmic Sciences, All India Institute of Medical Sciences, New Delhi, India.

Correspondence: Dr Pradeep Sharma, Strabismus and Neuro-
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