Table S1. Targeted genes captured by the custom-designed in-solution capture array

Gene	RefSeq	Diseases
ABCA4	NM_000350	recessive Stargardt disease, juvenile and late onset; recessive macular dystrophy; recessive retinitis pigmentosa; recessive fundus flavimaculatus; recessive cone-rod dystrophy
ABCC6	NM_001171	recessive pseudoxanthoma elasticum; dominant pseudoxanthoma elasticum
ABHD12	NM_001042472	recessive syndromic PHARC; recessive Usher syndrome
ADAM9	NM_003816	recessive cone-rod dystrophy
AHI1	NM_001134831	recessive Joubert syndrome
AIPL1	NM_014336	recessive Leber congenital amaurosis; dominant cone-rod dystrophy
ALMS1	NM_015120	recessive Alström syndrome; RP (cone-rod dystrophy)
ARL6	NM_001278293.1	recessive Bardet-Biedl syndrome; recessive retinitis pigmentosa
ARMS2	NM_001099667	age-related macular degeneration
ATXN7	NM_000333	dominant spinocerebellar ataxia w/ macular dystrophy or retinal degeneration
BBS1	NM_024649	recessive Bardet-Biedl syndrome; recessive limb-girdle muscular dystrophy
BBS10	NM_024685	recessive Bardet-Biedl syndrome
BBS12	NM_152618	recessive Bardet-Biedl syndrome
BBS2	NM_031885	recessive Bardet-Biedl syndrome; recessive retinitis pigmentosa
BBS4	NM_033028	recessive Bardet-Biedl syndrome
BBS5	NM_152384	recessive Bardet-Biedl syndrome
BBS7	NM_176824	recessive Bardet-Biedl syndrome
BBS9	NM_014451	recessive Bardet-Biedl syndrome
BEST1	NM_004183	dominant macular dystrophy, Best type; dominant vitreoretinochoroidopathy; recessive bestrophinopathy; recessive retinitis pigmentosa; dominant retinitis pigmentosa
C1QTNF5/ MFRP	NM_015645	dominant macular dystrophy, late onset; dominant macular dystrophy with lens zonules

C2	NM_000063	age-related macular degeneration
C2orf71	NM_001029883	recessive retinitis pigmentosa
C3	NM_000064	age-related macular degeneration
C8ORF37	NM_177965	recessive cone-rod dystrophy; recessive retinitis pigmentosa with early macular involvement
CA4	NM_000717	dominant retinitis pigmentosa
CABP4	NM_145200	recessive congenital stationary night blindness; recessive congenital cone-rod synaptic disease; recessive Leber congenital amaurosis
CACNA1F	NM_005183	X-linked congenital stationary night blindness, incomplete; AIED-like disease; severe congenital stationary night blindness; X-linked progressive cone-rod dystrophy
CACNA2D4	NM_172364	recessive cone dystrophy
CAPN5	NM_033002	dominant neovascular inflammatory vitreoretinopathy
CC2D2A	NM_001080522	recessive retinitis pigmentosa and mental retardation; recessive Joubert syndrome
CD2BP2	NM_001243646.1	Candidate gene
CDH23	NM_022124	recessive Usher syndrome, type 1d; recessive deafness without retinitis pigmentosa; digenic Usher syndrome with PCDH15
CDH3	NM_001793	recessive macular dystrophy, juvenile with hypotrichosis
CDHR1	NM_001171971	recessive cone-rod dystrophy
CEP164	NM_001271933	recessive nephronophthisis with retinal degeneration
CEP290	NM_025114	recessive Senior-Loken syndrome; recessive Joubert syndrome; recessive Leber congenital amaurosis; recessive Meckel syndrome
CERKL	NM_001160277	recessive retinitis pigmentosa; recessive cone-rod dystrophy with inner retinopathy
CFB	NM_001710	age-related macular degeneration
CFH	NM_000186	age-related macular degeneration
CHM	NM_000390	choroideremia
CIB2	NM_001271888	recessive Usher syndrome, type 1J

