

Metabolikus betegségek (Metabolic disorders) génpanel v:2023.10.12.

Panel tartalma 1510 gén:

A4GALT, AAAS, AARS1, AARS2, AASS, ABAT, ABCA1, ABCA12, ABCA3, ABCB11, ABCB4, ABCB7, ABCC2, ABCC8, ABCD1, ABCD3, ABCD4, ABCG2, ABCG5, ABCG8, ABHD12, ABHD5, ACAA1, ACAD8, ACAD9, ACADL, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACAT2, ACBD5, ACO2, ACOX1, ACOX2, ACTA1, ACY1, ACSF3, ADA, ADAMTS10, ADAMTS13, ADAMTSL2, ADAR, ADCK3, ADGRG1, ADK, ADNP, ADSL, AFG3L2, AGA, AGK, AGL, AGPAT2, AGPS, AGRN, AGXT, AHCY, AICDA, AIFM1, AIMP1, AIMP2, AKAP9, AKR1D1, AKT2, AKT3, ALAD, ALAS2, ALDH18A1, ALDH1B1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALOX12B, ALOXE3, ALPL, ALS2, AMACR, AMN, AMPD1, AMPD2, AMT, ANK1, ANKRD26, ANKS6, ANO10, ANO5, ANTXR1, ANTXR2, AOX1, AP2S1, AP3B1, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APOA1, APOA2, APOA5, APOB, APOC2, APOE, APOPT1, APP, APPL1, APRT, APTX, AQP2, ARCN1, ARG1, ARL6, ARL6IP1, ARSA, ARSB, ARSE, ARSG, ARSK, ARX, ASAH1, ASL, ASNS, ASPA, ASPM, ASS1, ATAD3A, ATAD3B, ATIC, ATM, ATP13A2, ATP1A3, ATP2A1, ATP5A1, ATP5B, ATP5C1, ATP5D, ATP5E, ATP5F1E, ATP5G1, ATP5G2, ATP5G3, ATP5I, ATP5J, ATP5O, ATP6AP1, ATP6AP2, ATP6V0A2, ATP6V1A, ATP6V1B1, ATP6V1E1, ATP7A, ATP7B, ATP8B1, ATPAF2, ATR, ATRX, ATXN7, AUH, AVP, AVPR2, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B4GAT1, BAAT, BCAP31, BCAT1, BCAT2, BCKDHA, BCKDHB, BCKDK, BCS1L, BEST1, BICD2, BIN1, BLK, BLNK, BMP6, BOLA1, BOLA2, BOLA3, BRAF, BRAT1, BRCA2, BSCL2, BSND, BTB, BTK, C10ORF2, C12orf65, C19orf12, C19orf70, C1GALT1C1, C1QBP, CA12, CA2, CA5A, CACNA1C, CACNA1D, CACNA1S, CACNB2, CAD, CALM1, CAMTA1, CANT1, CAPN1, CAPN3, CARS2, CASK, CASP10, CASP8, CASQ1, CASR, CAST, CAT, CAV1, CAV3, CAVIN1, CBLIF, CBS, CCDC103, CCDC115, CCDC78, CCT5, CD19, CD247, CD320, CD3D, CD3E, CD3G, CD40, CD40LG, CD59, CD79A, CD79B, CD81, CD96, CDAN1, CDK5RAP2, CDKL5, CDKN1C, CEL, CENPJ, CEP152, CEP290, CEP89, CERS1, CERS3, CETP, CFAP298, CFH, CFHR3, CFL2, CFTR, CHAT, CHCHD10, CHD7, CHKB, CHM, CHRNA1, CHRN1, CHRND, CHRNE, CHST14, CHST3, CHST6, CHSY1, CIDEA, CIDEA2, CIDEA3, CIDEA4, CIDEA5, CIDEA6, CIDEA7, CIDEA8, CIDEA9, CIDEA10, CIDEA11, CIDEA12, CIDEA13, CIDEA14, CIDEA15, CIDEA16, CIDEA17, CIDEA18, CIDEA19, CIDEA20, CIDEA21, CIDEA22, CIDEA23, CIDEA24, CIDEA25, CIDEA26, CIDEA27, CIDEA28, CIDEA29, CIDEA30, CIDEA31, CIDEA32, CIDEA33, CIDEA34, CIDEA35, CIDEA36, CIDEA37, CIDEA38, CIDEA39, CIDEA40, CIDEA41, CIDEA42, CIDEA43, CIDEA44, CIDEA45, CIDEA46, CIDEA47, CIDEA48, CIDEA49, CIDEA50, CIDEA51, CIDEA52, CIDEA53, CIDEA54, CIDEA55, CIDEA56, CIDEA57, CIDEA58, CIDEA59, CIDEA60, CIDEA61, CIDEA62, CIDEA63, CIDEA64, CIDEA65, CIDEA66, CIDEA67, CIDEA68, CIDEA69, CIDEA70, CIDEA71, CIDEA72, CIDEA73, CIDEA74, CIDEA75, CIDEA76, CIDEA77, CIDEA78, CIDEA79, CIDEA80, CIDEA81, CIDEA82, CIDEA83, CIDEA84, CIDEA85, CIDEA86, CIDEA87, CIDEA88, CIDEA89, CIDEA90, CIDEA91, CIDEA92, CIDEA93, CIDEA94, CIDEA95, CIDEA96, CIDEA97, CIDEA98, CIDEA99, CIDEA100, CIDEA101, CIDEA102, CIDEA103, CIDEA104, CIDEA105, CIDEA106, CIDEA107, CIDEA108, CIDEA109, 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CSGALNACT1, CSTB, D2HGDH, DAG1, DARS, DARS1, DARS2, DBH, DBT, DCAF17, DCC, DCLRE1C, DCXR, DDC, DDHD1, DDHD2, DDOST, DDR2, DEPDC5, DES, DGUOK, DHCR24, DHCR7, DHDDS, DHFR, DHFR2, DHODH, DHTKD1, DIABLO, DIAPH1, DKC1, DLAT, DLD, DLL3, DLST, DLX4, DMD, DMGDH, DNA2, DNAH11, DNAH5, DNAI1, DNAI2, DNAJB6, DNAJC12, DNAJC19, DNAJC5, DNM1L, DNM2, DOCK7, DOCK8, DOK7, DOLK, DPAGT1, DPEP1, DPM1, DPM2, DPM3, DPYD, DPYS, DRC1, DSE, DSP, DST, DSTYK, DTD1, DUOX2, DUOX2A2, DYM, DYSF, EARS2, EBP, ECHS1, ECSIT, EDN3, EEF1A2, EGF, EGR2, EHBP1L1, EHHADH, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, ELAC2, ELANE, EMD, ENO3, ENPP1, ENTPD1, EOGT, EPB42, EPCAM, EPG5, EPHX2, EPM2A, EPRS1, ERAL1, ERCC6, ERCC6L2, ERCC8, ERLIN1, ERLIN2, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOSC3, EXT1, EXT2, EXTL3, EYA1, EYA4, F10, F11, F13A1, F2, F5, F7, F8, F9, FA2H, FADD, FAH, FAM111A, FAM126A, FANCA, FANCB, FANCC, FANCD2, FANCL, FAR1, FARS2, FARSB, FAS, FASLG, FASTKD2, FBN1, FBP1, FBP2, FBXL4, FDX1L, FDX2, FDXR, FECH, FGA, FGB, FGF23, FGFR2, FGFR3, FGG, FH, FHL1, FIG4, FKBP14, FKRP, FKTN, FLAD1, FLNA, FLNB, FMO3, FOLR1, FOLR2, FOLR3, FOXC1, FOXE1, FOXG1, FOXP3, FOXRED1, FRAS1, FTCD, FTL