INSTITUTE OF IMMUNOLOGY AND GENETICS

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Requesting physician:	**Patient information:**

Request Form Eye diseases / Hearing impairment

Cause of examination: ☐ diagnostic ☐ predictive for familial mutation _____

Clinical information:

Family history:

(Check all that apply)

☐ Patient clinically affected

☐ No previous molecular genetic examinations existent

☐ The following previous molecular genetic examinations have been done:

_____

Date / Time of sampling:	**Signature:**

Specimen requirements and logistics:

- **Patient Consent Form for Genetic Testing according to the German Law and Request Form**
- **Billing information (Insurance-, Institutional- or Selfpay-Billing)**
- **5 ml EDTA blood collection tube labeled with patient name and date of birth**

Patient name: _____ **DOB:** _____

Eye diseases

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

☐ **External ophthalmoparesis**
DGUOK, DNA2, MGME1, OPA1, POLG, POLG2, RNASEH1, RRM2B, SLC25A4, TK2, TWNK, TYMP

☐ **Flecked retina disease**
ABCA4, CHM, CYP4V2, EFEMP1, ELOVL4, KCNJ13, OAT, PLA2G5, PROM1, PRPH2, RDH5, RHO, RLBP1, RPE65, RS1, VPS13B

☐ **Glaucoma**
ADAMTS10, ADAMTS17, ASB10, BEST1, COL18A1, CYP1B1, FOXC1, FOXE3, GPATCH3, LOXL1, LTBP2, MYOC, NTF4, OPTN, PAX6, PITX2, SBF2, TEK, WDR36

☐ **Corneal dystrophy**
AGBL1, CHST6, COL17A1, CYP4V2, DCN, GSN, KRT12, KRT3, LOXHD1, MIR184, OVOL2, PAX6, PIKFYVE, PRDM5, SLC4A11, TACSTD2, TCF4, TUBA3D, UBIAD1, VSX1, ZEB1, ZNF469

☐ **Cataract**
ABCN6, ABHD12, ADAMTSL4, AGK, BCOR, BFSP1, BFSP2, CHMP4B, CLPB, COL11A1, COL2A1, COL4A1, COL4A2, CRYAA, CRYAB, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGB, CRYGC, CRYGD, CRYGS, CTDP1, CYP27A1, CYP51A1, EPG5, EPHA2, ERCC1, ERCC2, ERCC5, ERCC6, EYA1, FAM126A, FOXC1, FOXE3, FTL, FYCO1, GALK1, GALT, GCNT2, GEMIN4, GJA3, GJA8, GLA, HMX1, HSF4, JAM3, LEMD2, LIM2, LONP1, LSS, LTBP2, MAF, MIP, MIR184, MYH9, NDP, NF2, NHS, OCRL, OPA3, P3H2, PAX6, PEX7, PITX2, PITX3, PXDN, RAB18, RAB3GAP1, RAB3GAP2, RECQL4, SC5D, SIL1, SIPA1L3, SIX6, SLC16A12, SLC33A1, TBC1D20, TDRD7, TMEM114, VIM, VSX2, WFS1, WRN

☐ **Leber congenital amaurosis**
AIPL1, ALMS1, CABP4, CEP290, CLUAP1, CNGA3, CRB1, CRX, DTHD1, GUCY2D, IDH3A, IFT140, IMPDH1, IQCB1, KCNJ13, LCA5, LRAT, MERTK, NMNAT1, OTX2, PRPH2, RD3, RDH12, RDH5, ROM1, RPE65, RPGRIP1, SNRNP200, SPATA7, TULP1, USP45

☐ **Retinal dystrophy**
*(Gene panel with 161 Genes *)*

☐ **Ocular malformation**
ABCB6, ACTB, ACTG1, ALDH1A3, ATOH7, BCOR, BMP4, BMP7, C12orf57, CHD7, CRYBA4, CYP1B1, ERCC1, ERCC2, ERCC5, ERCC6, FOXE3, FOXL2, FRAS1, FREM1, FZD4, GDF3, GDF6, GJA1, GRIP1, HCCS, HESX1, HMX1, MAB21L2, MFRP, NDP, NDUFB11, OCRL, OTX2, PAX2, PAX6, PIGL, PRSS56, PXDN, RAB18, RAB3GAP1, RAB3GAP2, RARB, RAX, RBP4, SALL2, SHH, SIX3, SIX6, SMOC1, SOX1, SOX2, STRA6, TBC1D20, TENM3, TFAP2A, TMEM98, VAX1, VPS13B, VSX2, ZIC2

☐ **Ocular und oculocutaneous albinism**
C10orf11, FRMD7, GPR143, LRMDA, LYST, MC1R, OCA2, SLC24A5, SLC38A8, SLC45A2, TYR, TYRP1

☐ **Optic atrophy**
ACO2, AFG3L2, ANTXR1, ATP1A3, C12orf65, CISD2, DNM1L, FDXR, MCAT, MFN2, OPA1, OPA3, RTN4IP1, SLC25A46, SPG7, SSBP1, TIMM8A, TMEM126A, WFS1, YME1L1