CISD2	NM_001008388	Candidate gene
CLN3	NM_001042432	recessive Batten disease
CLRN1	NM_174878	recessive Usher syndrome, type 3; recessive retinitis pigmentosa
CNGA1	NM_000087	recessive retinitis pigmentosa
CNGA3	NM_001298	recessive achromatopsia; recessive cone-rod dystrophy
CNGB1	NM_001297	recessive retinitis pigmentosa
CNGB3	NM_019098	recessive achromatopsia Pingelapese; recessive, progressive cone dystrophy
CNNM4	NM_020184	recessive cone-rod dystrophy and amelogenesis imperfecta syndrome
COL11A1	NM_080630	dominant Stickler syndrome, type II; dominant Marshall syndrome
COL2A1	NM_001844	dominant Stickler syndrome, type I; dominant bone dysplasias, developmental disorders, osteoarthritic diseases, and syndromic disorders
COL9A1	NM_001851	recessive Stickler syndrome; dominant multiple epiphyseal dysplasia (MED)
CRB1	NM_201253	recessive retinitis pigmentosa with para-arteriolar preservation of the RPE (PPRPE); recessive retinitis pigmentosa; recessive Leber congenital amaurosis; dominant pigmented paravenous chorioretinal atrophy
CRX	NM_000554	dominant cone-rod dystrophy; recessive, dominant and *de novo* Leber congenital amaurosis; dominant retinitis pigmentosa
CYP4V2	NM_207352	recessive Bietti crystalline corneoretinal dystrophy; recessive retinitis pigmentosa
DDX23	NM_004818.2	Candidate gene
DFNB31	NM_015404	recessive Usher syndrome, type 2; recessive deafness without retinitis pigmentosa
DHDDS	NM_024887	recessive retinitis pigmentosa
DMD	NM_004006	Oregon eye disease (probably)
EFEMP1	NM_001039349	dominant radial, macular drusen; dominant Doyne honeycomb retinal degeneration (Malattia Leventinese)
EFTUD2	NM_001142605.1	Candidate gene

ELOVL4	NM_022726	dominant macular dystrophy, Stargardt-like; recessive spinocerebellar ataxia; recessive ichthyosis, quadriplegia and retardation
ERCC6	NM_000124	age-related macular degeneration, complex etiology; Cockayne syndrome, recessive
EYS	NM_001142800	recessive retinitis pigmentosa
FAM161A	NM_032180	recessive retinitis pigmentosa
FBLN5	NM_006329	familial macular dystrophy, age-related
FLVCR1	NM_014053	recessive retinitis pigmentosa with posterior column ataxia (PCARP)
FSCN2	NM_012418	dominant retinitis pigmentosa; dominant macular dystrophy
FZD4	NM_012193	dominant familial exudative vitreoretinopathy
GNAT1	NM_144499	dominant congenital stationary night blindness, Nougaret type; recessive congenital stationary night blindness
GNAT2	NM_005272	recessive achromatopsia
GNPTG	NM_032520	recessive retinitis pigmentosa and skeletal abnormalities; recessive mucolipidosis III gamma
GPR179	NM_001004334	recessive complete congenital stationary night blindness
GPR98	NM_032119	recessive Usher syndrome, type 2; dominant/recessive febrile convulsions
GRK1	NM_002929	recessive congenital stationary night blindness, Oguchi type
GRM6	NM_000843	recessive congenital stationary night blindness
GUCA1A	NM_000409	dominant cone dystrophy; dominant cone-rod dystrophy
GUCA1B	NM_002098	dominant retinitis pigmentosa; dominant macular dystrophy
GUCY2D	NM_000180	recessive Leber congenital amaurosis; dominant cone-rod dystrophy
HARS	NM_001258040	recessive Usher syndrome
HMCN1	NM_031935	dominant macular dystrophy, age-related
HTRA1	NM_002775	age-related macular degeneration
IDH3B	NM_174855	recessive retinitis pigmentosa
IMPDH1	NM_000883	dominant retinitis pigmentosa; dominant Leber congenital amaurosis

