

## Augenerkrankungen

**Achromatopsie [GP001] (~10 kb)**

*ATF6, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H*

**Albinismus, okulärer [GP002] (~21 kb)**

1. *GPR143*; 2. *C10orf11, LYST, MC1R, OCA2, SLC45A2, TYR, TYRP1*

**umfassende Diagnostik [GP002XL] (69,5kb)**

*AP3B1, BLOC1S3, BLOC1S6, C10orf11, DTNBP1, EPG5, EDN3, EDNRB, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LYST, MC1R, MITFSNAI2, MLPH, MYO5A, OCA2, PAX3, RAB27A, SLC24A5, SLC45A2, SOX10, TYR, TYRP1*

**Albinismus, okulokutaner [GP168] (~10 kb)**

*MC1R, OCA2, SLC24A5, SLC45A2 (MATP), TYR, TYRP1*

**Axenfeld-Rieger-Syndrom [GP172]**

*ASPH, B3BLCT, COL4A1, COL4A2, CYP1B1, EYA1, FOXC1, FOXE3, HMX1, LTBP2, MIR184, MYOC, PAX6, PITX2, PXDN, SLC38A8*

**Hermansky-Pudlak-Syndrom [GP167] (~22 kb)**

*AP3B1, AP3D1, BLOC1S3, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6, PLDN*

**Bardet-Biedl-Syndrom [GP003] (~19 kb)**

*BBS1, BBS10, BBS12, BBS2, BBS4, BBS7, BBS9, MKKS, MKS1, TTC8*

**umfassende Diagnostik [GP003XL]**

*ALMS1, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CCDC28B, CEP290, LZTFL1, MKKS, MKS1, NPHP1, SDCCAG8, TMEM67, TRIM32, TTC21B, TTC8, WDPCP*

**Joubert-Syndrom [GP004] (~25 kb)**

*AHI1, CC2D2A, CEP290, NPHP, RPGRIP1L, TMEM67*

**umfassende Diagnostik [GP004XL]**

*AHI1, ARL13B, B9D1, C5orf42, CC2D2A, CEP290, CEP41, CSPP1, KIF7, MKS1, NPHP1, OFD1, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM237, TMEM67, TTC21B*

**Lebersche hereditäre Optikusneuropathie (LHON) [GP165]**

*MT-ATP6, MT-CO1, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-TE, MT-TL1, MT-TM, MT-TQ, MT-TT*

**Katarakt, kongenitale [GP005] (~23 kb)**

*BFSP1, BFSP2, CRYAA, CRYAB, CRYBA1, CRYBB1, CRYBB2, CRYBB3, CRYGC, CRYGD, CRYGS, EPHA2, FYCO1, GJA3, GJA8, HSF4, LIM2, MAF, MIP*

**Lebersche kongenitale Amaurose (LCA) [GP006] (~23 kb)**

*AIPL1, CEP290, CRX, GUCY2D, LCA5, RDH12, RPE65, RPGRIP1*

**umfassende Diagnostik [GP006XL]**

*ABCA4, AHI1, AIPL1, ALMS1, CABP4, CEP290, CRB1, CRX, GDF6, GUCY2D, IFT140, IMPDH1, IQCB1, KCNJ13, KCNV2, LCA5, LRAT, MERTK, MPDZ, NMNAT1, NPHP1, NXNL1, OTX2, PRPH2, RD3, RDH12, RDH5, RPE65, RPGRIP1, RPGRIP1L, SPATA7, TULP1*

**Anophthalmia/Microphthalmia/Coloboma (MAC-Spektrum) [GP007] (~25 kb)**

*BCOR, BMP4, CHD7, FOXE3, GDF6, OTX2, PAX6, RAX, SOX2, STRA6*

**umfassende Diagnostik [GP007XL]**

*BCOR, BMP4, CHD7, FOXE3, FREM1, GDF3, GDF6, HCCS, MFRP, MITF, NAA10, OTX2, PAX6, PRSS56, RAX, SHH, SIX6, SMOG1, SOX2, STRA6, VAX1, VSX2*

**Optikusatrophie [GP009] (~19 kb)**

1. *OPA1*, 2. *CISD2, MFN2, NDUFS1, NR2F1, OPA3, POLG, RTN4IP1, SPG7, TIMM8A, TMEM126A, WFS1*

**Peters Anomalie [GP171] (~6kb)**

*CYP1B1, FOXC1, PAX6, PITX2, PITX3*

**Refsum-Syndrom [GP010] (7,7kb)**

*PEX1, PEX2, PEX26, PEX7, PHYH*

**Retinitis pigmentosa [GP011]**

**AD/XL:** *IMPDH1, KLHL7, NR2E3, PRPF31, PRPF8, PRPH2, RHO, RP1, RP2, RPGR* (~ 25 kb)

**AR:** *EYS, USH2A* (~ 24 kb)

**umfassende Diagnostik [GP011XL]**

*ABCA4, AIPL1, ARL6, BEST1, C2orf71, C8orf37, CA4, CDHR1, CERKL, CLRN1, CNGA1, CNGB1, CRB1, CRX, DHDDS, EYS, FAM161A, FLVCR1, FSCN2, GUCA1B, IDH3B, IMPDH1, IMPG2, KLHL7, LRAT, MAK, MERTK, NR2E3, NRL, PDE6A, PDE6B, PDE6G, PRCD, PROM1, PRPF3, PRPF31, PRPF6, PRPF8, PRPH2, RBP3, RDH12, RGR, RHO, RLBP1, ROM1, RP1, RP1L, RP2, RP9, RPE65, RPGR, SAG, SEMA4A, SLC7A14, SNRNP200, SPATA7, TOPORS, TTC8, TULP1, USH2A, ZNF513*

**Stargardt / Makuladystrophie [GP008] (~24 kb)**

1. *ABCA4*, 2. *BEST1, CDH3, CNGB3, CRB1, ELOVL4, PROM1, PRPH2, RDH12, RP1L1, TIMP3*

**Stickler-Syndrom [GP013] (~22 kb)**

*COL11A1, COL2A1, COL9A2, COL9A3, COL11A2, COL9A1*

**USHER-Syndrom [GP091] (~24 kb)**

1. *USH2A*, 2. *MYO7A, USH1C*

**umfassende Diagnostik [GP091XL]**

*CDH23, CIB2, CLRN1, DFNB31, HARS, MYO7A, PCDH15, PDZD7, USH1C, USH1G, USH2A*

**Zapfen/Zapfen-Stäbchen-Dystrophie (ZD/ZSD) [GP014] (~25 kb)**

*ABCA4, ADAM9, CERKL, CNGA3, KCNV2, PDE6C, RPGRIP1, RDH5*

**umfassende Diagnostik [GP014XL]**

*ABCA4, ACBD5, ADAM9, AIPL1, ALMS1, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8orf37, CABP4, CACNA1F, CACNA2D4, CCDC28B, CDHR1, CEP290, CERKL, CNGA3, CNGB3, CNNM4, CRB1, CRX, CYP4V2, GNAT2, GUCA1A, GUCY2D, KCNV2, LZTFL1, MKKS, MKS1, NPHP1, PDE6C, PDE6H, PIPNMM3, PROM1, PRPH2, RAB28, RAX2, RDH5, RGS9, RGS9BP, RIMS1, RPGR, RPGRIP1, SDCCAG8, SEMA4, TMEM67, TRIM32, TTC21B, TTC8, WDPCP*