

## Metabolikus betegségek (Metabolic disorders) génpanel v:2025-05-22

Panel tartalma 1170 gén:

A4GALT, AAAS, AARS2, AASS, ABAT, ABCA1, ABCB11, ABCB4, ABCB7, ABCC2, ABCC8, ABCD1, ABCD3, ABCD4, ABCG5, ABCG8, ABHD12, ABHD5, ACAA1, ACACA, ACAD8, ACAD9, ACADL, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACBD5, ACO2, ACOX1, ACOX2, ACY1, ACSF3, ADA, ADAMTS10, ADAMTSL2, ADAR, ADGRG1, ADK, ADSL, AFG3L2, AGA, AGK, AGL, AGPAT2, AGPS, AGXT, AHCY, AIFM1, AIMP1, AIMP2, AKR1D1, AKT2, AKT3, ALAD, ALAS2, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALPL, ALS2, AMACR, AMN, AMPD1, AMPD2, AMT, ANK1, ANO10, ANO5, ANTXR2, AP1S1, AP3B1, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APOA1, APOA2, APOA5, APOB, APOC2, APOE, APP, APPL1, APRT, APTX, AQP2, ARCN1, ARG1, ARL6IP1, ARSA, ARSB, ARSG, ARSK, ARSL, ASAH1, ASL, ASNS, ASPA, ASS1, ATAD1, ATAD3A, ATIC, ATM, ATP13A2, ATP2A1, ATP5F1A, ATP5F1B, ATP5F1D, ATP5F1E, ATP5MC3, ATP5PO, ATP6AP1, ATP6AP2, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, ATP7B, ATP8B1, ATPAF2, ATRX, AUH, AVP, AVPR2, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B4GAT1, BAAT, BCAP31, BCAT2, BCKDHA, BCKDHB, BCKDK, BCS1L, BEST1, BICD2, BLK, BMP6, BOLA3, BRAT1, BSCL2, BSND, BTD, C19orf12, C1GALT1C1, C1QBP, CA2, CA5A, CACNA1D, CACNA1S, CAD, CANT1, CAPN1, CAPN3, CARS2, CASP10, CASP8, CASQ1, CASR, CAT, CAV1, CAV3, CAVIN1, CBLIF, CBS, CCDC115, CCT5, CD320, CEL, CERS1, CETP, CFTR, CHCHD10, CHKB, CHST14, CHST3, CHST6, CHSY1, CIDEA, CISD2, CKMT1A, CKMT1B, CKMT2, CLCN1, CLCN2, CLCN7, CLCNKB, CLDN16, CLDN19, CLN3, CLN5, CLN6, CLN8, CLPB, CLPP, CNM1, CNM2, CNM4, COA3, COA6, COA7, COA8, COASY, COG1, COG2, COG3, COG4, COG5, COG6, COG7, COG8, COL11A2, COL2A1, COL4A1, COL4A2, COLGALT1, COPA, COPB2, COQ2, COQ4, COQ5, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX14, COX15, COX20, COX4I2, COX5A, COX6A1, COX6B1, COX7B, CP, CPOX, CPS1, CPT1A, CPT1B, CPT1C, CPT2, CRAT, CREB3L3, CRLS1, CRPPA, CTC1, CTDP1, CTH, CTLA4, CTNS, CTSA, CTSC, CTSD, CTSF, CTSK, CUBN, CYC1, CYCS, CYP11B1, CYP17A1, CYP19A1, CYP27A1, CYP2U1, CYP7B1, CSF1R, CSGALNACT1, CSTB, D2HGDH, DAG1, DARS1, DARS2, DBH, DBT, DCAF17, DCC, DCXR, DDC, DDHD1, DDHD2, DDOST, DGUOK, DHCR24, DHCR7, DHDDS, DHFR, DHODH, DHRSX, DHTKD1, DIABLO, DKC1, DLAT, DLD, DLL3, DMD, DMPK, DNA2, DNAJB6, DNAJC12, DNAJC19, DNAJC5, DNMT1, DNMT2, DOLK, DPAGT1, DPM1, DPM2, DPM3, DPYD, DPYS, DSE, DSTYK, DYM, DYSF, EARS2, EBP, ECHS1, EDEM3, EGF, EHBP1L1, EHHADH, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, ELAC2, EMD, ENO3, ENPP1, ENTPD1, EOGT, EPB42, EPG5, EPHX2, EPM2A, EPRS1, ERAL1, ERCC6, ERCC8, ERLIN1, ERLIN2, ETFA, ETFB, ETFDH, ETHE1, EXOSC3, EXT1, EXT2, EXTL3, F2, F5, FA2H, FADD, FAH, FAM111A, FAM126A, FAR1, FARS2, FARSB, FAS, FASLG, FASTKD2, FBN1, FBP1, FBXL4, FCSK, FDX2, FDXR, FECH, FGF23, FGFR2, FH, FHL1, FKRP, FKTN, FLAD1, FLNA, FLNB, FMO3, FOLR1, FOXA2, FOXP3, FOXRED1, FTCD, FTL, FUCA1, FUT8, FXN, FXND2, G6PC1, G6PC3, G6PD, GAA, GABRB2, GABRG2, GAD1, GALC, GALE, GALK1, GALM, GALNS, GALNT2, GALNT3, GALT, GAMT, GAN, GANAB, GARS1, GATA4, GATA6, GATC, GATM, GBA, GBA2, GBE1, GCDH, GCGR, GCH1, GCK, GCLC, GCSH, GDAP1, GFAP, GFER, GFM1, GFM2, GFPT1, GHR, GJA1, GJB1, GJC2, GK, GLA, GLB1, GLDC, GLIS3, GLRA1, GLRB, GLRX5, GLS, GLUD1,

