

Hallásvesztés (Hearing loss) génpanel v:2023.10.12.

Panel tartalma 558 gén:

ABCC1, ABHD12, ABHD5, ABR, ACAN, ACOX1, ACTB, ACTG1, ADCY1, ADGRV1, AIFM1, ALDH1A2, ALMS1, AMMECR1, ANKH, ANLN, AP1S1, AP3D1, APAF1, APOPT1, AQP4, ARSB, ARSG, ASAH1, ATF2, ATOH1, ATP1A2, ATP1A3, ATP2B2, ATP6V0A4, ATP6V1B1, ATP6V1B2, ATP8B1, AXIN1, BARHL1, BBS1, BBS4, BCAP31, BCR, BCS1L, BDNF, BDP1, BLOC1S5, BLOC1S6, BMP4, BMP5, BSN, BSND, BTD, C10ORF2, CABP2, CACNA1D, CACNB2, CACNG2, CASP3, CATSPER2, CCDC50, CD151, CD164, CDC14A, CDC42, CDC6, CDH23, CDK9, CDKN1B, CDKN1C, CDKN2D, CDT1, CEACAM16, CELSR1, CEP250, CEP78, CHD7, CHRNA9, CHSY1, CIB2, CISD2, CKB, CLDN11, CLDN14, CLDN9, CLIC5, CLNS1A, CLPP, CLRN1, CLRN2, CNRIP1, COCH, COG4, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, CPLX1, CRLS1, CRYL1, CRYM, DACT1, DCAF17, DCDC2, DDB2, DDR1, DFNA5, DFNB31, DFNB59, DHODH, DIABLO, DIAPH1, DIAPH3, DIO2, DIO3, DLX2, DLX5, DMD, DMXL2, DNMT1, DNRB, DSPP, DVL1, DVL2, DVL3, DWS, EDN3, EDNRA, EDNRB, EFTUD2, EIF3F, EIF4A3, ELMOD3, EPHA10, EPHB1, EPHB2, EPHB3, EPS8, EPS8L2, ERAL1, ERBB4, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ESPN, ESR2, ESRP1, ESRRB, EVC, EXOSC2, EYA1, EYA4, FABP4, FAM136A, FAM65B, FAS, FBXO2, FDXR, FGF10, FGF3, FGFR1, FGFR2, FGFR3, FIGN, FITM2, FKBP14, FOXC1, FOXF2, FOXG1, FOXI1, FOXI3, FRAS1, FREM2, FZD3, FZD6, GAB1, GATA3, GBX2, GDF6, GFER, GFI1, GGPS1, GIPC3, GJA1, GJA1P1, GJB1, GJB2, GJB3, GJB4, GJB5, GJB6, GLI3, GNAI3, GPR98, GPRASP2, GPSM2, GPX1, GRAP, GREB1L, GRHL2, GRID1, GRIP1, GRXCR1, GRXCR2, GSC, GSDME, GSTM1, GSTP1, GSTT1, GUSB, HAAO, HAL, HARS, HARS1, HARS2, HES1, HES5, HGF, HMX1, HMX2, HMX3, HOMER2, HOXA1, HOXA2, HOXB1, HSD17B4, HSPA9, HTRA2, IARS2, IFT88, IGF1, ILDR1, ITGA8, JAG1, JAG2, KARS, KARS1, KCNE1, KCNJ10, KCNJ16, KCNMA1, KCNQ1, KCNQ4, KDM3B, KDM6A, KIT, KITLG, KMT2D, LAMA2, LARGE1, LARS2, LEMD3, LFNG, LHFPL5, LHX3, LMO4, LMX1A, LOXHD1, LRIG3, LRP2, LRTOMT, MAFB, MAN2B1, MANBA, MAP1A, MARVELD2, MASP1, MCM2, MCOLN3, MET, MGP, MIR182, MIR183, MIR96, MITF, MKKS, MN1, MORC2, MOS, MPV17, MPZL2, MSRB3, MSX2, MTAP, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MYH14, MYH9, MYO15A, MYO1A, MYO1C, MYO1F, MYO3A, MYO6, MYO7A, NARS2, NAV2, NCOA3, NDP, NDRG1, NEFL, NEU1, NEUROD1, NEUROG1, NF1, NF2, NKX3-2, NLRP3, NOG, NOTCH1, NOX3, NOXO1, NR2F1, NR4A3, NTF3, NTN1, NTRK2, NTRK3, OC90, OFD1, OGDHL, OPA1, ORC1, ORC4, ORC6, OSBPL2, OTOA, OTOF, OTOG, OTOGL, OTOP1, OTOR, OTX1, OTX2, OXR1, P2RX2, PAX1, PAX2, PAX3, PBX1, PCDH15, PCGF2, PDE1C, PDSS1, PDZD7, PET100, PEX1, PEX11B, PEX12, PEX13, PEX14, PEX2, PEX26, PEX5, PEX6, PEX7, PHEX, PHYH, PIK3C2A, PISD, PITX2, PJVK, PLCB4, PLEK, PLS1, PMP22, PNOC, PNPT1, POLD1, POLH, POLR1A, POLR1C, POLR1D, PORCN,

