

Table S1. Clinical characteristics

| P | Sex | ABCA4 variant 1 | ABCA4 variant 2 | Age at onset [years] | Follow-up period [years] | Duration between onset and the first exam [years] | Age at the first and last exams [years] | VA at the first and last exams [Snellen] | VA loss per year [Snellen] | DDAF area at the first and last exams [mm ²] | Increase of DDAF area per year [mm ²] | ERG group at the first and last exams | Fishman stage at the first and last exams |
|-----|-----|--|--|----------------------|--------------------------|---|---|--|----------------------------|--|---|---------------------------------------|---|
| 1* | F | c.4234C>T; p.(Gln1412*) | c.5882G>A; p.(Gly1961Glu) | 46 | 26 | 0 | 46 72 | 0.8 0.16 | 0.025 | 0.47 8.18 | 0.296 | normal ERG 1 | II III |
| 2 | F | c.2041C>T; p.(Arg681*) | c.5413A>G; p.(Asn1805Asp) | 15 | 25 | 14 | 29 54 | 0.1 0.05 | 0.002 | 13.70 78.61 | 2.596 | 3 3 | III IV |
| 3* | M | c.5882G>A; p.(Gly1961Glu) | c.[5461-10T>C;5603A>T]; p.[Thr1821Aspfs*6,Thr1821Valfs*13;(Asn1868Ile)] | 7 | 13 | 15 | 22 35 | 0.2 0.15 | 0.004 | 0.02 1.85 | 0.141 | 1 3 | I I |
| 4* | F | c.1648G>A; p.(Gly550Arg) | c.5882G>A; p.(Gly1961Glu) | 14 | 21 | 30 | 44 65 | 0.2 0.02 | 0.009 | 0.20 0.43 | 0.011 | normal ERG normal ERG | I I |
| 5 | F | c.2041C>T; p.(Arg681*) | c.6089G>A; p.(Arg2030Gln) | 15 | 23 | 4 | 19 42 | 0.15 0.03 | 0.005 | 1.43 17.69 | 0.707 | 1 2 | I III |
| 6 | F | c.5175dup(;)5603A>T; p.(Thr1726Aspfs*61)(;)(Asn1868Ile) | c.5714+5G>A; p.[=,Glu1863Leufs*33] | 18 | 11 | 9 | 27 38 | 0.4 0.03 | 0.034 | 11.60 21.82 | 0.929 | 2 2 | III III |
| 7 | F | c.5175dup(;)5603A>T; p.(Thr1726Aspfs*61)(;)(Asn1868Ile) | c.5714+5G>A; p.[=,Glu1863Leufs*33] | 25 | 11 | 6 | 31 42 | 0.8 0.02 | 0.071 | 11.18 59.14 | 4.361 | 2 3 | III III |
| 8 | M | c.688T>A; p.(Cys230Ser) | c.4539+2T>C; p.(?) | 11 | 19 | 2 | 13 32 | 0.3 0.03 | 0.015 | 0.24 9.81 | 0.504 | 2 2 | I III |
| 9* | M | c.2041C>T; p.(Arg681*) | c.5603A>T; p.(Asn1868Ile) | 30 | 17 | 0 | 30 47 | 0.8 0.2 | 0.035 | 0.12 0.96 | 0.050 | normal ERG 1 | I II |
| 10* | F | c.1918C>G(;)5603A>T; p.(Pro640Ala)(;)(Asn1868Ile) | c.5882G>A; p.(Gly1961Glu) | 38 | 20 | 1 | 39 59 | 0.8 0.2 | 0.030 | 0.30 0.71 | 0.021 | 1 1 | I I |
| 11* | F | c.5175dup(;)5603A>T; p.(Thr1726Aspfs*61)(;)(Asn1868Ile) | c.5882G>A; p.(Gly1961Glu) | 19 | 20 | 6 | 25 45 | 0.2 0.3 | -0.005 | 0.00 0.03 | 0.002 | normal ERG normal ERG | I I |

| | | | | | | | | | | | | | |
|-----|---|--|---|----|----|---|----|------|-------|-------|--------|------------|-----|
| 12* | F | c.4919G>A; p.(Arg1640Gln) | c.5603A>T; p.(Asn1868Ile) | 20 | 26 | 1 | 21 | 0.6 | 0.022 | 2.43 | 0.269 | 1 | I |
| | | | | | | | 47 | 0.03 | | 9.43 | | 2 | III |
| 13* | F | c.2041C>T; p.(Arg681*) | c.5603A>T; p.(Asn1868Ile) | 20 | 16 | 0 | 20 | 0.2 | 0.003 | 0.90 | 0.083 | 1 | I |
| | | | | | | | 36 | 0.16 | | 2.23 | | 1 | I |
| 14 | F | c.6445C>T; p.(Arg2149*) | c.6445C>T; p.(Arg2149*) | 7 | 10 | 0 | 7 | 0.1 | 0.008 | 0.03 | 1.152 | 3 | II |
| | | | | | | | 17 | 0.02 | | 11.54 | | 3 | II |
| 15 | F | c.6445C>T; p.(Arg2149*) | c.6445C>T; p.(Arg2149*) | 9 | 12 | 1 | 10 | 0.3 | 0.024 | 0.91 | 1.574 | 2 | II |
| | | | | | | | 22 | 0.02 | | 19.80 | | 3 | III |
| 16 | M | c.5977del; p.(Ser1993fs) | c.5461-10T>C; p.[Thr1821Aspfs*6,Thr1821Valfs*13] | 8 | 11 | 1 | 9 | 0.1 | 0.005 | 3.81 | 1.129 | 3 | II |
| | | | | | | | 20 | 0.04 | | 16.23 | | 3 | III |
| 17 | M | c.[5461-10T>C;5603A>T]; p.[Thr1821Aspfs*6,Thr1821Valfs*13;(Asn1868Ile)] | c.5714+5G>A; p.[=,Glu1863Leufs*33] | 11 | 18 | 5 | 16 | 0.2 | 0.007 | 0.16 | 0.1628 | 1 | II |
| | | | | | | | 34 | 0.03 | | 3.09 | | 3 | III |
| 18 | F | c.1570A>G; p.(Lys524Glu) | c.5714+5G>A; p.[=,Glu1863Leufs*33] | 17 | 11 | 1 | 18 | 0.3 | 0.009 | 0.15 | 0.4109 | normal ERG | II |
| | | | | | | | 29 | 0.2 | | 4.67 | | normal ERG | III |

Patients 6 and 7, as well as 14 and 15 are siblings. Eight patients with p.(Gly1961Glu) or p.(Asn1868Ile) allele are marked with a star. Abbreviation explanation: F—female, M—male.