Supplement 1. Genes, the expression of which is regulated by the analyzed nucleotide substitutions

| SNP | Gen | Characteristic | Number of tissues |
|---|---|---|---|
| rs560191 | *AC011330.13* | Uncharacterized transcript, direct intron sequence of the *CATSPER2* gene | 4 |
| | *AC011330.5* | Pseudogene of the *HISPPD2A* gene | 40 |
| | *ADAL* | Adenosine deaminase-like protein. Predicted to be involved in adenosine catabolic process and inosine biosynthetic process. | 37 |
| | *CATSPER2* | Cation channel, sperm associated 2. This gene encodes a member of a family of cation channel proteins that localize to the flagellum of spermatozoa | 1 |
| | *CATSPER2P1* | Cation Channel, Sperm Associated 2 Pseudogene 1 2 | 14 |
| | *CCNDBP1* | Cyclin D1 binding protein 1. Gene product inhibit E2F1-mediated transcription activity. Negative regulator of the cell cycle. | 4 |
| | *CKMT1A* | Two genes located near each other on chromosome 15 have been identified which encode identical mitochondrial creatine kinase proteins. Mitochondrial creatine (MtCK) kinase is responsible for the transfer of high energy phosphate from mitochondria to the cytosolic carrier, creatine. | 4 |
| | *CKMT1B* | | 1 |
| | *LCMT2* | Leucine carboxyl methyltransferase 2 | 5 |
| | *MAP1A* | This gene encodes a protein that belongs to the microtubule-associated protein family. Expression of this gene is almost exclusively in the brain. microtubule-associated protein 1A gene play a role in early events of spinal cord development. | 2 |
| | *PDIA3* | Protein disulfide isomerase family A member 3. This gene encodes a protein of the endoplasmic reticulum, have protein disulfide isomerase activity. This protein also functions as a molecular chaperone that prevents the formation of protein aggregates. | 3 |
| | *RNU6-554P* | RNA, U6 Small Nuclear 554, Pseudogene | 1 |
| | *STRC* | Stereocilin. This gene encodes a protein that is associated with the hair bundle of the sensory hair cells in the inner ear. | 27 |
| | *STRCP1* | *STRC* gene pseudogene 1. Predicted to be involved in cell-matrix adhesion. Predicted to be located in extracellular region. Predicted to be active in cell surface; kinocilium; and stereocilium tip. | 25 |
| | *TGM5* | This gene encodes a member of the transglutaminase family. This protein is stabilize protein assemblies. | 4 |

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| | *TGM7* | This gene encodes a member of the transglutaminase family. This protein is stabilize protein assemblies | 3 |
| | ***TP53BP1*** | Tumor protein P53 binding protein 1. This gene encodes a protein that functions in the DNA double-strand break repair pathway choice, promoting non-homologous end joining (NHEJ) pathways, and limiting homologous recombination. T | 15 |
| | *TTBK2* | This gene encodes a serine-threonine kinase that putatively phosphorylates tau and tubulin proteins. | 2 |
| | *TUBGCP4* | This gene encodes a component of the gamma-tubulin ring complex, which is required for microtubule nucleation. | 2 |
| | *ZSCAN29* | Zinc finger family gene. Predicted to be involved in regulation of transcription by RNA polymerase II. | 29 |
| rs1805800/ rs709816 | *CALB1* | Calbindin 1. The protein encoded by this gene is a member of the calcium-binding protein superfamily that includes calmodulin and troponin C. This protein is thought to buffer entry of calcium upon stimulation of glutamate receptors. | 6/7 |
| | *DECR1* | This gene encodes an accessory enzyme which participates in the beta-oxidation and metabolism of unsaturated fatty enoyl-CoA esters. Mitochondrial. | 5/6 |
| | ***NBN*** | Nibrin. This gene product is thought to be involved in DNA double-strand break repair and DNA damage-induced checkpoint activation.. | 18/14 |
| | *OSGIN2* | Oxidative Stress Induced Growth Inhibitor Family Member 2. Predicted to be involved in negative regulation of cell growth.. | 4/3 |
| | *RP11-662G23.1* | lncRNA. Uncharacterized transcript. | 1/1 |
| rs473297 | *GPR83* | G Protein-Coupled Receptor 83. Predicted to enable neuropeptide receptor activity. Predicted to be involved in neuropeptide signaling pathway. Predicted to act upstream of or within response to glucocorticoid. Located in cilium. | 4 |
| | *IZUMO1R* | IZUMO1 receptor, JUNO. Enables signaling receptor activity. Predicted to be involved in cell adhesion | 1 |
| | ***MRE11*** | This gene encodes a nuclear protein involved in homologous recombination, telomere length maintenance, and DNA double-strand break repair. By itself, the protein has 3' to 5' exonuclease activity and endonuclease activity. | 49 |
| | *RP11-685N10.1* | lncRNA. Uncharacterized transcript. | 37 |
| rs189037/ | *ACAT1* | Acetyl-CoA Acetyltransferase 1. This gene encodes a mitochondrially localized enzyme | 9/4 |

