**Appendix 3**

Clinical description of CD patients with rare variants in the *CFH* gene

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| **Number** | **Gender** | **Age** | **Age at onset** | **Variants** | **Family**  | **VA**  | **Retinal phenotype** |
|  |  |  |  | **DNA level** | **Protein level** | **history** | **OD** | **OS** |  |
| 1 | F | 48 | 45 | c.7C>G | p.Leu3Val | NO | 0,3+ | 0,1+ | ODS: Confluent large drusen in macula and numerous small drusen in posterior pole. OS: Small geographic atrophic lesion |
| 2 | F | 57 | 51 | c.428-2A>G | NA | NO | 0,5/60 | 1,0 | OD: Central fibrotic scar. OS: Confluent large drusen in macula. ODS: Small drusen en punched out lesions in peripheral retina |
| 3 | F |  48 |  47 | c.481G>T | p.Ala161Ser | YES | 1.0 | 0.8+  | ODS: Confluent large drusen in macula and some small drusen in posterior pole |
| 4 | M | 74 | 69 | c.518C>G | p.Ala173Gly | NO | 0.8 | 0,32- | ODS: Extensive small and large drusen in macula and mid-peripheral retina. OS: Large occult subfoveal CNV |
| 5 | M | 77 | 75 | c.524G>A  | p.Arg175Gln | YES | 0,6++ | 0.25 | ODS: Extensive hard macular drusen and temporal to the macula small areas of chorioretinal atrophy  |
| 6 | M | 73 | No visual loss | c.647T>C | p.Ile216Thr | YES | 1,25-- | 0,8-- | ODS: Some small drusen in posterior pole and numerous small drusen in peripheral retina |
| 7 | F | 56 | 44 | c.901\_902del | p.Ala301AsnfsX25 | YES |  NA |  NA | ODS: Innumerable small drusen in posterior pole. OD: Hard exudates, fluid pocket and CNV. OS: Juxta foveal confluent soft drusen |
| 8 | F | 86 | 81 | c.1198C>A | p.Gln400Lys | NA | 0,4+ | 2/30 | OD: Pigmentary changes, numerous reticular drusen in posterior pole and occult CNV. OS: Central fibrotic scar |
| 9 | M | 44 | NA | c.1198C>A | p.Gln400Lys |  NA |  NA |  NA | ODS: Extensive soft confluent and small hard drusen in posterior pole |
| 10 | F | 46 | 20 | c.2850G>C | p.Gln950His | NA | 1,25-- | 1,0+ | ODS: Numerous hard drusen, especially from the border of the macula to the vascular arcade; fundoscopic changes at the age of 20 years  |
| 11 | M | 80 | 73 | c.2850G>C | p.Gln950His | NO | 2/60 | 0,1- | ODS: Para macular drusen. OD: Large fibrotic scar in macula. OS: Macular pigmentary changes, occult CNV |
| 12 | F | 77 | 71 | c.2867C>T | p.Thr956Met | NO | 0,8++ | 0,6 | ODS: Extensive small drusen in posterior pole and peripheral retina |
| 13 | F | 55 | 54 | c.1135C>T  | p.Trp379Arg | YES | 0,9 | 0,5+ | ODS: Extensive small drusen in posterior pole and mid-peripheral retina OD: Soft large drusen in macula. OS: Classic exudative CNV in macula |
| 14 | F | 54 | 50 | c.3628C>T | p.Arg1210Cys | YES | 0.6 | 0.4 | ODS: Numerous small drusen in posterior pole and some drusen in peripheral retina |
| 15 | F | 76 | 64 | c.3628C>T | p.Arg1210Cys | NA | 1/60 | 0,5 | ODS: Innumerable small drusen in posterior pole. OD: Large fibrotic scar and hard exudates; OS: Pigmentary changes and occult CNV |