

A Patients	Age (years)	BCVA LogMAR	OCT		Full-field ERG						
			Foveal thickness (μm)	Total macular volume (mm^3)	Scotopic response						
					Rod b-wave amplitude (μV)	Rod b-wave implicit time (ms)	St. combined a-wave amplitude (μV)	St. combined a-wave implicit time (ms)	St. combined b-wave amplitude (μV)	St. combined b-wave implicit time (ms)	St. combined b/a-wave amplitude ratio
Family 1 IV/2	12	0.3	525	8.89	167	85	290	20	330	58	1.14
		0.25	484	9.35	157	82	268	19	368	65	1.37
Family 1 II/3	58	0.38	455	7.11	83	94	219	26	250	58	1.14
		2	121	5	41	95	143	30	186	59	1.3
Family 2 III/1	7	0.6	505	8.5	115	84	253	25	339	50	1.34
		0.52	591	9.05	129	89	253	25	360	50	1.43
Family 2 II/2	25	0.38	339	6.97	211	124	336	20	340	47	1.01
		0.38	621	8.41	116	97	302	20	304	46	1
Family 3 III/2	4	0.15	330	7.17	-	-	-	-	-	-	-
		0.15	355	7.56	-	-	-	-	-	-	-
Family 3 III/3	7	0.7	1923	34.06	138	92	272	21	197	40	0.72
		0.3	1003	20.75	185	88	308	20	258	42	0.83
Family 4 II/2	18	0.25	392	7.79	101	129	198	20	190	40	0.96
		0.6	468	8.39	81.7	86	271	19	263	39	0.97
Family 5 III/2	34	0.7	41	6.09	57.9	77	250	21	258	44	1.03
		1	58	5.96	60.2	87	208	20	201	39	0.97
Family 6 III/1	6	0.6	433	9.69	58.6	96	364	36	286	51	0.79
		0.6	460	9.55	84.1	105	306	35	249	51	0.81
Family 7 II/1	13	0.4	564	9.18	120	83	318	18	276	39	0.87
		0.52	568	9.03	111	89	327	18	294	38	0.9
Family 8 II/1	11	0.22	429	6.69	25.2	59	218	17	290	36	1.33
		0.3	423	6.84	26	63	195	17	356	38	1.83
Family 9 II/4	22	0.48	621	8.67	102	94	269	18	292	39	1.09
		0.48	647	9.17	72.6	88	253	18	249	39	0.98
Family 10 II/3	26	0.38	589	8.56	64	98	220	21	236	58	1.07
		0.38	619	9.23	75.4	113	189	20	245	60	1.3
Family 11 III/2	13	0.3	376	7.17	158	103	285	19	401	51	1.41
		0.38	312	7.54	119	107	263	23	347	51	1.32
Family 12 III/4	21	0.38	347	7.35	256	134	236	18	208	39	0.88
		0.15	330	7.18	162	113	243	21	220	38	0.9

Family 12 II/1	31	0.25	91	5.71	93.3	97	275	20	231	38	0.84
		0.3	97	5.74	57.6	94	247	20	208	36	0.84
Family 12 II/2	33	0.7	126	5	89	84	335	22	316	41	0.94
		0	115	5.89	119	74	303	23	368	41	1.21
Family 12 II/5	46	0.3	136	6.13	31.7	75	224	23	158	36	0.71
		0.3	119	6.09	37.6	75	200	21	143	37	0.72
Family 13 IV/2	13	0.3	607	9.7	36.6	111	219	19	165	39	0.75
		0.3	638	10.27	66.7	114	203	19	164	39	0.81
Family 13 III/3	26	0.2	421	6.51	127.9	80	387.7	14	411.1	63.5	1.06
		0.15	128	5.22	124.5	80.5	345.7	13.5	376	63	1.09
B Members / eyes	Age (years)	BCVA LogMAR	OCT		Full-field ERG						
			Foveal thickness (μm)	Total macular volume (mm^3)	Scotopic response						St. combined b/a-wave amplitude ratio
					Rod b-wave amplitude (μv)	Rod b-wave implicit time (ms)	St. combined a-wave amplitude (μv)	St. combined a-wave implicit time (ms)	St. combined b-wave amplitude (μv)	St. combined b-wave implicit time (ms)	
Controls 27/54	20.1 $\pm$ 8.2	0	156.7 $\pm$ 16.5	6.96 $\pm$ 0.26	320.2 $\pm$ 48.2	79.6 $\pm$ 5.4	313.6 $\pm$ 64.5	22.5 $\pm$ 1.4	592.5 $\pm$ 85.6	45.6 $\pm$ 3.4	1.91 $\pm$ 0.35
Carriers 13/26	37.7 $\pm$ 13.3	0.01 $\pm$ 0.03	153.2 $\pm$ 14.9	6.84 $\pm$ 0.26	282.2 $\pm$ 82.7	70.3 $\pm$ 21.3	336.5 $\pm$ 76.7	20.5 $\pm$ 3.6	635.9 $\pm$ 109.8	45.9 $\pm$ 2.2	1.94 $\pm$ 0.35
Patients Grp. I (<25 yrs) 12/24	14 $\pm$ 6.1	0.4 $\pm$ 0.15	566.9 $\pm$ 324.9	10 $\pm$ 5.8	116.6 $\pm$ 57.1	96.5 $\pm$ 18.5	268.8 $\pm$ 45	21 $\pm$ 4.9	281.1 $\pm$ 66.9	44.4 $\pm$ 7.5	1.06 $\pm$ 0.28
Grp. II (>25 yrs) 8/16	40.1 $\pm$ 15.3	0.43 $\pm$ 0.3	210.9 $\pm$ 191.8	6.12 $\pm$ 1.26	75.9 $\pm$ 31.8	87.4 $\pm$ 11.5	253.3 $\pm$ 68.3	21 $\pm$ 4.2	256.2 $\pm$ 82.5	48.1 $\pm$ 11.2	1.02 $\pm$ 0.19

