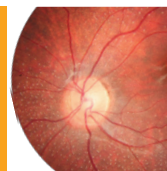


# Ophthalmology



Information for health professionals

## MEDICAL GENETIC TESTING FOR OPHTHALMOLOGY

Recent technologies, in particularly Next Generation Sequencing (NGS), allows fast, accurate and valuable diagnostic tests.

For Ophthalmology, CGC Genetics has an extensive list of medical genetic tests with clinical integration of results by our Medical Geneticists.

### 1. EXOME SEQUENCING:

Exome Sequencing is a very efficient strategy to study most exons of a patient's genome, unraveling mutations associated with specific disorders or phenotypes. With this diagnostic strategy, patients can be studied with a significantly reduced turnaround time and cost.

CGC Genetics has available 2 options for Exome Sequencing:

- Whole Exome Sequencing (WES), which analyzes the entire exome (about 20 000 genes);
- Disease Exome by CGC Genetics, which analyzes about 6 000 clinically-relevant genes.

Any of these can be performed in the index case or in a Trio.

### 2. NGS PANELS

For NGS panels, several genes associated with the same phenotype are simultaneously sequenced. These panels provide increased diagnostic capability with a significantly reduced turnaround time and cost. CGC Genetics has several NGS panels for Ophthalmology that are constantly updated ([www.cgcbgenetics.com](http://www.cgcbgenetics.com)).

Any gene studied in exome or NGS panel can also be individually sequenced and analyzed for deletion/duplication events.

### 3. EXPERTISE IN MEDICAL GENETICS

CGC Genetics has Medical Geneticists specialized in genetic counseling for ophthalmological diseases who may advice in choosing the most appropriate genetic test or help in the interpretation of a result.

## NGS PANELS AVAILABLE FOR OPHTHALMOLOGY:

- |   |  |  |   |
|---|--|--|---|
| • Cone-rod dystrophy                      | • Retinite pigmentosa, AR and X-linked | • Marfan and Loeys-Dietz syndromes and aortic aneurysm | • Neurofibromatosis type 1 and type 2 and Schwannomatosis |
| • Stargardt disease and macular dystrophy | • Stickler syndrome                    | • Albinism   | • Bardet-Biedl syndrome                                   |
| • Microphthalmia                          | • Usher syndrome                       | • Leber congenital amaurosis                           | • Senior-Loken syndrome                                   |
| • Retinitis pigmentosa                    | • Marfan and Loeys-Dietz syndromes     | • Cataracts  |   |
| • Retinitis pigmentosa, AD and X-linked   |  | • Ciliopathies   |   |

