Appendix 6 Gene-based association results for predicted functional partners of *PTCHD2, CRISP3, NAP1L4, FSCB,* and *AP3B2*

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Genea,b** | **Chromosome** | **Confidence Score** | **SKAT P value** | **Burden P value** |
| ***PTCHD2* (0/2)** | | | | |
| -- | -- | -- | -- | -- |
| ***CRISP3* (2/3)** | | | | |
| *KNG1* | 3 | 0.800 | 0.7644 | 0.9097 |
| *MTDH* | 8 | 0.751 | 0.8262 | 0.5669 |
| ***NAP1L4* (10/27)** | | | | |
| *PES1* | 22 | 0.978 | 0.4759 | 0.1178 |
| *EIF3A* | 10 | 0.917 | 0.3239 | 0.4515 |
| *UBC* | 12 | 0.907 | 0.1454 | 0.0519 |
| *CSNK2A2* | 16 | 0.898 | 0.9680 | 0.8017 |
| *SDAD1* | 4 | 0.844 | 0.3682 | 0.9285 |
| *TSPYL4* | 6 | 0.729 | 0.3375 | 0.0512 |
| *CYFIP1* | 15 | 0.728 | 0.5990 | 0.9344 |
| *CYFIP2* | 5 | 0.724 | 0.3222 | 0.3775 |
| *WASF3* | 13 | 0.723 | 0.8066 | 0.7887 |
| *STAG3* | 7 | 0.715 | 0.3579 | 0.2207 |
| ***FSCB* (1/1)** | | | | |
| *CABYR* | 18 | 0.974 | 0.4877 | 0.1166 |
| ***AP3B2* (11/20)** | | | | |
| *AP3D1* | 19 | 0.934 | 0.0904 | 0.1387 |
| *AP3M1* | 10 | 0.917 | 0.5912 | 0.7360 |
| *AP4M1* | 7 | 0.908 | 0.2385 | 0.2845 |
| *AP4E1* | 15 | 0.882 | 0.1882 | 0.3055 |
| *AP4S1* | 14 | 0.842 | 0.8600 | 0.6469 |
| *AP2A2* | 11 | 0.787 | 0.8672 | 0.8275 |
| *AP1G1* | 16 | 0.76 | 0.8564 | 0.7385 |
| *AP1G2* | 14 | 0.76 | 0.3710 | 0.9008 |
| *RAB38* | 11 | 0.729 | 0.5684 | 0.8313 |
| *AP1M2* | 19 | 0.722 | 0.9993 | 0.8650 |
| *AP5M1* | 14 | 0.722 | 0.3148 | 0.4068 |
| a Genes in the STRING database with strong evidence (confidence score > 0.70) of protein-protein interactions with reported genes. The number in the parentheses indicates the proportion of genes tested in our gene-based analysis.  b The number in parentheses represents the number of genes out of the total number of genes identified as functional partners that were tested in our gene-based analysis. | | | | |