				Multifocal ERG	
Photopic response				Photopic response	
Cone b-wave amplitude (μV)	Cone b-wave implicit time (ms)	30 Hz flicker amplitude (μV)	30 Hz flicker implicit time (ms)	Summed amplitude (μV)	Summed implicit time (ms)
67.9	33	66.8	35	38	36
77.2	33	66.1	33	42	35
60	40	48.3	41	36	43
36	43	35.8	43	24	49
74	36	72	34	49	37
59	37	70	36	42	37
70.3	36	75.3	35	46	40
71.1	37	69.6	34	42	42
-	-	-	-	-	-
-	-	-	-	-	-
89	39	71.4	40	37	41.2
109	38	84.2	38	55	40
40.4	38	46.6	39	24	35
61	36	72	36	60	38
87.7	36	83.2	40	37	40
72.4	37	67.1	38	50	39
58.8	52	45	40	21	39
48.8	53	57.1	38	16	39
109	35	89	35	57	36
110	34	89	35	58	33
134	30	56.3	26	75	37
146	29	57.6	26	57	37
106	40.5	75.6	30	53	33
89.8	37.9	60	35	38	35
66.6	41	86.1	40	23	40
47.4	39	83.3	40	32	40
107	33	92.5	33	51	34
79.9	32	65.3	34	44	36
90.2	32	74	31	43	34
82.2	31	83.7	29	40	33

139	31	114	32	59	29
123	32	96.9	32	56	30
105	30	105	29	52	33
143	29	120	27	59	33
92.4	35	72.1	35	43	35
77.5	36	69.6	36	30	36
87.6	35	72.1	35	34	35
77.8	34	66.1	35	28	36
39.6	52.5	-	-	-	-
38.1	69.5	-	-	-	-

				Multifocal ERG	
Photopic response				Photopic response	
Cone b-wave amplitude (μ V)	Cone b-wave implicit time (ms)	30 Hz flicker amplitude (μ V)	30 Hz flicker implicit time (ms)	Summed amplitude (μ V)	Summed implicit time (ms)
191.4 $\pm$ 36.5	31.6 $\pm$ 1.1	149.5 $\pm$ 29.5	28 $\pm$ 1.5	117.9 $\pm$ 24.5	33.4 $\pm$ 0.9
220.4 $\pm$ 53.2	31.8 $\pm$ 1.3	172.9 $\pm$ 41.8	28.4 $\pm$ 1.3	123.4 $\pm$ 17.9	34.5 $\pm$ 1.8
85.3 $\pm$ 25.6	36.3 $\pm$ 5.8	69.9 $\pm$ 12.4	34.3 $\pm$ 3.7	42.6 $\pm$ 9.1	36.5 $\pm$ 2.7
87.5 $\pm$ 34.5	39.4 $\pm$ 10.6	81.8 $\pm$ 25.2	36.1 $\pm$ 5.1	41.8 $\pm$ 13.2	37.3 $\pm$ 5.7