Praxis für Humangenetik
Dr. med. Ines Schönbuchner
Fachärztin für Humangenetik

in Kooperation mit

Institut für Humangenetik, UR
Prof. Dr. rer. nat. Bernhard Weber
Fachhumangenetiker GfH

Genpanel „hereditäre Hörstörungen“ (OtoNet v.01)

ABHD12, ACTG1, ADCY1, ADGRV1, AIFM1, ALMS1, ANKH, ATP6V1B1, BCAP31, BCS1L, BDP1, BSND, CABP2, CACNA1D, CCDC50, CD151, CD164, CDC14A, CDH23, CEACAM16, CEP250, CHD7, CHSY1, CIB2, CLDN14, CLDN9, CLIC5, CLPP, CLRN1, COCH, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, CRYM, DCDC2, DIABLO, DIAPH1, DIAPH3, DMXL2, DNMT1, DSPP, EDN3, EDNRB, ELMOD3, EPS8, EPS8L2, ERAL1, ESPN, ESRP1, ESRRB, EXOSC2, EYA1, EYA4, FGF3, FOXI1, GAB1, GATA3, GIPC3, GJB2, GJB3, GJB6, GPRASP2, GPSM2, GRAP, GRHL2, GRXCR1, GRXCR2, GSDME, HARS1, HARS2, HGF, HOMER2, HSD17B4, ILDR1, KARS1, KCNE1, KCNJ10, KCNQ1, KCNQ4, KITLG, LARS2, LHFPL5, LMX1A, LOXHD1, LRTOMT, MANBA, MARVELD2, MCM2, MET, MITF, MPZL2, MSRB3, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NARS2, NDP, NLRP3, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX3, PCDH15, PDE1C, PDZD7, PEX1, PEX6, PHYH, PJVK, PLS1, PNPT1, POLR1C, POLR1D, POU3F4, POU4F3, PPIP5K2, PRPS1, PTPRQ, RDX, REST, RIPOR2, ROR1, S1PR2, SALL1, SERPINB6, SIX1, SIX5, SLC17A8, SLC19A2, SLC26A4, SLC26A5, SLC44A4, SLITRK6, SMPX, SNAI2, SOX10, SPATA5, SPNS2, STRC, SYNE4, TBC1D24, TCOF1, TECTA, TIMM8A, TMC1, TMEM132E, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TSPEAR, TWNK, USH1C, USH1G, USH2A, WBP2, WFS1, WHRN

- **Sub-Genpanel Nicht-syndromale Hörstörungen**

ACTG1, ADCY1, ALMS1, ANKH, BCAP31, BCS1L, BDP1, CABP2, CCDC50, CD151, CD164, CDC14A, CDH23, CEACAM16, CHD7, CHSY1, CIB2, CLDN14, CLDN9, CLIC5, COCH, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, CRYM, DCDC2, DIABLO, DIAPH1, DIAPH3, DMXL2, DNMT1, DSPP, ELMOD3, EPS8, EPS8L2, ESPN, ESRP1, ESRRB, EXOSC2, EYA4, FGF3, FOXI1, GAB1, GATA3, GIPC3, GJB2, GJB3, GJB6, GPRASP2, GRAP, GRHL2, GRXCR1, GRXCR2, GSDME, HGF, HOMER2, ILDR1, KARS1, KCNQ4, KITLG, LHFPL5, LMX1A, LOXHD1, LRTOMT, MANBA, MARVELD2, MCM2, MET, MPZL2, MSRB3, MYH14, MYO15A, MYO3A, MYO6, MYO7A, NARS2, NDP, NLRP3, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PCDH15, PDE1C, PDZD7, PEX1, PEX6, PHYH, PJVK, PLS1, PNPT1, POLR1C, POLR1D, POU3F4, POU4F3, PPIP5K2, PRPS1, PTPRQ, RDX, REST, RIPOR2, ROR1, S1PR2, SALL1, SERPINB6, SLC17A8, SLC19A2, SLC26A5, SLC44A4, SLITRK6, SMPX, SPATA5, SPNS2, STRC, SYNE4, TBC1D24, TCOF1, TECTA, TIMM8A, TMC1, TMEM132E, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TSPEAR, USH1C, WBP2, WHRN

- **Sub-Genpanel Syndromale Hörstörungen**

ABHD12, ADGRV1, AIFM1, ALMS1, ANKH, ATP6V1B1, BCAP31, BCS1L, BSND, CACNA1D, CD151, CDH23, CEP250, CHD7, CHSY1, CIB2, CLPP, CLRN1, COL2A1, COL4A3, COL4A4, COL4A5, COL9A1, COL9A2, DNMT1, DSPP, EDN3, EDNRB, ERAL1, EXOSC2, EYA1, FGF3, GATA3, GPRASP2, GPSM2, HARS1, HARS2, HSD17B4, KCNE1, KCNJ10, KCNQ1, LARS2, MANBA, MITF, MYH9, MYO7A, NDP, PAX3, PCDH15, PDZD7, PEX1, PEX6, PHYH, POLR1C, POLR1D, SALL1, SIX1, SIX5, SLC19A2, SLC26A4, SLC44A4, SNAI2, SOX10, SPATA5, TCOF1, TIMM8A, TWNK, USH1C, USH1G, USH2A, WFS1, WHRN

Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Sequence capture (Agilent), Sequencing-by synthesis (Illumina), ggf. Bestätigung mit PCR, Sanger-Sequenzierung	Target Enrichment and Amplification (Twist), Sequencing-by synthesis (Illumina), ggf. Bestätigung mit PCR, Sanger-Sequenzierung	6.2.21 AM Twist Custom Ansatz, 6.1.14 AG Anleitung Illumina MiSeq, 6.1.15 AG Anleitung Illumina NextSeq, 6.2.3 AM PCR, 6.2.6 AM Durchführung einer genetischen Untersuchung mittels Sanger-Sequenzierung	Nein	Ja

Alle Analyte sind flexibel akkreditiert und erfüllen die Anforderungen der EA (European Accredidation) bzw. DAkkS (Deutsche Akkreditierungsstelle) an die Akkreditierung flexibler Geltungsbereiche.