Gen-Panels Augenerkrankungen | LI

Hinweise:

- Gen-Panels mit mehr als 10 Genen dürfen gemäss Analysenliste des BAG nur durch Ärzte mit einem FMH-Titel Medizinische Genetik verordnet werden.
- Die Gen-Panels sind nicht endgültig und können je nach Fragestellung auf Wunsch angepasst werden.
- Die Gen-Panels können abhängig von der aktuellen Datenlage zum Zeitpunkt der Analysen von denjenigen auf dieser Liste abweichen.

Achromatopsie v1 (6 Gene)

ATF6, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H

Anterior segment dysgenesis v2 (179 Gene)

ABCB6, ABHD12, ACTB, ADAMTS10, ADAMTS17, ADAMTSL4, AGBL1, AGK, ALDH1A3, ASPH, ATOH7, B3GLCT, BCOR, BEST1, BFSP1, BFSP2, BMP4, C12orf57, C2CD3, CC2D2A, CHD7, CHMP4B, CHRDL1, CHST6, CLDN19, CLPB, COL17A1, COL18A1, COL4A1, COL5A1, COL8A2, CPAMD8, CRYAA, CRYAB, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGA, CRYGB, CRYGC, CRYGD, CRYGS, CTDP1, CYP1B1, CYP27A1, CYP4V2, DCN, EPG5, EPHA2, EYA1, FAM111A, FAM126A, FGF5, FOXC1, FOXE3, FRAS1, FREM1, FREM2, FTL, FYCO1, GALK1, GALT, GCNT2, GDF3, GDF6, GJA1, GJA3, GJA8, GLI2, GRHL2, GRIP1, GSN, HCCS, HDAC6, HMGB3, HMX1, HSF4, IGBP1, INPP5E, IRX5, KIF11, KRT12, KRT3, LCAT, LGR4, LIM2, LOXHD1, LOXL1, LRP2, LTBP2, MAB21L2, MAF, MASP1, MEF2C, MFRP, MIP, MIR184, MITF, MPDZ, MYOC, NAA10, NDP, NHS, NMNAT1, OCRL, OPA3, OSGEP, OTX2, P3H2, PAX2, PAX3, PAX6, PDGFRB, PEX7, PIGL, PIKFYVE, PITX2, PITX3, POMGNT1, POMT1, PORCN, PQBP1, PRDM5, PRSS56, PUF60, PXDN, RAB3GAP1, RARB, RAX, RAX2, RBP4, RECQL4, RPGRIP1L, SALL2, SALL4, SEMA3E, SH3PXD2B, SHH, SIL1, SIPA1L3, SIX3, SIX6, SLC16A12, SLC38A8, SLC4A11, SMCHD1, SMG9, SMO, SMOC1, SOX2, SRD5A3, STRA6, TACSTD2, TBX1, TCF4, TDRD7, TENM3, TFAP2A, TGFBI, TMEM67, TMEM98, TMX3, UBIAD1, VAX1, VAX2, VIM, VPS13B, VSX1, VSX2, WASHC5, WDR37, WRN, YAP1, ZEB1, ZEB2, ZIC2, ZNF469

Cornea-Dystrophie v1 (27 Gene)

AGBL1, CHST6, COL17A1, COL5A1, COL8A2, CYP4V2, DCN, FOXE3, GJA8, GRHL2, GSN, KRT12, KRT3, LCAT, LOXHD1, MAF, PIKFYVE, PITX2, PRDM5, SLC4A11, TACSTD2, TCF4, TGFBI, UBIAD1, VSX1, ZEB1, ZNF469

Hermansky-Pudlak-Syndrom v1 (9 Gene)

AP3B1, BLOC1S3, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6

Katarakte v2 (61 Gene)

