

Patient ID	Age at Diagnostic Exam (yr)	Visual field (Hx)	Visual acuity OD	Visual acuity OS	Spherical Equivalent OD	Spherical Equivalent OS	ERG	Flash VEP	AO performed	OCT/RL	AI (ganglion cell) hyperfucrescence (muV)	Fundus RL	First symptoms	Age of onset	Hypertension	Strabismus	PSS	Comment	
0001: 15 patients																			
0007b	39	not recordable	0.03	0.04	-1.5	-1.75	extinguished	-	-	loss of photoreceptor layer, LHM wrinkling	yes	RP	right blindness	childhood	no	no	total color discrimination impairment	RL, ICL at age of 39	
0004b	16	RL, ICL: concentric constriction 8-10°	0.03	0.03	-	-	extinguished	-	-	loss of photoreceptor layer, deposits adjacent to Bruch's membrane, foveal atrophy	yes	RP	right blindness, light photophobia	childhood	no	no	total color discrimination impairment	at age of 15 (unilateral)	
0003b	34	RL, ICL: 3-4° at age of 34	0.1	0.08	-	-	extinguished	good amplitude	-	loss of photoreceptor layer, deposits adjacent to Bruch's membrane, LHM wrinkling	yes	RP	right blindness	20 yr	no	no	total color discrimination impairment	RL, ICL at age of 34	
0007b	46	not recordable	HM	HM	-	-	extinguished	severe reduction in amplitude	-	loss of photoreceptor layer, deposits adjacent to Bruch's membrane, LHM wrinkling	yes	RP	right blindness	childhood	no	yes, LD CP	total color discrimination impairment	RL, ICL at age of 46	
0005b	33	RL, ICL: 2°	0.2	0.25	-1.75	-2	extinguished	good amplitude	yes	loss of photoreceptor layer, foveal atrophy	yes	RP	right blindness	childhood	no	no	total color discrimination impairment	RL, ICL at age of 33	
0005b-0	36	RL, ICL: 3°	0.25	0.05	-0.25	-0.5	extinguished	pattern VEP: severe reduction in amplitude	yes	loss of photoreceptor layer, deposits adjacent to Bruch's membrane, LHM wrinkling	yes	RP	right blindness	childhood	no	yes, LD CP	total color discrimination impairment	RL, ICL at age 36 (unilateral)	
0006b	34	not recordable	0.03	0.03	2	1.5	extinguished	severe reduction in amplitude	-	loss of photoreceptor layer, deposits adjacent to Bruch's membrane, LHM wrinkling	yes	cone-rod dystrophy	light photophobia, right blindness	6 yr	no	no	total color discrimination impairment	at age of 34 (unilateral)	
0001b	43	not recordable	HM	HM	-	-	extinguished	severe reduction in amplitude	-	loss of photoreceptor layer, foveal atrophy, LHM wrinkling	yes	RP	right blindness	30 yr	yes	no	total color discrimination impairment	RL, ICL at age of 43 (unilateral)	
0000b	14	RL, ICL: 5-7°	0.05	0.05	0.5	0.5	extinguished	good amplitude	-	loss of photoreceptor layer, severe macular atrophy, LHM wrinkling (epithelial membrane)	yes	RP	right blindness	childhood	no	no	total color discrimination impairment	RL, ICL at age of 14	
0007b-0	56	not recordable	HM	HM	-	-	extinguished	good amplitude	-	loss of photoreceptor layer, foveal atrophy, LHM wrinkling (epithelial membrane)	yes	RP	light photophobia	14 yr	yes	yes	total color discrimination impairment	RL, ICL at age of 56	
0007b	15	RL, ICL: 1.5-7°	0.2	0.25	-7	-6.75	extinguished	good amplitude	-	loss of photoreceptor layer, foveal atrophy, LHM wrinkling	no	RP	right blindness	4 yr	no	no	Normal	no	
0004b-0	18	Perimetry 10°, 10° nasal, superior, 10° temporal and 60° inferior; good function (no peripheral)	0.01	0.05	-4.75	-3.5	extinguished	good amplitude	-	persistence of the photoreceptor layer cells outside the fovea	yes	cone dystrophy (age 18)	light photophobia	7 yr	no	no	Normal	no	
0007b-0	30	not recordable	0.02	0.025	4.5	3.5	extinguished	good amplitude	-	loss of photoreceptor layer, LHM wrinkling	yes	RP	right blindness	childhood	yes	no	not possible	at age of 30	
0004b	13	RL, ICL: 6-8°	0.05	0.05	1	1	extinguished	reduction in amplitude	-	loss of photoreceptor layer, foveal atrophy, LHM wrinkling	yes	rod-cone dystrophy	right blindness, nyctalopia	1 yr	yes	no	total color discrimination impairment	RL, ICL at age of 13	