# LIST OF TESTS

- Achromatopsia 2 (sequence analysis of CNGA3 gene)
- Achromatopsia 2 (sequence analysis of PDE6C gene)
- Achromatopsia 3 (sequence analysis of CNGB3 gene)
- Achromatopsia 4 (sequence analysis of GNAT2 gene)
- Achromatopsia 6 (sequence analysis of PDE6H gene)
- Alagille syndrome (deletion/duplication analysis on JAG1 gene)
- Alagille syndrome (sequence analysis of JAG1 gene)
- Alagille syndrome 2 (sequence analysis of NOTCH2 gene)
- Aland Island eye disease (sequence analysis of CACNA1F gene)
- Albinism (NGS panel for 12 genes)
- Albinism oculocutaneous type II (deletion/duplication analysis on OCA2 gene)
- Albinism oculocutaneous type II (sequence analysis of OCA2 gene)
- Albinism oculocutaneous type VI (sequence analysis of SLC24A5 gene)
- Albinism oculocutaneous type VII (sequence analysis of C10orf11 gene)
- Albinism, oculocutaneous type I (sequence analysis of TYR gene)
- Albinism, oculocutaneous type IB (deletion/duplication analysis on TYR gene)
- Alstrom syndrome (sequence analysis of ALMS1 gene)
- Alstrom syndrome (sequence analysis of exons 8, 10 and 16 of ALMS1 gene)
- Amish infantile epilepsy syndrome (sequence analysis of ST3GAL5 gene)
- Amyloidosis, finnish type (sequence analysis of GSN gene)
- Aniridia (deletion/duplication analysis on PAX6 gene)
- Aniridia (sequence analysis of PAX6 gene)
- Anterior segment mesenchymal dysgenesis (sequence analysis of FOXE3 gene)
- Anterior segment mesenchymal dysgenesis (sequence analysis of PITX3 gene)
- Aplasia of lacrimal and salivary glands (sequence analysis of FGF10 gene)
- Apolipoprotein E deficiency (sequence analysis of APOE gene)
- Asphyxiating thoracic dystrophy of the newborn type 3 (deletions / duplications in the DYNC2H1 gene)
- Ataxia with oculomotor apraxia (sequence analysis of APTX gene)
- Ataxia, posterior column, with retinitis pigmentosa (sequence analysis of FLVCR1 gene)
- Ataxia-ocular apraxia 2 (AOA2, sequence analysis of SETX gene)
- Ataxia-oculomotor apraxia type 3 (sequence analysis of PIK3R5 gene)
- Ataxia-oculomotor apraxia type 4 (sequence analysis of PNKP gene)
- Athabaskan brainstem dysgenesis syndrome (sequence analysis of HOXA1 gene)
- Axenfeld-Rieger syndrome (deletion/duplication analysis on PITX2 and FOXC1 genes)
- Axenfeld-Rieger syndrome (sequence analysis of FOXC1 gene)
- Axenfeld-Rieger syndrome (sequence analysis of PITX2 gene)
- Bardet-Biedl syndrome (NGS panel for 22 genes)
- Bardet-Biedl syndrome 10 (sequence analysis of BBS10 gene)
- Bardet-Biedl syndrome 12 (sequence analysis of BBS12 gene)
- Bardet-Biedl syndrome 15 (sequence analysis of WDPCP gene)
- Bardet-Biedl syndrome 17 (sequence analysis of LZTFL1 gene)
- Bardet-Biedl syndrome 4 (sequence analysis of BBS4 gene)
- Bardet-Biedl syndrome 7 (sequence analysis of BBS7 gene)
- Bardet-Biedl syndrome 9 (sequence analysis of BBS9 gene)
- Bardet-Biedl syndrome CCDC28B related (sequence analysis of CCDC28B gene)
- Bardet-Biedl syndrome type 2 (sequence analysis of BBS2 gene)
- Bardet-Biedl syndrome type 5 (sequence analysis of BBS5 gene)
- Bardet-Biedl syndrome type 6 (sequence analysis of MKKS gene)
- Blepharophimosis, epicanthus inversus and ptosis, types 1 and 2 (BPES 1 and 2, deletion/duplication analysis on FOXL2 gene)
- Bothnia retinal dystrophy (sequence analysis of RLBP1 gene)
- Bradyopsia (sequence analysis of RGS9 gene)
- Brittle cornea syndrome 1 (sequence analysis of ZNF469 gene)
- Capillary malformations, congenital (mutation p. Arg183Gln on GNAQ gene)
- Carney complex (deletion/duplication analysis on PRKAR1A gene)
- Cataract 1, multiple types (sequence analysis of GJA8 gene)
- Cataract 13 with adult i phenotype (sequence analysis of GCNT2 gene)
- Cataract 17, multiple types (sequence analysis of CRYBB1 gene)
- Cataract 18, AR (sequence analysis of FYCO1 gene)
- Cataract 19 (sequence analysis of CRYBB3 gene)
- Cataract 22, AR (sequence analysis of CRYBB3 gene)
- Cataract 23 (sequence analysis of CRYBA4 gene)
- Cataract 33 (sequence analysis of BFSP1 gene)
- Cataract 36 (sequence analysis of TDRD7 gene)
- Cataract 38, AR (sequence analysis of AGK gene)
- Cataract 4, multiple types (sequence analysis of CRYGD gene)
- Cataract 40, X-linked (sequence analysis of NHS gene)
- Cataract 5, multiple types (sequence analysis of HSF4 gene)
- Cataract 9, multiple types (sequence analysis of CRYAA gene)
- Cataract type 12 (sequence analysis of BFSP2 gene)
- Cataracts (NGS panel for 41 genes)
- Cataracts with facial dysmorphism and neuropathy (sequence analysis of CTD1P1 gene)
- Central areolar choroidal dystrophy (sequence analysis of GUCY2D gene)
- Cerebro oculo facio skeletal syndrome 4 (sequence analysis of ERCC1 gene)
- Cerebrotendinous xanthomatosis (sequence analysis of CYP27A1 gene)
- CHARGE syndrome (deletion/duplication analysis on CHD7 gene)
- CHARGE syndrome (sequence analysis of CHD7 gene)
- CHARGE syndrome (sequence analysis of SEMA3E gene)
- Chediak-Higashi syndrome (deletion/duplication analysis on LYST gene)
- Chediak-Higashi syndrome (sequence analysis of LYST gene)
- CHIME syndrome (sequence analysis of PIGL gene)
- Chondrodysplasia punctata, X-linked dominant (sequence analysis of EBP gene)
- Choroidal dystrophy, central areolar 2 (sequence analysis of PRPH2 gene)