ABHD12, AGK, BCOR, BFSP1, BFSP2, CHMP4B, CLPB, COL4A1, CRYAA, CRYAB, CRYBA1, CRYBA2, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGA, CRYGB, CRYGC, CRYGD, CRYGS, CTDP1, CYP27A1, CYP51A1, EPG5, EPHA2, EYA1, FAM126A, FOXE3, FTL, FYCO1, GALK1, GALT, GCNT2, GJA3, GJA8, HSF4, LEMD2, LIM2, LSS, MAF, MIP, MIR184, NDP, NHS, OCRL, OPA3, P3H2, PAX6, PEX7, PITX3, PXDN, RAB3GAP1, RECQL4, SIL1, SIPA1L3, SLC16A12, TDRD7, VIM, VSX2, WRN

Kongenitale stationäre Nachtblindheit (CSNB) v1 (15 Gene)

CABP4, CACNA1F, GNAT1, GNB3, GPR179, GRK1, GRM6, LRIT3, NYX, PDE6B, RHO, RIMS2, SAG, SLC24A1, TRPM1

Lebersche kongenitale Amaurose (LCA) v2 (27 Gene)

AIPL1, BCOR, CABP4, CCT2, CEP290, CLUAP1, CRB1, CRX, DTHD1, GDF6, GUCY2D, IFT140, IMPDH1, IQCB1, KCNJ13, LCA5, LRAT, NMNAT1, OTX2, PRPH2, RD3, RDH12, RPE65, RPGRIP1, SLC6A6, SPATA7, TULP1

Makuladegeneration, altersbedingt (AMD) v1 (10 Gene)

ARMS2, C2, C3, CFB, CFH, ERCC6, FBLN5, HTRA1, TLR3, TLR4

Makuladystrophie, umfassendes Panel v1 (49 Gene)

ABCA4, ACBD5, ADAM9, AIPL1, ATXN7, BEST1, C1QTNF5, C8orf37, CABP4, CACNA1F, CACNA2D4, CDH3, CDHR1, CEP78, CERKL, CFAP410, CNNM4, CRX, CTNNA1, DRAM2, EFEMP1, ELOVL4, GUCA1A, GUCA1B, GUCY2D, HMCN1, IFT81, IMPG1, IMPG2, KCNV2, MFSD8, OPN1LW, OPN1MW, OTX2, PCYT1A, PDE6C, PITPNM3, POC1B, PRDM13, PROM1, PRPH2, RAB28, RAX2, RDH5, RIMS1, RPGRIP1, SEMA4A, TTLL5, UNC119

Makuladystrophie, autosomal rezessiv v1 (29 Gene)

ABCA4, ACBD5, ADAM9, BEST1, C8orf37, CABP4, CACNA1F, CACNA2D4, CDH3, CDHR1, CEP78, CERKL, CFAP410, CNNM4, DRAM2, IFT81, IMPG1, KCNV2, MFSD8, PCYT1A, PDE6C, POC1B, PROM1, PRPH2, RAB28, RAX2, RDH5, RPGRIP1, TTLL5

Makuladystrophie, autosomal dominant v1 (22 Gene)

AIPL1, ATXN7, BEST1, C1QTNF5, CRX, CTNNA1, EFEMP1, ELOVL4, GUCA1A, GUCA1B, GUCY2D, HMCN1, IMPG1, IMPG2, OTX2, PITPNM3, PRDM13, PRPH2, RAX2, RIMS1, SEMA4A, UNC119

Okulärer Albinismus v1 (9 Gene)

GPR143, LRMDA, LYST, MC1R, OCA2, SLC24A5, SLC45A2, TYR, TYRP1

Optikusatrophie / Ophthalmoparese v2 (56 Gene)

