Test Name	Genes involved	Method
Achromatopsia	*CNGA3, CNGB3, GNAT2, PDE6C*	Exome
Albinism	*AP3B1, AP3D1, BLOC1S3, BLOC1S6, DTNBP1, EDN3, EDNRB, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, KIT, LRMDA, LYST, MC1R, MITF, MLPH, MYO5A, OCA2, PAX3, RAB27A, SLC24A5, SLC45A2, SNAI2, SOX10, TYR, TYRP1*	Exome
Alport Syndrome	*CD151, COL4A3, COL4A4, COL4A5, COL4A6, MYH9*	Exome
Ataxia with Oculomotor Apraxia	*APTX, PIK3R5, PNKP, SETX*	Exome
Bardet-Biedl	*ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP290, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP*	Exome
Ciliopathies	*ADGRV1, AHI1, AIPL1, ALMS1, ARL13B, ARL6, ATXN10, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, #C2orf71, CC2D2A, CCDC28B, CCDC39, CCDC40, CDH23, CEP104, CEP290, CFTR, CLRN1, CPLANE1, CRB1, CRX, DNAAF1, DNAAF2, DNAH11, DNAH5, DNAI1, DNAI2, DNAJB11, DNAL1, DYNC2H1, DZIP1L, EVC, EVC2, GANAB, GLIS2, GUCY2D, HYLS1, IFT43, IFT80, IMPDH1, INVS, IQCB1, KCNJ13, KIF7, LCA5, LRAT, MKKS, MKS1, MYO7A, NEK1, NEK8, NKX2-5, NME8, NODAL, NPHP1, NPHP3, NPHP4, OFD1, PCDH15, PKD1, PKD2, PKHD1, RD3, RDH12, RPE65, RPGR, RPGRIP1, RPGRIP1L, RSPH4A, RSPH9, SDCCAG8, SPATA7, TCTN1, TCTN2, TMEM216, TMEM231, TMEM67, TOPORS, TRIM32, TTC21B, TTC8, TULP1, UMOD, USH1C, USH1G, USH2A, VHL, WDPCP, WDR19, WDR35, WHRN, XPNPEP3, ZIC3*	Exome
Comprehensive Eye Disorders	*ABCA4, ABCB6, ABCC6, ABCD1, ABHD12, ACBD5, ACO2, ACTB, ACVR1, ADAM9, ADAMTS18, ADGRA3, ADGRV1, ADIPOR1, AGBL1, AGBL5, AGK, AHI1, AIPL1, ALDH1A3, ALMS1, AMACR, ARHGEF18, ARL13B, ARL2BP, ARL3, ARL6, ATF6, ATOH7, ATXN7, AUH, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCOR, BEST1, BFSP1, BFSP2, BMP4, BMP7, #C12orf57, #C12orf65, C1QTNF5, #C21orf2, #C2orf71, #C8orf37, CA2, CA4, CABP4, CACNA1F, CACNA2D4, CANT1, CAPN5, CAV1, CC2D2A, CDH23, CDH3, CDHR1, CEP164, CEP250, CEP290, CEP41, CERKL, CFH, CHD7, CHM, CHMP4B, CHN1, CHST6, CIB2, CISD2, CLDN19, CLN3, CLN5, CLN6, CLN8, CLPB, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL17A1, COL18A1, COL2A1, COL4A1, COL5A1, COL8A2, COL9A1, CPLANE1, CRB1, CRX, CRYAA, CRYAB, CRYBA1, CRYBA2, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGB, CRYGC, CRYGD, CRYGS, *CRYL1, CSPP1, CTDP1, CTNNA1, CTNNB1, CTSD, CTSF, CYP1B1, CYP4V2, DCN, DGKQ, DHDDS, DHX38, DNAJC5, DRAM2, DTHD1, EFEMP1, ELOVL4, EMC1, EPHA2, ERCC1, ERCC2, ERCC5, ERCC6, EYA1, EYS, FAM126A, FAM161