**S10 Table:** Enrichment analysis-based candidate genes in discovery cohort versus ‘Control cohort 2’.

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| Gene | Number of variants | Uncorrectedc | Three gene correctiond | Exome-wide correctione | Replication cohort (*n*=174) |
| **Discovery cohort (*n*=55)** | **Control cohort 2 (*n*=2,329)** |
| *EMR3* | 2 | 12a | OR: 7.16CI: 0.769-32.733*P* = 0.04006 | *P* = 0.040060  | *P* = 1 | 0 |
| *PTPN12* | 3 | 11b | OR: 11.82CI: 2.088-45.615*P* = 0.003613 | *P* = 0.026100 | *P* = 1 | 1f |
| *LRP6* | 3 | 18b | OR: 7.218CI: 1.342-25.278*P* = 0.0117 | *P* = 0.010839 | *P* = 1 | 0 |

aTotal number of loss-of-function variants (e.g. nonsense, frameshift or splice site variants) with a MAF of ≤0.001 in NHLBI-EVS database in ‘Control cohort 2’. bTotal number of highly conserved missense variants (PhyloP ≥3.0) with a MAF of ≤0.001 in NHLBI-EVS database in ‘Control cohort 2’. cFisher’s exact test between Discovery cohort and ‘Control cohort 2’ not corrected for multiple testing. *dP*-values Fisher’s exact test between Discovery cohort and ‘Control cohort 2’ corrected for three genes. *eP*-values Fisher’s exact test between Discovery cohort and ‘Control cohort 2’ corrected for (exome-wide) multiple testing. fFisher’s exact test between Replication cohort and ‘Control cohort 2’ was not significant. NHLBI-EVS: Exome variant server.