, FUCA1, FUK, FUT8, FXN, FXYD2, G6PC, G6PC1, G6PC3, G6PD, GAA, GABRB2, GABRG2, GALC, GALE, GALK1, GALM, GALNS, GALNT12, GALNT2, GALNT3, GALT, GAMT, GAN, GANAB, GARS, GARS1, GATA1, GATA4, GATA6, GATB, GATC, GATM, GBA, GBA2, GBE1, GCDH, GCCR, GCH1, GCK, GCLC, GCSH, GDAP1, GFAP, GFER, GFM1, GFM2, GFPT1, GGT1, GHR, GIF, GJA1, GJB1, GJB2, GJB4, GJC2, GK, GLA, GLB1, GLDC, GLIS3, GLRA1, GLRB, GLRX5, GLS, GLUD1, GLUL, GLYCTK, GM2A, GMPPA, GMPPB, GNAS, GNE, GNMT, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GOSR2, GP1BA, GP1BB, GP9, GPAA1, GPC3, GPD1, GPHN, GPSM2, GRHPR, GRN, GSS, GTPBP2, GTPBP3, GUSB, GYG1, GYS1, GYS2, HAAO, HACE1, HADH, HADHA, HADHB, HAL, HAMP, HARS2, HAX1, HBA1, HBA2, HBB, HCFC1, HCCS, HEPACAM, HESX1, HEXA, HEXB, HFE, HFE2, HGD, HGF, HGSNAT, HIBCH, HIKESHI, HJV, HK1, HLCS, HMBS, HMGCL, HMGCS2, HNF1A, HNF1B, HNF4A, HOGA1, HPD, HPGD, HPRT1, HPS1, HRAS, HS2ST1, HSD17B10, HSD17B4, HSD3B2, HSD3B7, HSPA9, HSPD1, HSPG2, HTRA1, HTRA2, HYAL1, HYKK, IARS2, IBA57, ICOS, IDH1, IDH2, IDH3B, IDS, IDUA, IER3IP1, IFIH1, IFT172, IGF1, IGF1R, IGHMBP2, IGLL1, IGSF1, IKKB, IL12RB1, IL2RA, IL2RG, IL7R, INS, INSR, INVS, IRF8, IRS4, ISCA1, ISCA2, ISCU, ISPD, ITGA2B, ITGA6, ITGA7, ITGB3, ITGB4, ITK, ITPA, IVD, IYD, JAG1, JAGN1, JAK3, JAM3, KARS, KAT6A, KAT6B, KBTBD13, KCNA1, KCNC1, KCNE1, KCNH1, KCNH2, KCNJ10, KCNJ11, KCNJ2, KCNQ1, KCNQ2, KCNQ3, KCNT1, KCTD7, KDM6A, KHK, KIDINS220, KIF1A, KIF1B, KIF1C, KIF5A, KLF1, KLF11, KLHL40, KLHL41, KLHL7, KMT2D, KRAS, KRT5, KYNU, L1CAM, L2HGDH, LACTB, LAMA2, LAMA3, LAMB1, LAMB3, LAMC2, LAMP2, LAMTOR2, LARGE, LARGE1, LARS, LARS2, LAS1L, LAT, LBR, LCAT, LCT, LDB3, LDHA, LDLR, LDLRAP1, LETM1, LFNG, LHX3, LHX4, LIAS, LIG4, LIPA, LIPC, LIPE, LIPI, LIPN, LIPT1, LIPT2, LMBRD1, LMNA, LMNB1, LONP1, LPA, LPIN1, LPL, LRBA, LRPPRC, LRRC8A, LYRM4, LYRM7, LYST, MAG, MAGEL2, MAGT1, MALT1, MAN1B1, MAN2B1, MANBA, MAOA, MAP2K1, MAP2K2, MARS1, MARS2, MAT1A, MCCC1, MCCC2, MCEE, MCM4, MCOLN1, MCPH1, MDH2, MECP2, MECP2, MED12, MEF2C, MEGF10, MFF, MFN2, MFSD8, MGAT2, MGME1, MICU1, MIPEP, MITF, MKKS, MLC1, MLPH, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMT1, MMUT, MOCOS, MOCS1,