A, FLVCR1, FOXC1, FOXE3, FOXL2, FRAS1, FREM1, FREM2, FSCN2, FTL, FYCO1, FZD4, GALK1, GCNT2, GDF3, GDF6, GFER, GJA1, GJA3, GJA8, GJB2, GJB6, GLI2, GNAT1, GNAT2, GNB3, GNPTG, GPR179, GRIP1, GRK1, GRM6, GRN, GSN, GUCA1A, GUCA1B, GUCY2D, #HARS, HCCS, HCN1, HESX1, HGSNAT, HK1, HMCN1, HMX1, HOXA1, HOXB1, HSF4, IARS2, IDH3A, IDH3B, IFT140, IFT172, IFT27, IFT81, IGBP1, IMPDH1, IMPG1, IMPG2, INPP5E, INVS, IQCB1, #ISPD, ITM2B, JAG1, JAM3, KCNJ13, KCNV2, KCTD7, KERA, KIAA1549, KIF11, KIF21A, KIF7, KIZ, KLHL7, KRT12, KRT3, LAMA1, LCA5, LCAT, LEMD2, LIM2, LMX1B, LOXHD1, LOXL1, LRAT, LRIT3, LRP5, LSS, LTBP2, LZTFL1, MAB21L2, MAF, MAK, MECR, MERTK, MFN2, MFRP, MFSD8, MIP, #MIR184, MITF, MKKS, MKS1, MMACHC, MSMO1, MTPAP, MTTP, MVK, MYO7A, MYOC, NAA10, NDP, NDUFS1, NEK2, NEUROD1, NGLY1, NHS, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NR2F1, NRL, NTF4, NYX, OAT, OCRL, OFD1, OPA1, OPA3, OPTN, OR2W3, OTX2, OVOL2, P3H2, PANK2, PAX2, PAX6, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6D, PDE6G, PDE6H, PDZD7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGK1, PHOX2A, PHYH, PIGL, PIKFYVE, PITPNM3, PITX2, PITX3, PLA2G5, PLK4, PNPLA6, POC1B, POLG, POMGNT1, POMT1, PORCN, PPT1, PQBP1, PRCD, PRDM5, PRKCG, PROM1, PRPF3, PRPF31, PRPF4, PRPF6,*	Exome

	*PRPF8, PRPH2, PRPS1, PRSS56, PXDN, RAB18, RAB28, RAB3GAP1, RAB3GAP2, RARB, RAX, RAX2, RB1, RBP3, RBP4, RD3, RDH11, RDH12, RDH5, REEP6, RERE, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, ROBO3, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, RPGRIP1, RPGRIP1L, RRM2B, RS1, RTN4IP1, SAG, SALL2, SALL4, SBF2, SDCCAG8, SEMA3E, SEMA4A, SH3PXD2B, SHH, SIL1, SIPA1L3, SIX3, SIX6, SLC16A12, SLC24A1, SLC25A46, SLC33A1, SLC38A8, SLC4A11, SLC4A4, *SLC4A7, SLC7A14, SMOC1, SNRNP200, SOX2, SOX5, SPATA7, SPG7, SPP2, SRD5A3, STRA6, TACSTD2, TBC1D20, TBK1, TCF4, TCTN1, TCTN2, TCTN3, TDRD7, TEAD1, TENM3, TFAP2A, TGFBI, TGIF1, TIMM8A, TIMP3, TMEM126A, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TOPORS, TPP1, TREX1, TRIM32, TRNT1, TRPM1, TSPAN12, TTC21B, TTC8, TTLL5, TTPA, TTR, TUB, TUBB3, TUBGCP4, TUBGCP6, TULP1, UBIAD1, UNC119, UNC45B, USH1C, USH1G, USH2A, VAX1, VCAN, VIM, VPS13B, VSX1, VSX2, WDPCP, WDR19, WDR36, WFS1, WHRN, YAP1, ZEB1, ZIC2, ZNF408, ZNF423, ZNF469, ZNF513*	