IMPG1	NM_001282368	dominant macular dystrophy, vitelliform; recessive macular dystrophy, vitelliform
IMPG2	NM_016247	recessive retinitis pigmentosa
INPP5E	NM_019892	recessive Joubert syndrome; recessive MORM syndrome
INVS	NM_183245	recessive Senior-Loken syndrome; recessive nephronophthisis
IQCB1	NM_001023571	recessive Senior-Loken syndrome; recessive Leber congenital amaurosis
JAG1	NM_000214	dominant Alagille syndrome
KCNJ13	NM_002242	dominant vitreoretinal degeneration, snowflake; recessive Leber congenital amaurosis
KCNV2	NM_133497	recessive cone dystrophy with supernormal rod electroretinogram
KIF11	NM_004523	dominant microcephaly, lymphedema and chorioretinopathy
KSS	YP_003024036.1	Kearns-Sayre syndrome including retinal pigmentary degeneration
LCA5	NM_001122769	recessive Leber congenital amaurosis
LHON	YP_003024026.1;YP_003024035.1;YP_003024037.1	Leber hereditary optic neuropathy
LRAT	NM_004744	recessive retinitis pigmentosa, severe early-onset; recessive Leber congenital amaurosis
LRIT3	NM_198506	recessive congenital stationary night blindness
LRP5	NM_002335	dominant familial exudative vitreoretinopathy; dominant high bone mass trait; recessive osteoporosis-pseudoglioma syndrome; recessive familial exudative vitreoretinopathy
LZTFL1	NM_001276378	recessive Bardet-Biedl syndrome with developmental anomalies
MAK	NM_005906	recessive retinits pigmentosa
MERTK	NM_006343	recessive retinitis pigmentosa; recessive rod-cone dystrophy
MFN2	NM_001127660	dominant optic atrophy with neuropathy and myopathy; dominant Charcot-Marie-Tooth disease
MFRP	NM_031433	recessive microphthalmos and retinal disease syndrome; recessive nanophthalmos
MKKS	NM_170784	recessive Bardet-Biedl syndrome
MKS1	NM_001165927	recessive Bardet-Biedl syndrome; recessive Meckel syndrome

MT-ATP6	YP_003024031	retinitis pigmentosa with developmental and neurological abnormalities; Leigh syndrome; Leber hereditary optic neuropathy
MT-TH	ENST00000387441	pigmentary retinopathy and sensorineural hearing loss
MT-TL1	ENST00000386347	macular pattern dystrophy with type II diabetes and deafness
MTTP	NM_000253	recessive abetalipoproteinemia
MT-TP	ENST00000387461	retinitis pigmentosa with deafness and neurological abnormalities
MT-TS2	ENST00000387449	retinitis pigmentosa with progressive sensorineural hearing loss
MYO7A	NM_000260	recessive Usher syndrome, type 1b; recessive congenital deafness without retinitis pigmentosa; recessive atypical Usher syndrome (USH3-like)
NDP	NM_000266	Norrie disease; familial exudative vitreoretinopathy; Coats disease
NHP2L1	NM_001003796.1	Candidate gene
NMNAT1	NM_022787	recessive Leber congenital amaurosis
NPHP1	NM_207181	recessive Senior-Loken syndrome; recessive nephronophthisis, juvenile; recessive Joubert syndrome; recessive Bardet-Biedl syndrome
NPHP3	NM_153240	recessive Senior-Loken syndrome; recessive nephronophthisis, adolescent
NPHP4	NM_015102	recessive Senior-Loken syndrome; recessive nephronophthisis, juvenile
NR2E3	NM_014249	recessive enhanced S-cone syndrome (ESC); recessive retinitis pigmentosa in Portuguese Crypto Jews; recessive Goldmann-Favre syndrome; dominant retinitis pigmentosa; combined dominant and recessive retinopathy
NRL	NM_006177	dominant retinitis pigmentosa; recessive retinitis pigmentosa
NYX	NM_022567	X-linked congenital stationary night blindness
OAT	NM_001171814	recessive gyrate atrophy
OFD1	NM_003611	Jobert syndrome; orofaciodigital syndrome 1, Simpson-Golabi-Behmel syndrome 2; X-linked retinitis pigmentosa, severe
OPA1	NM_015560	dominant optic atrophy, Kjer type; dominant optic atrophy with sensorineural hearing loss

