**Table S2. Mis-spliced genes affected by *PRPF31* mutations.**

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| --- | --- | --- | --- | --- |
|  |  |  |  |  |
| **Function** | **Affected genes** | **Associated disease** | **OMIM** | **Reference** |
| **pre-mRNA splicing** | *PRPF3* | Retinitis Pigmentosa | 601414 | (Buskin et al., 2018;Valdes-Sanchez et al., 2019;Azizzadeh Pormehr et al., 2020) |
| *PRPF4* | Retinitis Pigmentosa | 615922 |
| *PRPF8* | Retinitis Pigmentosa | 600059 |
| *PRPF19* | Retinitis Pigmentosa | 608330 |
| *PRPF31* | Retinitis Pigmentosa | 600138 |
| *SF1* | Multiple Endocrine Neoplasia | [601516](http://omim.org/entry/601516) |
| *SART1* | Squamous Cell Carcinoma and Renal Cell Carcinoma, Nonpapillary. | [605941](http://omim.org/entry/605941) |
| *DDX5* | Lung Cancer and Cartilage-Hair Hypoplasia. | [180630](http://omim.org/entry/180630) |
| *LSM2* | Mixed Connective Tissue Disease and Spinal Muscular Atrophy. | [607282](http://omim.org/entry/607282) |
| *CPSF1* | Myopia | [606027](http://omim.org/entry/606027) |
| *U2AF1L4* | Myelophthisic Anemia and Atypical Chronic Myeloid Leukemia. | [601080](http://omim.org/entry/601080) |
| *DDX39B* | Plasmodium Vivax Malaria and Rheumatoid Arthritis. | [142560](http://omim.org/entry/142560) |
| *PTPB1* | Human T-Cell Leukemia Virus Type 2 and Patellar Tendinitis. | [600693](http://omim.org/entry/600693) |
| **phototransduction** | *RHO* | retinitis pigmentosa; dominant congenital stationary night blindness | 180380 | (Yuan et al., 2005;Mordes et al., 2007;Valdes-Sanchez et al., 2019;Azizzadeh Pormehr et al., 2020) |
| *FSCN2* | retinitis pigmentosa; dominant macular dystrophy | 607643 |
| *RDS* | dominant retinitis pigmentosa; dominant macular dystrophy; digenic RP with ROM1; dominant adult vitelliform macular dystrophy; dominant cone-rod dystrophy; dominant central areolar choroidal dystrophy; recessive LCA. | 179605 |
| *ROM1* | retinitis pigmentosa | 180721 |
| *GNAT1* | dominant congenital stationary night blindness, Nougaret type; recessive congenital stationary night blindness | 139330 |
| *PDE6A* | retinitis pigmentosa | 180071 |
| *PDE6B* | retinitis pigmentosa; dominant congenital stationary night blindness | 180072 |
| *ABCA4* | Age-Related Macular Degeneration, Stargardt Disease. | 601691 |
| **ciliogenesis** | *ARL6* | retinitis pigmentosa; recessive Bardet-Biedl syndrome | 608845 | (Ivings et al., 2008;Buskin et al., 2018;Valdes-Sanchez et al., 2019;Kaukonen et al., 2021) |
| *IFT122* | Retinitis Pigmentosa | 606045 |
| *IFT27* | recessive Bardet-Biedl syndrome | 615870 |
| *IFT88* | Retinitis Pigmentosa; Polycystic Kidney Disease | 600595 |
| *SAG* | Retinitis Pigmentosa; recessive Oguchi disease | 181031 |
| *RPGR* | Retinitis Pigmentosa, X-Linked, and Sinorespiratory Infections, With or Without Deafness. | 312610 |
| *RPGRIP1L* | Meckel Syndrome, Type 5 and Joubert Syndrome 7. | 610937 |