POU1F1, POU3F4, POU4F3, PPIP5K2, PPP3R1, PRKCB, PROP1, PRPS1, PRRX1, PRRX2, PTK7, PTPRQ, RAI1, RARA, RARB, RARG, RASA1, RDX, REST, RIPOR2, RMND1, RNF220, ROR1, RPGR, RPS28, RPS6KA3, S1PR2, SALL1, SALL4, SARS, SCARB2, SCD5, SCRIB, SDHD, SEMA3E, SERAC1, SERPINB6, SF3B4, SGPL1, SH3TC2, SIX1, SIX2, SIX5, SLC12A1, SLC12A2, SLC12A6, SLC12A7, SLC17A8, SLC19A2, SLC1A3, SLC22A4, SLC26A4, SLC26A5, SLC29A3, SLC30A4, SLC33A1, SLC44A4, SLC4A11, SLC4A7, SLC52A2, SLC52A3, SLC9A1, SLITRK6, SMAD4, SMPX, SMS, SNAI2, SOBP, SOD1, SOX10, SOX2, SOX9, SPATA5, SPATA5L1, SPATC1L, SPINK5, SPNS2, SPRY2, SPTBN4, ST3GAL5, STAG2, STRC, STXBP3, SUCLA2, SUCLG1, SYNE4, SYNJ2, SYT2, TBC1D24, TBL1X, TBX1, TBX10, TCF21, TCOF1, TECTA, TFAP2A, TGFA, TGFB2, THOC1, THRA, THRB, TIMM8A, TJP2, TMC1, TMEM126A, TMEM132E, TMIE, TMPRSS3, TMPRSS5, TMTC2, TNC, TNFRSF11B, TOP2B, TPRN, TRIOBP, TRMU, TRPV4, TRRAP, TSHR, TSHZ1, TSPEAR, TUB, TUBB4B, TWNK, TWSG1, TYR, TYRP1, UBR1, UCN, USH1C, USH1G, USH2A, USP48, VANG2, VCAN, WBP2, WFS1, WHRN, WIF1, XPA, XPC, XYLT2, YAP1, YARS, ZPR1

Forrás:

Monogenic hearing loss (Version 4.9)

<https://panelapp.genomicsengland.co.uk/panels/126/> (Megtekintve: 2023.05.16.)

Deafness and congenital structural abnormalities (Version 1.22)

<https://panelapp.genomicsengland.co.uk/panels/251/> (Megtekintve: 2023.05.16.)

Comprehensive Hearing Loss and Deafness Panel

<https://blueprintgenetics.com/tests/panels/ear-nose-throat/comprehensive-hearing-loss-and-deafness-panel/> (Megtekintve: 2023.05.16.)

Hearing Loss

<https://cegat.com/diagnostics/rare-diseases/hearing-loss/> (Megtekintve: 2023.07.19.)

Hearing Loss Panel

<https://www.genedx.com/tests/detail/hearing-loss-panel-925> (Megtekintve: 2023.05.16.)

Comprehensive Hearing Loss NGS Panel

<https://www.fulgentgenetics.com/comprehensive-hearing-loss> (Megtekintve: 2023.05.16.)

CentoHear

<https://www.centogene.com/diagnostics/our-tests/ngs-panels/ear-nose-throat> (Megtekintve: 2023.07.19.)