- Choroideremia (deletion/duplication analysis on CHM, RPGR and RP2 gene)
- Choroideremia (sequence analysis of CHM gene)
- Ciliopathies (NGS panel for 91 genes)
- Coloboma of optic disc (deletions/duplications analysis of PAX6 gene)
- Coloboma of optic nerve (sequence analysis of PAX6 gene)
- Combined oxidative phosphorylation deficiency type 15 (sequence analysis of MTMT gene)
- Cone-rod dystrophy (NGS panel for 36 genes)
- Cone-rod dystrophy (sequence analysis of AIPL1 gene)
- Cone-rod dystrophy (sequence analysis of UNC119 gene)
- Cone-rod dystrophy 11 (sequence analysis of RAX2 gene)
- Cone-rod dystrophy 14 (sequence analysis of GUCA1A gene)
- Cone-rod dystrophy 15 (sequence analysis of CDHR1 gene)
- Cone-rod dystrophy 5 (sequence analysis of PITPNM3 gene)
- Cone-rod dystrophy 7 (sequence analysis of RIMS1 gene)
- Cone-rod dystrophy 9 (sequence analysis of ADAM9 gene)
- Cone-rod dystrophy, X-linked (sequence analysis of exon 15a of RPGR gene)
- Congenital disorder of glycosylation type Ia (deletion/duplication analysis on PMM2 gene)
- Congenital disorder of glycosylation type Ia (sequence analysis of PMM2 gene)
- Congenital fibrosis of extraocular muscles (sequence analysis of TUBB2B gene)
- Congenital fibrosis of extraocular muscles (sequence analysis of TUBB3 gene)
- Congenital glaucoma (deletion/duplication analysis on CYP1B1 gene)
- Congenital glaucoma (sequence analysis of CYP1B1 gene)
- Congenital muscular dystrophy and hypoglycosylation of a-dystroglycan (sequence analysis of B3GALNT2 gene)
- Congenital stationary night blindness 1C, AR (deletion/duplication analysis of TRPM1 gene)
- Cornea plana 2, autosomal recessive (sequence analysis of KERA gene)
- Corneal dystrophy, Avellino type (mutations pArg555Trp and pArg124His on TGFB1 gene)
- Corneal endothelial dystrophy 2, AR (sequence analysis of SLC4A11 gene)
- Corpus callosum agenesis - cataract - immunodeficiency (sequence analysis of EPG5 gene)
- Corpus callosum agenesis of with mental retardation, ocular coloboma and micrognathia (sequence analysis of IGBP1 gene)
- Cortical malformations, occipital (sequence analysis of LAMC3 gene)
- Culler-Jones syndrome | Holoprosencephaly 9 (sequence analysis of GLI2 gene)
- Cystinosis (deletion/duplication analysis on CTNS gene)
- Cystinosis (sequence analysis of CTNS gene)
- Diabetes mellitus neonatal (sequence analysis of GLIS3 gene)
- Disease exome by CGC Genetics
- Disease exome-Trio by CGC Genetics
- Doyme honeycomb retinal dystrophy (R345W mutation on EFEMP1 gene)
- Doyme honeycomb retinal dystrophy (sequence analysis of EFEMP1 gene)
- Duane retraction syndrome (sequence analysis of SALL4 gene)
- Duane retraction syndrome 2 (sequence analysis of CHN1 gene)
- Duane-radial ray syndrome (deletion/duplication analysis on SALL4 gene)
- Duane-radial ray syndrome (sequence analysis of SALL4 gene)
- Dyskeratosis congenita (sequence analysis of CTC1 gene)
- Ectodermal dysplasia, ectrodactyly and macular dystrophy (sequence analysis of CDH3 gene)
- Ectodermal dysplasia-ectrodactyly-macular dystrophy syndrome (deletion/duplication analysis of CDH3 gene)
- Ehlers-Danlos, Marfan and Loeys-Dietz syndromes, aortic aneurysm and differential diagnosis (NGS panel for 44 genes)
- Encephalomyopathy, mitochondrial (sequence analysis of MT-TL2 gene)
- Exudative vitreoretinopathy 5 (sequence analysis of TSPAN12 gene)
- Familial ectopia lentis (sequence analysis of ADAMTSL4 gene)
- Fibrosis of extraocular muscles, congenital 2 (sequence analysis of PHOX2A gene)
- Fibrosis of extraocular muscles, congenital type 1 (sequence analysis of KIF21A gene)
- Fleck retina, familial benign (sequence analysis of PLA2G5 gene)
- Frank-ter Haar syndrome (sequence analysis of SH3PXD2B gene)
- Fraser syndrome (sequence analysis of GRIP1 gene)
- Fukuyama congenital muscular dystrophy (sequence analysis of FKTN gene)
- Fundus albipunctatus (sequence analysis of RLBP1 gene)
- Fundus albipunctatus and related disorders (NGS panel for 7 genes)
- Galactosemia type II (sequence analysis of GALK1 gene)
- Gillespie syndrome (sequence analysis of ITPR1 gene)
- Glaucoma, open angle type 1G (sequence analysis of WDR36 gene)
- Glaucoma, primary congenital (sequence analysis of LTBP2 gene)
- Glycosylation disorder type Id (sequence analysis of ALG3 gene)
- Glycosylation disorder type Ii (sequence analysis of ALG2 gene)
- GM1 Gangliosidosis (sequence analysis of GLB1 gene)
- GM2-gangliosidosis type 2 (Sandhoff disease, sequence analysis of HEXB gene)
- Gyrate atrophy of choroid and retina with or without ornithinemia (sequence analysis of OAT gene)
- Hamamy syndrome (sequence analysis of IRX5 gene)
- Hereditary optic neuropathy, including Leber's hereditary optic neuropathy (NGS panel for 5 genes and 3 frequent mutations on mt DNA)
- Hermansky-Pudlak syndrome 1 (sequence analysis of HPS1 gene)
- Hermansky-Pudlak syndrome 2 (sequence analysis of AP3B1 gene)
- Hermansky-Pudlak syndrome 4 (sequence analysis of HPS4 gene)
- Hermansky-Pudlak syndrome 5 (sequence analysis of HPS5 gene)
- Hermansky-Pudlak syndrome 6 (sequence analysis of HPS6 gene)
- Hermansky-Pudlak syndrome 7 (sequence analysis of DTNBP1 gene)
- Hermansky-Pudlak syndrome 8 (sequence analysis of BLOC1S3 gene)
- Hermansky-Pudlak syndrome type 3 (sequence analysis of HPS3 gene)

