**Supplemental Table S4.** Significantly overrepresented GO terms related to the genes in the top 200 dataset SNPs in the TGen dataset.

|  |  |  |  |
| --- | --- | --- | --- |
|  | **GO term identifier** | **GO term description** | **Gene Symbol** |
| BP | GO:0030516 | regulation of axon extension | APOE,NRCAM |
| *GO:0010976* | *positive regulation of neuron projection development* | *DCC,CAMK1D* |
| *GO:0045162* | *clustering of voltage-gated sodium channels* | *NRCAM* |
| *GO:0090050* | *positive regulation of cell migration involved in sprouting angiogenesis* | *HDAC9* |
| *GO:0007409* | *axonogenesis* | *DCC,NRCAM* |
| *GO:0003007* | *heart morphogenesis* | *DLC1,COL11A1* |
| *GO:0035767* | *endothelial cell chemotaxis* | *PRKD1* |
| *GO:0045823* | *positive regulation of heart contraction* | *GLP1R* |
| *GO:0072358* | *cardiovascular system development* | *APOE* |
| *GO:0045666* | *positive regulation of neuron differentiation* | *NRCAM,VWC2* |
| *GO:0048842* | *positive regulation of axon extension involved in axon guidance* | *DSCAM* |
| *GO:0045617* | *negative regulation of keratinocyte differentiation* | *TP63* |
| *GO:0002021* | *response to dietary excess* | *APOE* |
| *GO:0060513* | *prostatic bud formation* | *TP63* |
| *GO:0045597* | *positive regulation of cell differentiation* | *GLP1R,SMAD9* |
| *GO:0060529* | *squamous basal epithelial stem cell differentiation involved in prostate gland acinus development* | *TP63* |
| *GO:0050773* | *regulation of dendrite development* | *CAMK1D* |
| *GO:0002064* | *epithelial cell development* | *TP63* |
| *GO:0002063* | *chondrocyte development* | *COL11A1* |
| *GO:0001736* | *establishment of planar polarity* | *TP63* |
| *GO:0021575* | *hindbrain morphogenesis* | *DLC1* |
| *GO:2000171* | *negative regulation of dendrite development* | *DCC* |
| *GO:0048671* | *negative regulation of collateral sprouting* | *DCC* |
| *GO:0014706* | *striated muscle tissue development* | *EYA2* |
| *GO:0070593* | *dendrite self-avoidance* | *DSCAM* |
| *GO:0043589* | *skin morphogenesis* | *TP63* |
| *GO:0051153* | *regulation of striated muscle cell differentiation* | *HDAC9* |
| *GO:0014012* | *peripheral nervous system axon regeneration* | *APOE* |
| *GO:0048742* | *regulation of skeletal muscle fiber development* | *HDAC9* |
| *GO:0030859* | *polarized epithelial cell differentiation* | *TP63* |
| GO:0033700 | phospholipid efflux | APOC1,APOE |
| *GO:0033344* | *cholesterol efflux* | *APOC1,APOE* |
| *GO:0032368* | *regulation of lipid transport* | *APOC1* |
| *GO:0032374* | *regulation of cholesterol transport* | *APOC1* |
| *GO:0032375* | *negative regulation of cholesterol transport* | *APOC1* |
| GO:0051410 | detoxification of nitrogen compound | MOSC1 |
| GO:0030029 | actin filament-based process | ELMO1 |
| GO:0042157 | lipoprotein metabolic process | APOC1,APOE |
| GO:0042126 | nitrate metabolic process | MOSC1 |
| GO:0016266 | O-glycan processing | GXYLT2 |
| *GO:0006029* | *proteoglycan metabolic process* | *COL11A1* |
| GO:0042159 | lipoprotein catabolic process | APOE |
| *GO:0042158* | *lipoprotein biosynthetic process* | *APOE* |
| GO:0046717 | acid secretion | SLC22A16 |
| GO:0010873 | positive regulation of cholesterol esterification | APOC1,APOE |
| *GO:0042981* | *regulation of apoptotic process* | *DCC,CAMK1D,ALK* |
| *GO:0045833* | *negative regulation of lipid metabolic process* | *APOC1* |
| *GO:0008203* | *cholesterol metabolic process* | *APOC1,APOE* |