Comprehensive Glaucoma	*ADAMTS10, ASB10, ATOH7, BEST1, BMP4, CANT1, CNTNAP2, COL18A1, COL4A1, *COL8A1, COL8A2, CREBBP, CRYAA, CRYBA4, CRYGC, CRYGD, CYP1B1, FBN1, FOXC1, FOXE3, GDF6, GJA1, GJA8, #ISPD, LMX1B, LOXL1, LTBP2, MAF, MFRP, MYOC, NOTCH2, NTF4, OPA1, OPA3, OPTC, OPTN, PAX6, PIK3R1, PITX2, PITX3, POMT1, PRSS56, PXDN, RPS19, RRM2B, RS1, SBF2, SH3PXD2B, SIX6, SLC4A4, TBK1, TEK, TMEM126A, TTR, VSX1, VSX2, WDR36*	Exome
Cone-Rod Dystrophy	*ABCA4, ADAM9, AIPL1, BEST1, #C8orf37, CACNA1F, CACNA2D4, CDHR1, CERKL, CNGB3, CNNM4, CRX, DRAM2, GUCA1A, GUCY2D, KCNV2, PDE6C, PDE6H, PITPNM3, POC1B, PROM1, RAB28, RAX2, RDH5, RIMS1, RPGRIP1, SEMA4A, TTLL5, UNC119*	Exome
Corneal Dystrophy	*AGBL1, CHST6, COL17A1, COL8A2, CYP4V2, DCN, GSN, KERA, KRT12, KRT3, LCAT, OVOL2, PAX6, PIKFYVE, PITX2, PRDM5, SLC4A11, TACSTD2, TCF4, TGFBI, UBIAD1, VSX1, ZEB1, ZNF469*	Exome
Comprehensive Cataracts	*ABCB6 *,PTPRU ,SLC25A13 ,SEMA3A ,SEC23A ,POMT1 ,SPINT2 ,LENG8 ,ERCC8 ,COL2A1 ,COL4A1 ,COL4A2 ,TBC1D20 ,CHMP4B ,COL11A1 ,*FAM131A ,CRYAA ,CRYAB ,CRYBA1 ,CRYBA4 ,CRYBB1 ,CRYBB2 ,CRYBB3 ,CRYGB ,CRYGC ,CRYGD ,CRYGS ,B3GLCT ,SIX5 ,*TTC14 ,ESCO2 ,*AKAP14 ,CYP27A1 ,DHCR7 ,EPHA2 ,ERCC1 ,ERCC2 ,ERCC5 ,ERCC6 ,EYA1 ,FBN1 ,RAB3GAP1 ,RAB18 ,FOXC1 ,FOXE3 ,SIPA1L3 ,RHOBTB2 ,*TMED3 ,TDRD7 ,PLD3 ,ALPL ,FTL ,RAB3GAP2 ,GALK1 ,*NECTIN3 ,*STEAP2 ,GCNT2 ,ELP4 ,*STEAP1 ,GJA1 ,*GPR160 ,GJA3 ,GJA8 ,GLA ,TMEM114 ,POMT2 ,HCCS ,HMX1 ,HSF4 ,VSX2 ,GJC3 ,INPP5B ,*TNPO1 ,SLC16A12 ,LAMB1 ,LCT ,LIM2 ,LMX1B ,LRP5 ,LTBP2 ,LTBP3 ,**MIR184 ,MAB21L1 ,MAF ,MAN2B1 ,*MEIS1 ,MIP ,MMP1 ,MSRA ,MYH9 ,NDUFA1 ,NF2 ,NHS ,*NRCAM ,*YBX1 ,OCRL ,SIX6 ,OTX2 ,PAX6 ,MECR ,PEX1 ,PEX6 ,PEX7 ,PEX10 ,PEX12 ,PEX13 ,PITX2 ,PITX3 ,PLD1 ,PON2 ,MXRA8 ,BCOR ,P3H2 ,PEX26 ,AGK ,*PRKCI ,SLC25A40 ,*PROX1 ,*MAN1C1 ,*NIPAL3 ,PTCH1 ,PRX ,EPG5 ,*NECTIN2 ,PEX2 ,ALDH18A1 ,SC5D ,BFSP1 ,SIL1 ,SIX3 ,*SLC1A5 ,UPF3B ,SLC2A1 ,BMP4 ,BMP7 ,*CAPN15 ,SOX1 ,SOX2 ,SREBF2 ,*TACR1 ,VIM ,BEST1 ,WFS1 ,PXDN ,TRAPPC6A ,FYCO1 ,SRD5A3 ,*PEAK1 ,*MAP6D1 ,#CCNP ,OPA3 ,COL18A1 ,ADAMTS10 ,FZD4 ,JAM3 ,*GRWD1 ,*EVA1A ,BFSP2 ,*SLC25A33 ,GNPAT ,#HYCC1 ,PEX3 ,AGPS ,PEX11B ,*MTMR7 ,CTDP1 ,SLC33A1 ,LARGE1 ,RECQL4 ,PEX16 ,*KLHL21*	Exome
Congenital Extraocular Muscles Fibrosis	*TUBB3, PHOX2A ,KIF21A ,COL25A1 ,*	Exome
Congenital Stationary Night Blindness	*GNAT1, GRM6 ,LRIT3 ,TRPM1 ,GPR179 ,PDE6B ,CABP4 ,RDH5 ,RHO ,GRK1 ,NYX ,SAG ,CACNA1F ,SLC24A1*	Exome