- Homocystinuria due to cystathionine beta-synthase deficiency (sequence analysis of CBS gene)
- Hyperferritinemia with or without cataract (sequence analysis of the IRE region of the FTL gene)
- Hyperferritinemia-cataract syndrome (sequence analysis of FTL gene)
- Hypomyelination and congenital cataract (sequence analysis of FAM126A gene)
- Hypotrichosis simplex, type 8 (sequence analysis of LPAR6 gene)
- Hypotrichosis with juvenile macular degeneration (deletion/duplication analysis of CDH3 gene)
- IFAP syndrome (sequence analysis of MBTPS2 gene)
- Jalili syndrome (sequence analysis of CNM4 gene)
- Joubert syndrome 3 (sequence analysis of AHI1 gene)
- Joubert syndrome type 14 (JBTS14, sequence analysis of TMEM237 gene)
- Joubert syndrome type 15 (JBTS15, sequence analysis of CEP41 gene)
- Joubert syndrome type 16 (JBTS16, sequence analysis of TMEM138 gene)
- Joubert syndrome type 20 (JBTS20, sequence analysis of TMEM231 gene)
- Joubert syndrome type 5 (sequence analysis of CEP290/NPHP6 gene)
- Joubert syndrome type 8 (JBTS8, sequence analysis of ARL13B gene)
- Joubert syndrome type 9 (sequence analysis of CC2D2A gene)
- Juvenile Paget disease (sequence analysis of TNFRSF11B gene)
- Kearns-Sayre syndrome (KSS, 4977bp deletion in mtDNA)
- Keratoconus type 1 (sequence analysis of VSX1 gene)
- Knobloch syndrome type 1 (sequence analysis of COL18A1 gene)
- LADD syndrome (sequence analysis of FGF10 gene)
- LCAT deficiency (sequence analysis of LCAT gene)
- Leber congenital amaurosis (deletion/duplication analysis on GUCY2D, RDH12, RRGRI1 and CEP290 genes)
- Leber congenital amaurosis (NGS panel for 20 genes)
- Leber congenital amaurosis (sequence analysis of LRAT gene)
- Leber congenital amaurosis (sequence analysis of RPE65 gene)
- Leber congenital amaurosis (sequence analysis of RRGRI1 gene)
- Leber congenital amaurosis 1 (sequence analysis of GUCY2D gene)
- Leber congenital amaurosis 10 (LCA10, sequence analysis of CEP290 gene)
- Leber congenital amaurosis 11 (sequence analysis of IMPDH1 gene)
- Leber congenital amaurosis 12 (LCA12, sequence analysis of RD3 gene)
- Leber congenital amaurosis 13 (sequence analysis of RDH12 gene)
- Leber congenital amaurosis 16 (LCA16, sequence analysis of KCNJ13 gene)
- Leber congenital amaurosis 3 (LCA3, sequence analysis of SPATA7 gene)
- Leber congenital amaurosis 5 (LCA5, sequence analysis of LCA5 gene)
- Leber congenital amaurosis 7 (sequence analysis of CRX gene)
- Leber congenital amaurosis 9 (LCA9, sequence analysis of MNM11 gene)
- Leber congenital amaurosis type 8 (sequence analysis of CRB1 gene)
- Leber congenital neuropathy (sequence analysis of MTCYB gene)
- Leber optic atrophy (sequence analysis of MTCO1 gene)
- Leber optic atrophy (sequence analysis of MTCO3 gene)
- Leber optic atrophy (sequence analysis of MTND1 gene)
- Leber optic atrophy (sequence analysis of MTND2 gene)
- Leber optic atrophy (sequence analysis of MTND4 gene)
- Leber optic atrophy (sequence analysis of MTND4L gene)
- Leber optic atrophy (sequence analysis of MTND5 gene)
- Leber optic atrophy (sequence analysis of MTND6 gene)
- Leber's Hereditary Optic Neuropathy (LHON, 3 frequent mutations)
- Legg-Calve-Perthes disease (sequence analysis of COL2A1 gene)
- Leukodystrophy and leukoencephalopathy (NGS panel for 90 genes)
- Lowe syndrome (sequence analysis of OCRL gene)
- Lysosomal and peroxisomal diseases (NGS panel for 109 genes)
- Macular dystrophy (sequence analysis of PRPH2 gene)
- Macular dystrophy vitelliform (deletion/duplication analysis on BEST1 and PRPH2 genes)
- Macular Dystrophy Vitelliform (sequence analysis of BEST1 and PRPH2 genes)
- Macular dystrophy vitelliform (sequence analysis of BEST1 gene)
