Supp. Table S1. Study Sample

Initial Clinical Dx	No. of independent cases/families	Cases with KCNV2 mutations (Σ affected family members)	Families with KCNV2 Deletions (see Fig.1)
Achromatopsia/Blue Cone Monochromacy	167	7 (9)	CHRO8, CHRO158, CHRO390, CHRO417
Cone Dystrophy	154	4 (4)	ZD283
Cone-Rod Dystrophy	43	1 (1)	RCD307
CDSRR	3	3 (7)	-
Σ	367	15 (21)	
CDSRR (Wissinger et al.2008)*	1	1 (1)	BD114

* Patient included for deletion mapping

Supp. Table S2. Clinical diagnosis and genotypes of patients with *KCNV2* mutations

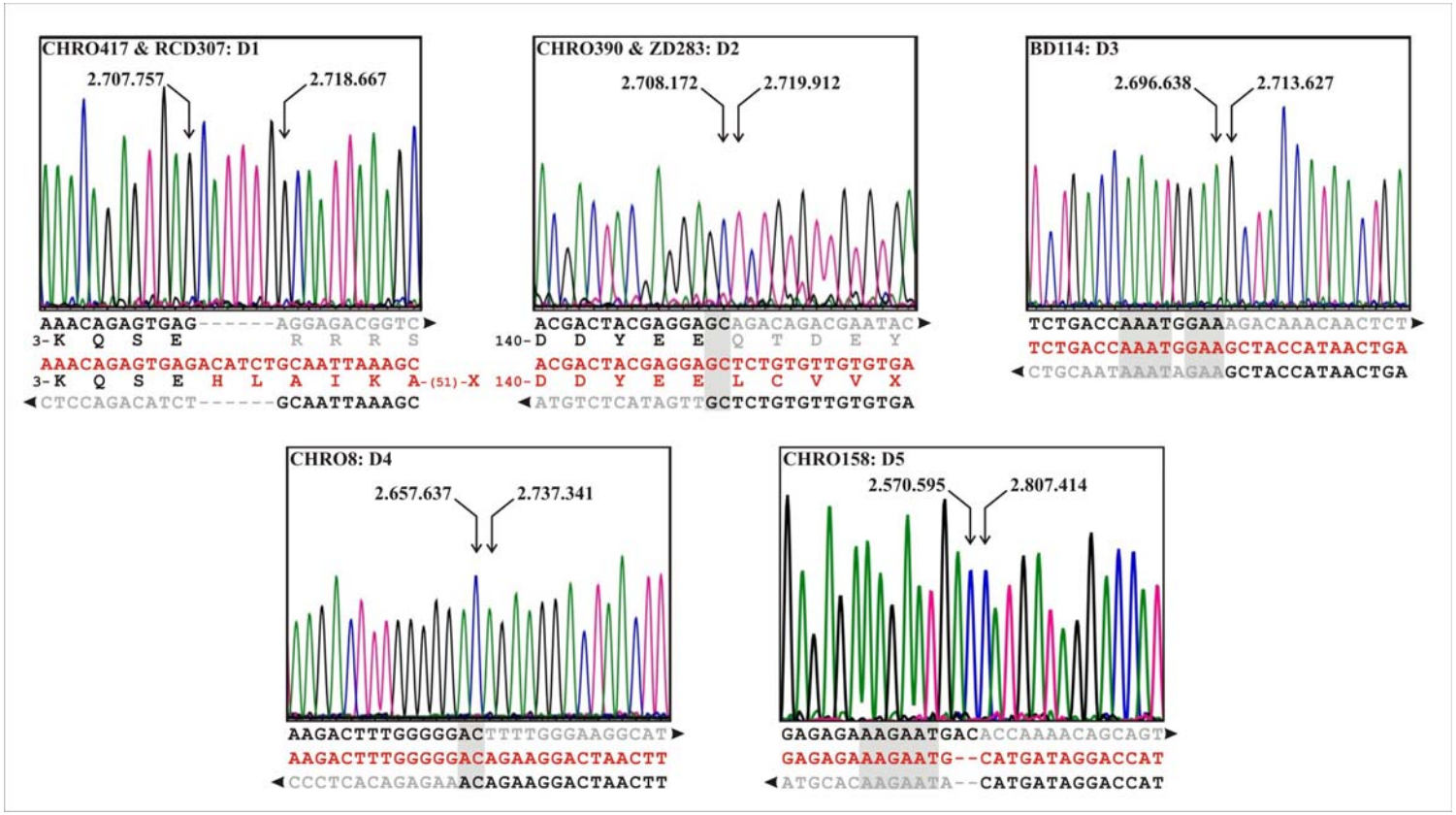
Family	Subject	YOB/G	Initial Dx	Allele 1	Allele 2	Dx after re-evaluation
RCD307	HPM	1949/M	Cone-Rod Dystrophy	D1	D1	CDSRR
ZD164	AS	1968/M	Cone Dystrophy	p.Lys3ArgfsX96	p.Asp339_Val341del	CDSRR
ZD220	KW	1997/M	Cone Dystrophy	p.Phe164Ser	p.Gly461Arg	ERG b/a quotient not sign. elevated in high intensity
RCD272	RO	1999/M	Cone Dystrophy	p.Glu148X	p.Gly461Arg	CDSRR
ZD283	ET	1991/F	Cone Dystrophy	p.Glu148X	D2	CDSRR
CHRO417	BS	1965/F	Achromatopsia/Cone Dystrophy	D1	D1	CDSRR
CHRO450	LK	1990/M	Achromatopsia / cong. Cone Dysfunction	p.Lys3ArgfsX96	p.Glu148X	CDSRR
CHRO8	VH	1975/F	Achromatopsia	p.Leu404Pro	D4	n.d.
CHRO8	MAH	1973/M	Achromatopsia	p.Leu404Pro	D4	n.d.
CHRO158	W07-1637	n.k./F	Achromatopsie	p.Tyr108TrpfsX14	D5	n.d.
CHRO246	ACH13-01	1987/F	Achromatiopsia	p.Trp450Arg	p.Trp450Arg	n.d.
CHRO271	AA	1965/F	Achromatopsia	p.Ala261Asp	p.Ala261Asp	CDSRR
CHRO390	GL	1968/F	Achromatopsie	p.Lys260X	D2	CDSRR
CHRO390	AT	1964/F	Achromatopsia	p.Lys260X	D2	n.d.
RCD382	MB	1990/M	CDSRR	p.Lys3ArgfsX96	p.Ala259_Ala265dup7	-
RCD382	AL	1961/F	Retinal Dystrophy	p.Phe330Ser	p.Ala259_Ala265dup7	n.d.
RCD382	IF	1966/F	Achromatopsia / Cone (Rod) Dystrophy	p.Phe330Ser	p.Ala259_Ala265dup7	CDSRR
RCD382	HU	1967/F	Retinal Dystrophy	p.Phe330Ser	p.Ala259_Ala265dup7	n.d.
RCD425	WM	n.k./M	CDSRR	p.Arg243Trp	p.Ser266ProfsX57	-
ZD389	PGS	1957/M	CDSRR	p.Gly461Arg	p.Gly461Arg	-
ZD389	AS	1961/F	CDSRR	p.Gly461Arg	p.Gly461Arg	-
BD114*	PW	n.k./M	CDSRR	p.Lys120fsX371	D3	-

