seemed to be dark. The narrowing of the retinal vessels and disc pallor diminished gradually over time. In the late phase of the attack (after 1 minute 28 seconds), the retinal vessels were dilated and the disc was hyperemic. Video 2 and Video 3 show the reperfusion of the retinal circulation in the late phase. The images obtained immediately after the attack (Figure 2) show the dilated retinal vessels in the right eye. The images show that the retinal veins and arteries in the right eye were more dilated than those in the left eye (not shown) or those obtained under normal conditions 1 month after the attack.

No hypercoagulability was identified with hematologic and serologic testing. Findings on neurologic tests and magnetic resonance imaging of the brain were normal. The patient was treated with propranolol hydrochloride because of an allergy to lomerizine hydrochloride, and the retinal migraines have not recurred.

Discussion  Most cases of previously documented retinal vasospasms have been associated with emboli or systemic diseases.1-3,4 Most cases reported to be retinal migraine were cases of presumed retinal vasospasm, and this disorder is exceedingly rare.5 Because our patient had no systemic disease except migraine and no embolus, the attacks were likely to have been primary vasospasms. The images clearly show vasospasm in a patient with a history of migraine with aura, which supports the belief that retinal migraine is a distinct entity. Individual cases of transient monocular vision loss have varied in the main component of vasoconstriction.6 In our case, diffuse narrowing of the retinal vessels may have represented decreased blood flow in the central retinal artery, and disc pallor and a dark choroid may have indicated decreased posterior ciliary circulation. Therefore, we speculated that the main component of the vasoconstriction may be the ophthalmic artery and that marked retinal vasodilatation at the end and immediately after the attack may represent compensation for hypoxia of the retinal tissues.

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Author Contributions: Study concept and design: All authors. Acquisition of data: Ota, Kuroshima. Analysis and interpretation of data: Ota, Kuroshima. Drafting of the manuscript: All authors. Critical revision of the manuscript for important intellectual content: Ota. Obtained funding: Ota. Administrative, technical, or material support: Ota, Kuroshima. Study supervision: Nagaoka.

Conflict of Interest Disclosures: None reported.


Novel Compound Heterozygous Mutations Resulting in Cone Dystrophy With Supernormal Rod Response

Cone dystrophy with supernormal rod response (CDSRR) (RCD3B, OMIM #610356) was first described in 2 siblings by Gouras et al1 in 1983. In subsequent reports, it has been characterized as a rare autosomal recessive retinal disorder associated with a delayed and markedly decreased cone and rod response that exhibits an exaggerated, or superthreshold, rod electroretinogram (ERG) in response to higher stimulus levels.2 Reports of CDSRR commonly describe an early onset of dyschromatopsia, photophobia, and central scotoma with poor best-corrected visual acuity.3 Associated signs and symptoms include nyctalopia, nystagmus, and macular retinal pigment epithelium changes.3,4

Genetic studies have linked CDSRR to mutations in the potassium channel, subfamily V, member 2 gene (KCNV2), which is predominantly expressed in retinal photoreceptors and encodes a modulatory subunit of the Kv8.2 voltage-gated potassium channel.4 Mutations in KCNV2 may inhibit proper assembly of heteromeric voltage-gated potassium channels with a subsequent pathologically prolonged outward potassium current in the dark, causing an abnormality in photoreceptor membrane potentials.5 The exclusive link of CDSRR to KCNV2 mutations is a notable contrast from the majority of inherited retinal disorders, which display genetic heterogeneity.

Methods  A complete ophthalmic examination by a retinal physician (S.H.T.) was performed, including fundus autofluorescence using scanning laser ophthalmoscopy (Heidelberg retinal angiograph; Heidelberg Engineering), microperimetry (MP1; Nidek Technologies), and spectral-domain optical coherence tomography (Spectralis optical coherence tomography/ scanning laser ophthalmoscopy; Heidelberg Engineering). An electrophysiological assessment was performed using the Espion 5 system (Diagnosys). Full-field electroretinograms (ERGs) were recorded according to the standards of the International Society for Clinical Electrophysiology of Vision.