| *GO:2000107* | *negative regulation of leukocyte apoptotic process* | *CXCL12* |
| *GO:0045541* | *negative regulation of cholesterol biosynthetic process* | *APOE* |
| *GO:0043523* | *regulation of neuron apoptotic process* | *TP63* |
| *GO:0006917* | *induction of apoptosis* | *DCC,DLC1,APOE,TP63* |
| *GO:0045717* | *negative regulation of fatty acid biosynthetic process* | *APOC1* |
| GO:0032793 | positive regulation of CREB transcription factor activity | PRKD1,CAMK1D |
| *GO:0060158* | *activation of phospholipase C activity by dopamine receptor signaling pathway* | *GNA14* |
| *GO:0051005* | *negative regulation of lipoprotein lipase activity* | *APOC1* |
| *GO:0007202* | *activation of phospholipase C activity* | *DLC1,PDE1C* |
| GO:0055085 | transmembrane transport | SLC22A16,KCNH7,HK1,RYR3,ATP6V1C2,SLCO3A1 |
| GO:0046069 | cGMP catabolic process | PDE1C |
| *GO:0030828* | *positive regulation of cGMP biosynthetic process* | *APOE* |
| GO:0007186 | G-protein coupled receptor signaling pathway | GABBR2,ENPP2,APOE,CXCL12,GNA14 |
| *GO:0007165* | *signal transduction* | *DLC1,PRKD1,PDE1C,ARHGAP23,ALK,CXCL12,PRKG1,GNA14* |
| GO:0006396 | RNA processing | CELF2,RCL1 |
| GO:0030203 | glycosaminoglycan metabolic process | CHST9 |
| GO:0034638 | phosphatidylcholine catabolic process | ENPP2 |
| *GO:0006641* | *triglyceride metabolic process* | *APOC1,APOE* |
| *GO:0010900* | *negative regulation of phosphatidylcholine catabolic process* | *APOC1* |
| GO:0071205 | protein localization to juxtaparanode region of axon | NFASC |
| GO:0032581 | ER-dependent peroxisome organization | PEX16 |
| *GO:0016557* | *peroxisome membrane biogenesis* | *PEX16* |
| *GO:0016558* | *protein import into peroxisome matrix* | *PEX16* |
| *GO:0006625* | *protein targeting to peroxisome* | *PEX16* |
| *GO:0045046* | *protein import into peroxisome membrane* | *PEX16* |
| GO:0015879 | carnitine transport | SLC22A16 |
| GO:0044245 | polysaccharide digestion | SI |
| GO:0019934 | cGMP-mediated signaling | APOE |
| GO:0032488 | Cdc42 protein signal transduction | APOE |
| *GO:0035024* | *negative regulation of Rho protein signal transduction* | *DLC1* |
| *GO:0016601* | *Rac protein signal transduction* | *ELMO1* |
| GO:0007499 | ectoderm and mesoderm interaction | TP63 |
| *GO:0007501* | *mesodermal cell fate specification* | *EYA2* |
| GO:0006376 | mRNA splice site selection | CELF2 |
| GO:0034447 | very-low-density lipoprotein particle clearance | APOC1,APOE |
| *GO:0034375* | *high-density lipoprotein particle remodeling* | *APOC1,APOE* |
| *GO:0034380* | *high-density lipoprotein particle assembly* | *APOE* |
| *GO:0034379* | *very-low-density lipoprotein particle assembly* | *APOC1* |
| *GO:0034384* | *high-density lipoprotein particle clearance* | *APOE* |
| *GO:0034382* | *chylomicron remnant clearance* | *APOC1,APOE* |
| *GO:0034372* | *very-low-density lipoprotein particle remodeling* | *APOE* |
| *GO:0034369* | *plasma lipoprotein particle remodeling* | *APOC1* |
| *GO:0010916* | *negative regulation of very-low-density lipoprotein particle clearance* | *APOC1* |
| GO:0002175 | protein localization to paranode region of axon | NFASC |
| GO:0000173 | inactivation of MAPK activity involved in osmosensory signaling pathway | MBIP |
| GO:0050910 | detection of mechanical stimulus involved in sensory perception of sound | COL11A1 |
| GO:0071622 | regulation of granulocyte chemotaxis | CAMK1D |