0007b	11	not recordable	0.25	0.25	4	3.5	extinguished	good amplitude	-	loss of layer of photoreceptor cells outside the fovea, LHM wrinkling	no	cone-rod dystrophy	right photophobia, right blindness	childhood	no	no	not possible	no	
0002: 3 patients																			
0003b	21	not recordable	0.125	0.1	-3	-3	extinguished	-	-	loss of photoreceptor layer, foveal atrophy, internal limiting membrane wrinkling	no	RP	right blindness, light photophobia	childhood	yes	childhood	total color discrimination impairment	RL, ICL at age of 21	
0005b	25	RL, ICL: 5-7°	0.1	0.2	-3.25	-3.5	extinguished	-	-	loss of photoreceptor layer outside the fovea, internal limiting membrane wrinkling	yes, built's eye	cone-rod dystrophy	light photophobia, right blindness	8 yr	no	yes	total color discrimination impairment	RL, ICL at age of 25	
0007b-0	8	not recordable	0.4	0.5	0.4	0.5	extinguished	-	-	loss of photoreceptor layer outside the fovea, internal limiting membrane wrinkling	no	cone-rod dystrophy	light photophobia	5 yr	no	no	not done	no	
0003: 3 patients																			
0004b-0	23	RL, ICL: 30°	0.125	0.125	-4.5	-4	remaining scotopic response	-	-	loss of photoreceptors in fovea with good layer of photoreceptors towards periphery	yes	cone dystrophy	light photophobia	childhood	no	no	total color discrimination impairment	no	
0004b-0	13	central scotoma ex	0.25	0.25	-6.75	-7.25	remaining scotopic response	-	-	loss of photoreceptor layer towards periphery	yes	cone-rod dystrophy	visual acuity impairment, light photophobia	childhood	no	no	total color discrimination impairment	no	
0006b	17	not recordable	0.03	0.05	-10.5	-10.5	extinguished	good amplitude	-	LD: loss of photoreceptor layer outside the fovea, internal limiting membrane wrinkling	no	cone-rod dystrophy	right blindness, nyctalopia	1 yr	yes	yes, RD	total color discrimination impairment	at age of 17 yrs (bilateral)	
0004: 3 patients																			
0003b	23	not recordable	HM	HM	-3	-3.25	extinguished	-	-	loss of photoreceptor layer, foveal atrophy, internal limiting membrane wrinkling	yes	RP	right blindness	childhood	yes	yes	total color discrimination impairment	no	
0004b-0	30	not recordable	0.03	0.05	-8.5	-9	extinguished	reduction in amplitude	-	loss of photoreceptor layer, foveal atrophy, internal limiting membrane wrinkling	yes, built's eye	RP	right blindness	childhood	yes	no	total color discrimination impairment	RL, ICL at age of 29 (bilateral)	
0005b	22	not recordable	0.006	0.06	10	10	extinguished	-	-	LD: loss of photoreceptor layer, foveal atrophy	yes	RP	right blindness, nyctalopia	1 yr	yes	childhood	total color discrimination impairment	at age of 22	
0005: 3 patients																			
0006b-0	20	RA about 4°, LA about 3°, peripheral temporal scotoma remaining islands at age of 20	0.16	0.16	-0.75	-0.75	extinguished	Pattern VEP: severe reduction in amplitude	yes	loss of photoreceptor cell layer outside the fovea, internal limiting membrane wrinkling	yes	rod-cone dystrophy	right blindness	childhood	yes	yes	total color discrimination impairment	at age of 20 (bilateral)	
0006b-0	7	not recordable	0.5	0.1	-3.5	-1	NP	-	-	loss of photoreceptor cell layer outside the fovea, internal limiting membrane wrinkling	no	rod-cone dystrophy	visual acuity impairment at age 1 yr	no	no	not possible	no		
0004b	37	not recordable	NLP	NLP	-	-	extinguished	-	-	RL: severe loss of photoreceptor layer	yes at age 37 yr	RP	right blindness	10 yr	no	no	not possible	no	
0006: 1 patient																			
0003b	48	not recordable	0.05	NLP	-8.5	(profound)	extinguished	-	-	severe loss of photoreceptor layer in RE	no	RP	light photophobia, right blindness	childhood	yes	LD: enucleation	not possible	RL, ICL at age of 48	
0007: 3 patients																			
0007b	20	RL, ICL: 30°	0.07	0.05	1.5	1	extinguished	good amplitude	-	loss of photoreceptor layer outside the fovea	yes	RP	right blindness, light photophobia	childhood	no	no	total color discrimination impairment	at age of 20	