*reported in Wissinger et al., 2008.; YOB/G – Year ob birth / Gender; n.k. – not known; n.d. – not done

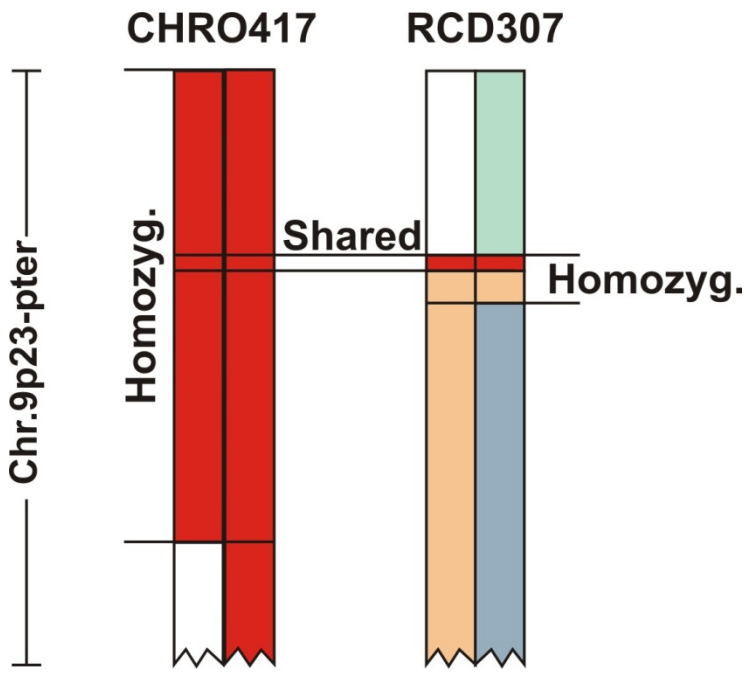
Supp. Table S3. Clinical findings in family RCD382

