

Szemészeti (Ophtalmology) génpanel v:2023.10.12.

Panel tartalma 817 gén:

ABCA3, ABCA4, ABCB6, ABHD12, ACBD5, ACO2, ACTB, ACTG1, ACVR1, ADAM9, ADAMTS10, ADAMTS17, ADAMTS18, ADAMTSL4, ADGRA3, ADGRV1, ADIPOR1, AFG3L2, AGBL1, AGBL5, AGK, AGPS, AHI1, AHR, AIPL1, AIRE, AKR1E2, ALDH18A1, ALDH1A3, ALDH3A2, ALG3, ALMS1, ALPK1, AMACR, AMN, ANAPC1, AP3B1, AP3B2, AP4B1, APOA1, APTX, ARHGEF18, ARL13B, ARL2BP, ARL3, ARL6, ARMC9, ARMS2, ARR3, ARSG, ASB10, ASRGL1, ATAD3A, ATF6, ATG7, ATOH7, ATP13A2, ATXN7, AUH, B3GALNT2, B3GLCT, B4GAT1, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCOR, BEST1, BFSP1, BFSP2, BLOC1S1, BLOC1S3, BLOC1S6, BMP4, BMP7, BMPR1A, BUB1B, C12orf57, C12orf65, C16orf62, C19orf12, C1QTNF5, C1QTNF9B, C2, C21orf2, C2CD3, C2orf71, C3, C5orf42, C8orf37, CA4, CABP4, CACNA1F, CACNA2D4, CANT1, CAPN15, CAPN5, CBS, CC2D2A, CCDC111, CCDC28B, CCT2, CCZ1B, CDH10, CDH12, CDH15, CDH23, CDH3, CDH6, CDHR1, CDK9, CDKN2A, CENPF, CEP104, CEP120, CEP164, CEP19, CEP250, CEP290, CEP41, CEP78, CERKL, CFAP418, CFB, CFH, CHAD, CHD7, CHM, CHMP4B, CHRDL1, CHST6, CIB2, CISD2, CLCC1, CLCN7, CLDN19, CLN3, CLN5, CLN6, CLN8, CLPB, CLRN1, CLUAP1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COG4, COL11A1, COL11A2, COL18A1, COL1A1, COL2A1, COL4A1, COL6A3, COL8A2, COL9A1, COL9A2, COL9A3, COPB1, COQ2, COQ4, COQ5, COX7B, CPAMD8, CPLANE1, CRB1, CREBBP, CROCC, CRX, CRYAA, CRYAB, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGA, CRYGB, CRYGC, CRYGD, CRYGFP, CRYGS, CTC1, CTDP1, CTNNA1, CTNNB1, CTSD, CTSF, CUBN, CWC27, CYP1B1, CYP27A1, CYP2R1, CYP4V2, CYP51A1, CSPP1, DAG1, DCN, DDB1, DDB2, DDX58, DDX59, DGKD, DGUOK, DHCR7, DHDDS, DHX38, DKC1, DMD, DNA2, DNAJC19, DNAJC30, DNM1L, DNMBP, DRAM2, DSPG3, DTHD1, DTNBP1, DYNC2H1, DYRK1A, EDN3, EDNRB, EED, EFEMP1, EIF2B2, ELOVL1, ELOVL4, EMC1, ENPP1, EPHA2, EPRS, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, ESCO2, ESPN, EVR3, EXOC8, EXOSC2, EYA1, EYS, FAM126A, FAM149B1, FAM161A, FAM57B, FAM71A, FAR1, FAT1, FBLN5, FBN1, FDXR, FKRP, FKTN, FLNB, FLVCR1, FOXC1, FOXD3, FOXE3, FOXI2, FOXL2, FRAS1, FREM1, FREM2, FRMD7, FSCN2, FTL, FUT5, FXN, FYCO1, FZD4, FZD5, GALE, GALK1, GALM, GALT, GBA, GCNT2, GDF3, GDF6, GEMIN4, GFER, GJA1, GJA3, GJA8, GJC3, GJE1, GLI3, GLS, GMPPB, GNAT1, GNAT2, GNB3, GNPAT, GNPTAB, GNPTG, GOLPH3, GP1BA, GPR143, GPR179, GRIP1, GRK1, GRM6, GRN, GSN, GTF2H5, GUCA1A, GUCA1B, GUCY2D, HARS, HARS1, HCCS, HDAC6, HESX1, HEXA, HGSNAT, HIKESHI, HK1, HKDC1, HMCN1, HMGB3, HMX1, HPS1, HPS3, HPS4, HPS5, HPS6, HSF4, HTRA1, HTRA2, HYLS1, IDH3A, IDH3B, IDH3B, IFIH1, IFT140, IFT172, IFT27, IFT74, IFT81, IGBP1, IKBKG, IMPDH1, IMPG1, IMPG2, INPP5E, INPP5K, INTS1, INTS8, INVS, IQCB1, IRX5, IRX6, ISCA2, ISPD, ITIH2, ITM2B, JAG1, JAM3, KATNIP, KCNJ13, KCNV2, KCTD7, KERA, KIAA0556, KIAA0586, KIAA1109, KIAA1549, KIF11, KIF3B, KIF7, KIT, KIZ, KLHL7, KMT2D, KRT12, KRT3, LAMA1, LAMB2, LARGE1, LCA5, LCAT, LCT, LEMD2, LEP, LEPR, LIG3, LIM2, LMX1B, LONP1, LOXL1, LRAT, LRIT3, LRMDA, LRP1, LRP2, LRP5, LRPAP1,