OPA3	NM_001017989	recessive optic atrophy with ataxia and 3-methylglutaconic aciduria; dominant optic atrophy with cataract, ataxia and areflexia
OPN1LW	NM_020061	deuteranopia and rare macular dystrophy in blue cone monochromacy with loss of locus control element
OPN1MW	NM_000513	protanopia and rare macular dystrophy in blue cone monochromacy with loss of locus control element
OPN1SW	NM_001708	dominant tritanopia
OTX2	NM_021728	dominant Leber congenital amaurosis and pituitary dysfunction; recessive microphthalmia
PANK2	NM_024960	recessive HARP (hypoprebetalipoproteinemia, acanthocytosis, retinitis pigmentosa, and palladial degeneration); recessive Hallervorden-Spatz syndrome
PAX2	NM_000278	dominant renal-coloboma syndrome
PCDH15	NM_033056	recessive Usher syndrome, type 1f; recessive deafness without retinitis pigmentosa; digenic Usher syndrome with CDH23
PDE6A	NM_000440	recessive retinitis pigmentosa
PDE6B	NM_001145291	recessive retinitis pigmentosa; dominant congenital stationary night blindness
PDE6C	NM_006204	recessive cone dystrophy, early onset; recessive complete and incomplete achromatopsia
PDE6G	NM_002602	recessive retinitis pigmentosa
PDE6H	NM_006205	recessive achromatopsia, incomplete
PDZD7	NM_024895	recessive non-syndromic deafness
PEX1	NM_000466	recessive Refsum disease, infantile form
PEX2	NM_000318	recessive Refsum disease, infantile form
PEX7	NM_000288	recessive Refsum disease, adult form
PGK1	NM_000291	retinitis pigmentosa with myopathy
PHYH	NM_006214	Candidate gene
PITPNM3	NM_031220	dominant cone-rod dystrophy

PLA2G5	NM_001122954	recessive benign fleck retina
PLXNA1	NM_032242.3	Candidate gene
PPIH	XM_005270336.3	Candidate gene
PRCD	NM_001077620	recessive retinitis pigmentosa
PROM1	NM_001145848	recessive retinitis pigmentosa with macular degeneration; dominant Stargardt-like macular dystrophy; dominant macular dystrophy, bull's-eye; dominant cone-rod dystrophy
PRPF3	NM_004698	dominant retinitis pigmentosa
PRPF31	NM_015629	dominant retinitis pigmentosa
PRPF4	NM_001244926.1	dominant retinitis pigmentosa
PRPF6	NM_012469	Candidate gene
PRPF8	NM_006445	dominant retinitis pigmentosa
PRPH2	NM_000322	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
RAX2	NM_032753	cone-rod dystrophy, isolated; age-related macular degeneration, isolated
RB1	NM_000321	dominant germline or somatic retinoblastoma; benign retinoma; pinealoma; osteogenic sarcoma
RBP3	NM_002900	Candidate gene
RBP4	NM_006744	recessive RPE degeneration
RD3	NM_183059	recessive Leber congenital amaurosis
RDH12	NM_152443	recessive Leber congenital amaurosis with severe childhood retinal dystrophy; dominant retinitis pigmentosa
RDH5	NM_002905	recessive fundus albipunctatus; recessive cone dystrophy, late onset
RGR	NM_002921	recessive retinitis pigmentosa; dominant choroidal sclerosis
RGS9	NM_001081955	recessive delayed cone adaptation

