**Supplementary table 1. Target genes were listed.**

|  |  |  |  |
| --- | --- | --- | --- |
| Gene HGNC | Disease/Phenotype | OMIM Phenotype ID | Gene ID (OMIM#/GC) |
| *ABCA4* | Stargardt disease 1 | 248200 | 601691 |
| *ADAM9* | Cone-rod dystrophy 9 | 612775 | 602713 |
| *ADAMTS18* | Microcornea, chorioretinal atrophy, and telecanthus | 615458 | 607512 |
| *AHI1* | Joubert syndrome 3 | 608629 | 608894 |
| *AIPL1* | Cone-rod dystrophy, Leber congenital amaurosis 4, RP juvenile | 604393 | 604392 |
| *ALMS1* | Alstrom syndrome | 203800 | 606844 |
| *ARL13B* | Joubert syndrome 8 | 612291 | 608922 |
| *ATF6* | Achromatopsia 7 | 616517 | 605537 |
| *ATXN7* | Spinocerebellar ataxia 7 | 164500 | 607640 |
| *BEST1* | Bestrophinopathy, autosomal recessive | 611809 | 607854 |
| *C10orf11* | Albinism, oculocutaneous, type VII | 615179 | 614537 |
| *C1QTNF5* | Retinal degeneration, late-onset, autosomal dominant | 605670 | 608752 |
| *C21orf2* | Axial Spondylometaphyseal dysplasia | - | - |
| *C5orf42* | Joubert syndrome 17 | 614615 | 614571 |
| *C8orf37* | Cone-rod dystrophy 16, RP 64 | 614500 | 614477 |
| *CABP4* | Cone-rod synaptic disorder, congenital nonprogressive | 610427 | 608965 |
| *CACNA1F* | Night blindness, congenital stationary (incomplete), 2A | 300071 | 300110 |
| *CACNA2D4* | Retinal cone dystrophy 4 | 610478 | 608171 |
| *CAPN5* | Vitreoretinopathy, neovascular inflammatory | 193235 | 602537 |
| *CC2D2A* | COACH syndrome | 216360 | 612013 |
| *CDH3* | Hypotrichosis, congenital, with juvenile macular dystrophy | 601553 | 114021 |
| *CDHR1* | Cone-rod dystrophy 15, RP 65 | 613660 | 609502 |
| *CEP290* | Leber congenital amaurosis 10,  | 611755 | 610142 |
| *CEP41* | Joubert syndrome 15 | 614464 | 610523 |
| *CERKL* | RP 26 | 608380 | 608381 |
| *CFH* | Macular degeneration, age-related, 4 | 610698 | 134370 |
| *CHM* | Choroideremia | 303100 | 300390 |
| *CNGA3* | Achromatopsia 2 | 216900 | 600053 |
| *CNGB3* | Achromatopsia 3 | 262300 | 605080 |
| *CNNM4* | Jalili syndrome | 217080 | 607850 |
| *COL11A1* | Stickler syndrome, type II | 604841 | 120280 |
| *COL11A2* | Stickler syndrome, type III | 184840 | 120290 |
| *COL2A1* | Stickler syndrome, type 1 | 108300 | 120140 |
| *CRB1* | Leber congenital amaurosis 8 | 613835 | 604210 |
| *CRX* | Leber congenital amaurosis 7 | 613829 | 602225 |
| *CSPP1* | Joubert syndrome 21 | 615636 | 611654 |
| *CYP27A1* | Cerebrotendinous xanthomatosis | 213700 | 606530 |
| *DRAM2* | Cone-rod dystrophy 21 | 616502 | 613360 |
| *DTHD1* | Leber congenital amaurosis | - | 616979 |
| *EFEMP1* | Doyne honeycomb degeneration of retina | 126600 | 601548 |
| *ELOVL4* | Stargardt disease 3 | 600110 | 605512 |
| *FRMD7* | Nystagmus 1, congenital, X-linked | 310700 | 300628 |
| *FSCN2* | RP 30 | 607921 | 607643 |
| *FZD4* | Exudative vitreoretinopathy 1 | 133780 | 604579 |
| *GDF6* | Leber congenital amaurosis 17 | 615360 | 601147 |
| *GNAT2* | Achromatopsia 4 | 613856 | 139340 |
| *GPR143* | Ocular albinism, type I | 300500 | 300808 |
| *GUCA1A* | Cone-rod dystrophy 14 | 602093 | 600364 |
| *GUCA1B* | RP 48 | 613827 | 602275 |
| *GUCY2D* | Leber congenital amaurosis 1 | 204000 | 600179 |
| *HMCN1* | Macular degeneration, age-related, 1 | 603075 | 608548 |
| *IFT140* | Short-rib thoracic dysplasia 9 with or without polydactyly | 266920 | 614620 |
| *IMPDH1* | Leber congenital amaurosis 11 | 613837 | 146690 |
| *IMPG1* | Macular dystrophy, vitelliform, 4 | 616151 | 602870 |
| *INPP5E* | Joubert syndrome 1 | 213300 | 613037 |
| *IQCB1* | Senior-Loken syndrome 5 | 609254 | 609237 |
| *JAG1* | Alagille syndrome 1 | 118450 | 601920 |
| *KCNJ13* | Leber congenital amaurosis 16 | 614186 | 603208 |