MOCS2, MOGS, MPC1, MPDU1, MPI, MPV17, MRM2, MRPL12, MRPL3, MRPL40, MRPL44, MRPS14, MRPS16, MRPS2, MRPS22, MRPS23, MRPS34, MRPS7, MSMO1, MSTO1, 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, MTFMT, MTHFD1, MTHFR, MTO1, MTPAP, MTR, MTRFR, MTRR, MTPP, MUT, MVK, MYH3, MYO5A, MYOT, NADK2, NAGA, NAGLU, NAGS, NANS, NARS2, NAT8L, NAXD, NAXE, NBAS, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA3, NDUFA4, NDUFA5, NDUFA6, NDUFA7, NDUFA8, NDUFA9, NDUFAB1, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF8, NDUFB1, NDUFB10, NDUFB11, NDUFB2, NDUFB3, NDUFB4, NDUFB5, NDUFB6, NDUFB7, NDUFB8, NDUFB9, NDUFC1, NDUFC2, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS5, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NDUFV3, NEU1, NEUROD1, NEUROG3, NFE2L2, NFS1, NFU1, NGLY1, NHLRC1, NIPA1, NIPA2, NKX6-2, NNT, NOTCH3, NPC1, NPC2, NPR2, NR0B1, NRAS, NSDHL, NSUN3, NT5C, NT5C2, NT5C3A, NUBPL, NUP62, NUS1, OAT, OCLN, OCRL, OGDH, OGT, OPA1, OPA3, OPLAH, OSGEP, OTC, OXA1L, OXCT1, PAH, PANK2, PAPSS2, PARS2, PAX4, PC, PCBD1, PCCA, PCCB, PCK1, PCK2, PCYT2, PCSK9, PDHA1, PDHB, PDHX, PDK1, PDK2, PDK3, PDK4, PDP1, PDP2, PDPR, PDSS1, PDSS2, PDX1, PDXK, PEPD, PET100, PET117, PEX1, PEX10, PEX11A, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGAM2, PGAP1, PGAP2, PGAP3, PGK1, PGM1, PGM3, PHGDH, PHKA1, PHKA2, PHKB, PHKG1, PHKG2, PHYH, PHYKPL, PIGA, PIGB, PIGC, PIGG, PIGL, PIGM, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGU, PIGV, PIGW, PIGY, PIK3R1, PINK1, PITRM1, PKLR, PLA2G6, PLAA, PLCG2, PLIN1, PLP1, PLPBP, PMM2, PMPCA, PMPCB, PNLIP, PNP, PNPLA2, PNPLA4, PNPLA6, PNPLA8, PNPO, PNPT1, POFUT1, POGLUT1, POLG, POLG2, POLR1C, POLR3A, POLR3B, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POP1, POR, PPA2, PPARG, PPM1B, PPM1K, PPOX, PPP1R17, PPT1, PRDX1, PREPL, PRF1, PRICKLE1, PRKAG2, PRKAG3, PRKCD, PRKCSH, PRODH, PROSC, PRPS1, PSAP, PSAT1, PSEN1, PSPH, PTCD1, PTCD3, PTF1A, PTPRZ1, PTRF, PTS, PUS1, PYCR1, PYCR2, PYGL, PYGM, PYY, QARS, QDPR, QRSL1, RAB11B, RAB27A, RAB3GAP2, RAI1, RANBP2, RARS1, RARS2, RBCK1, RBP4, REEP1, REEP2, REN, RFT1, RFX6, RMND1, RNASEH1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF216, ROBO3, RPIA, RPL10, RPN2, RRM2B, RTN2, RTN4IP1, RXYLT1, RYR1, SACS, SAMHD1, SAR1B, SARDH, SARS2, SC5D, SCARB1, SCARB2, SCN4A, SCO1, SCO2, SCP2, SDHA, SDHAF1, SDHAF2, SDHB, SDHC, SDHD, SEC23A, SEC23B, SEC24D, SEC63, SERAC1, SERPINA1, SERPINI1, SETX, SFXN4, SGCA, SGCB, SGCD, SGCG, SGSH, SHMT2, SHPK, SI, SIL1, SKIV2L, SLC10A7, SLC12A3, SLC13A3, SLC16A1, SLC16A2, SLC17A5, SLC18A2, SLC19A2, SLC19A3, SLC1A4, SLC22A4, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A2, SLC25A20, SLC25A22, SLC25A26, SLC25A3, SLC25A32, SLC25A38, SLC25A4, SLC25A40, SLC25A42, SLC25A46, SLC26A2, SLC26A6, SLC27A5, SLC2A1, SLC2A2, SLC30A10, SLC31A1, SLC33A1, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC35D1, SLC36A2, SLC37A4, SLC39A14, SLC39A4, SLC39A8, SLC3A1, SLC40A1, SLC41A2, SLC41A3, SLC46A1, SLC4A1, SLC52A1, SLC52A2, SLC52A3, SLC5A1,