- Macular dystrophy vitelliform 4 (sequence analysis of IMPG1 gene)
- Marfan and Loeys-Dietz syndromes (NGS panel for 3 genes)
- Marfan and Loeys-Dietz syndromes and aortic aneurysm (NGS panel for 10 genes)
- Marfan syndrome (deletion/duplication analysis on FBN1 gene)
- Marfan syndrome (sequence analysis of FBN1 gene)
- Marfan syndrome type 2 (sequence analysis of TGFBR1 and TGFBR2 genes)
- Marzolf syndrome (sequence analysis of RAB3GAP2 gene)
- Megalocornea 1, X-linked (sequence analysis of CHRD1 gene)
- Microcephaly and chorioretinopathy with or without mental retardation (sequence analysis of TUBGCP6 gene)
- Microcephaly with or without chorioretinopathy, lymphedema or mental retardation (sequence analysis of KIF11 gene)
- Microcornea, myopic chorioretinal atrophy and telecanthus (sequence analysis of ADAMTS18 gene)
- Microphthalmia (deletion/duplication analysis of BCOR gene)
- Microphthalmia (NGS panel for 26 genes)
- Microphthalmia isolated 2 (sequence analysis of VSX2 gene)
- Microphthalmia isolated 3 (sequence analysis of RAX gene)
- Microphthalmia isolated 8 (sequence analysis of ALDH1A3 gene)
- Microphthalmia syndromic 3 (deletion/duplication analysis on SOX2 gene)
- Microphthalmia syndromic 6 (sequence analysis of BMP4 gene)
- Microphthalmia syndromic 7 (sequence analysis of HCCS gene)
- Microphthalmia syndromic 9 (sequence analysis of STRA6 gene)
- Microphthalmia, Lenz type (sequence analysis of BCOR gene)
- Microphthalmia, syndromic 3 (sequence analysis of SOX2 gene)
- Microphthalmia, syndromic type 1 (deletion/duplication analysis of NAA10 gene)
- Microphthalmia, syndromic type 1 (sequence analysis of NAA10 gene)
- Microvascular complications of diabetes 1 (sequence analysis of VEGFA gene)
- Mitochondrial complex I deficiency (sequence analysis of NDUFB3 gene)
- Mitochondrial complex I deficiency (sequence analysis of NUBPL gene)
- Mitochondrial DNA depletion syndrome 2 (sequence analysis of TK2 gene)
- Mohr-Tranebjaerg syndrome (sequence analysis of TIMM8A gene)
- MOTA syndrome (sequence analysis of FREM1 gene)
- Mucopolipidosis type IV (sequence analysis of MCOLN1 gene)
- Mucopolysaccharidosis IVA (sequence analysis of GALNS gene)
- Mucopolysaccharidosis IVB (sequence analysis of GLB1 gene)
- Mucopolysaccharidosis type Ih (deletion/duplication analysis of IDUA gene)
- Mucopolysaccharidosis type VI (Maroteaux-Lamy, sequence analysis of ARSB gene)
- Mulibrey nanism (sequence analysis of TRIM37 gene)
- Muscle-eye-brain disease (sequence analysis of POMGNT1 gene)
- Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6 (sequence analysis of LARGE gene)
- Myasthenic syndrome, congenital (sequence analysis of CHRNB1 gene)
- Myopathy, mitochondrial progressive with congenital cataract, hearing loss and developmental delay (sequence analysis of GFER gene)
- Myotonic dystrophy type 2 (CTG expansion on CNBP/ZNF9 gene)
- Myotonic dystrophy type I (Steinert disease, DM1, CTG expansion on DMPK gene)
- Nance-Horan syndrome (sequence analysis of NHS gene)
- Nanophthalmia (sequence analysis of MFRP gene)
- Nephronophthisis 14 (sequence analysis of ZNF423 gene)
- Nephronophthisis 15 (sequence analysis of CEP164 gene)
- Neurofibromatosis type 1 (sequence analysis of NF1 gene)
- Neurofibromatosis type 2 (sequence analysis of NF2 gene)
- Neurofibromatosis type I (deletion/duplication analysis on NF1 gene)
- Neurofibromatosis type II (deletion/duplication analysis on NF2 gene)
- Neurofibromatosis types 1 and 2, Legius syndrome and schwannomatosis (NGS panel for 4 genes)
- Neuronal ceroid lipofuscinosis 2 (sequence analysis of TPP1 gene)