LSS, LTBP2, LTBP3, LUM, LZTFL1, LYST, MAB21L2, MAF, MAFIP, MAG, MAK, MAN2A1, MAN2B1, MAPKAPK3, MC1R, MCAT, MECR, MED12, MED25, MED27, MERTK, MFF, MFN2, MFRP, MFSD6L, MFSD8, MIP, MIPEP, MIR184, MIR204, MITF, MKKS, MKS1, MLPH, MMACHC, MMP1, MPLKIP, MSMO1, MSTO1, MT-ATP6, MT-ND1, MT-ND4, MT-ND6, MT-TH, MT-TL1, MT-TP, MT-TS2, MTPAP, MTRFR, MTTP, MVK, MYH9, MYO5A, MYO7A, MYOC, MYRF, NAA10, NAALADL1, NACC1, NBAS, NDP, NDUFA12, NDUFS1, NDUFS2, NDUFS3, NECTIN3, NEFH, NEK2, NEUROD1, NF2, NHS, NMNAT1, NPHP1, NPHP3, NPHP4, NR, NR0B2, NR2E3, NR2F1, NRL, NSUN2, NTF4, NUMB, NUP188, NYX, OAT, OCA2, OCRL, OFD1, OPA1, OPA2, OPA3, OPA4, OPA5, OPA6, OPA8, OPN1LW, OPN1MW, OPN1SW, OPTN, OR2M7, OSTM1, OTX2, OVOL2, P3H2, P4HA2, PAK2, PANK2, PANK4, PAX2, PAX3, PAX6, PCARE, PCDH15, PCYT1A, PDAP1, PDE6A, PDE6B, PDE6C, PDE6D, PDE6G, PDE6H, PDSS1, PDZD2, PDZD7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX5L, PEX6, PEX7, PGK1, PGRMC1, PHF6, PHYH, PIBF1, PIGL, PIK3C2A, PIK3R1, PIK3R5, PIKFYVE, PISD, PITPNM3, PITX2, PITX3, PLA2G5, PLD4, PLK4, PLOD3, PMM2, PNKP, PNPLA6, POC1B, POC5, PODNL1, POLG, POLG2, POLH, POMC, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POMZP3, PORCN, PPARG, PPT1, PQBP1, PRCD, PRDM13, PRDM5, PRKCG, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, PRSS56, PRTFDC1, PRX, PSMC3, PTCH1, PUF60, PXDN, RAB18, RAB27A, RAB28, RAB3GAP1, RAB3GAP2, RARB, RAX, RAX2, RB1, RBP3, RBP4, RCBTB1, RD3, RDH11, RDH12, RDH5, RECQL4, REEP6, RGR, RGS9, RGS9BP, RHO, RIC1, RIMS1, RIMS2, RLBP1, RNLS, RNPC3, RNU4ATAC, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, RPGRIP1, RPGRIP1L, RRH, RRM2B, RS1, RTN4IP1, SAG, SALL2, SALL4, SAMD11, SBF2, SC5D, SCAPER, SCLT1, SCO2, SDCCAG8, SDHA, SEC23A, SEMA4A, SERAC1, SETX, SGSH, SH3PXD2B, SHH, SIL1, SIPA1L3, SIX3, SIX5, SIX6, SLC16A12, SLC24A1, SLC24A5, SLC25A4, SLC25A46, SLC2A1, SLC33A1, SLC37A3, SLC38A8, SLC39A5, SLC44A1, SLC45A2, SLC4A11, SLC52A2, SLC6A6, SLC7A14, SLC9A6, SMCHD1, SMG8, SMO, SMOC1, SNAI2, SNRNP200, SNX10, SOX10, SOX2, SPATA13, SPATA7, SPG7, SPINT2, SPP2, SPTLC1, SPTLC2, SRD5A3, SREBF1, SREBF2, SSBP1, STN1, STRA6, SUFU, TACSTD2, TAF1A, TAPT1, TBC1D20, TBC1D32, TBK1, TCF4, TCIRG1, TCTN1, TCTN2, TCTN3, TDRD7, TEAD1, TEK, TENM3, TEX28, TFAP2A, TFG, TGFBI, TGIF, TIMM50, TIMM8A, TIMP3, TINF2, TK2, TKFC, TMEM107, TMEM114, TMEM126A, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TMEM5, TMEM67, TMEM70, TMEM98, TNFRSF11A, TNFRSF11, TOPORS, TP53BP2, TPP1, TRAF3IP1, TREX1, TRIM32, TRNT1, TRPM1, TSFM, TSPAN12, TTC21B, TTC8, TTLL5, TTPA, TUB, TUBB4B, TUBGCP4, TUBGCP6, TULP1, TWNK, TYMP, TYR, TYRP1, UBAP1L, UBE2U, UBIAD1, UCHL1, UNC119, USH1C, USH1G, USH2A, USP45, VAX1, VCAN, VIM, VIPR2, VPS13B, VPS4A, VSX1, VSX2, WASF3, WDPCP, WDR19, WDR36, WFS1, WHRN, WRN, WT1, XPA, XPC, XYLT2, YAP1, YME1L1, ZEB1, ZFYVE26, ZIC2, ZNF408, ZNF423, ZNF469, ZNF513, ZNF526, ZNF644, ZNHIT3, ZPR1, ZWINT