| *SORBS1* | Erythematosquamous Dermatosis and Body Mass Index Quantitative Trait Locus 11. | 605264 |
| *PTGS1* | Gastric Ulcer; Aspirin Resistance | 176805 |
| *ODF2* | Spermatogenic Failure; Infertility | 602015 |
| *IFT80* | Short-Rib Thoracic Dysplasia | 611177 |
| *PKD2* | Polycystic Kidney Disease 2 With Or Without Polycystic Liver Disease; Autosomal Dominant Polycystic Kidney Disease | 173910 |
| *RAB15* | Tylosis With Esophageal Cancer; Griscelli Syndrome | 610848 |
| *DYNC2H1* | Short-Rib Thoracic Dysplasia | 603297 |
| *BBS1* | Bardet-Biedl Syndrome | 209901 |
| *BBS4* | Bardet-Biedl Syndrome | 600374 |
| *BBS5* | Bardet-Biedl Syndrome | 603650 |
| *BBS7* | Bardet-Biedl Syndrome | 607590 |
| *BBS9* | Bardet-Biedl Syndrome | 607968 |
| **lysosome** | *CLN3* | Juvenile Neuronal Ceroid Lipofuscinosi, non-syndromic Retinitis Pigmentosa. | 607042 | (Buskin et al., 2018) |
| *AGA* | Aspartylglucosaminuria; Lysosomal Storage Disease | 613228 |
| *ANK3* | Mental Retardation; Autosomal Recessive 37; Neuroma | 600465 |
| *AP5S1* | Spastic Paraplegia | 614824 |
| *CLN5* | Neuronal Ceroid-Lipofuscinoses | 608102 |
| *IL4I1* | Striatonigral Degeneration, Infantile and Primary Mediastinal Large B-Cell Lymphoma. | 609742 |
| *ITM2C* | Cerebral Amyloid Angiopathy, Itm2b-Related, 1 and Cerebral Amyloid Angiopathy, Itm2b-Related, 2. | 609554 |
| *LAMP2* | Danon Disease and Hypertrophic Cardiomyopathy. | 309060 |
| *LDLR* | Hypercholesterolemia, Familial, 1 and Homozygous Familial Hypercholesterolemia. | 606945 |
| *LYN* | Sarcoma and Mastocytosis, Cutaneous. | 165120 |
| *NAGA* | Kanzaki Disease and Schindler Disease, Type I. | 104170 |
| *RPTOR* | Tuberous Sclerosis 1 and Tuberous Sclerosis. | 607130 |
| *SNAP23* | Tetanus and Hemophagocytic Lymphohistiocytosis. | 602534 |
| *STX3* | Diarrhea 2, With Microvillus Atrophy and Tetanus. | 600876 |
| *STXBP2* | Hemophagocytic Lymphohistiocytosis | 601717 |
| *SYT7* | Fetal Akinesia Deformation Sequence 4 and Prostate Cancer. | 604146 |
| *TIAL1* | Salpingitis Isthmica Nodosa and Ulcerative Blepharitis. | 603413 |
| *USP4* | Oculopharyngeal Muscular Dystrophy | 603486 |
| *VPS33B* | Arthrogryposis, Renal Dysfunction, And Cholestasis 1 and Cholestasis. | 608552 |
| **transcription regulation** | *CNOT3* | Intellectual Developmental Disorder With Speech Delay, Autism, And Dysmorphic Facies and Precursor T-Cell Acute Lymphoblastic Leukemia. | 604910 | (Buskin et al., 2018) |
| **endoplasmic reticulum** | *RSK* | Coffin-Lowry Syndrome and Tuberous Sclerosis. | 601684 | (Buskin et al., 2018) |
| *DNAJC10* | Cutis Laxa, Autosomal Dominant 3 and Cutis Laxa. | 607987 |
| *FKBP14* | Ehlers-Danlos Syndrome, Kyphoscoliotic Type, 2 and Muscular Dystrophy. | 614505 |
| *FKBP7* |  | 607062 |
| *OS9* | Overhydrated Hereditary Stomatocytosis and Bone Cancer. | 609677 |
| *P4HA2* | Myopia 25, Autosomal Dominant and Myopia. | 600608 |