| *GO:0009750* | *response to fructose stimulus* | *SI* |
| *GO:0009744* | *response to sucrose stimulus* | *SI* |
| *GO:0090331* | *negative regulation of platelet aggregation* | *PRKG1* |
| *GO:0048261* | *negative regulation of receptor-mediated endocytosis* | *APOC1* |
| *GO:0090026* | *positive regulation of monocyte chemotaxis* | *CXCL12* |
| *GO:0032869* | *cellular response to insulin stimulus* | *HDAC9,DLC1* |
| *GO:0045806* | *negative regulation of endocytosis* | *PRKD1* |
| *GO:0051929* | *positive regulation of calcium ion transport via voltage-gated calcium channel activity* | *GLP1R* |
| *GO:0070098* | *chemokine-mediated signaling pathway* | *CXCL12* |
| *GO:0010544* | *negative regulation of platelet activation* | *APOE* |
| *GO:0006911* | *phagocytosis, engulfment* | *ELMO1* |
| GO:0031571 | mitotic cell cycle G1/S transition DNA damage checkpoint | TP63 |
| GO:0000302 | response to reactive oxygen species | APOE |
| GO:0006874 | cellular calcium ion homeostasis | RYR3,APOE,CXCL12 |
| GO:0030913 | *paranodal junction assembly* | *NFASC* |
| GO:0022011 | *myelination in peripheral nervous system* | *ARHGEF10* |
| GO:0050658 | RNA transport | RBFOX1 |
| GO:0060060 | post-embryonic retina morphogenesis in camera-type eye | DSCAM |
| GO:0048807 | *female genitalia morphogenesis* | *TP63* |
| GO:0001880 | *Mullerian duct regression* | *SMAD9* |
| GO:0032805 | positive regulation of low-density lipoprotein particle receptor catabolic process | APOE |
| *GO:0035307* | *positive regulation of protein dephosphorylation* | *DLC1* |
| *GO:0033126* | *positive regulation of GTP catabolic process* | *ARHGEF10* |
| GO:0030036 | actin cytoskeleton organization | DLC1,ELMO1,PRKG1 |
| *GO:0090307* | *spindle assembly involved in mitosis* | *ARHGEF10* |
| *GO:0051497* | *negative regulation of stress fiber assembly* | *DLC1* |
| *GO:0016568* | *chromatin modification* | *HDAC9,HIRA,EYA2* |
| *GO:0008064* | *regulation of actin polymerization or depolymerization* | *CXCL12* |
| GO:0060197 | cloacal septation | TP63 |
| *GO:0060157* | *urinary bladder development* | *TP63* |
| *GO:0030850* | *prostate gland development* | *TP63* |
| GO:0051651 | maintenance of location in cell | APOE |
| GO:2001044 | regulation of integrin-mediated signaling pathway | PRKD1 |
| GO:0060267 | positive regulation of respiratory burst | CAMK1D |
| GO:0010468 | regulation of gene expression | APOE,PRDM14 |
| GO:0016576 | histone dephosphorylation | EYA2 |
| *GO:0034983* | *peptidyl-lysine deacetylation* | *HDAC9* |
| *GO:0070932* | *histone H3 deacetylation* | *HDAC9* |
| *GO:0070933* | *histone H4 deacetylation* | *HDAC9* |
| MF | GO:0004551 | nucleotide diphosphatase activity | ENPP2 |
| GO:0005275 | amine transmembrane transporter activity | SLC22A16 |
| GO:0008528 | G-protein coupled peptide receptor activity | GLP1R |
| GO:0060228 | phosphatidylcholine-sterol O-acyltransferase activator activity | APOC1,APOE |
| GO:0071813 | lipoprotein particle binding | APOE |
| GO:0001537 | N-acetylgalactosamine 4-O-sulfotransferase activity | CHST9 |
| GO:0003963 | RNA-3'-phosphate cyclase activity | RCL1 |
| GO:0003997 | acyl-CoA oxidase activity | ACOXL |
| GO:0070326 | very-low-density lipoprotein particle receptor binding | APOE |
| GO:0030151 | molybdenum ion binding | MOSC1 |
| GO:0043546 | molybdopterin cofactor binding | MOSC1 |
| GO:0000166 | nucleotide binding | CELF2,KIF5C,HK1,PRKD1,RAB3C,RBM20,CAMK1D,ALK,KIAA0564,RBFOX1,C8orf80,PRKG1,GNA14 |