Blood samples were genetically screened at Casey Eye Institute Laboratory, Portland, Oregon, for KCNV2 coding region mutations that cause disease. All exons and flanking introns of KCNV2 were directly sequenced on the ABI 3100XL DNA sequencer (Applied Biosystems), and detected variants were analyzed for evolutionary conservation by the prediction programs PolyPhen-2 and SIFT.

Report of a Case  A 47-year-old man presented with decreased visual acuity and a history of hemeralopia and photophobia.
for several years. Medical, ocular, and family histories were unremarkable. Best-corrected visual acuity was 20/150 OD and 20/100 OS. Pupils, extraocular movements, confrontational visual fields, intraocular pressures, and anterior segment examination findings were within normal limits. Color vision was diminished to 3/6 on Hardy-Rand-Rittler plates in both eyes.

On fundus examination, there was bilateral macular atrophy with retinal pigment epithelial changes and pale optic nerves with peripapillary atrophy (Figure 1A and B). The right eye had moderately dense asteroid hyalosis (Figure 1A). Fundus autofluorescence demonstrated bilateral patchy, central hypoa autofluorescence surrounded by a ring of high-density hyperautofluorescence in the macula (Figure 1C and D). Spectral-domain optical coherence tomography revealed diffuse outer retinal atrophy as evidenced by loss of inner segment–outer segment continuity and loss of outer nuclear layer–outer plexiform layer normal architecture as well as granular changes corresponding to the hyperreflective crystalline lesions seen on fundus examination (Figure 1E and F). Mean full-field ERG results and respective stereotypical tracings showed significant delays and amplitudinal loss in the photopic cone and 30-Hz flicker ERGs indicating generalized cone system dysfunction (Figure 2). Bipolar cell–corresponding b-waves were reduced compared with the photoreceptor–specific a-wave. The dim-flash, scotopic rod ERGs (0.01 candela · s/m²) showed profoundly delayed b-waves, and the bright-flash, maximum cone
and rod ERGs (11 candelas · s/m²) showed a significantly increased amplitude along with a prolonged b-wave.

The diagnosis of CDSRR was suspected based on reports of an association between this unique ERG finding and KCNV2 mutations. Genetic testing confirmed the diagnosis and also revealed 2 novel mutations, p.W46X:c.137G>A and p.C177X:c.531T>A, in KCNV2 to possibly account for this novel disease phenotype.

Discussion | There are several novel conclusions to be drawn from this case of CDSRR. Testing with ERG can focus candidate gene screening in patients with dyschromatopsia, nyctalopia, or hemeralopia presenting with distinct bilateral macular atrophy with retinal pigment epithelial changes and diffuse outer retinal atrophy. This case highlights specific optical coherence tomographic findings of diffuse outer retinal atrophy that correspond with previously described mechanistic theories detailing preservation of the inner nuclear layer.\(^3\) Additionally, these findings confirm reports in the literature of older patients presenting with fundus autofluorescence images with a ringlike area of high density encircling retinal pigment epithelial atrophy.\(^2\) It is likely that in patients who present later, milder symptoms and more subtle examination findings are consistent with slowly progressing atrophy outward from the macula, highlighted by complete degradation centrally and incomplete degradation of photoreceptor outer segments in a peripheral ring.

Ensembl analysis revealed that p.C177X:c.531T>A is likely a nonsense truncating mutation in the N-terminal, cytoplasmic tetramerization domain of the Kv8.2 protein, thus impairing its ability to form functional α-unit tetrameric potassium channels (http://www.ensembl.org/). The p.W46X:c.137G>A mutation is a nonsense mutation that can also lead to a truncated protein. Both are loss-of-function alleles.\(^6\)

The 2 mutations identified are novel and thus expand the current knowledge of CDSRR genotype-phenotype descrip-
mutations in the literature. This patient uniquely presented with a more dramatic macular pattern at a much later age than an average patient with CDSRR and had neither nyctalopia nor dyschromatopsia. This would suggest not only that the heterogeneity of phenotypes for CDSRR is much broader than the literature indicates but also that the patient’s 2 novel mutations are mild, having resulted in compound heterozygosity associated with a late presenting phenotype.