0002b	17	not recordable	0.038	0.02	-4.25	-5	extinguished	Pattern VEP: good amplitude	-	loss of photoreceptor layer, foveal atrophy, internal limiting membrane wrinkling	yes, built's eye	RP	right blindness, light photophobia	childhood	no	no	normal	no	
0006b	17	not recordable	0.04	0.06	3	2.5	extinguished	good amplitude	-	loss of photoreceptor layer, foveal atrophy, internal limiting membrane wrinkling	yes, built's eye	RP	light photophobia	12 yr	yes	yes	total color discrimination impairment	at age of 17 (bilateral)	
0008: 1 patient																			
0007b	17	RL, ICL: 20-25°	0.125	0.05	3	3	extinguished	-	yes	loss of photoreceptor layer, foveal atrophy, internal limiting membrane wrinkling	yes	RP	right blindness	6 yr	yes	yes, LD	not possible	RL, ICL at age of 17	
0009: 5 patients																			
0004b-0	43	not recordable	LS	LS	-0.25	-0.5	extinguished	severe reduction in amplitude	-	LD: loss of photoreceptor layer, foveal atrophy, internal limiting membrane wrinkling	no	rp	right blindness	childhood	yes	yes, RA	total color discrimination impairment	RL, Hemianopia	
0004b-0	30	not recordable	HM	CP	1.25	2	extinguished	-	-	loss of photoreceptor layer, foveal atrophy	yes, built's eye	RP	right blindness, light photophobia	childhood	yes	yes	total color discrimination impairment	at age of 30	
0004b-0	20	RL, ICL: peripheral remaining islands	0.25	0.25	1	1.25	extinguished	-	-	loss of photoreceptor layer, foveal atrophy	yes, built's eye	rod-cone dystrophy	right blindness	childhood	yes	yes	total color discrimination impairment	RL, ICL at age of 16	
LC470	39	not recordable	LS	LS	-1.5	-1	NP	-	-	loss of photoreceptor layer, foveal atrophy	yes, built's eye	RP	right blindness, visual acuity impairment at age 10 yr	childhood	yes	yes	total color discrimination impairment	no	
0007b	6	not recordable	0.63	-0.3	-3.25	-3.25	extinguished	-	-	NP	no	cone-rod dystrophy	right blindness at age 1 yr	childhood	no	no	total color discrimination impairment	no	
0010: 20 patients																			
0001b	11	RE 3-9°, LE 7-10°	0.25	0.2	-4	-4	extinguished	pattern VEP: severe reduction in amplitude	-	loss of photoreceptor layer, foveal atrophy, LHM wrinkling	yes	RP	right blindness	2 yr	until age of 5	yes	total color discrimination impairment	no	
0004b	38	not recordable	LP	LP	-	-	extinguished	-	-	loss of photoreceptor layer, foveal atrophy, LHM wrinkling	yes	RP	right blindness, light photophobia	childhood	yes	yes	total color discrimination impairment	RL, ICL at age of 38	
0004b	34	poor fixation	0.06	0.06	-2	-2.25	extinguished	-	-	loss of photoreceptor layer, foveal atrophy, LHM wrinkling	yes	RP	right blindness	10 yr	yes	no	total color discrimination impairment	at age of 34	
0001b	37	not recordable	0.02	0.02	-9	-1.25	extinguished	good amplitude	-	loss of photoreceptor layer, foveal atrophy	no	rod-cone dystrophy	right blindness	childhood	yes	no	total color discrimination impairment	at age of 37 (bilateral)	
0003b	23	not recordable	0.02	LS	-3.25	-3.5	extinguished	good amplitude	-	loss of photoreceptor layer, foveal atrophy, LHM wrinkling	yes	RP	right blindness, light photophobia	childhood	yes	no	total color discrimination impairment	at age of 23	
0004b	5	not recordable	0.4	0.2	3.25	4.75	extinguished	-	-	loss of photoreceptor layer outside the fovea	yes	cone-rod dystrophy	visual acuity impairment at age 1 yr, nyctalopia since first year	no	no	yes	total color discrimination impairment	no	
0003b	28	no fixation, nyctalopia, not recordable	HM	0.1	-5	-3.5	extinguished	good amplitude	-	loss of photoreceptor layer, foveal atrophy, LHM wrinkling	yes	RP	right blindness	childhood	yes	no	total color discrimination impairment	RL, ICL at age of 28	
0005b	13	RL, ICL: 1.5-10°	0.3	0.3	-2.5	-2.5	extinguished	-	yes	loss of photoreceptor layer outside the fovea, LHM wrinkling	no	RP	strabismus at age 1 yr	childhood	no	yes, RD	not possible	no	
