Supplementary Table 1: Pathogenicvariants of the Stargardt patients

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| --- | --- | --- | --- | --- | --- | --- |
| **Pat** | **Sex** | **Gene** | **Allele 1** | | **Allele 2** | |
| Nt change | Protein change | Nt change | Protein change |
| STGD 1 | M | *ABCA4* | c.5714+5G>A | p.? | c.5917delG | p.Val1973\* |
| STGD 2 | M | *ABCA4* | n.a. | n.a. | n.a. | n.a. |
| STGD 3 | M | *ABCA4* | c.428del | p.Pro143Argfs\*11 | c.5882G>A | p.Gly1961Glu |
| STGD 4 | F | *ABCA4* | c.2588G>C | p.Gly863Ala | c.5653G>A | p.Glu1885Lys |
| STGD 5 | M | *ABCA4* | c.4958G>A | p.Gly1653Glu | c.5882G>A | p.Gly1961Glu |
| STGD 6 | M | *ABCA4* | c.2588G>C | p.Gly863Ala | c.5653G>A | p.Glu1885Lys |
| STGD 7 | F | *ABCA4* | c.5329A>T | p.Met1777Leu | c.5461-10T>C | p.? |
| STGD 8 | F | *ABCA4* | c.4958G>A | p.Gly1653Glu | c.5882G>A | p.Gly1961Glu |
| STGD 9 | F | *ABCA4* | c.2401G>A | p.Ala801Thr | c.5381C>A | p.Ala1794Asp |
| STGD 10 | M | *ABCA4* | c.2588G>C | p.Gly863Ala | c.5653G>A | p.Glu1885Lys |
| STGD 11 | F | *ABCA4* | c.3179A>C | p.Gln1060Pro | c.3179A>C | p.Gln1060Pro |
| STGD 12 | F | *ELOVL4* | c.810C>G | p.Tyr270\* | + | + |

Nucleotide (Nt) numbering is according to the GenBank entry NM\_000350.2 for *ABCA4* and NM\_022726.3 for *ELOVL4*. Amino acid numbering is in accordance to the GenBank entry NP\_000341.2 for ABCA4 and NP\_073563.1 for ELOVL4. Patient S2 was not available (n.a.) for genetic testing. Patients S4,S7 and S14, respectively patients S6 and S10 are siblings. Patient S11 also harbors a pathogenic c.659T>G variant (p.Phe220Cys) in *RHO*.