ACO2, AFG3L2, AUH, CISD2, CLPB, DGUOK, DNA2, DNAJC19, DNM1L, EARS2, ELOVL1, FDXR, FH, GYG2, ISCA2, MFF, MFN2, MGME1, MIEF1, MMP19, MTFMT, MTO1, MTPAP, MTRFR, NARS2, NBAS, NDUFAF3, NDUFS1, NR2F1, OPA1, OPA3, PAX2, PDHX, PDSS1, POLG, POLG2, PRPS1, RNASEH1, RRM2B, RTN4IP1, SLC19A2, SLC19A3, SLC25A4, SLC25A46, SNX10, SPG7, SUCLA2, TACO1, TIMM8A, TK2, TMEM126A, TSFM, TWNK, TYMP, VARS2, WFS1

Optikusneuropathie ohne mtDNA v2

ACO2, AFG3L2, ANTXR1, ATOH7, ATP1A3, CISD2, DNAJC30, DNM1L, FA2H, FDXR, HMBS, MFF, MFN2, MIEF1, MTPAP, MTRFR, NBAS, NDUFAF5, NDUFS2, NR2F1, OPA1, OPA3, PAX2, PRPS1, RTN4IP1, SLC25A46, SPG7, SSBP1, TBCE, TIMM8A, TMEM126A, UCHL1, WFS1, YME1L1

Peters-Anomalie v1 (7 Gene)

B3GLCT, CYP1B1, FOXC1, FOXE3, PAX6, PITX2, PITX3

Retinadystrophien v3 (292 Gene)

ABCA4, ABCC6, ABHD12, ACBD5, ACO2, ADAM9, ADAMTS18, ADGRA3, ADGRV1, ADIPOR1, AFG3L2, AGBL5, AHI1, AHR, AIPL1, AIRE, ALMS1, ARHGEF18, ARL2BP, ARL3, ARL6, ARMS2, ARSG, ASRGL1, ATF6, ATOH7, ATXN7, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCOR, BEST1, C1QTNF5, C2, C3, C8orf37, CA4, CABP4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CCDC28B, CCT2, CDH23, CDH3, CDHR1, CEP164, CEP19, CEP250, CEP290, CEP78, CERKL, CFAP410, CFB, CFH, CHM, CIB2, CLCC1, CLN3, CLRN1, CLUAP1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL11A2, COL18A1, COL2A1, COL9A1, COL9A2, COL9A3, CRB1, CRX, CSPP1, CTC1, CTNNA1, CTNNB1, CYP4V2, DHDDS, DHX38, DMD, DRAM2, DTHD1, EFEMP1, ELOVL1, ELOVL4, EMC1, ERCC6, ESPN, EXOSC2, EYS, FAM161A, FBLN5, FLVCR1, FRMD5, FSCN2, FZD4, GDF6, GNAT1, GNAT2, GNB3, GNPTG, GPR179, GRK1, GRM6, GUCA1A, GUCA1B, GUCY2D, HARS1, HGSNAT, HK1, HMCN1, HMX1, HTRA1, IDH3B, IFT140, IFT172, IFT27, IFT81, IMPDH1, IMPG1, IMPG2, INPP5E, INVS, IQCB1, ITM2B, JAG1, KCNJ13, KCNV2, KIAA1549, KIF11, KIZ, KLHL7, LAMA1, LCA5, LRAT, LRIT3, LRP5, LZTFL1, MAK, MAPKAPK3, MERTK, MFN2, MFRP, MFSD8, MIR204, MKKS, MKS1, MMP19, MTRFR, MTTP, MVK, MYO7A, NBAS, NDP, NEK2, NEUROD1, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NR2F1, NRL, NYX, OAT, OFD1, OPA1, OPA3, OPN1LW, OPN1MW, OPN1SW, OTX2, PANK2, PAX2, PAX6, PCARE, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6G, PDE6H, PDZD7, PEX1, PEX2, PEX7, PGK1, PHYH, PITPNM3, PLA2G5, PLK4, PNPLA6, POC1B, POC5, POMGNT1, PRCD, PRDM13, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, PYGM, RAB28, RAX2, RB1, RBP3, RBP4, RCBTB1, RD3, RDH11, RDH12, RDH5, REEP6, RGR, RGS9, RGS9BP, RHO, RIMS1, RIMS2, RLBP1, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, RPGRIP1, RPGRIP1L, RS1, RTN4IP1, SAG, SAMD11, SCAPER, SDCCAG8, SEMA4A, SLC24A1, SLC25A46, SLC6A6, SLC7A14, SNRNP200, SPATA7, SPP2, TEAD1, TIMM8A, TIMP3, TLR3, TLR4, TMEM126A, TMEM216, TMEM237, TOPORS, TREX1, TRIM32, TRNT1, TRPM1, TSPAN12, TTC8, TTLL5, TTPA, TUB, TUBGCP4, TUBGCP6, TULP1, UNC119, USH1C, USH1G, USH2A, VCAN, WASF3, WDPCP, WDR19, WFS1, WHRN, ZNF408, ZNF423, ZNF513