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Conflict of Interest Disclosures: None reported.

Funding/Support: This work was supported by the LuEsther T. Mertz Retinal Research Center, Manhattan Eye, Ear, and Throat Hospital, Macular Foundation, Inc, core grant SP30EY019007 from the National Eye Institute, core grant SP30CA136696 from the National Cancer Institute, and unrestricted funds from Research to Prevent Blindness. The Bernard and Shirlie Brown Glaucoma Laboratory, Department of Ophthalmology, Columbia University is supported by grant RO1 EY018213 from the National Institutes of Health, the Foundation Fighting Blindness, Schneeweiss Stem Cell Fund, and the Tistou and Charlotte Kerstan Foundation. Tsang is a member of the RD-CURE Consortium, is a fellow of the Burroughs-Wellcome Program in Biomedical Sciences, and has been supported by the Bernard Becker Association of University Professors in Ophthalmology Award from Research to Prevent Blindness, the Dennis W. Jahnigen Award from the American Geriatrics Society, the Joel Hoffman Scholarship, the Barbara and Donald Jonas Family Fund, and the Professor Gertrude Rothschild Stem Cell Foundation.

Role of the Sponsors: The sponsors had no role in the design and conduct of the study; collection, management, analysis, and interpretation of the data; or preparation, review, or approval of the manuscript.


Peripapillary Chorioretinal Lacunae in a Girl With 3q21.3 to 3q22.1 Microdeletion With Features of Aicardi Syndrome

Aicardi syndrome is characterized by the classic triad of agenesis of the corpus callosum, seizures, and peripapillary chorioretinal lacunae. This disorder occurs exclusively in girls and XXY boys and is presumed to be inherited in an X-linked dominant pattern, although the causative genes have not been identified. We examined a girl with a microdeletion on chromosome 3 who was found to have bilateral peripapillary chorioretinal lacunae with other features of Aicardi syndrome.

Report of a Case A girl was born at 32 weeks’ gestation to a mother with preeclampsia via induced vaginal delivery. At birth, she was noted to have preaxial polydactyly involving her left hand as well as an atrial-septal defect with mild pulmonary valvular stenosis. At age 18 months, she was evaluated for speech and motor delay and staring spells. Examination disclosed a broad flat nasal bridge, epicanthal folds, broad and wide mouth, retromicrognathia, and truncal hypotonia. A chromosomal microarray study revealed a 6-megabase deletion of chromosome 3, spanning from 3q21.3 to 3q22.1.

Ophthalmologic examination showed normal optokinetic responses and reactive pupils with no relative afferent pupillary defect. She had no strabismus, nystagmus, anterior segment anomalies, or refractive error. Retinal examination showed bilateral peripapillary chorioretinal lacunae. In the right eye, a normal right optic disc was abutted by a cluster of poorly circumscribed chorioretinal lacunae with hyperpigmented borders superotemporally (Figure 1). In the left eye, a dysplastic optic disc was encircled by a cluster of well-circumscribed de-pigmented chorioretinal lacunae with variably dense fine pigmentation around the borders (Figure 1). The midperipheral retinas showed multiple additional streaky areas of focal retinal pigment epithelial depigmentation (Figure 1). Magnetic resonance imaging showed thinning of the corpus callosum, mildly decreased white matter volume with dilation of the posterior aspect of the left lateral ventricle, mild cortical thickening with abnormal deep sulcation involving the right parasagittal lobe, and a cavum septum pellucidum (Figure 2).

Discussion In 1946, Krause first described the ocular findings of Aicardi syndrome in an infant girl with seizures, developmental delay, and gray-white plaques in the retina bilaterally. In 1965, Aicardi et al documented the classic findings of this