

## Genpanel „hereditäre Netzhauterkrankung“ (RetNet v.04)

ABCA4, ABCC6, ABHD12, ACBD5, ACO2, ADAM9, ADAMTS18, ADGRA3, ADGRV1, ADIPOR1, AFG3L2, AGBL5, AHI1, AHR, AIPL1, ALMS1, AP5Z1, ARHGEF18, ARL2BP, ARL3, ARL6, ARSG, ASRGL1, ATF6, ATXN7, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C12ORF65, C1QTNF5, C3, C8ORF37, CABP4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CCDC51, CCT2, CDH23, CDH3, CDHR1, CEP164, CEP19, CEP250, CEP290, CEP78, CERKL, CFAP410, CFH, CHM, CIB2, CISD2, CLCC1, CLN3, CLRN1, CLUAP1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL2A1, COL9A1, CRB1, CRX, CSPP1, CTNNA1, CTNNB1, CWC27, CYP4V2, DHDDS, DHX38, DNAJC30, DNM1L, DRAM2, DTHD1, DYNC2H1, EFEMP1, ELOVL1, ELOVL4, EMC1, ENSA, ESPN, EXOSC2, EYS, FAM161A, FBLN5, FDXR, FLVCR1, FZD4, GDF6, GNAT1, GNAT2, GNB3, GNPTG, GPR179, GRK1, GRM6, GUCA1A, GUCA1B, GUCY2D, HADHA, HARS, HGSNAT, HK1, HMX1, IDH3A, IDH3B, IFT140, IFT172, IFT27, IFT74, IFT81, IMPDH1, IMPG1, IMPG2, INPP5E, INVS, IQCB1, ITM2B, JAG1, KCNJ13, KCNV2, KIAA1549, KIF11, KIF3B, KIZ, KLHL7, LAMA1, LCA5, LRAT, LRIT3, LRP5, LZTFL1, MAK, MAPKAPK3, MCAT, MERTK, MFN2, MFRP, MFSD8, MIEF1, MKKS, MKS1, MTPAP, MVK, MYO7A, NAALADL1, NBAS, NDP, NDUFS2, NEK2, NEUROD1, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NR2F1, NRL, NYX, OAT, OFD1, OPA1, OPA3, OTX2, PANK2, PAX2, PCARE, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6G, PDE6H, PDZD7, PEX1, PEX2, PEX26, PEX7, PGK1, PHYH, PITPNM3, PLA2G5, PLK4, PNPLA6, POC1B, POC5, POMGNT1, PRCD, PRDM13, PROM1, PROS1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, RAB28, RAX2, RB1, RBP3, RBP4, RCBTB1, RD3, RDH11, RDH12, RDH5, REEP6, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, RPGRIP1, RPGRIP1L, RS1, RTN4IP1, SAG, SAMD11, SCAPER, SCLT1, SDCCAG8, SEMA4A, SEMA6B, SLC24A1, SLC25A46, SLC4A7, SLC7A14, SNRNP200, SPATA7, SPG7, SPP2, SSBP1, TEAD1, TIMM8A, TIMP3, TMEM126A, TMEM216, TMEM237, TOPORS, TREX1, TRIM32, TRNT1, TRPM1, TSPAN12, TTC8, TTLL5, TTPA, TUB, TUBGCP4, TUBGCP6, TULP1, UNC119, USH1C, USH1G, USH2A, USP45, VCAN, WDPCP, WDR19, WDR34, WFS1, WHRN, YME1L1, ZNF408, ZNF423, ZNF513

- **Sub-Genpanel Achromatopsie**  
ATF6, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H
- **Sub-Genpanel Bardet-Biedl-Syndrom**  
ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8ORF37, CEP19, CEP290, IFT27, IFT74, INPP5E, LZTFL1, MKKS, MKS1, NPHP1, SDCCAG8, TRIM32, TTC8, WDPCP
- **Sub-Genpanel Familiäre exsudative Vitreoretinopathie**  
CAPN5, CTNNB1, FZD4, KIF11, LRP5, NDP, RCBTB1, TREX1, TSPAN12, VCAN, ZNF408
- **Sub-Genpanel Kongenitale stationäre Nachtblindheit**  
CABP4, CACNA1F, GNAT1, GNB3, GPR179, GRK1, GRM6, LRIT3, NYX, PDE6B, RDH5, RHO, RPE65, SAG, SLC24A1, TRPM1
- **Sub-Genpanel Leber kongenitale Amaurose**  
AGBL5, AIPL1, ALMS1, CABP4, CCT2, CEP290, CLUAP1, CRB1, CRX, DTHD1, GDF6, GUCY2D, IFT140, IMPDH1, IQCB1, KCNJ13, LCA5, LRAT, NMNAT1, OTX2, PRPH2, RD3, RDH12, RPE65, RPGRIP1, SPATA7, TULP1, USP45