Developmental Eye Disease	*ABCB6 ,POMT1 ,MAB21L2 ,VAX1 ,#C12orf57 ,COL4A1 ,COX7B ,CRX ,B3GLCT ,CYP1B1 ,ALDH1A3 ,ATOH7 ,FKTN ,RAB3GAP1 ,RAB18 ,FOXC1 ,FOXE3 ,FOXC2 ,RAB3GAP2 ,TMEM98 ,ELP4 ,POMT2 ,RAX ,HMGB3 ,HMX1 ,VSX2 ,*DCDC1 ,LAMB2 ,GDF6 ,NDP ,SIX6 ,OTX2 ,PAX2 ,PAX6 ,PITX2 ,PITX3 ,BCOR ,CHD7 ,TENM3 ,RARB ,STRA6 ,SHH ,PRSS56 ,SIX3*	Exome

	*,BMP4 ,SLC25A1 ,SOX2 ,SOX3 ,CRPPA ,FKRP ,NAA10 ,MFRP ,CASK ,*SNX3 ,HESX1 ,LARGE1 ,*LHX2 ,GDF3 ,ZEB2*	
Diabetes and Obesity	*GNE ,ALG3 ,ZMPSTE24 ,COG5 ,SLC35A1 ,CEL ,SLC19A2 ,RBCK1 ,CETP ,RAI1 ,SDCCAG8 ,ADCY3 ,PNPLA6 ,IFT27 ,CHD2 ,MAN1B1 ,MRAP2 ,APOA5 ,TTC8 ,CANT1 ,BBS5 ,CP ,CPE ,CREBBP ,#CFAP418 ,VPS13B ,CYP27A1 ,BBS12 ,GLIS3 ,AGL ,AGRP ,ABCA1 ,MEGF8 ,EIF2B1 ,EIF2S3 ,ACSF3 ,ARL13B ,ENO3 ,AKT2 ,FBP1 ,RFX6 ,ALDOA ,DOLK ,CEP164 ,ALDOB ,TRIM32 ,CNOT1 ,MYT1L ,RPGRIP1L ,ATP6V0A2 ,#G6PC1 ,GAA ,PCSK9 ,PTF1A ,COG4 ,SH2B1 ,APPL1 ,LDLRAP1 ,ZBTB20 ,TRAF3IP1 ,IFT172 ,GATA6 ,GBE1 ,GCK ,GCKR ,B4GALT1 ,GH1 ,GHR ,GHRHR ,NPHP3 ,AFF4 ,GPC3 ,BBS9 ,ANGPTL3 ,GLI3 ,GLUD1 ,GNAS ,GPD1 ,SETD2 ,GMPPA ,ALG6 ,GYS1 ,GYS2 ,HADH ,HEXA ,HMGCL ,HMGCS2 ,HNF4A ,AIRE ,HSD11B1 ,APOA1 ,APOB ,GPIHBP1 ,APOC2 ,APOC3 ,ZFP57 ,APOE ,IGF1R ,AQP2 ,INS ,INSR ,PDX1 ,KIF7 ,KCNJ11 ,NHLRC1 ,ACAT1 ,LAMP2 ,LDHA ,LDLR ,LEP ,LEPR ,LIPA ,LIPC ,LIPE ,LMNA ,LPL ,MC3R ,MC4R ,MGAT2 ,MPI ,MPV17 ,ALG11 ,MTNR1B ,MTTP ,MYO5A ,MYO7A ,NDN ,NEUROD1 ,NPHP1 ,NTRK2 ,CISD2 ,OXCT1 ,NEUROG3 ,PAX4 ,PAX6 ,PC ,PCBD1 ,PDE11A ,FOXP3 ,PCK1 ,IER3IP1 ,PCNT ,PCSK1 ,PRKAG2 ,PDE4D ,ENPP1 ,RAB23 ,GHRL ,PFKM ,PGAM2 ,PGK1 ,PGM1 ,PHKA1 ,PHKA2 ,PHKB ,PHKG2 ,PIK3R1 ,PMM2 ,POLD1 ,DPM3 ,POMC ,PRMT7 ,MAGEL2 ,LZTFL1 ,PPARG ,MKS1 ,PHIP ,AVP ,BBS7 ,SLC29A3 ,SLC35C1 ,AVPR2 ,PRKAR1A ,NGLY1 ,TMEM165 ,ALG1 ,DNAJC3 ,INPP5E ,THOC2 ,PTEN ,KIDINS220 ,COG6 ,CC2D2A ,BBS1 ,BBS2 ,PYGL ,PYGM ,BBS4 ,KMT2C ,RDH5 ,PRPH2 ,RHO ,RLBP1 ,RPS6KA3 ,BDNF ,BLK ,XYLT1 ,ABCG5 ,ABCG8 ,NSD1 ,LMF1 ,SIM1 ,SLC2A2 ,SLC16A1 ,SNRPN ,SSR4 ,STAT1 ,STAT3 ,ABCC8 ,TBX3 ,HNF1A ,HNF1B ,THRA ,UCP2 ,UCP3 ,SLC35A2 ,WFS1 ,XRCC4 ,ZNF711 ,MKRN3 ,MOGS ,ALG8 ,ALG12 ,CCDC28B ,EPM2A ,SRD5A3 ,BBS10 ,ALG9 ,ARMC5 ,TTC21B ,EHMT1 ,TUSC3 ,DCAF17 ,CEP290 ,SPG11 ,LAS1L ,MKKS ,TRAPPC9 ,ITCH ,ARL6 ,PHF6 ,NR0B2 ,COG8 ,CUL4B ,KLF11 ,OFD1 ,PPP1R15B ,CEP19 ,DPM1 ,DPM2 ,PROM1 ,AIP ,TMEM67 ,DYRK1B ,RFT1 ,COG7 ,LARGE1 ,TRMT10A ,COG1 ,EIF2AK3 ,MPDU1 ,H6PD*	Exome
Dystroglycanopathy via the LARGE1/LARGE Gene	*LARGE1*	Exome