| *KCNV2* | Retinal cone dystrophy 3B | 610356 | 607604 |
| *KIF7* | Joubert syndrome 12 | 200990 | 611254 |
| *LCA5* | Leber congenital amaurosis 5 | 604537 | 611408 |
| *LRAT* | Leber congenital amaurosis 14 | 613341 | 604863 |
| *MFN2* | Charcot-Marie-Tooth disease, axonal, type 2A2A | 609260 | 608507 |
| *NDP* | Norrie disease | 310600 | 300658 |
| *NMNAT1* | Leber congenital amaurosis 9 | 608553 | 608700 |
| *NPHP1* | Senior-Loken syndrome-1 | 266900 | 607100 |
| *OCA2* | Albinism, oculocutaneous type II | 203200 | 611409 |
| *OFD1* | Joubert syndrome 10 | 300804 | 300170 |
| *OPA1* | Optic atrophy 1 | 165500 | 605290 |
| *OPA3* | Optic atrophy 3 with cataract | 165300 | 606580 |
| *OTX2* | Retinal dystrophy, early-onset, with or withour pituitary dysfunction | 610125 | 600037 |
| *PANK2* | Neurodegeneration with brain iron accumulation 1 | 234200 | 606157 |
| *PAX2* | Papillorenal syndrome | 120330 | 167409 |
| *PAX6* | Foveal hypoplasia 1 | 136520 | 607108 |
| *PDE6C* | Cone dystrophy 4 | 613093 | 600827 |
| *PDE6H* | Achromatopsia 6 | 610024 | 601190 |
| *PITPNM3* | Cone-rod dystrophy 5 | 600977 | 608921 |
| *POC1B* | Cone-rod dystrophy 20 | 615973 | 614784 |
| *PRDM13* | North Carolina macular dystrophy? | - | 616741 |
| *PROM1* | Cone-rod dystrophy 12 | 612657 | 604365 |
| *PRPH2* | Leber congenital amaurosis 18 | 608133 | 179605 |
| *RAB28* | Cone-rod dystrophy 18 | 615374 | 612994 |
| *RAX2* | Cone-rod dystrophy 11 | 610381 | 610362 |
| *RD3* | Leber congenital amaurosis 12 | 610612 | 180040 |
| *RDH12* | Leber congenital amaurosis 13 | 612712 | 608830 |
| *RDH5* | Fundus albitunctatus | 136880 | 601617 |
| *RGS9* | Bradyopsia | 608415 | 604067 |
| *RGS9BP* | Bradyopsia | 608415 | 607814 |
| *RIMS1* | Cone-rod dystrophy 7 | 603649 | 606629 |
| *RP1L1* | Occult macular dystrophy | 613587 | 608581 |
| *RPE65* | Leber congenital amaurosis 2 | 204100 | 180069 |
| *RPGR* | Cone-rod dystrophy, X-linked, 1 | 304020 | 312610 |
| *RPGRIP1* | Leber congenital amaurosis 6 | 613826 | 605446 |
| *RPGRIP1L* | Joubert syndrome 7 | 611560 | 610937 |
| *SEMA4A* | Cone-rod dystrophy 10 | 610283 | 607292 |
| *SLC24A5* | Albinism, oculocutaneous, type VI | 113750 | 609802 |
| *SLC45A2* | Albinism, oculocutaneous, type IV | 606574 | 606202 |
| *SPATA7* | Leber congenital amaurosis 3 | 604232 | 609868 |
| *TCTN3* | Joubert syndrome 18 | 614815 | 613847 |
| *TIMP3* | Sorby fundus dystrophy | 136900 | 188826 |
| *TMEM126A* | Optic atrophy 7 | 612989 | 612988 |
| *TMEM138* | Joubert syndrome 16 | 614465 | 614459 |
| *TMEM216* | Joubert syndrome 2 | 608091 | 613277 |
| *TMEM231* | Joubert syndrome 20 | 614970 | 614949 |
| *TMEM237* | Joubert syndrome 14 | 614424 | 614423 |
| *TMEM67* | COACH syndrome | 216360 | 609884 |
| *TTLL5* | Cone-rod dystrophy 19 | 615860 | 612268 |
| *TULP1* | Leber congenital amaurosis 15 | 613843 | 602280 |
| *TYR* | Albinism, oculocutaneous, type IA | 203100 | 606933 |
| *TYRP1* | Albinism, oculocutaneous, type III | 203290 | 115501 |
| *UNC119* | ?Cone-rod dystrophy | - | 604011 |
| *WT1* | Wilm’s tumor, type I | 194070 | 607102 |
| *ZNF423* | Joubert syndrome 19 | 614844 | 604557 |

Table 1: Genes included in infantile nystagmus syndrome target enrichment. Listed are the genes included in the custom designed target enrichment along with the disease or phenotype associated with the gene according to Online Medelian Inheritance in Man (OMIM), OMIM phenotype identification number, and OMIM or Gene Cards gene identification number. Genes are named according HUGO Gene Nomenclature Committee (HUGO, [http://www.genenames.org/)](http://www.genenames.org/%29) approved nomenclature.