Retinitis pigmentosa, umfassendes Panel v1 (101 Gene)

ABCA4, ADIPOR1, AGBL5, AHR, ARHGEF18, ARL2BP, ARL3, ARL6, BBS1, BBS2, BEST1, CA4, CC2D2A, CERKL, CFAP418, CLCC1, CLRN1, CNGA1, CNGB1, CRB1, CRX, CYP4V2, DHDDS, DHX38, EMC1, EXOSC2, EYS, FAM161A, FLVCR1, GNPTG, GPR125, GUCA1B, HGSNAT, HK1, IDH3B, IFT140, IFT172, IMPDH1, IMPG1, IMPG2, KIAA1549, KIZ, KLHL7, LRAT, MAK, MAPKAPK3, MERTK, MVK, NEK2, NEUROD1, NR2E3, NRL, PANK2, PCARE, PDE6A, PDE6B, PDE6G, PGK1, POC5, POMGNT1, PRCD, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, RBP3, RBP4, RDH11, RDH12, REEP6, RGR, RHO, RLBP1, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, SAG, SAMD11, SCAPER, SEMA4A, SLC7A14, SNRNP200, SPATA7, SPP2, TOPORS, TRNT1, TTC8, TTPA, TULP1, USH2A, WDR19, ZNF408, ZNF513

Retinitis pigmentosa, autosomal-rezessiv v1 (77 Gene)

ABCA4, ADGRA3, ADIPOR1, AGBL5, AHR, ARHGEF18, ARL2BP, ARL3, ARL6, BBS1, BBS2, BEST1, C8orf37, CC2D2A, CERKL, CLCC1, CLRN1, CNGA1, CNGB1, CRB1, CYP4V2, DHDDS, DHX38, EMC1, EXOSC2, EYS, FAM161A, FLVCR1, GNPTG, HGSNAT, IDH3B, IFT140, IFT172, IMPG2, KIAA1549, KIZ, LRAT, MAK, MERTK, MVK, NEK2, NEUROD1, NR2E3, NRL, PANK2, PCARE, PDE6A, PDE6B, PDE6G, POC5, POMGNT1, PRCD, PROM1, RBP3, RBP4, RDH11, REEP6, RGR, RHO, RLBP1, ROM1, RP1, RP1L1, RPE65, SAG, SAMD11, SCAPER, SLC7A14, SPATA7, TRNT1, TTC8, TTPA, TULP1, USH2A, WDR19, ZNF408, ZNF513

Retinitis pigmentosa, autosomal-dominant v1 (33 Gene)

ADIPOR1, ARL3, BEST1, CA4, CRB1, CRX, GUCA1B, HK1, IMPDH1, IMPG1, KLHL7, MAPKAPK3, NR2E3, NRL, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RDH12, RGR, RHO, ROM1, RP1, RP1L1, RP9, RPE65, SAG, SEMA4A, SNRNP200, SPP2, TOPORS

Retinitis pigmentosa inkl. LCA (RP_total_LCA) v2 (118 Gene)

