|  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Number of subjects** | **Variant** | **Genomic coordinates (GRCh37)** | **Reference allele** | **Alternative allele** | **Variant class** | **Consequence** | **HGVSc** | **HGVSp** | **AF (gnomAD),**  **%** | **ACMG/**  **AMP2015** | **Clinical Significance** |
| 5 | rs121918393  *APOE2* Heidelberg | chr19:45412013 | C | T | SNV | missense\_variant | NM\_000041.4:c.460C>T | NP\_000032.1: p.Arg154Cys | 0.008979 | PM1 [1], PM2, PP1\_Moderate [1], PP3 (REVEL = 0.898), PP4, PP5 | likely pathogenic |
| 4 | rs267606664 | chr19:45411987 | G | A | SNV | missense\_variant | NM\_000041.4:c.434G>A | NP\_000032.1: p.Gly145Asp | 0.01532 | BS1, PP4, PP1  REVEL = 0.581 | variant of uncertain significance |
| 6 | rs199768005 | chr19:45412314 | T | A | SNV | missense\_variant | NM\_000041.4:c.761  T>A | NP\_000032.1: p.Val254Glu | 0.04515 | BS1, PP41,  REVEL score = 0.260 | variant of uncertain significance |
| 1 | rs267606661 | chr19:45412358 | C | G | SNV | missense\_variant | NM\_000041.4:c.805C>G | NP\_000032.1: p.Arg269Gly | 0.03605 | BS1, PP4 [2], REVEL = 0.581 | variant of uncertain significance |
| 1 | rs1969839083 | chr19:45411157-45411160 | CTGT | - | deletion | frameshift\_variant | NM\_000041.4:c.184\_187del | NP\_000032.1: p.Glu63ArgfsTer15 | - | PVS1, PM2, PP42 | pathogenic |
| 1 | - | chr19:45411985-45412012 | CGGCCAGAGCACCGAGGAGCTGCGGGTG | - | deletion | frameshift\_variant | NM\_000041.4: c.432\_459del | NP\_000032.1: p.Gly145AlafsTer97 | - | PVS1, PM2 | likely pathogenic |

**Supplementary Table S2**. The clinical interpretation of the detected *APOE* variants, associated with the autosomal dominant FD.

1—3 subjects in present study had a clinical data (1 – male, 48 years old, TG level 3.27 mmol/L, glucose 6.72 mmol/L, BMI 28.82 kg/m2, carotid atherosclerosis (number of plaques 3 and maximum stenosis 54.0%), femoral atherosclerosis (number of plaques 3 and maximum stenosis 27.0%), coronary heart disease – no, diabetes mellitus – not known;

2 – woman, 51 years old, TG level 1.70 mmol/L, BMI 39.85 kg/m2, carotid and femoral atherosclerosis, coronary heart disease – no; 3 – woman, 53 years old, TG level 1.43 mmol/L, BMI 28.88 kg/m2, carotid and femoral atherosclerosis, coronary heart disease – no. 2—data of the present study (woman, 57 years old, TG level 4.75 mmol/L, Achilles tendon xanthomas). AF— allele frequency; ACMG/AMP2015— the American College of Medical Genetics and Genomics/Association for Molecular Pathology; gnomAD— Genome Aggregation Database; HGVSc —Human Genome Variation Society coding sequence name; HGVSp — Human Genome Variation Society protein sequence name; SNV – single nucleotide variant.

References

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