RGS9BP	NM_207391	recessive delayed cone adaptation
RHO	NM_000539	dominant retinitis pigmentosa; dominant congenital stationary night blindness; recessive retinitis pigmentosa
RIMS1	NM_014989	dominant cone-rod dystrophy
RLBP1	NM_000326	recessive retinitis pigmentosa; recessive Bothnia dystrophy; recessive retinitis punctata albescens; recessive Newfoundland rod-cone dystrophy
ROM1	NM_000327	dominant retinitis pigmentosa; digenic retinitis pigmentosa with PRPH2
RP1	NM_006269	dominant retinitis pigmentosa; recessive retinitis pigmentosa
RP1L1	NM_178857	dominant occult macular dystrophy; recessive retinitis pigmentosa
RP2	NM_006915	X-linked retinitis pigmentosa; X-linked retinitis pigmentosa, dominant
RP9	NM_203288.1	dominant retinitis pigmentosa
RPE65	NM_000329	recessive Leber congenital amaurosis; recessive retinitis pigmentosa; dominant retinits pigmentosa with choroidal invlovlement
RPGR	NM_000328	X-linked retinitis pigmentosa, recessive; X-linked retinitis pigmentosa, dominant; X-linked cone dystrophy 1; X-linked atrophic macular dystrophy, recessive
RPGRIP1	NM_020366	recessive Leber congenital amaurosis; recessive cone-rod dystrophy
RPGRIP1L	NM_015272	recessive Joubert syndrome; recesssive Meckel syndrome
RS1	NM_000330	Retinoschisis
SAG	NM_000541	recessive Oguchi disease; recessive retinitis pigmentosa
SART1	NM_005146.4	Candidate gene
SDCCAG8	NM_006642	recessive nephronophthisis, ciliopathy-related; recessive Bardet-Biedl syndrome
SEMA4A	NM_022367	dominant retinitis pigmentosa; dominant cone-rod dystrophy
SLC24A1	NM_004727	recessive congenital stationary night blindness
SLC4A5	NM_021196.3	glaucoma; cataract; band keratopathy; intellectual disability
SLC9A8	NM_001260491.1	Candidate gene
SNRNP200	NM_014014.4	dominant retinitis pigmentosa

SNRNP27	NM_006857.2	Candidate gene
SNRNP40	NM_004814.2	Candidate gene
SPATA7	NM_018418	recessive Leber congenital amaurosis; recessive RP
SPP2	NM_006944	dominant retinitis pigmentosa
SQLE	NM_003129	Candidate gene
TEAD1	NM_021961	dominant atrophia areata
TIMM8A	NM_004085	optic atrophy with deafness-dystonia syndrome
TIMP3	NM_000362	dominant Sorsby's fundus dystrophy
TLR3	NM_003265	age-related macular degeneration
TLR4	NM_003266	age-related macular degeneration
TMEM126A	NM_032273	recessive non-syndromic optic atrophy
TMEM216	NM_016499	Joubert Syndrome; Nephronophthisis
TMEM237	NM_001044385	recessive Jobert syndrome
TOPORS	NM_005802	dominant retinitis pigmentosa
TREX1	NM_016381	dominant retinal vasculopathy with cerebral leukodystrophy; dominant Aicardi-Goutiere syndrome 1, dominant chilblain lupus
TRIM32	NM_012210	recessive Bardet-Biedl syndrome; recessive limb-girdle muscular dystrophy
TRPM1	NM_002420	recessive congenital stationary night blindness
TSPAN12	NM_012338	dominant familial exudative vitreoretinopathy
TTC8	NM_198309	recessive Bardet-Biedl syndrome; recessive retinitis pigmentosa
TTPA	NM_000370	recessive retinitis pigmentosa and/or recessive or dominant ataxia
TULP1	NM_003322	recessive retinitis pigmentosa; recessive Leber congenital amaurosis
TXNL4A	NM_001303471.2	Candidate gene
UNC119	NM_005148	dominant cone-rod dystrophy
USH1C	NM_153676	recessive Usher syndrome, Acadian; recessive deafness without retinitis pigmentosa
USH1G	NM_173477	recessive Usher syndrome

USH2A	NM_206933	recessive Usher syndrome, type 2a; recessive retinitis pigmentosa
USP39	NM_001256725.1	Generalized Anxiety Disorder; Agoraphobia
VCAN	NM_001126336	dominant Wagner disease and erosive vitreoretinopathy
WDPCP	NM_015910	recessive Bardet-Biedl syndrome
WDR19	NM_025132	recessive renal, skeletal and retinal anomalies
WFS1	NM_006005	recessive Wolfram syndrome; dominant low frequency sensorineural hearing loss
ZNF423	NM_001271620	recessive Jobert syndrome; recessive nephronophthisis
ZNF513	NM_144631	recessive retinitis pigmentosa