GLUL, GLYCTK, GM2A, GMPPA, GMPPB, GNE, GNMT, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GOSR2, GOT2, GPAA1, GPC3, GPD1, GPHN, GPIHBP1, GRHPR, GRN, GSS, GSTZ1, GTPBP2, GTPBP3, GUSB, GYG1, GYS1, GYS2, HAAO, HACE1, HADH, HADHA, HADHB, HAMP, HARS2, HCFC1, HCCS, HEPACAM, HEXA, HEXB, HFE, HGD, HGSNAT, HIBCH, HIKESHI, HJV, HK1, HLCS, HMBS, HMGCL, HMGCS2, HNF1A, HNF1B, HNF4A, HOGA1, HPD, HPRT1, HPS1, HRAS, HS2ST1, HSD17B10, HSD17B4, HSD3B2, HSD3B7, HSPA9, HSPD1, HTRA1, HTRA2, HYAL1, IARS2, IBA57, IDH1, IDH2, IDS, IDUA, IER3IP1, IFIH1, IL2RA, INS, INSR, ISCA1, ISCA2, ISCU, ITIH4, ITK, ITPA, IVD, JAG1, JAGN1, JAM3, KARS1, KCNA1, KCNC1, KCNJ10, KCNJ11, KCNJ2, KCNT1, KCTD7, KDM6A, KHK, KIDINS220, KIF1A, KIF1C, KIF5A, KLF11, KMT2D, KRAS, KYNU, L1CAM, L2HGDH, LAMA2, LAMB1, LAMP2, LARGE1, LARS1, LARS2, LAT, LBR, LCAT, LCT, LDB3, LDHA, LDHD, LDLR, LDLRAP1, LETM1, LFNG, LIAS, LIPA, LIPC, LIPE, LIPI, LIPT1, LIPT2, LMBRD1, LMF1, LMNA, LMNB1, LONP1, LPIN1, LPL, LRBA, LRPPRC, LYRM4, LYRM7, LYST, MAG, MAGT1, MAN1B1, MAN2B1, MAN2B2, MANBA, MAOA, MARS1, MARS2, MAT1A, MCCC1, MCCC2, MCEE, MCOLN1, MDH2, MECR, MFF, MFN2, MFSD8, MGAT2, MGME1, MICOS13, MICU1, MIPEP, MLC1, MLPH, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMGT1, MMUT, MOCOS, MOCS1, MOCS2, MOGS, MPC1, MPDU1, MPI, MPV17, MRM2, MRPL3, MRPL44, MRPS14, MRPS16, MRPS2, MRPS22, MRPS34, MRPS7, MSMO1, MSTO1, MTFMT, MTHFR, MTO1, MTPAP, MTR, MTRFR, MTRR, MVK, MYH3, MYO5A, MYOT, NADK2, NAGA, NAGLU, NAGS, NANS, NARS2, NAXD, NAXE, NBAS, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA4, NDUFA6, NDUFA8, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF8, NDUFB10, NDUFB11, NDUFB3, NDUFB7, NDUFB8, NDUFB9, NDUFC2, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEU1, NEUROD1, NEUROG3, NFE2L2, NFS1, NFU1, NGLY1, NHLRC1, NIPA1, NIPA2, NKX2-2, NKX6-2, NNT, NOTCH3, NPC1, NPC2, NPR2, NR0B1, NRAS, NSDHL, NSUN3, NT5C2, NT5C3A, NT5E, NUBPL, NUS1, OAT, OCLN, OCRL, OGDH, OGT, OPA1, OPA3, OPLAH, OSGEP, OTC, OXCT1, PAH, PANK2, PAPSS2, PARS2, PAX4, PC, PCBD1, PCCA, PCCB, PCK1, PCK2, PCYT2, PCSK9, PDHA1, PDHB, PDHX, PDK3, PDP1, PDSS1, PDSS2, PDX1, PEPD, PET100, PET117, PEX1, PEX10, 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, 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, PNP, PNPLA2, PNPLA6, PNPLA8, PNPO, PNPT1, POFUT1, POGLUT1, POLG, POLG2, POLR1C, POLR3A, POLR3B, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POR, PPA2, PPARG, PPM1K, PPOX, PPP1R17, PPT1, PRDX1, PRF1, PRICKLE1, PRKAG2, PRKAG3, PRKCD, PRKCSH, PRODH, PRPS1, PSAP, PSAT1, PSEN1, PSPH, PTCD3, PTF1A, PTS, PUS1, PYCR1, PYCR2, PYGL, PYGM, PYY, QARS1, QDPR, QRSL1, RAB11B, RAB27A, RAB3GAP2, RAI1, RANBP2, RARS1, RARS2, RBCK1, RBP4, REEP1, REEP2, REN, RFT1, RFX6, RMND1, RNASEH1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF216, RPIA, RPL10, RPN2, RRM2B, RTN2, RTN4IP1, RXYLT1, RYR1, SACS, SAMHD1, SAR1B, SARS2, SC5D, 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, SI, SIL1, SKIV2L, SLC10A7, SLC12A3, SLC13A3, SLC13A5, SLC16A1, SLC16A2, SLC17A5, SLC18A2, SLC19A1, SLC19A2, SLC19A3, SLC1A3, SLC1A4, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A26,