- Neuropathy, ataxia, and retinitis pigmentosa (NARP, sequence analysis of MTATP6 gene)
- Night blindness, congenital stationary 1C, AR (sequence analysis of TRPM1 gene)
- Night blindness, congenital stationary (NGS panel for 13 genes)
- Night blindness, congenital stationary 1A, X-linked (sequence analysis of NYX gene)
- Night blindness, congenital stationary 1B, AR (sequence analysis of GRM6 gene)
- Night blindness, congenital stationary 1E, AR (sequence analysis of GPR179 gene)
- Night blindness, congenital stationary 2, AD (sequence analysis of PDE6B gene)
- Night blindness, congenital stationary 2B, AR (sequence analysis of CABP4 gene)
- Night blindness, congenital stationary 3, AD (sequence analysis of GNAT1 gene)
- Norrie disease (deletion/duplication analysis on NDP gene)
- Norrie disease (sequence analysis of NDP gene)
- Nystagmus 1, congenital idiopathic (sequence analysis of FRMD7 gene)
- Occult macular dystrophy (sequence analysis of RP11L1 gene)
- Ocular albinism type 1 (deletion/duplication analysis on GPR143 gene)
- Ocular albinism type I (sequence analysis of GPR143 gene)
- Ocular albinism type III (sequence analysis of TYRP1 gene)
- Ocular albinism type IV (OCA4, sequence analysis of SLC45A2 gene)
- Oculodentodigital dysplasia (sequence analysis of GJA1 gene)
- Oculopharyngeal muscular dystrophy (GCG expansion on PABPN1 gene)
- Oguchi disease 1 (sequence analysis of SAG gene)
- Oguchi disease 2 (sequence analysis of GRK1 gene)
- Optic atrophy 1 (deletion/duplication analysis on OPA1 gene)
- Optic atrophy 10 with or without ataxia, mental retardation, and seizures (sequence analysis of RTN4IP1 gene)
- Optic atrophy 3 (sequence analysis of OPA3 gene)
- Optic atrophy 7 (sequence analysis of TMEM126A gene)
- Optic atrophy type 1 (sequence analysis of OPA1 gene)
- Optic nerve hypoplasia (deletion/duplication analysis on PAX6 gene)
- Osteopetrosis autosomal recessive 8 (sequence analysis of SNX10 gene)
- Osteoporosis - pseudoglioma (sequence analysis of LRP5 gene)
- Papillorenal syndrome (deletion/duplication analysis on PAX2 gene)
- Papillorenal syndrome (sequence analysis of PAX2 gene)
- Paroxysmal extreme pain disorder (sequence analysis of SCN9A gene)
- Peroxisome biogenesis disorder 14B (sequence analysis of PEX11B gene)
- Persistent hyperplastic primary vitreous, AR (sequence analysis of ATOH7 gene)
- Peters anomaly (sequence analysis of FOXC1 gene)
- Peters anomaly (sequence analysis of PAX6 gene)
- Peters anomaly (sequence analysis of PITX2 gene)
- Peters-plus syndrome (frequent mutations of B3GALT gene)
- Peters-plus syndrome (sequence analysis of B3GALT gene)
- Pharmacogenetics of antiangiogenics in ophthalmology (detection of p.Y402H variant in CFH gene)
- Pierson syndrome (sequence analysis of LAMB2 gene)
- Polymicrogyria with optic nerve hypoplasia (sequence analysis of TUBA8 gene)
- Primary adult open-angle glaucoma (sequence analysis of OPTN gene)
- Primary open angle glaucoma (sequence analysis of MYOC gene)
- Progressive external ophthalmoplegia - scoliosis (sequence analysis of ROBO3 gene)
- Progressive external ophthalmoplegia (CPEO, 4977bp deletion in mtDNA)
- Progressive external ophthalmoplegia (deletions / duplications in the POLG, POLG2, SLC25A4 and TWNK genes)
- Progressive external ophthalmoplegia (NGS panel of 12 genes)
- Progressive external ophthalmoplegia (sequence analysis of POLG gene)
- Progressive external ophthalmoplegia with mitochondrial deletions type 5 (sequence analysis of RRM2B gene)
- Prosaposin deficiency (sequence analysis of PSAP gene)
- Pseudoxanthoma elasticum (deletion/duplication analysis on ABCC6 gene)
- Pseudoxanthoma elasticum (sequence analysis of ABCC6 gene)