Early-Onset High Myopia	*IRX5 , POMT1 , PRDM5 , CHST14 , TTC8, CNGA3 , B3GALT6, COL2A1 , COL4A1 , COL5A1 , COL9A1, COL9A2 , COL9A3, COL11A1 , COL11A2 , SLC38A8 , VCAN , CTNNB1, CTSH, #CFAP418, VPS13B, KCNV2 , ADAMTS17 , ADAMTS18, ASXL1 , JAG1, EPHA2, PRIMPOL , EPHB2 , ERBB3, FBN1, FBN2, ATOH7, FGFR3, DZIP1, MYCBP2, CRB1, TSPAN12, PRPF6, NIPBL, LRRC32, GJA1, GJA8, TNFRSF21, MMADHC, GNAT1, GNB3 , SLC39A5, LAMA1, CYP4V2, GRM6 , CPSF1 , GUCY2D , VSX1, LRIT3 , ABCC6, KCNJ13 , KIF11 , ARL2, LRP2 , LRP5, LRPAP1 , LTBP2 , ARR3 , TRPM1*	Exome
Ectopia Lentis	*AASS, ADAMTS10, ADAMTS17, ADAMTSL4, ASPH, BCOR, CBS, COL18A1, FBN1, LTBP2, P3H2, PAX6, PORCN, SUOX, VSX2*	Exome
Flecked Retina	*ABCA4, CHM, CYP4V2, EFEMP1, ELOVL4, LRAT, PLA2G5, PROM1, PRPH2, RDH5, RHO, RLBP1, RS1, VPS13B*	Exome
Glaucoma and Neuro-Ophthalmology	*ASB10, CPAMD8, FOXC1, FOXD3, LTBP2, NTF4, OPA1, OPTN, PAX6, PXDN, SPATA13, SSBP1, TEK, WDR36*	Exome
Hermansky-Pudlak Syndrome	*AP3B1, BLOC1S3, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6*	Exome
Inherited Retinal Disorders	*NR2E3 ,CDH3 ,USH1C ,TOPORS ,CWC27 ,MERTK ,CIB2 ,CCT2 ,PRPF8 ,PLK4 ,SDCCAG8 ,PNPLA6 ,IFT27 ,CEP250 ,CHM ,IFT43 ,TREX1 ,C1QTNF5 ,TPP1 ,CLN3 ,TTC8 ,USH1G ,CNGB1 ,CNGA1 ,CNGA3 ,COL2A1 ,DRAM2 ,COL9A1 ,COL9A2 ,BBS5 ,COL9A3 ,COL11A1 ,COL11A2 ,ZNF513 ,SCLT1 ,HGSNAT ,CRX ,RDH12 ,VCAN ,SAMD11 ,CTNNA1 ,#CFAP418 ,VPS13B ,BBS12 ,ADGRA3 ,LCA5 ,TIMM8A ,KCNV2 ,ADAMTS18 ,JAG1 ,AHR ,ARL13B ,EFEMP1 ,ATOH7 ,CEP164 ,ARSG ,ATF6 ,TRIM32 ,RIMS1 ,SNRNP200 ,CLUAP1 ,EMC1 ,ZNF423 ,TTLL5 ,CLCC1 ,RPGRIP1L ,ARHGEF18 ,EXOSC2 ,CRB1 ,TSPAN12 ,ARL2BP ,AIPL1 ,ABCA4 ,PRPF6 ,FSCN2 ,WHRN ,ABHD12 ,PRPF31 ,TCTN3 ,TRAF3IP1 ,IFT172 ,NPHP4*	Exome

	,CNNM4 ,NPHP3 ,B9D1 ,INVS ,TUBGCP4 ,BBS9 ,GNAT1 ,GNAT2 ,GNB3 ,POC1B ,CYP4V2 ,IFT81 ,FLVCR1 ,GRM6 ,GUCA1A ,GUCA1B ,GUCY2D ,HARS1 ,HK1 ,HMX1 ,IDH3A ,IDH3B ,RD3 ,LRIT3 ,EYS ,IMPDH1 ,IMPG1 ,ABCC6 ,KIF7 ,CERKL ,KCNJ13 ,KIF11 ,RGS9BP ,PCARE ,GDF6 ,DTHD1 ,ARL3 ,LRP2 ,LRP5 ,MAK ,TRPM1 ,GPR179 ,MTTP ,MYO7A ,NDP ,NEK2 ,NEUROD1 ,NPHP1 ,NRL ,GPR143 ,CISD2 ,OAT ,OCA2 ,OPA1 ,ACO2 ,OTX2 ,PAX2 ,PAX6 ,IMPG2 ,WDPCP ,ADIPOR1 ,TRNT1 ,RDH11 ,CEP83 ,SLC45A2 ,TMEM216 ,PCYT1A ,PDE6A ,PDE6C ,PDE6D ,PDE6G ,PDE6H ,TMEM138 ,PDE6B ,PEX1 ,PEX6 ,PEX7 ,PEX10 ,PEX12 ,PEX13 ,PEX14 ,PHYH ,PLA2G5 ,LZTFL1 ,CNGB3 ,AHI1 ,MKS1 ,BBS7 ,RCBTB1 ,P3H2 ,POMGNT1 ,PEX26 ,SPATA7 ,KIZ ,KLHL7 ,PRPS1 ,INPP5E ,CABP4 ,RPGRIP1 ,CC2D2A ,IFT80 ,KIAA1549 ,SLC7A14 ,WDR19 ,BBS1 ,PEX19 ,PEX2 ,BBS2 ,PEX5 ,BBS4 ,PRDM13 ,RBP3 ,RBP4 ,RDH5 ,PRPH2 ,RGR ,RHO ,RLBP1 ,NYX ,AGBL5 ,ROM1 ,RP1 ,RP2 ,OPN1SW ,RPE65 ,RS1 ,SAG ,CDH23 ,SEMA4A ,NMNAT1 ,TMEM237 ,PCDH15 ,CPLANE1 ,SPP2 ,ELOVL4 ,TEAD1 ,NR2F1 ,TIMP3 ,CAPN5 ,TTPA ,TUB ,TULP1 ,TYR ,TYRP1 ,USH2A ,CLRN1 ,BEST1 ,WFS1 ,CFAP410 ,CA4 ,PRCD ,CACNA1F ,ALMS1 ,MAPKAPK3 ,TMEM231 ,TCTN1 ,BBS10 ,ZNF408 ,TTC21B ,CSPP1 ,TCTN2 ,DHDDS ,PDZD7 ,ASRGL1 ,CEP290 ,OPA3 ,ARMC9 ,COL18A1 ,MKKS ,FZD4 ,PITPNM3 ,MFRP ,ADGRV1 ,ARL6 ,CEP78 ,FAM161A ,TMEM126A ,TMEM107 ,GNPTG ,OFD1 ,RTN4IP1 ,RAX2 ,CEP19 ,PEX3 ,TUBGCP6 ,ADAM9 ,MPDZ ,RGS9 ,PEX11B ,PROM1 ,FRMD7 ,UNC119 ,TMEM67 ,PRPF4 ,PRPF3 ,ACBD5 ,SLC24A1 ,CDHR1 ,LRAT ,BBIP1 ,REEP6 ,CACNA2D4 ,RAB28 ,PEX16 ,RP1L1 ,CEP41 ,IQCB1 ,IFT140 ,DHX38 ,KIAA0586 ,MFN2	
Macular Degeneration	*ABCA4, C3, CFB, CFH, CFI, CNGB3, CST3, CX3CR1, EFEMP1, ELOVL4, ERCC6, FBLN5, HMCN1, HTRA1, PRPH2, RAX2, RLBP1, RPGR, TLR4*	Exome
Microphthalmia, Anophthalmia, and Coloboma	*ABCB6, ACTB, ACTG1, ADAMTS18, ALDH1A3, ATOH7, BCOR, BMP4, BMP7, #C12orf57, *CAPN15, CC2D2A, CDK9, CHD7, CLDN19, COL4A1, COX7B, CRYAA, CRYBA4, CYP1B1, DHX38, ERCC1, ERCC2, ERCC5, ERCC6, FAT1, FIBP, FOXC1, FOXE3, FOXL2, FRAS1, FREM1, FREM2, FZD5, GDF3, GDF6, GJA1, GJA8, GLI2, GRIP1, HCCS, HESX1, HMGB3, HMX1, IGBP1, IPO13, LRP5, MAB21L2, MAF, MFRP, MITF, NAA10, NDP, NDUFB11, NHS, OCRL, OTX2, PAX2, PAX6, PIGL, PITX2, PITX3, POLR1C, POLR1D, PORCN, PQBP1, PRR12, PRSS56, PUF60, PXDN, RAB18, RAB3GAP1, RAB3GAP2, RARB, RAX, RBP4, RPGRIP1L, SALL1, SALL2, SALL4, SEMA3E, SHH, SIX3, SIX6, SLC38A8, SMCHD1, SMO, SMOC1, SOX2, SRD5A3, STRA6, TBC1D20, TCOF1, TENM3, TFAP2A, TGIF1, TMEM67, TMEM98, VAX1, VPS13B, VSX1, VSX2, YAP1, ZIC2*	Exome
Mottled Retinal Disorders	*CHM ,VPS13B ,ABCA4 ,CYP4V2 ,PLA2G5 ,RDH5 ,PRPH2 ,RHO ,RLBP1 ,RS1 ,ELOVL4 ,PROM1*	Exome
mtDNA Depletion Syndrome	*AGK, DGUOK, FBXL4, MGME1, MPV17, OPA1, POLG, POLG2, RRM2B, SLC25A4,SUCLA2, SUCLG1, TFAM, TK2, TWNK, TYMP*	Exome
Oculocutaneous Albinism	*AP3D1, GPR143, HPS6, LRMDA, LYST, MC1R, MITF, MYO5A, OCA2, RAB27A, SLC24A5, SLC45A2, TYR, TYRP1*	Exome
Open Angle Glaucoma	*#CYPIB1,PITX2*	Exome
Refsum Disease	*PEX1, PEX2, PEX26, PEX7, PHYH*	Exome
Ring dermoid of cornea	*PITX2*	Exome