☐ **Retinitis pigmentosa**
ABCA4, ADGRA3, AGBL5, AHI1, ARGEF18, ARL2BP, ARL3, ARL6, BBS1, BBS2, BEST1, C2orf71, C8orf37, CA4, CACNA1F, CDHR1, CEP290, CERKL, CLN3, CLRN1, CNGA1, CNGB1, CRB1, CRX, CQC27, CYP4V2, DHDDS, DHX38, EMC1, EYS, FAM161A, FLVCR1, FSCN2, GNAT1, GNPTG, GUCA1B, GUCY2D, HGSNAT, HK1, IDH3B, IFT140, IFT172, IMPDH1, IMPG2, KIAA1549, KIF11, KIZ, KLHL7, LRAT, MAK, MERTK, MFRP, NEK2, NMNAT1, NR2E3, NRL, PCARE, PDE6A, PDE6B, PDE6G, POMGNT1, POPORS, PRCD, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RBP3, RBP4, RCBTB1, RDH11, RDH12, REEP6, RGR, RHO, RLBP1, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, RPGRIP1, SAG, SEMA4A, SLC7A14, SNRNP200, SPATA7, TOPORS, TRNT1, TTC8, TUB, TULP1, USH2A, ZNF408, ZNF513

☐ **Septo-optic dysplasia**
FGFR1, HESX1, OTX2, PROKR2, SOX2, SOX3

☐ **Syndromal albinism**
AP3B1, AP3D1, BLOC1S3, BLOC1S6, DTNBP1, EDN3, EDNRB, EPG5, HPS1, HPS3, HPS4, HPS5, HPS6, KIT, LYST, MITF, MLPH, MYO5A, PAX3, RAB27A, SNAI2, SOX10, TYP

☐ **Usher syndrome**
ABHD12, ADGRV1, ARSG, CDH23, CEP250, CEP78, CIB2, CLRN1, ESPN, HARS1, MYO7A, PCDH15, PDZD7, PEX1, PEX6, USH1C, USH1G, USH2A, WHRN

☐ **Walker-Warburg-syndrome**
B3GALNT2, B4GAT1, DAG1, FKRP, FKTN, GMPPB, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1

☐ **Cone dystrophy**
ABCA4, ADAM9, AIPL1, ALMS1, ATF6, BEST1, C12orf2, C2orf71, C8orf37, CABP4, CACNA1F, CACNA2D4, CDHR1, CEP290, CEP78, CERKL, CFAP140, CNGA3, CNGB3, CNNM4, CRB1, CRX, CYP4V2, DRAM2, EYS, GNAT2, GUCA1A, GUCY2D, KCNV2, NMNAT1, NPHP4, PAX6, PCARE, PCYT1A, PDE6C, PDE6H, PITPNM3, POC1B, PROM1, PRPH2, RAB28, RAX2, RBP4, RDH12, RDH5, RGS9, RGS9BP, RIMS1, RPGR, RPGRIP1, SEMA4A, TTLL5, TULP1, UNC119, WDR19

* A detailed gene list can be provided on request.

Patient name: _____ **DOB:** _____

Hearing impairment

☐ **Isolated deafness**
ACTG1, ADCY1, BDP1, BSND, CABP2, CCDC50, CD164, CDC14A, CDH23, CEACAM16, CIB2, CLDN14, CLIC5, COCH, COL11A2, COL4A6, CRYM, DCDC2, DIABLO, DIAPH1, DIAPH3, EPS8, EPS8L2, ESPN, ESRRB, EYA4, GIPC3, GJB2, GJB3, GJB6, GPRASP2, GPSM2, GRHL2, GRXCR1, GRXCR2, GSDME, HGF, ILDR1, KARS, KCNQ4, KITLG, LHFPL5, LOXHD1, LRTOMT, MARVELD2, MIR96, MSRB3, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NARS2, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PCDH15, PDZD7, PJVK, PNPT1, POU3F4, PRPS1, PTPRQ, RDX, RIPOR2, S1PR2, SERPINB6, SLC17A8, SLC26A4, SLC26A5, SLITRK6, SMPX, STRC, SYNE4, TBC1D24, TECTA, TJP2, TMC1, TMC2, TMEM132E, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TSPEAR, USH1C, WBP2, WFS1

☐ **Syndromal deafness**
ABHD12, ADGRV1, AIFM1, ALMS1, ANKH, ATP6V1B1, BCAP31, BCS1L, BSND, C10orf2, CACNA1D, CATSPER2, CD151, CDH23, CDKN1C, CHD7, CHSY1, CIB2, CISD2, CLPP, CLRN1, COL11A1, COL11A2, AOL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, DFNB31, DIAPH3, DNMT1, EDN3, EDNRB, ERAL1, EXOSC2, EYA1, FGF3, FOXI1, GATA3, GPR98, GPSM2, HARS, HARS2, HOXB1, HSD17B4, KCNE1, KCNJ10, KCNQ1, KITLG, LARS2, MANBA, MITF, MYH9, MYO7A, NDP, NLRP3, PAX3, PCDH15, PDZD7, PEX1, PEX6, POLR1D, SALL1, SEMA3E, SIX1, SIX5, SLC19A2, SLC26A4, SLITRK6, SNAI2, SOX10, SPATA5, TCOF1, TFAP2A, TIMM8A, TWNK, TYP, USH1C, USH1G, USH2A, WFS1, WHRN