|  |  |  |  |
| --- | --- | --- | --- |
| Cluster | Exemplary Diseases | Genes | Exemplary GO\_terms |
| 3 | Charcot-Marie-Tooth disease type 2C; Metatropic dysplasia; Brachyolmia type 3; Parastremmatic dwarfism; Spondylometaphyseal dysplasia; Centronuclear myopathy; King Denborough syndrome; Myopathy congenital; Cap myopathy; Congenital fiber type disproportion; Freeman-Sheldon syndrome; Distal arthrogryposis type 1; Spinocerebellar ataxia 15/29; Tubular aggregate myopathy; Rigid spine syndrome; Dysferlinopathy; Becker muscular dystrophy; Duchenne muscular dystrophy | TRPV4; RYR1; TPM3; NALCN; ITPR1; NEB; MYH3; TTN; AK9; CHRNA1; CHRNE; CHRNB1; CACNA1S; CHRNG; ORAI1; BIN1; TPM2; ACTA1; KLHL41; TCAP; CHRND; MYPN; STIM1; MYH7; SELENON; DYSF; DMD; LMOD3; RAPSN | Muscle filament sliding; Muscle organ development; Myofibril assembly; Synaptic transmission, cholinergic; Skeletal muscle tissue development; Neuromuscular synaptic transmission; Skeletal muscle thin filament assembly; Sarcomere organization; Regulation of heart contraction; Calcium ion transport; Regulation of membrane potential; Ligand-gated cation channel activity; |
| 17 | Leber congenital amaurosis; Cone-rod dystrophy; Achromatopsia 2/3; Stargardt disease; Usher syndrome type 1/2A/3A; Corneal dystrophy; Coats disease; Norrie disease; Retinal cone dystrophy 1; | NMNAT1; SAG; RPGR; SPATA7; ZNF408; CDHR1; RHO; PDE6B; RPE65; CRX; TULP1; ABCA4; BEST1; USH2A; PROM1; TGFBI; IMPG2; NDP; OPN1MW; GUCA1A; PDE6H; GNAT2; CNGA3; PDE6C; CNGB3; ATF6; AIPL1; TIMP3; RDH12; IMPDH1; GNAQ; GRK1; PRPH2; LRAT; CLRN1; LRP5; CACNA1F; MYO7A; GUCY2D; TYR; CACNA2D4; FZD4; ADCY5; OPN1LW | Visual perception; Retina homeostasis; Phototransduction, visible light; Photoreceptor cell maintenance; Regulation of rhodopsin mediated signaling pathway; |
| 28 | Autosomal recessive pseudohypoaldosteronism type 1; Liddle syndrome; Brugada syndrome; Thomsen and Becker disease; Familial hemiplegic migraine; Familial infantile convulsions and paroxysmal choreoathetosis; Benign familial infantile epilepsy; Paroxysmal kinesigenic choreoathetosis; Early Infantile Epileptic Encephalopathy; West syndrome; Familial primary hypomagnesemia; Dravet syndrome; Familial atrial fibrillation; Long QT syndrome 1; Progressive familial heart block type 1B/1A/2; Andersen-Tawil syndrome; Hyperkalemic periodic paralysis; Potassium aggravated myotonia; Rapid-onset dystonia-parkinsonism; Congenital insensitivity to pain; Paroxysmal extreme pain disorder; Erythromelalgia; | SCNN1A; CLCN1; PRRT2; CACNA1A; SCNN1G; SIK1; KCNA1; SCN2A; SCN2B; SCN4B; SCN3B; EEF1A2; SCN1B; KCNJ2; SCN4A; PLCB1; TRPM4; ATP1A3; KCNE2; KCNQ2; SCN5A; SCN11A; NKX2-5; SCNN1B; ATP1A2; KCNQ3; SCN1A; KCNT1; SCN8A; SCN9A; SCN10A; AKAP9; ABCC9; DNM1; GRIN1; GRIN2B; KCND3; SYNGAP1; KCNJ8; KCNQ1; GABRA1; KCNJ10; GRIN2A; SLC25A22; KCNE3; CHD2; GABRG2; KCNE1; SLC4A11 | Sodium ion transmembrane transport; Cardiac muscle contraction; Transmission of nerve impulse; Blood circulation; Regulation of ventricular cardiac muscle cell membrane repolarization; Ventricular cardiac muscle cell action potential; Sensory perception of pain; Ion channel binding; Ligand-gated cation channel activity; Potassium channel regulator activity; Calmodulin binding; Glutamate-gated calcium ion channel activity |