Forrás:

CentoVision

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

Anophthalmia or microphthalmia (Version 1.51)

<https://panelapp.genomicsengland.co.uk/panels/34/> (Megtekintve: 2023.05.18.)

Bilateral congenital or childhood onset cataracts (Version 4.1)

<https://panelapp.genomicsengland.co.uk/panels/230/> (Megtekintve: 2023.07.20.)

Corneal abnormalities (Version 1.12)

<https://panelapp.genomicsengland.co.uk/panels/250/> (Megtekintve: 2023.07.20.)

Glaucoma (developmental) (Version 1.42)

<https://panelapp.genomicsengland.co.uk/panels/249/> (Megtekintve: 2023.07.20.)

Ocular coloboma (Version 1.46)

<https://panelapp.genomicsengland.co.uk/panels/294/> (Megtekintve: 2023.07.20.)

Ophthalmological ciliopathies (Version 3.1)

<https://panelapp.genomicsengland.co.uk/panels/722/> (Megtekintve: 2023.07.20.)

Optic neuropathy (Version 4.2)

<https://panelapp.genomicsengland.co.uk/panels/186/> (Megtekintve: 2023.07.20.)

Retinal disorders (Version 4.10)

<https://panelapp.genomicsengland.co.uk/panels/307/> (Megtekintve: 2023.07.20.)

Cai XB, Shen SR, Chen DF, Zhang Q, Jin ZB. An overview of myopia genetics. Exp Eye Res. 2019;188:107778. doi: 10.1016/j.exer.2019.107778. (Megtekintve: 2023.07.20.)