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|---|---|---|---|
| rs1801516 | | that catalyzes the reversible formation of acetoacetyl-CoA from two molecules of acetyl-CoA. | |
| | ***ATM*** | ATM serine/threonine kinase. The protein encoded by this gene belongs to the PI3/PI4-kinase family. This protein is an important cell cycle checkpoint kinase that phosphorylates; thus, it functions as a regulator of a wide variety of downstream proteins, including tumor suppressor proteins p53 and BRCA1, checkpoint kinase CHK2, checkpoint proteins RAD17 and RAD9, and DNA repair protein NBS1. | 15/1 |
| | *C11orf65* | Uncharacterized transcript. Predicted to be involved in negative regulation of mitochondrial fission and negative regulation of protein targeting to mitochondrion. Predicted to be located in cytosol and mitochondrial outer membrane | 1/3 |
| | *KDELC2* | Protein O-Glucosyltransferase 3. Endoplasmic reticulum protein. Targets extracellular EGF repeat proteins, may regulate Notch-signaling. | 5/0 |
| | *NPAT* | Nuclear protein, coactivator of histone. enables protein C-terminus binding activity; transcription coactivator activity; and transcription corepressor activity. Involved in positive regulation of transcription by RNA polymerase II and regulation of transcription involved in G1/S transition of mitotic cell cycle. | 11/2 |
| rs1799977 | *EPM2AIP1* | EPM2A (Laforin) interacting protein 1, its function is not known. This gene is intronless. | 1 |
| | *GOLGA4* | Golgin A4. This gene encodes one of the golgins, a family of proteins localized to the Golgi. Involved in vesicular trafficking at the Golgi apparatus level. Involved in endosome-to-Golgi trafficking | 3 |
| | *ITGA9* | This gene encodes an alpha integrin. Integrins are mediate cell-cell and cell-matrix adhesion. | 1 |
| | *LRRFIP2* | May function as activator of the canonical Wnt signaling pathway, in association with DVL3, upstream of CTNNB1/beta-catenin. Positively regulates TLR signaling in response to agonist | 17 |
| | ***MLH1*** | Miss-match DNA repair protein. | 1 |
| | *PRADC1P1* | Protease Associated Domain Containing 1 (*PRADC1*) Pseudogene 1 | 1 |
| | *RP11-129K12.1* | lncRNA. | 3 |
| | *RP11-259K5.2* | lncRNA. | 1 |
| | *RP11-285J16.1* | lncRNA. | 2 |
| | *UBE2FP1* | UBE2F pseudogene 1 | 2 |

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| rs1805321 | *ANKRD61* | Ankyrin repeat domain-containing protein 61. Located in nucleoplasm. | 7 |
| | *CCZ1* | Enables guanyl-nucleotide exchange factor activity. Predicted to be involved in vesicle-mediated transport. Located in intracellular membrane-bounded organelle | 8 |
| | *CCZ1B* | Enables guanyl-nucleotide exchange factor activity. Predicted to be involved in vesicle-mediated transport. Located in intracellular membrane-bounded organelle. | 11 |
| | *EIF2AK1* | The protein encoded by this gene acts at the level of translation initiation to downregulate protein synthesis in response to stress. The encoded protein is a kinase that can be inactivated by hemin. | 18 |
| | ***PMS2*** | The endonuclease encoded by this gene is a key component of the mismatch repair system that functions to correct DNA mismatches and small insertions and deletions that can occur during DNA replication and homologous recombination. | 30 |
| | *RAC1* | The protein encoded by this gene is a GTPase which belongs to the RAS superfamily of small GTP-binding proteins. Members of this superfamily to regulate a diverse array of cellular events | 1 |
| | *SNORA42* | small nucleolar RNA, H/ACA box 80E | 4 |
| rs20579 | *AC022154.7* | Uncharacterized transcript | 1 |
| | *CARD8* | This protein may be a component of the inflammasome, a protein complex that plays a role in the activation of proinflammatory caspases. | 2 |
| | *CTC-453G23.5* | Uncharacterized transcript | 1 |
| | ***LIG1*** | This gene encodes a member of the ATP-dependent DNA ligase protein family. The encoded protein functions in DNA replication, recombination, and the base excision repair process. | 1 |
| | *PLA2G4C* | Calcium-independent phospholipase, lysophospholipase and O-acyltransferase involved in phospholipid remodeling with implications in endoplasmic reticulum membrane homeostasis and lipid droplet biogenesis. | 22 |
| | *PLA2G4C-AS1* | Is an RNA gene, and is affiliated with the lncRNA class.. | 1 |