SLC5A6, SLC6A19, SLC6A20, SLC6A3, SLC6A8, SLC6A9, SLC7A7, SLC7A9, SLC9A7, SLCO1B1, SLCO1B3, SMPD1, SNTA1, SOX10, SPART, SPAST, SPG11, SPG21, SPG7, SPR, SPTA1, SPTB, SPTLC1, SPTLC2, SRD5A3, SRRT, SSBP1, SSR3, SSR4, ST3GAL3, ST3GAL5, STAC3, STAT1, STAT2, STAT3, STS, STT3A, STT3B, STX11, STXBP2, SUCLA2, SUCLG1, SUCLG2, SUGCT, SUMF1, SUOX, SURF1, SYNE1, TACO1, TAFAZZIN, TALDO1, TANGO2, TARS2, TAT, TAZ, TBC1D24, TBC1D4, TCAP, TCF4, TCN1, TCN2, TDO2, TECPR2, TFAM, TFG, TFR2, TGDS, TGFB1, TH, THAP11, TIMM44, TIMM50, TIMM8A, TIMMDC1, TINF2, TK2, TKFC, TM6SF2, TMEM106B, TMEM126A, TMEM126B, TMEM165, TMEM199, TMEM5, TMEM65, TMEM70, TNPO3, TOP3A, TPK1, TPMT, TPP1, TRAK1, TRAP1, TRAPPC11, TRAPPC12, TRAPPC2, TRAPPC6B, TRAPPC9, TREH, TREM2, TREX1, TRIM32, TRIM37, TRIP11, TRIT1, TRMT10A, TRMT10C, TRMT5, TRMU, TRNT1, TRPM6, TRPM7, TRPV4, TSFM, TTC19, TTC37, TTPA, TUBB4A, TUFM, TUSC3, TWNK, TXN2, TYMP, TYROBP, UBAP1, UCHL1, UCP2, UFM1, UGT1A1, UMOD, UMPS, UNC13D, UNC80, UPB1, UQCC2, UQCC3, UQCRB, UQCRC1, UQCRC2, UQCRFS1, UQCRH, UQCRQ, UROC1, UROD, UROS, USF1, USH1C, VAMP1, VARS2, VCP, VIPAS39, VKORC1, VMA21, VPS11, VPS13B, VPS13C, VPS33B, VPS37A, WARS2, WASHC5, WDR45, WDR45B, WFS1, XDH, XPNPEP3, XYLT1, XYLT2, YARS2, YME1L1, ZFP57, ZFYVE26, ZFYVE27, ZMPSTE24, ZNF143

Forrás:

Likely inborn error of metabolism - targeted testing not possible (Version 4.34)

<https://panelapp.genomicsengland.co.uk/panels/467/> (Megtekintve: 2023.05.17.)

Comprehensive Metabolism NGS Panel

<https://www.fulgentgenetics.com/Metabolism-Comprehensive> (Megtekintve: 2023.05.17.)

CentoICU[®]

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