## Table S9: OMIM disorders associated with the genes located within the replicated loci

|  |  |  |  |
| --- | --- | --- | --- |
| **Gene** | **OMIM number** | **OMIM disorder** | **Mechanism** |
| ATG16L1 | #611081 | Inflammatory bowel disease 10 | non-synonymous polymorphism associated with reduced protein function |
| BCAS1 | NONE |  |  |
| C11orf36  (MRGPRG antisense RNA) | NONE |  |  |
| C2orf16 | NONE |  |  |
| CARS | NONE |  |  |
| CASR | #145980 | Hypocalciuric hypercalcemia, familial, type I | heterozygous loss-of-function mutations |
| CASR | #239200 | Neonatal severe hyperparathyroidism | loss-of-function |
| CCDC121 | NONE |  |  |
| CDKN1C | **#**130650 | Beckwith-Wiedemann syndrome | imprinting |
| CYP24A1 | #143880 | Hypercalcemia, idiopathic, of infancy | homozygous or compound heterozygous mutation |
| DGKD | NONE |  |  |
| DGKH | NONE |  |  |
| DNAJC5G | NONE |  |  |
| [EIF2B4](#RANGE!_ENREF_21) | #603896 | Leukoencephaly with vanishing white matter ; Ovarioleukodystrophy | autosomal recessive transmission. |
| FNDC4 | NONE |  |  |
| FTHL3P | NONE |  |  |
| GATA3 | #146255 | Hypoparathyroidism, sensorineural deafness, and renal dysplasia | haploinsufficiency |
| GCKR | #61463 | fasting plasma glucose level quantitative trait locus |  |
| [GPN1](#RANGE!_ENREF_30) | NONE |  |  |
| GTF3C2 | NONE |  |  |
| [IFT172](#RANGE!_ENREF_18) | NONE |  |  |
| INPP5D | NONE |  |  |
| KCNQ1 | #607554 | Atrial fibrillation, familial, 3 | missense mutation |
| KCNQ1 | #220400 | Jervell and Lange-Nielsen syndrome | homozygous frameshift mutation or deletion |
| KCNQ1 | #192500 | Long QT syndrome-1 | various mechanisms described including missense mutation |
| KCNQ1 | #609621 | Short QT syndrome-2 | missense mutation |
| KCNQ1OT1 | **#**130650 | Beckwith-Wiedemann syndrome | imprinting |
| KIAA0564 | NONE |  |  |
| KRTCAP3 | NONE |  |  |
| MPV17 | #256810 | Mitochondrial DNA depletion syndrome 6 (hepatocerebral type) |  |
| MRGPRE | NONE |  |  |
| MRGPRG | NONE |  |  |
| NAP1L4 | NONE |  | located 100 kb centromeric to the proximal Beckwith-Wiedemann breakpoint cluster region |
| [NRBP1](#RANGE!_ENREF_19) | NONE |  |  |
| OSBPL5 | NONE |  |  |
| PFDN4 | NONE |  |  |
| PHLDA2 | NONE |  | near Beckwith-Wiedemann syndrome region |
| PPM1G | NONE |  |  |
| SAG | #258100 | Oguchi disease-1 | homozygous deletion resulting in functional null alleles |
| SAG | #613758 | Retinitis pigmentosa 47 | deletion |
| SCARNA5 | NONE |  |  |
| SCARNA6 | NONE |  |  |
| [SLC4A1AP](#RANGE!_ENREF_31) | NONE |  |  |
| [SNX17](#RANGE!_ENREF_20) | NONE |  |  |
| SUPT7L | NONE |  |  |
| [TRIM54](#RANGE!_ENREF_27) | NONE |  |  |
| UCN | NONE |  |  |
| ZNF512 | NONE |  |  |
| [ZNF513](#RANGE!_ENREF_22) | #61367 | Retinitis pigmentosa 58 | homozygous missense mutation |