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## Novel *KCNV2* Mutations in Cone Dystrophy with Supernormal Rod Electroretinogram

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### Purpose

To describe patients with cone dystrophy and supernormal rod electroretinogram (ERG) and search for mutations in the recently described *KCNV2* gene.

### Design

Clinical and molecular study.

### Methods

Patients from three families originating from France, Morocco, and Algeria had standard ophthalmologic examination and color vision analysis, Goldmann perimetry, International Society for Clinical Electrophysiology of Vision (ISCEV) protocol in accordance with ERG testing, autofluorescence evaluation, and optical coherence tomography 3 scanning. The two coding exons of *KCNV2* were polymerase chain reaction amplified and sequenced.

### Results

All patients had the characteristic features of supernormal, delayed rod ERG responses at the highest levels of stimulation and markedly reduced cone responses. In the French family, two affected sisters were compound heterozygotes for the recurrent c.1381G>A (Gly461Arg) mutation and for a novel c.442G>T (Glu148Stop) mutation. In the Moroccan family, affected members were homozygotes for the novel c.1404delC mutation (His468fsX503) and in the Algerian family, the proband was homozygote for the novel c.1001delC mutation (Ala334fsX453). In the three families, parents were unaffected heterozygote carriers. None of the mutations were present in 50 control chromosomes.

### Conclusions

The three novel truncative mutations are likely to be null mutations leading to loss of function, with no difference in the phenotype presentation. Amino acid changes are found exclusively in the N-terminal fragment of the protein and in the P-loop, indicating the importance of those regions for the function of the *KCNV2* protein.

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