- Pseudoxanthoma elasticum (sequence analysis of exons 24 and 28 of ABCC6 gene)
- Recessives ataxias (deletion/duplication analysis on SETX, APTX and FXN genes)
- Refsum disease (sequence analysis of PEX7 gene)
- Retinal cone dystrophy 4 (sequence analysis of CACNA2D4 gene)
- Retinal cone dystrophy type 3B (sequence analysis of KCNV2 gene)
- Retinal degeneration late-onset, AD (sequence analysis of C1QTNF5 gene)
- Retinitis pigmentosa (deletion/duplication analysis on BEST1 and PRPH2 genes)
- Retinitis pigmentosa (deletion/duplication analysis on RHO, PRPF31, RP1 and IMPDH1 genes)
- Retinitis pigmentosa (NGS panel for 72 genes)
- Retinitis pigmentosa (p.R283 mutation on CERKL gene)
- Retinitis pigmentosa 1, AD (sequence analysis of RP1 gene)
- Retinitis pigmentosa 10 (sequence analysis of IMPDH1 gene)
- Retinitis pigmentosa 11 (sequence analysis of PRPF31 gene)
- Retinitis pigmentosa 12 (sequence analysis of CRB1 gene)
- Retinitis pigmentosa 13, AD (sequence analysis of PRPF8 gene)
- Retinitis pigmentosa 14, AR (sequence analysis of TULP1 gene)
- Retinitis pigmentosa 17 (sequence analysis of CA4 gene)
- Retinitis pigmentosa 18, AD (sequence analysis of PRPF3 gene)
- Retinitis pigmentosa 19 (deletion/duplication analysis on ABCA4 gene)
- Retinitis pigmentosa 2, X-linked (sequence analysis of RP2 gene)
- Retinitis pigmentosa 25, AR (deletion/duplication analysis on EYS gene)
- Retinitis pigmentosa 25, AR (sequence analysis of EYS gene)
- Retinitis pigmentosa 26 (sequence analysis of CERKL gene)
- Retinitis pigmentosa 27 (sequence analysis of NRL gene)
- Retinitis pigmentosa 28 (sequence analysis of FAM161A gene)
- Retinitis pigmentosa 3 (sequence analysis of RPGR gene)
- Retinitis pigmentosa 30 (sequence analysis of FSCN2 gene)
- Retinitis pigmentosa 31 (sequence analysis of TOPORS gene)
- Retinitis pigmentosa 33 (sequence analysis of SNRNP200 gene)
- Retinitis pigmentosa 35 (sequence analysis of SEMA4A gene)
- Retinitis pigmentosa 36 (sequence analysis of PRCD gene)
- Retinitis pigmentosa 38 (deletion/duplication analysis of MERTK gene)
- Retinitis pigmentosa 38 (sequence analysis of MERTK gene)
- Retinitis pigmentosa 4 (sequence analysis of RHO gene)
- Retinitis pigmentosa 40 (sequence analysis of PDE6B gene)
- Retinitis pigmentosa 42 (sequence analysis of KLHL7 gene)
- Retinitis pigmentosa 43 (sequence analysis of PDE6A gene)
- Retinitis pigmentosa 44 (sequence analysis of RGR gene)
- Retinitis pigmentosa 45 (sequence analysis of CNGB1 gene)
- Retinitis pigmentosa 46 (sequence analysis of IDH3B gene)
- Retinitis pigmentosa 47 (deletions/duplications analysis of SAG gene)
- Retinitis pigmentosa 48 (sequence analysis of GUCA1B gene)
- Retinitis pigmentosa 49 (sequence analysis of CNGA1 gene)
- Retinitis pigmentosa 50 (sequence analysis of BEST1 gene)
- Retinitis pigmentosa 51 (sequence analysis of TTC8 gene)
- Retinitis pigmentosa 54 (sequence analysis of C2ORF71 gene)
- Retinitis pigmentosa 55 (sequence analysis of ARL6 gene)
- Retinitis pigmentosa 56 (sequence analysis of IMPG2 gene)
- Retinitis pigmentosa 57 (sequence analysis of PDE6G gene)
- Retinitis pigmentosa 58 (sequence analysis of ZNF513 gene)
- Retinitis pigmentosa 59 (sequence analysis of DHDDS gene)
- Retinitis pigmentosa 60 (sequence analysis of PRPF6 gene)
- Retinitis pigmentosa 61 (sequence analysis of CLRN1 gene)
- Retinitis pigmentosa 62 (sequence analysis of MAK gene)
- Retinitis pigmentosa 64 (sequence analysis of C8ORF37 gene)
- Retinitis pigmentosa 66 (sequence analysis of RBP3 gene)
- Retinitis pigmentosa 7 (sequence analysis of ROM1 gene)
- Retinitis pigmentosa 9 (sequence analysis of RP9 gene)
- Retinitis pigmentosa type 7 (sequence analysis of PRPH2 gene)