ABCA4, ADGRA3, ADIPOR1, AGBL5, AHR, AIPL1, ARHGEF18, ARL2BP, ARL3, ARL6, BBS1, BBS2, BCOR, BEST1, C8orf37, CA4, CABP4, CC2D2A, CCT2, CEP290, CERKL, CLCC1, CLRN1, CLUAP1, CNGA1, CNGB1, CRB1, CRX, CYP4V2, DHDDS, DHX38, DTHD1, EMC1, EXOSC2, EYS, FAM161A, FLVCR1, GDF6, GNPTG, GUCA1B, GUCY2D, HGSNAT, HK1, IDH3B, IFT140, IFT172, IMPDH1, IMPG1, IMPG2, IQCB1, KCNJ13, KIAA1549, KIZ, KLHL7, LCA5, LRAT, MAK, MAPKAPK3, MERTK, MVK, NEK2, NEUROD1, NMNAT1, NR2E3, NRL, OTX2, PANK2, PCARE, PDE6A, PDE6B, PDE6G, PGK1, POC5, POMGNT1, PRCD, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, RBP3, RBP4, RD3, RDH11, RDH12, REEP6, RGR, RHO, RLBP1, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, RPGRIP1, SAG, SAMD11, SCAPER, SEMA4A, SLC6A6, SLC7A14, SNRNP200, SPATA7, SPP2, TOPORS, TRNT1, TTC8, TTPA, TULP1, USH2A, WDR19, ZNF408, ZNF513

Retinitis pigmentosa inklusive LCA und CNSB (RP_total_LCA_CSNB) v2 (129 Gene)

ABCA4, ADGRA3, ADIPOR1, AGBL5, AHR, AIPL1, ARHGEF18, ARL2BP, ARL3, ARL6, BBS1, BBS2, BCOR, BEST1, C8orf37, CA4, CABP4, CACNA1F, CC2D2A, CCT2, CEP290, CERKL, CLCC1, CLRN1, CLUAP1, CNGA1, CNGB1, CRB1, CRX, CYP4V2, DHDDS, DHX38, DTHD1, EMC1, EXOSC2, EYS, FAM161A, FLVCR1, GDF6, GNAT1, GNB3, GNPTG, GPR179, GRK1, GRM6, GUCA1B, GUCY2D, HGSNAT, HK1, IDH3B, IFT140, IFT172, IMPDH1, IMPG1, IMPG2, IQCB1, KCNJ13, KIAA1549, KIZ, KLHL7, LCA5, LRAT, LRIT3, MAK, MAPKAPK3, MERTK, MVK, NEK2, NEUROD1, NMNAT1, NR2E3, NRL, NYX, OTX2, PANK2, PCARE, PDE6A, PDE6B, PDE6G, PGK1, POC5, POMGNT1, PRCD, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, RBP3, RBP4, RD3, RDH11, RDH12, REEP6, RGR, RHO, RIMS2, RLBP1, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, RPGRIP1, SAG, SAMD11, SCAPER, SEMA4A, SLC24A1, SLC6A6, SLC7A14, SNRNP200, SPATA7, SPP2, TOPORS, TRNT1, TRPM1, TTC8, TTPA, TULP1, USH2A, WDR19, ZNF408, ZNF513

Usher-Syndrom v1 (18 Gene)

ABHD12, ADGRV1, ARSG, CDH23, CEP250, CEP78, CIB2, CLRN1, ESPN, EXOSC2, HARS1, MYO7A, PCDH15, PDZD7, USH1C, USH1G, USH2A, WHRN

Vitreoretinopathien inkl. FEVR v1 (24 Gene)

ATOH7, BEST1, CAPN5, COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, CRB1, CTC1, CTNNB1, FZD4, JAG1, KCNJ13, KIF11, LRP5, NDP, RCBTB1, TIMP3, TREX1, TSPAN