- **Sub-Genpanel Makuladystrophie**

ABCA4, BEST1, C1QTNF5, C3, CDH3, CDHR1, CFH, CNGB3, CRB1, CTNNA1, DRAM2, EFEMP1, ELOVL4, FBLN5, GUCA1B, IMPG1, IMPG2, KIF11, MAPKAPK3, MFSD8, PRDM13, PROM1, PRPH2, RDH5, RP1, RP1L1, RPE65, RPGR, RS1, TIMP3

- **Sub-Genpanel Morbus Stargardt**

ABCA4, CNGB3, ELOVL4, PROM1, PRPH2

- **Sub-Genpanel Optikusatrophie**

ACO2, AFG3L2, C12ORF65, CISD2, DNM1L, ELOVL1, FDXR, MCAT, MFN2, MIEF1, MTPAP, NBAS, NDUFS2, NR2F1, OPA1, OPA3, PRPS1, RTN4IP1, SLC25A46, SPG7, SSBP1, TIMM8A, TMEM126A, WFS1, YME1L1

- **Sub-Genpanel Retinitis pigmentosa**

ABCA4, ADGRA3, ADIPOR1, AGBL5, AHI1, AHR, AP5Z1, ARHGEF18, ARL2BP, ARL3, ARL6, ASRGL1, BBS1, BBS2, BEST1, C8ORF37, CACNA1F, CC2D2A, CCDC51, CDHR1, CEP290, CEP78, CERKL, CHM, CLCC1, CLN3, CLRN1, CNGA1, CNGB1, CRB1, CRX, CWC27, CYP4V2, DHDDS, DHX38, DYNC2H1, EMC1, ENSA, EXOSC2, EYS, FAM161A, FLVCR1, GNAT1, GUCA1B, HGSNAT, HK1, IDH3A, IDH3B, IFT140, IFT172, IMPDH1, IMPG2, KIAA1549, KIF3B, KIZ, KLHL7, LRAT, MAK, MERTK, MFRP, MVK, NAALADL1, NEK2, NEUROD1, NR2E3, NRL, OAT, PCARE, PCYT1A, PDE6A, PDE6B, PDE6G, POC1B, POMGNT1, PRCD, PROM1, PROS1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RBP3, RBP4, RCBTB1, RDH11, RDH12, REEP6, RGR, RHO, RLBP1, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, SAG, SAMD11, SCAPER, SCLT11, SEMA4A, SLC7A14, SNRNP200, SPATA7, SPP2, TEAD1, TOPORS, TRNT1, TTC8, TTPA, TUB, TULP1, USH2A, WDR34, ZNF408, ZNF513

- **Sub-Genpanel Usher-Syndrom**

ABHD12, ADGRV1, ARSG, CDH23, CEP250, CIB2, CLRN1, ESPN, HARS, MYO7A, PCDH15, PDZD7, USH1C, USH1G, USH2A, WHRN

- **Sub-Genpanel Zapfen-Stäbchen-Dystrophie**

ABCA4, ADAM9, AIPL1, ATF6, C8ORF37, CABP4, CACNA1F, CACNA2D4, CDHR1, CEP250, CEP78, CERKL, CFAP410, CNGA3, CNGB3, CNM4, CRX, GNAT2, GUCA1A, GUCY2D, IFT81, KCNV2, KIF11, PCYT1A, PDE6C, PDE6H, PITPNM3, POC1B, PROM1, PRPH2, RAB28, RAX2, RDH5, RGS9, RGS9BP, RIMS1, RP1, RP1L1, RPE65, RPGR, RPGRIP1, SEMA4A, SEMA6B, SLC4A7, TLL5, UNC119