| GO:0004438 | phosphatidylinositol-3-phosphatase activity | MTMR14 |
| GO:0016763 | transferase activity, transferring pentosyl groups | GXYLT2 |
| GO:0004558 | alpha-glucosidase activity | SI |
| GO:0008940 | nitrate reductase activity | MOSC1 |
| GO:0004396 | hexokinase activity | HK1 |
| GO:0048156 | tau protein binding | APOE |
| GO:0030618 | transforming growth factor beta receptor, pathway-specific cytoplasmic mediator activity | SMAD9 |
| GO:0015278 | calcium-release channel activity | RYR3 |
| GO:0004692 | cGMP-dependent protein kinase activity | PRKG1 |
| GO:0034190 | apolipoprotein receptor binding | KIF5C |
| GO:0004528 | phosphodiesterase I activity | ENPP2 |
| GO:0045236 | CXCR chemokine receptor binding | CXCL12 |
| GO:0055102 | lipase inhibitor activity | APOC1 |
| GO:0048101 | calcium- and calmodulin-regulated 3',5'-cyclic-GMP phosphodiesterase activity | PDE1C |
| GO:0005042 | netrin receptor activity | DCC |
| GO:0035252 | UDP-xylosyltransferase activity | GXYLT2 |
| GO:0004965 | G-protein coupled GABA receptor activity | GABBR2 |
| CC | GO:0005576 | extracellular region | APOC1,ENPP2,CHST9,APOE,DSCAM,IGSF21,KIAA0564,COL11A1,CXCL12,VWC2 |
| GO:0033010 | paranodal junction | NFASC |
| GO:0042627 | chylomicron | APOC1,APOE |
| *GO:0034363* | *intermediate-density lipoprotein particle* | *APOE* |
| *GO:0034364* | *high-density lipoprotein particle* | *APOC1,APOE* |
| *GO:0034361* | *very-low-density lipoprotein particle* | *APOC1,APOE* |
| GO:0043005 | neuron projection | GABBR2,KIF5C,NRCAM |
| GO:0045121 | membrane raft | DCC,DLC1,HK1,SI |
| GO:0005829 | cytosol | DCC,DLC1,HK1,ARHGEF10,PRKD1,ELMO1,RAB3C,ATP6V1C2,TP63,PDE1C,SMAD9,PRKG1 |
| GO:0005622 | intracellular | ZNF407,DLC1,ARHGEF10,PSD3,PRDM14,RBM20,ATF7,ARHGAP23,SMAD9,STAU2 |
| GO:0045211 | postsynaptic membrane | GABBR2,DLGAP1,PSD3 |
| GO:0005887 | integral to plasma membrane | GABBR2,GLP1R,ENPP2,PRKD1,DSCAM,ALK,NRCAM |
| GO:0030314 | junctional membrane complex | RYR3 |
| GO:0045334 | clathrin-coated endocytic vesicle | GLP1R |
| GO:0005954 | calcium- and calmodulin-dependent protein kinase complex | CAMK1D |
| GO:0005634 | nucleus | HDAC9,CELF2,ZNF407,DLC1,HK1,PRKD1,PRDM14,MBIP,RBM20,ATF7,CAMK1D,TP63,HIRA,RCL1,SMAD9,RBFOX1,C8orf80,STAU2,EYA2 |
| GO:0005737 | cytoplasm | GABBR2,MTMR14,HDAC9,CELF2,KIF5C,DLC1,PRKD1,PSD3,ELMO1,APOE,MBIP,RAB3C,ATF7,CAMK1D,TP63,SMAD9,RBFOX1,PRKG1,STAU2,MTUS2,EYA2 |
| GO:0005592 | collagen type XI | COL11A1 |
| GO:0034399 | nuclear periphery | ATF7 |
| GO:0016021 | integral to membrane | DCC,NFASC,GLP1R,SLC22A16,KCNH7,RYR3,PEX16,CHST9,THSD7A,TM4SF20,GXYLT2,LINGO2,TMEM132D,FNDC3B,SLC35F3,SI,SLCO3A1 |
| GO:0005874 | microtubule | KIF5C,APOE,STAU2 |
| GO:0033270 | paranode region of axon | NFASC |
| GO:0033268 | *node of Ranvier* | *NFASC* |
| GO:0005886 | plasma membrane | DCC,GABBR2,DLGAP1,NFASC,GLP1R,ENPP2,SLC22A16,KCNH7,PRKD1,PSD3,ELMO1,APOE,RAB3C,TM4SF20,DSCAM,ALK,PRKG1,NRCAM,SI,GNA14,SLCO3A1 |
| GO:0031232 | extrinsic to external side of plasma membrane | APOE |
| GO:0032584 | growth cone membrane | DCC |
| GO:0005667 | transcription factor complex | HDAC9,TP63,SMAD9 |

*MF:* GO molecular function; *CC:* GO cellular compartment; *BP:* biological process, *No:* number of genes from the list that have the relevant annotation. . Italicized entries represent the redundant terms; they are placed under their most informative common ancestor (in normal font).