0007b	7	not recordable	0.25	0.25	-2.25	-4.75	extinguished	-	yes	loss of photoreceptor layer outside the fovea, LHM wrinkling	yes	cone-rod dystrophy	right blindness	childhood	no	no	total color discrimination impairment	no	
0002b	6	RL, ICL: 3-2°	0.05	HM	-3.25	-3.25	extinguished	-	-	NP	yes, at age 25 yr	RP	right blindness	childhood	yes	no	total color discrimination impairment	no	
0007b	24	RL, ICL: concentric constriction 10° Crow	0.125	0.125	-4.5	-5.5	extinguished	-	-	loss of photoreceptor layer, foveal atrophy, LHM wrinkling	yes	cone-rod dystrophy	right blindness	6 yr	no	no	total color discrimination impairment	at age of 24	
0002b	12	severely asymmetric binocular hemianopia, RL: concentric constriction 10°, central 40°, inferior 40°	0.2	0.05	1	0.5	extinguished	reduction in amplitude	-	loss of photoreceptor layer, foveal atrophy, LHM wrinkling	yes	rod-cone dystrophy	right blindness, light photophobia	3 yr	yes	no	total color discrimination impairment	no	
0007b	41	not recordable	amaurotic	amaurotic	-	-	extinguished	-	-	loss of photoreceptor layer, foveal atrophy, LHM wrinkling	no	RP	light photophobia, right blindness	childhood	no	no	not done	RL, ICL at age of 41	
14095	41	not recordable	LP	HM	-	-	extinguished	reduction in amplitude	-	severe atrophy of photoreceptor layer	no	RP	right blindness	childhood	yes	yes	total color discrimination impairment	at age of 41 (bilateral)	
0005b	31	not recordable	HM	HM	1.5	2.25	extinguished	-	-	loss of photoreceptor layer, foveal atrophy, LHM wrinkling	yes	RP	right blindness, light photophobia	childhood	yes	no	perimetry was defect	at age of 31 (bilateral)	
0007b	9	not recordable	0.02	0.1	-0.5	-0.5	extinguished	-	-	loss of photoreceptor layer, foveal atrophy, LHM wrinkling	no	RP	right blindness, light photophobia	childhood	2 yr	yes	no	not possible	no
0006b	9	not recordable	0.125	0.1	2	2	extinguished	-	-	loss of photoreceptor layer, foveal atrophy, LHM wrinkling	no	cone-rod dystrophy	right blindness, strabismus at age 2 yr	2 yr	yes	yes, LD	total color discrimination impairment	no	
0004b	36	not recordable	LS, LS	LS, LS	-5	-5.5	extinguished	-	-	loss of photoreceptor layer, foveal atrophy, LHM wrinkling	no	RP	right blindness	childhood	yes	no	total color discrimination impairment	RL, ICL at age of 36	
0007b	19	not recordable	0.02	0.06	-2.5	-2	extinguished	good amplitude	-	LD: loss of photoreceptor layer, foveal atrophy, LHM wrinkling	yes	RP	right blindness	childhood	yes	no	total color discrimination impairment	no	
IC2768	29	RL, ICL: concentric constriction with beginning of ring systems, central scotoma	0.05	0.05	0.5	0.5	remaining scotopic response	-	-	foveal atrophy with persistence of the layer of photoreceptor cells outside the fovea	yes	cone-rod dystrophy	light photophobia	14 yr	no	no	total color discrimination impairment	no	
0012: 3 patients																			
0009b	21	relative central scotoma	0.8	0.63	0.8	0.63	scotopic response	-	-	An intact photoreceptors layer except for a small loss in fovea area (cone dystrophy)	yes	cone dystrophy	light photophobia, right blindness	17 yr	no	no	normal	no	
0005b	16	Perimetry 10°, 10° nasal, superior, 10° temporal and 60° inferior (good function in periphery)	0.4	0.3	-5	-4	scotopic response	-	-	little loss of photoreceptors in fovea with good layer of photoreceptors outside towards periphery	yes	cone dystrophy (age 16), pericapsular myopia (age 16)	visual acuity impairment	11 yr	no	no	normal	no	
0004b	26	not recordable	HM	LS	-3.5	-3.5	extinguished	-	-	severe loss of the layer of photoreceptors	yes	rod-cone dystrophy	right blindness	childhood	yes	yes	total color discrimination impairment	RL, ICL at age of 26	
0016: 1 patient																			
0009b	42	RL, ICL: 2-7° with large remaining islands nasal inferior	0.12	0.4	0.5	0.5	extinguished	-	-	loss of photoreceptor layer outside the fovea, LHM wrinkling	no	RP	right blindness	16 yr	no	no	total color discrimination impairment	RL, ICL at age of 42	