**Table 2. Summary of Genes with an Over-Representation of Ultra-Rare Variants with a MAF < 0.00005**

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **Gene ID** | **Gene/Protein Name** | **pValue** | **Odds Ratio (OR)** | **Number of Variant Positive Cases (n=278)** | **Number of Variant Positive Controls (n=973)** |
| *LRP1* | LDL Receptor Related Protein 1 | 0.000269 | 3.8948 | 16 (5.8%) | 15 (1.5%) |
| *LHX9* | LIM Homeobox 9 | 0.000527 | INF | 5 (1.8%) | 0 |
| *PDS5A* | PDS5 Cohesin Associated Factor A | 0.000657 | 21.389 | 6 (2.2%) | 1 (0.1%) |
| *ULK1* | Unc-51 Like Autophagy Activating Kinase 1 | 0.002137 | 10.68 | 6 (2.2%) | 2 (0.21%) |
| *NR3C2* | Nuclear Receptor Subfamily 3 Group C Member 2 | 0.002137 | 10.68 | 6 (2.2%) | 2 (0.21%) |
| *OR4C6* | Olfactory Receptor Family 4 Subfamily C Member 6 | 0.002398 | INF | 4 (1.4%) | 0 |
| *CHRM3* | Cholinergic Receptor Muscarinic 3 | 0.002398 | INF | 4 (1.4%) | 0 |
| *CYB5D1* | Cytochrome B5 Domain Containing 1 | 0.002398 | INF | 4 (1.4%) | 0 |
| *GBP2* | Guanylate Binding Protein 2 | 0.002398 | INF | 4 (1.4%) | 0 |
| *ZNF506* | Zinc Finger Protein 506 | 0.002398 | INF | 4 (1.4%) | 0 |
| *IFRD2* | Interferon-Related Developmental Regulator 2 | 0.002398 | INF | 4 (1.4%) | 0 |
| *BICD2* | BICD Cargo Adaptor 2 | 0.002398 | INF | 4 (1.4%) | 0 |
| *KDM4DL* | Lysine Demethylase 4E | 0.002584 | 17.751 | 5 (1.8%) | 1 (0.1%) |
| *TNFRSF10D* | Tumor Necrosis Factor Receptor Superfamily Member 10d | 0.002584 | 17.751 | 5 (1.8%) | 1 (0.1%) |
| *CTIF* | CBP80/20-Dependent Translation Initiation Factor | 0.003588 | 6.2452 | 7 (2.5%) | 4 (0.4%) |
| *COL20A1* | Collagen Type XX Alpha 1 | 0.003588 | 6.2452 | 7 (2.5%) | 4 (0.4%) |
| *ARHGEF16* | Rho Guanine Nucleotide Exchange Factor 16 | 0.004595 | 4.7675 | 8 (2.9%) | 6 (0.62%) |

Genes are listed in ascending order by their p-value. INF = infinite.