

**Table 2 : Clinical features in members from the 3 families with cone dystrophy with supernormal rod electroretinogram**

	KCNV2 protein	Functional signs	Age at exam	Refraction Visual acuity OD / OS	Fundus	Color vision	Goldman perimetry	Electroretinogram of right eye * Dark adapted maximum at 0 dB (rod cone) Light adapted 30-Hz flickers (cone)
<b>Family 1</b>								
III-2	G461R/ (E148X)	nystagmus at age 2 photophobia mild night blindness	11	-1.75(-2.25;115°): 5/10 -0.50(-1.25;65°): 4/10	loss of foveal reflex fine granular pigmentation of the macula	Subnormal	6° central scotoma at I4	supernormal at 634 μV severely decreased at 43 μV
III-3	G461R/ E148X	no nystagmus photophobia mild night blindness	8	-1.75(-1.25;160°): 3/10 -1.75(-1.00;10°): 3/10	loss of foveal reflex	red/green axis of dyschromatopsia	15° central scotoma at I4	supernormal at 662 μV severely decreased. at 47 μV
<b>Family 2</b>								
II-1	H468fsX503/+	none	56	+1.00(-0.75;105°): 10/10 +2.25(-3.00;150°): 10/10	normal	mild tritanopia	not done	normal at 352 μV normal. at 111 μV
II-2	H468fsX503/+	none	54	+1.00(-0.75;115°): 10/10 +1.50(-0.75;155°): 10/10	normal	mild tritanopia	not done	normal at 304 μV moderately decreased. at 60 μV
III-1	+/+	none	38	emmetropic : 10/10 emmetropic: 10/10	normal	subnormal	not done	normal at 359 μV subnormal at 89 μV
III-2	H468fsX503/+	none	33	+0.25(-0.75;165°): 10/10 +0.50(-1.75;140°): 10/10	few depigmented areas in mid periphery ; posterior pole normal	mild tritanopia	not done	normal at 339 μV moderately decreased. at 83 μV
III-4	+/+	none	31	emmetropic : 10/10 emmetropic : 10/10	normal	subnormal	not done	moderately decreased at 275 μV moderately decreased. at 69 μV
III-5	H468fsX503/+	none	25	emmetropic : 10/10 emmetropic : 10/10	normal	subnormal	not done	normal at 408 μV normal. at 107 μV
III-6	H468fsX503/ H468fsX503	no nystagmus intense photophobia no night blindness	22	+1.00(-1.50;160°): 1.5/10 -2.00(-1.25;175°): 1.5/10	loss of foveal reflex	red/green axis of dyschromatopsia	5° central scotoma at I4	supernormal at 690 μV highly decreased at 27 μV
III-7	H468fsX503/ H468fsX503	no nystagmus photophobia mild night blindness	17	-7.00(-1.25;170°): 1.5/10 -7.50(-1.00;30°): 1.5/10	loss of foveal reflex atrophy of peripapillary and inferior retina (myopia)	red/green axis of dyschromatopsia	20-30° central scotoma at I4	supernormal at 560 μV severely decreased. at 37 μV
III-8	H468fsX503/ H468fsX503	nystagmus at age 2	9	-15.75(-1.75;5°): 1/20 -16.00(-1.25;140°): 1/20	foveal depigmentation atrophy of retina (myopia)	scotopic axis of dyschromatopsia	not done	not done

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**Family 3**

<b>II-2</b>	affected not tested	no nystagmus photophobia no night blindness	54	+0.75(-2.25; 80°): 1/10 +0.50(-2.00; 20°): LP	round-shaped foveal atrophy	red green axis of dyschromatopsia	not done	supernormal at 650 $\mu$ V moderately decreased. at 54 $\mu$ V
<b>II-3</b>	A334fsX453/ A334fsX453	no nystagmus photophobia no night blindness	38	-8.00(-0.75; 5°): 1/10 -8.00(-1.25; 170°): 1/10	round-shaped foveal atrophy	red green axis of dyschromatopsia	central relative scotoma	supernormal at 588 $\mu$ V highly decreased at 15 $\mu$ V
<b>III-2</b>	affected not tested	no nystagmus photophobia no night blindness	23	-7.00(-1.00; 90°): 2/10 -10.50(-0.25; 165°): 1/20	round-shaped foveal atrophy	red green-scotopic axis of dyschromatopsia	no central scotoma on right eye	supernormal at 469 $\mu$ V highly decreased at 28 $\mu$ V

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\* : normal values are : dark adapted maximum at 0 dB = 300-450  $\mu$ V ; light adapted 30-Hz flickers = > 105  $\mu$ V. LP = light perception