- Retinitis pigmentosa type 37 (sequence analysis of NR2E3 gene)
- Retinitis pigmentosa, AD and X-linked (NGS panel for 27 genes)
- Retinitis pigmentosa, AR and X-linked (NGS panel for 53 genes)
- Retinitis pigmentosa, juvenile (sequence analysis of AIPL1 gene)
- Retinitis punctata albescens (sequence analysis of PRPH2 gene)
- Retinitis punctata albescens (sequence analysis of RDH5 gene)
- Retinoblastoma (deletion/duplication analysis on RB1 gene)
- Retinoblastoma (methylation and deletion/duplication analysis on RB1 gene)
- Retinoblastoma (sequence analysis of RB1 gene)
- Retinopathy of prematurity (sequence analysis of FZD4 gene)
- Retinoschisis (sequence analysis of RS1 gene)
- Riley-Day syndrome (sequence analysis of IKBKAP gene)
- Senior-Loken syndrome (NGS panel for 5 genes)
- Senior-Loken syndrome 4 (sequence analysis of NPHP4 gene)
- Senior-Loken syndrome 5 (sequence analysis of IQCB1 gene)
- Senior-Loken syndrome 7 (sequence analysis of SDCCAG8 gene)
- Septooptic dysplasia (deletion/duplication analysis on HESX1 gene)
- Septooptic dysplasia (NGS panel for 17 genes)
- Septooptic dysplasia (sequence analysis of HESX1 gene)
- SHORT syndrome (sequence analysis of PIK3R1 gene)
- Sorsby fundus dystrophy (sequence analysis of TIMP3 gene)
- Spinocerebellar ataxia 7 (SCA7, CAG expansion on ATXN7 gene)
- Spondyloepiphyseal dysplasia congenita (sequence analysis of COL2A1 gene)
- Stargardt disease 4 (sequence analysis of PROM1 gene)
- Stargardt disease and macular dystrophy (NGS panel for 14 genes)
- Stargardt disease type 1 (deletion/duplication analysis on ABCA4 gene)
- Stargardt disease type 1 (sequence analysis of ABCA4 gene)
- Stargardt disease type 3 (sequence analysis of ELOVL4 gene)
- Stickler syndrome (NGS panel for 6 genes)
- Stickler syndrome (sequence analysis of COL2A1 gene)
- Stickler syndrome type 1 (deletion/duplication analysis on COL2A1 gene)
- Stickler syndrome type 2 (deletion/duplication analysis on COL11A1 gene)
- Stickler syndrome type 2 (sequence analysis of COL11A1 gene)
- Stickler syndrome type 3 (sequence analysis of COL11A2 gene)
- Stickler syndrome type IV (sequence analysis of COL9A1 gene)
- Stickler syndrome type V (sequence analysis of COL9A2 gene)
- Sturge-Weber syndrome (mutation p.Arg183Gln on GNAQ gene)
- Syndromic microphthalmia type 5 (sequence analysis of OTX2 gene)
- Tangier disease (sequence analysis of ABCA1 gene)
- Temtamy syndrome (sequence analysis of C12ORF57 gene)
- Usher syndrome (NGS panel for 12 genes)
- Usher syndrome type 1D/F (deletion/duplication analysis on PCDH15 gene)
- Usher syndrome type 1D/F (sequence analysis of CDH23 gene)
- Usher syndrome type 1D/F (sequence analysis of PCDH15 gene)
- Usher syndrome type 1G (sequence analysis of USH1G gene)
- Usher syndrome type 2A (deletion/duplication analysis on USH2A gene)
- Usher syndrome type 2A (sequence analysis of USH2A gene)
- Usher syndrome type 3A (sequence analysis of CLRN1 gene)
- Usher syndrome type 3B (sequence analysis of HARS gene)
- Usher syndrome type II (sequence analysis of CIB2 gene)
- Van Buchem disease (sequence analysis of SOST gene)
- Wagner syndrome (sequence analysis of VCAN gene)
- WAGR syndrome (deletion/duplication analysis on WT1 gene)
- Walker-Warburg syndrome (sequence analysis of ISPD gene)
- Warburg micro syndrome 3 (sequence analysis of RAB18 gene)
- Warburg micro syndrome type 1 (sequence analysis of RAB3GAP1 gene)
- Weill-Marchesani syndrome (sequence analysis of ADAMTS10 gene)
- Werner syndrome (sequence analysis of WRN gene)
- Wolfram syndrome (sequence analysis of WFS1 gene)
- Wolfram syndrome type 2 (sequence analysis of CISD2 gene)

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