Patient	At age	VA OD / OS	Refraction	Visual field	Color vision	Fundus	OCT	FAF	ERG Scotopic	ERG Photopic	Mf-ERG
RCD382-MB, male	18.9	0.1 / 0.1	OD -0,25 – 1,25/ 165° OS –0,25 – 0,75/ 32°	Central scotoma for I/4e with 25° diameter; III/4e outer border OD normal, OS slightly constricted	Desaturated panel D15: moderate confusions; Saturated version: OD no mistake, OS 3 mistakes.	Macula slight pigmentary changes, faint yellow dots in periphery	Central thickness reduced to 132 µm	Smaller central area with reduced FAF; macula surrounded by very faint ring of increased FAF	Dim →→ Mixed *a* (→) Mixed *b* ↑→	30Hz↓→ *a* (↓→) *b* ↓→	Remaining responses for peripheral hexagons
RCD382-IF, female	32.5 1st visit	0.03 / 0.03	OD +1,0 -2,0/ 0° OS +1,0 -2,0/ 30°	concentric constriction to ~75° diameter Goldmann V/4e	Multiple confusions even of saturated panel D15; anomaloscope not possible	No macular reflex	—	—	Dim: ∅ Mixed *a* → Mixed *b* ↓→	*30Hz*↓→ *a* → *b* ↓→	—

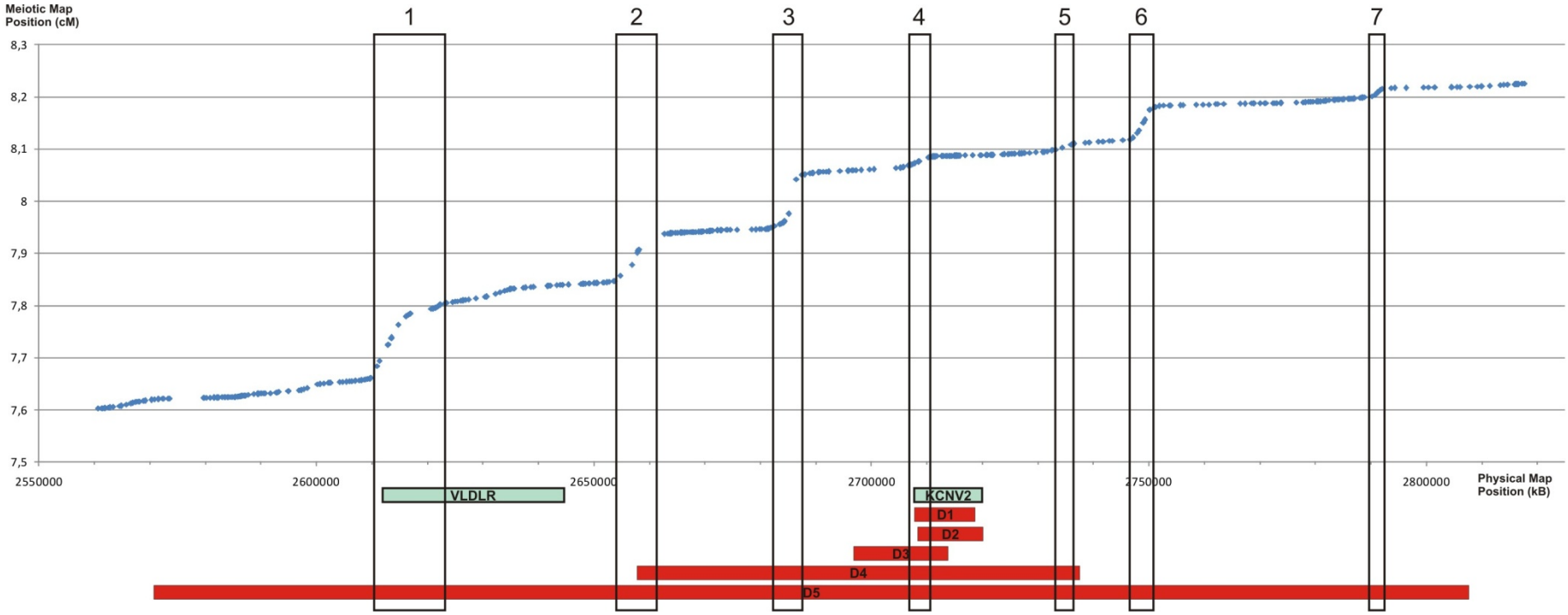
Compilation of clinical data including visual acuity (VA), refraction, Goldmann visual fields, fundus, Optical coherence tomography (OCT), Fundus autofluorescence (FAF), electroretinograms (ERG) for scotopic dim a- and combined *a*- and *b*-wave response, photopic 30Hz and *a*- and *b*-wave to single flashes (symbols: ↑ larger, ↓ smaller, → delayed or border line if in brackets, √ amplitude and peak time normal, ∅ not recordable with separate information on both eyes as required), and multifocal ERG (Mf-ERG). — denotes results that could not be obtained.