Retina Gene Curation	*ABCA4, ADAM9, ADAMTS18, AIPL1, ATF6, CACNA1F, CACNA2D4, CAPN5, CDH3, CEP290, CERKL, CHM, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, CYP4V2, EFEMP1, ELOVL4, EYS, FLVCR1, GNAT2, GPR143, GPR179, GRK1, GRM6, GUCY2D, GUCY2D, HMX1, IDH3B, IFT140, KIF11, KIZ, LCA5, LRP5, MAK, MERTK, NMNAT1, NYX, PCARE, PCYT1A, PDE6A, PPT1, PRPF31, PRPF8, RAB28, RCBTB1, RDH12, RDH5, RGS9, RLBP1, RP1, RP1, RP2, RPE65, RPE65, RPGR, RS1, SNRNP200, TIMP3, TOPORS, TRPM1, TSPAN12, VCAN*	Exome
Retinitis Pigmentosa	*ABCA4, ABHD12, ADGRA3, AIPL1, ARL2BP, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C1QTNF5, #C2orf71, #C8orf37, CA4, CACNA1F, CC2D2A, CDH23,*	Exome

	CDHR1, CEP290, CERKL, CLN3, CLRN1, CNGA1, CNGB1, CRB1, CRX, CYP4V2, DHDDS, DHX38, ELOVL4, EMC1, EYS, FAM161A, FLVCR1, FSCN2, GNPTG, GUCA1B, GUCY2D, HGSNAT, HK1, IDH3B, IFT172, IMPDH1, IMPG2, INPP5E, INVS, IQCB1, KIAA1549, KIZ, KLHL7, LCA5, LRAT, MAK, MERTK, MFRP, MKKS, NEK2, NEUROD1, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NRL, PCDH15, PDE6A, PDE6B, PDE6G, PEX1, PEX2, PEX26, PEX7, PHYH, PITPNM3, PLA2G5, PRCD, PRKCG, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RBP3, RBP4, RD3, RDH11, RDH12, RGR, RHO, RLBP1, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, RPGRIP1, RPGRIP1L, SAG, SEMA4A, SLC7A14, SNRNP200, SPATA7, SPP2, TOPORS, TRIM32, TRNT1, TTC8, TTPA, TUB, TULP1, USH1C, USH2A, WFS1, WHRN, ZNF408, ZNF513	
Retinopathy and Optic Atrophy	*ABCA4, ABCC6, ABHD12, ACBD5, ACO2, ADAM9, ADAMTS18, ADGRV1, AGBL5, AIPL1, ALMS1, AMACR, ARHGEF18, ARL2BP, ARL3, ARL6, ATF6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCOR, BEST1, #C12orf65, C1QTNF5, #C21orf2, #C2orf71, #C8orf37, CA2, CABP4, CACNA1F, CACNA2D4, CDH23, CDH3, CDHR1, CEP250, CEP290, CERKL, CFH, CHM, CIB2, CISD2, CLN3, CLN5, CLN6, CLN8, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL2A1, COL9A1, CRB1, CRX, CTNNA1, CTNNB1, CTSD, CTSF, CYP1B1, CYP4V2, *DGKQ, DHDDS, DHX38, DNAJC5, DRAM2, DTHD1, EFEMP1, ELOVL4, EMC1, EYS, FAM161A, FLVCR1, FOXC1, FSCN2, FZD4, GDF6, GNAT1, GNAT2, GNB3, GPR179, GRK1, GRM6, GRN, GUCA1A, GUCA1B, GUCY2D, #HARS, HCN1, HGSNAT, HK1, HMCN1, HMX1, IDH3A, IDH3B, IFT172, IFT27, IFT81, IMPDH1, IMPG1, IMPG2, IQCB1, ITM2B, KCNV2, KCTD7, KIAA1549, KIZ, KLHL7, LAMA1, LCA5, LRAT, LRIT3, LZTFL1, MAK, MECR, MERTK, MFN2, MFRP, MFSD8, MKKS, MKS1, MMACHC, MYO7A, MYOC, NDP, NEK2, NEUROD1, NMNAT1, NPHP1, NPHP4, NR2E3, NR2F1, NRL, NYX, OAT, OFD1, OPA1, OPA3, OPTN, OR2W3, OTX2, PANK2, PAX2, PAX6, PCDH15, PDE6A, PDE6B, PDE6C, PDE6G, PDE6H, PDZD7, PEX7, PGK1, PHYH, PITPNM3, PITX2, PLA2G5, PLK4, PNPLA6, POC1B, POMGNT1, PPT1, PRCD, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, RAB28, RAX2, RB1, RBP3, RBP4, RD3, RDH11, RDH12, RDH5, REEP6, RGR, RGS9, RHO, RIMS1, RLBP1, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, RPGRIP1, RS1, RTN4IP1, SAG, SDCCAG8, SEMA4A, SLC24A1, SLC25A46, *SLC4A7, SLC7A14, SNRNP200, SOX2, SPATA7, SPP2, TCTN3, TEAD1, TIMP3, TMEM126A, TOPORS, TPP1, TRIM32, TRNT1, TRPM1, TSPAN12, TTC21B, TTC8, TTLL5, TTPA, TUB, TUBGCP4, TUBGCP6, TULP1, UNC119, USH1C, USH1G, USH2A, VCAN, VSX2, WDPCP, WDR19, WHRN, ZNF408, ZNF513*	Exome
Senior-Loken Syndrome	*CEP290, NPHP1, NPHP3, NPHP4, SDCCAG8*	Exome
Septo-optic Dysplasia	*HESX1, OTX2, PAX6, PROKR2, PROP1, SOX2, SOX3, TAX1BP3*	Exome
Stargardt disease	*ABCA4, CNGB3 ,ELOVL4 ,PROM1*	Exome
Usher Syndrome	*ABHD12, ADGRV1, ARSG, CDH23, CEP250, CEP78, CIB2, CLRN1, #HARS, MYO7A, PCDH15, PDZD7, RPGR, USH1C, USH1G, USH2A, WHRN*	Exome
Vitreoretinopathy and Wagner Syndrome	*COL2A1, FZD4, LRP5, NDP, TSPAN12, VCAN*	Exome
Walker Warburg Syndrome	*FKRP, FKTN, #ISPD, LARGE1, POMGNT1, POMT1, POMT2*	Exome
Wolfram Syndrome Comprehensive	*CISD2, WFS1*	Exome
Xeroderma Pigmentosum	*DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, POLH, XPA, XPC*	Exome
Zellweger Syndrome	*PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6*	Exome
Clinical Exome gene panel (6161 genes)	Covering 6161 clinically relevant genes	CES

Whole Exome Sequencing	Covering all the coding regions (~24383)	WES
Whole genome Sequencing	Covering Exons, Introns, Noncoding regions	WGS
Specimen Type	Peripheral blood/purified genomic DNA/chorionic villus sample (CVS)/amniotic fluid/ Dried Blood Spots (FTA Cards)/ Product of Conception (POC)	
Container	EDTA anticoagulated peripheral blood; DNA in sealed eppendorf tube; amniotic fluid in a sterile falcon tube/cultured cells; CVS in a sterile 15ml falcon tube with RPMI1640+10% FBS+ 1% antibiotic. For Dried Blood Spots (FTA Cards) - Whatman FTA card in sealable plastic bag/Envelope cover (Add desiccant packets if available). For Product of conception (POC) - Wide mouth screw capped containers with plain RPMI, or sterile saline may be used for transportation of the specimen.	

*** Genes which are not covered in CES but present in WES.**

 # Genes which are not covered in both CES and WES.

**** Genes which are not covered in WES but present in CES**