

Table 3: clinical signs in the family with mutation c.730_731delATinsG (pedigree on figure 1B)

Family member	History of symptoms	Age on examination Visual acuity	Lens	Fundus	OCT-3	Visual field	Electroretinogram
IV1	No history of visual symptoms	57 OD: 4/10 with +1.00(-1.00;80°) OS: 10/10 with +0.75(-0.50;100°)	OD : clear OG : sectorial cataract	Normal	Not done	Not done	OD : hypovolted in photopic white, flat in scotopic blue OS : normal
IV2	No history of visual symptoms	54 OD: 10/10 with +1.75(-0.50;90°) OS: 10/10 with +1.75(-0.50;30°)	OU : inferior cataract	Normal	Not done	Not done	OU : Normal
V1	No history of visual symptoms	18 OD: 8/10 with -8.00(-0.25;85°) OS: 8/10 with -7.00(-0.25;95°)	OU : clear	Normal	Not done	Not done	OD : hypovolted in photopic white and scotopic blue OS : normal
V2	Night blindness from early childhood Moves normally No photophobia	20 OD : 7/10 with -2.50(-0.50;50°) OS : 7/10 with -0.75(-0.50;170)	OU: clear	Few bone spicule-shaped pigment deposits in periphery with retina looking unglossy and whitish Slight attenuation of retinal vessels	Moderate loss in photoreceptors, both foveal and periphery of the macula (210 μ M thickness) No macular edema	Horse shoe shaped scotoma open upwards, in mid periphery, between 10-20° and 40-50° (OD), and 10-17° and 50-60° (OS)	OU : highly attenuated response in photopic white Flat in scotopic blue and 30 Hz-flickers
V3	Night blindness began at 17 Difficulties in moving began at 20 Moderate photophobia	26 OD : 1/10 (-1.25) OS : 1/10 (-1.50)	OU : posterior central subcapsular cataract	Bone spicule-shaped pigment deposits on nasal retina Attenuation of retinal vessels Macular epiretinal membrane	Important loss in photoreceptors, both foveal and periphery of the macula (140-180 μ M thickness) No macular edema	Goldman V4e isopter at 10° OU	Unrecordable for all types of stimulation
V4	No history of visual symptoms	29 OD: 8/10 with +0.5(-0.75;110°) OS: 10/10	OU : clear	Normal	Not done	Not done	OU : Normal
V5	Night blindness Does not move by himself	31 OD: LP with -8.00 OS: LP with -8.00	OD : posterior and anterior central subcapsular cataract OG: posterior central subcapsular cataract	Dense bone spicule-shaped pigment deposits on 360° in mid periphery Attenuation of retinal vessels and waxy pale optic disc	Not done	Undetectable	Unrecordable for all types of stimulation
V6	No history of visual symptoms	33 OD: 10/10 with +0.50(-2.25; 85°) OS: 12/10 (-2.00;85°)	OU : clear	Normal	Not done	Not done	Not done