| *PDGFC* | Milker's Nodule and Age-Related Macular Degeneration. | 608452 |
| *PRKCSH* | Polycystic Liver Disease 1 With Or Without Kidney Cysts and Polycystic Liver Disease. | 177060 |
| *RDH5* | Fundus Albipunctatus and Fundus Dystrophy. | 601617 |
| *SUMF2* | Multiple Sulfatase Deficiency and Phosphoserine Phosphatase Deficiency. | 607940 |
| *TOR2A* | Dystonia 1, Torsion, Autosomal Dominant and Blepharospasm. | 608052 |
| *UGGT1* | Pulmonary Subvalvular Stenosis and Mitochondrial Complex Iv Deficiency, Nuclear Type 1. | 605897 |
| **unfolded protein response** | *ACADVL* | Acyl-Coa Dehydrogenase, Very Long-Chain, Deficiency Of and Encephalopathy. | 609575 | (Buskin et al., 2018) |
| *ADD1* | Hypertension, Essential and Tracheoesophageal Fistula. | 102680 |
| *ARFGAP1* | Ceroid Lipofuscinosis, Neuronal, 4B, Autosomal Dominant and Hypotrichosis-Lymphedema-Telangiectasia Syndrome. | 608377 |
| *CUL7* | Three M Syndrome 1 and Dubowitz Syndrome. | 609577 |
| *CXXC1* | Zinc Finger Protein 1 and Gait Apraxia. | 609150 |
| *DCTN1* | Perry Syndrome and Neuronopathy, Distal Hereditary Motor, Type Viib. | 601143 |
| *SEC31A* | Neurodevelopmental Disorder With Spastic Quadriplegia, Optic Atrophy, Seizures, And Structural Brain Anomalies and Pseudobulbar Palsy. | 610257 |
| *SEC61A2* |  | 618271 |
| *SEC62* | Polycystic Liver Disease and Hereditary Lymphedema I. | 602173 |
| *SYVN1* | Wolfram Syndrome 1 and Wolfram Syndrome. | 608046 |
| **serine/threonine-protein kinase that activates AMPK** | *SKT11* | Peutz-Jeghers Syndrome and Testicular Germ Cell Tumor. | 602216 | (Ivings et al., 2008) |
| **cell surface receptor and has been implicated as a regulator of synapse formation, neural plasticity, antimicrobial activity, and iron export.** | *APP* | Cerebral Amyloid Angiopathy, App-Related and Alzheimer Disease. | 104760 | (Tanackovic et al., 2011) |
| **Signaling Pathway** | *FGFR1* | Osteoglophonic Dysplasia and Encephalocraniocutaneous Lipomatosis. | 136350 | (Tanackovic et al., 2011) |
| *BMP4* | Microphthalmia, Syndromic 6 and Orofacial Cleft 11. | 112262 |
| *DDR1* | Meninges Sarcoma and Lymphangioleiomyomatosis. | 600408 |
| **apoptosis** | *TRAIL-R2* | Squamous Cell Carcinoma, Head And Neck and Squamous Cell Carcinoma. | 603612 | (Tanackovic et al., 2011) |
| *PTPN13* | Streptococcal Meningitis and Colorectal Cancer, Hereditary Nonpolyposis, Type 6. | 600267 |
| **post-synaptic membrane** | *UTRN* | Muscular Dystrophy, Becker Type and Muscular Dystrophy. | 128240 | (Tanackovic et al., 2011) |
| **autophagy** | *APG5L* | Spinocerebellar Ataxia, Autosomal Recessive 25 and Stomatitis. | 604261 | (Tanackovic et al., 2011) |
| **DNA repair** | *FANCA* | Fanconi Anemia, Complementation Group A and Pituitary Stalk Interruption Syndrome. | 607139 | (Tanackovic et al., 2011) |

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