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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 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 ophthalmological examination and color vision analysis, Goldman perimetry, ISCEV ERG testing, autofluorescence evaluation and OCT-3 scanning. The two coding exons of *KCNV2* were PCR-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.