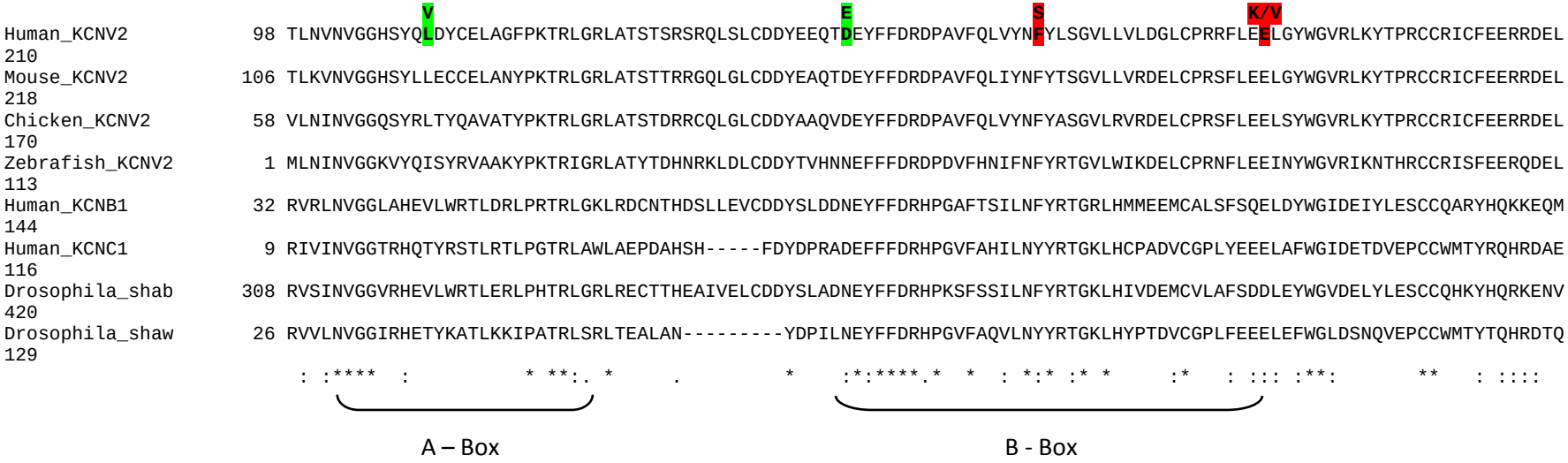
Supp. Figure S1. Breakpoint Sequences of Deletions at the *KCNV2* Locus. Electropherograms of breakpoint sequences as obtained from long-distance PCR amplifications covering the deletions. Left and right junction sequences are in bold and the deletion allele sequence is given in red. Small stretches of conserved sequences at the breakpoint junctions are boxed in grey. The telomeric junction of the deletions D1 and D2 are located within the coding sequence of *KCNV2* and the deduced amino acid sequence of the wildtype and mutant allele is depicted.



Supp. Figure S2. Common Origin of the Deletion D1 in Families CHRO417 and RCD307. Homozygosity intervals and haplotypes on chromosome 9p23-pter of the two affected family members of families CHRO417 and RCD307 as reconstructed from high density SNP arrays. Though homozygosity intervals are much larger in each individual, in particular for CHRO417, the shared region of identity containing 162 SNPs is rather small (about 300 kb). Breakpoints disrupting the shared region on both allelic chromosomes of RCD307 were reconstructed in close vicinity to the 5' end of *KCNV2*.



Supp. Figure S3. Some Deletion Breakpoints co-localize with recombinational Hotspots. The graph displays the non-linearity between recombination frequency and physical distance in the analyzed region on chromosome 9. Each datapoint represent a SNP that has been placed on the genetic map based on HapMap data. The region contains four strong recombinational hotspots (>20cM/Mb; boxes 1-3, 6) and three weak recombinational hotspots (>5cM/Mb ; boxes 4- 5, 7). The telomeric junctions of deletions (D1, D2 and D4) co-localize with such hotspots and the centromeric junction of deletion D4 maps close to hotspot #5.



Supp. Figure S4. Positioning and Evolutionary Conservation of Amino Acid Residues investigated by Yeast-two-Hybrid Assays. Alignment of N-terminal amino acid sequences of KCNV2 for various species, as well as human KCNB1 (Kv2.1) and KCNC1 (Kv3.1) and drosophila shab and shaw potassium channels. The position and deduced amino acid substitution of proposed *KCNV2* mutations (in red) and unknown rare variants (in green) that were investigated by the yeast-two-hybrid interaction assay are indicated. A-Box and B-Box denote sequence elements that are responsible for potassium channel subunit recognition and assembly.