SLC25A3, SLC25A32, SLC25A38, SLC25A4, SLC25A42, SLC25A46, SLC26A2, SLC2A1, SLC2A2, SLC30A10, SLC31A1, SLC33A1, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC35D1, SLC37A4, SLC39A14, SLC39A4, SLC39A8, SLC3A1, SLC40A1, SLC41A2, SLC41A3, SLC46A1, SLC4A1, SLC52A1, SLC52A2, SLC52A3, SLC5A1, SLC5A6, SLC6A19, SLC6A3, SLC6A5, SLC6A8, SLC6A9, SLC7A7, SLC7A9, SLC9A7, SLCO1B1, SLCO1B3, SMPD1, SNTA1, SOX10, SPART, SPAST, SPG11, SPG21, SPG7, SPR, SPTA1, SPTB, SPTLC1, SPTLC2, SRD5A3, SSBP1, SSR3, SSR4, ST3GAL3, ST3GAL5, STAC3, STAT1, STAT2, STAT3, STS, STT3A, STT3B, STX11, STXBP2, SUCLA2, SUCLG1, SUGCT, SUMF1, SUOX, SURF1, SYNE1, TACO1, TAFAZZIN, TALDO1, TANGO2, TARS2, TAT, TBC1D24, TBC1D4, TCAP, TCF4, TCN1, TCN2, TECPR2, TFAM, TFG, TFR2, TGDS, TGFB1, TH, THAP11, TIMM50, TIMM8A, TIMMDC1, TINF2, TK2, TKFC, TMEM106B, TMEM126A, TMEM126B, TMEM165, TMEM199, TMEM65, TMEM70, TNPO3, TOP3A, TPI1, TPK1, TPMT, TPP1, TRAK1, TRAP1, TRAPPC11, TRAPPC12, TRAPPC2, TRAPPC6B, TRAPPC9, TREM2, TREX1, TRIM32, TRIM37, TRIP11, TRIT1, TRMT10A, TRMT10C, TRMT5, TRMU, TRNT1, TRPM6, TRPM7, TRPV4, TSFM, TTC19, TTC37, TTPA, TUBB4A, TUFM, TUSC3, TWNK, TYMP, TYROBP, UBAP1, UCHL1, UCP2, UFM1, UGT1A1, UMOD, UMPS, UNC13D, UNC80, UPB1, UQCC2, UQCC3, UQCRB, UQCRC1, UQCRC2, UQCRFS1, UQCRQ, UROC1, UROD, UROS, USH1C, VAMP1, VARS2, VCP, VIPAS39, VKORC1, VMA21, VPS11, VPS13B, VPS16, VPS33A, VPS33B, VPS37A, WARS2, WASHC5, WDR45, WDR45B, WFS1, WWTR1, XDH, XPNPEP3, XYLT1, XYLT2, YARS2, YME1L1, ZFP57, ZFYVE26, ZFYVE27, ZMPSTE24, ZNF143

Forrás:

Likely inborn error of metabolism (Version 8.5)

<https://panelapp.genomicsengland.co.uk/panels/467/>

Undiagnosed metabolic disorders (Version 1.629)

<https://panelapp.genomicsengland.co.uk/panels/302/>

Comprehensive Metabolism NGS Panel

<https://www.fulgentgenetics.com/Metabolism-Comprehensive>

CentoIEM®

<https://www.centportal.com/order/new/products/analysis-method?queryType=TEST&query=CentoIEM>

Invitae Metabolic Newborn Screening Confirmation Panel

<https://www.invitae.com/en/physician/tests/06102/>

Invitae Supplemental Metabolic Newborn Screening Panel

<https://www.invitae.com/en/physician/tests/98003/>

Comprehensive Metabolism Panel

<https://blueprintgenetics.com/tests/panels/metabolic-disorders/comprehensive-metabolism-panel/>

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