

**Table 7: Subject data**

Nr.	Gender	Onset (y)	Duration (y)	BCVA (LogMAR)	fERG	Allele 1 c. Nomenclature	Allele 1 p. Nomenclature	Allele 2 c. Nomenclature	Allele 2 p. Nomenclature
1	F	11	10	1,30	3	c.3050+5G>T	Splice defect	c.3050+5G>T	Splice defect
2	F	10	16	1,30	ND	c.4667+1G>A	Splice defect	c.5882G>A	Gly1961Glu
3	F	11	8	1,30	1	c.5461-10T>C	Splice defect	c.5714+5G>A	Splice defect
4	F	7	34	2,00	3	c.4124C>A	Ala1375Glu	c.4124C>A	Ala1375Glu
5	M	32	9	1,00	3	c.2588G>C	Gly863Ala	c.5018+2T>C	Splice defect
6	F	6	15	2,00	3	c.5461-10T>C	Splice defect	c.5177C>A	Thr1726Asn
7	F	14	2	0,52	2	c.122G>A	Trp41Ter	c.5381C>A	Ala1794Asp
8	M	17	15	0,52	ND	c.2588G>C	Gly863Ala	c.4462T>C	Cys1488Arg
9	F	25	23	0,82	ND	c.5461-10T>C	Splice defect	c.5882G>A	Gly1961Glu
10	M	8	7	1,00	3	c.768G>T	Splice defect	c.5461-10T>C	Splice defect
11	M	4	11	1,00	3	c.768G>T	Splice defect	c.5461-10T>C	Splice defect
12	F	10	5	1,00	ND	c.3813G>A	Splice defect	c.6088C>T	Arg2030Ter
13	F	22	0	0,70	2	c.2588G>C	Gly863Ala	c.3259G>A	Gly1087Lys
14	M	48	0	0,00	1	c.1015T>G	Trp339Gly	c.5693G>A	Arg1898His
15	M	38	7	0,22	2	c.2588G>C	Gly863Ala	c.5461-10T>C	Splice defect
16	M	10	3	0,82	2	c.4139C>T	Pro1380Leu	c.6471del	Lys2158Serfs*12
17	F	16	1	0,70	1	c.2588G>C	Gly863Ala	c.6543_6578del	Leu2182_Phe193del
18	M	15	23	0,82	1	c.5882G>A	Gly1961Glu	c.5753A>G	Asp1918Gly
19	F	27	23	1,30	1	c.2588G>C	Gly863Ala	c.4918C>T	Arg1640Trp
20	M	8	1	0,77	2	c.2299_2305delinsTTAACC	Val767Leufs*20	c.5461-10T>C	Splice defect
21	F	29	16	0,82	1	c.2588G>C	Gly863Ala	c.5461-10T>C	Splice defect
22	M	7	0	1,00	3	c.160T>G	Cys54Gly	c.160T>G	Cys54Gly
23	M	22	0	1,00	ND	c.2588G>C	Gly863Ala	c.1822T>A	Phe608Ile
24	F	9	3	0,70	ND	c.1804C>T	Arg602Trp	c.1019A>G	Tyr340Cys
25	F	24	2	0,00	1	c.223T>G	Cys75Gly	c.5882G>A	Gly1961Glu
26	M	5	13	1,30	ND	c.5461-10T>C	Splice defect	c.717G>A	Trp239Ter
27	M	24	1	0,00	ND	c.5882G>A	Gly1961Glu	c.768G>T	Splice defect
28	F	20	9	0,82	ND	c.5882G>A	Gly1961Glu	c.768G>T	Splice defect
29	F	20	4	0,82	1	c.2588G>C	Gly863Ala	c.2299_2303delinsTTAA	Val767Leufs*20
30	F	35	7	0,60	2	c.5882G>A	Gly1961Glu	c.4253+5G>A	Splice defect
31	F	34	1	-0,08	2	c.1015T>G	Trp339Gly	c.2791G>C	Val931Met (corr)
32	F	12	22	0,82	2	c.5460+1G>A	Splice defect	c.5882G>A	Gly1961Glu
33	M	7	3	1,00	1	c.1798G>T	Asp600Tyr	c.1798G>T	Asp600Tyr
34	F	43	6	0,15	ND	c.3386G>T	Arg1129Leu	c.5882G>A	Gly1961Glu
35	F	36	11	1,00	ND	c.3386G>T	Arg1129Leu	c.5882G>A	Gly1961Glu
36	F	6	29	1,82	3	c.3322C>T; 6320G>A	Arg1108Cys; Arg2107His	c.6386+2C>G	Splice defect
37	F	12	13	0,22	ND	c.2588G>C	Gly863Ala	c.5461-10T>C	Splice defect
38	M	8	16	1,30	3	c.4918C>T	Arg1640Trp	c.6704C>G	Ser2235Ter
39	M	14	0	0,40	1	c.286A>G	Asn96Asp	c.1015T>G	Trp339Gly
40	F	21	3	0,30	3	c.286A>G	Asn96Asp	c.1015T>G	Trp339Gly
41	F	30	11	1,00	2	c.1622T>C; 3113C>T	Leu541Pro; Ala1038Val	c.5882G>A	Gly1961Glu
42	F	9	24	1,60	3	c.768G>T	Splice defect	c.5113C>T	Arg1705Trp
43	M	17	0	0,00	ND	c.1957C>T	Arg653Cys	c.2588G>C	Gly863Ala
44	M	4	9	0,70	1	c.1957C>T	Arg653Cys	c.2588G>C	Gly863Ala
45	M	11	1	0,40	1	c.286A>G	Asn96Asp	c.4253+5G>A	Splice defect
46	F	ND	ND	0,30	1	c.5714+5G>A	Splice defect	c.5413A>G	Asn1805Asp

47	F	13	9	0,15	1	c.5714+5G>A	Splice defect	c.5413A>G	Asn1805Asp
48	M	4	1	0,89	1	c.634C>T	Arg212Cys	c.4918C>T	Arg1640Trp
49	F	14	4	1,00	1	c.3386G>T	Arg1129Leu	c.5381C>A	Ala1794Asp
50	M	16	3	0,52	1	c.3386G>T	Arg1129Leu	c.5381C>A	Ala1794Asp
51	M	10	11	1,00	ND	c.5312+1G>A	Splice defect	c.5645T>C	Met1882Thr
52	F	43	10	0,10	1	c.2971G>C	Gly991Arg	c.5461-10T>C	Splice defect
53	M	4	9	1,00	2	c.3210_3211dup	Ser1071Cysfs*14	c.4918C>T	Arg1640Trp
54	M	17	1	0,82	1	c.6454G>T	Gly2152Cys	c.5692C>T	Arg1898Cys
55	F	10	1	0,52	3	c.6454G>T	Gly2152Cys	c.5692C>T	Arg1898Cys
56	M	8	15	0,82	ND	c.2588G>C	Gly863Ala	c.5461-10T>C	Splice defect
57	M	59	7	-0,08	3	c.1015T>G	Trp339Gly	c.1928T>G	Val643Gly
58	M	10	7	0,70	1	c.1015T>G	Trp339Gly	c.2588G>C	Gly863Ala
59	F	43	8	-0,16	ND	c.2588G>C	Gly863Ala	c.5714+5G>A	Splice defect
60	F	35	11	0,15	1	c.2588G>C	Gly863Ala	c.5714+5G>A	Splice defect
61	F	14	1	1,00	1	c.768G>T	Splice defect	c.2588G>C	Gly863Ala
62	F	15	4	1,00	ND	<i>Data lost</i>	<i>Data lost</i>		
63	F	18	4	0,82	ND	c.5461-10T>C	Splice defect	2588G>C	Gly863Ala
64	F	45	12	0,30	1	c.3113C>T	Ala1038Val	6543_6578del	Leu2182_Phe193del
65	M	12	13	1,00	1	c.634C>T	Arg212Cys	6543_6578del	Leu2182_Phe193del
66	F	27	20	1,30	1	c.634C>T	Arg212Cys	6079C>T	Leu2027Phe
67	M	45	0	-0,08	1	c.1015T>G	Trp339Gly	3608-7G>A	Splice defect
68	F	48	3	0,02	1	c.1015T>G	Trp339Gly	3608-7G>A	Splice defect
69	F	6	15	1,30	2	c.5714+5G>A	Splice defect	5461-10T>C	Splice defect
70	M	8	15	1,30	3	c.4139C>T	Pro1380Leu	5929G>A	Gly1977Ser
71	M	6	45	2,30	ND	c.768G>T	Splice defect	4539+1G>T	Splice defect
72	M	13	13	0,82	ND	c.2041C>T	Arg681Ter	5817G>T	Arg1939Ser
73	F	35	9	0,00	ND	c.2588G>C	Gly863Ala	3386G>T	Arg1129Leu

Nr.= patient number; Pat.=patient; M= male, F=female; y= year ; BCVA= best-corrected visual acuity; ffERG= Full Field Flash electroretinogram; ND= not determined