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Klinika pediatrie a dědičných poruch metabolismu – Diagnostické laboratoře DMP
Ke Karlovu 455/2, 128 08 Praha 2

NGS PANELY - SEZNAM VYŠETŘOVANÝCH GENŮ

Seznam genů analyzovaných v metabolickém panelu

Poruchy metabolismu glykogenu (29 genů): *AGL, ALDOA, ALDOB, ALDOC, ENO3, FBP1, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, KHK, PC, PFKM, PGAM2, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAB1, PRKAB2, PRKAG2, PYGL, PYGM, RBCK1, SLC2A2, SLC37A4*

Rhabdomyolýzy a poruchy metabolismu mastných kyselin (47 genů): *ACADM, ACADVL, AGL, ALDOA, AMPD1, ANO5, ATP2A1, CACNA1S, CASQ1, CAV3, CHKB, CPT1A, CPT2, CTDP1, DGUOK, DYSF, ENO3, ETFA, ETFB, ETFDH, FKRP, FLAD1, HADHA, HADHB, ISCU, LAMP2, LDHA, LPIN1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PYGM, RYR1, SCN4A, SIL1, SLC16A1, SLC22A5, SLC25A20, SLC25A32, SLC52A1, SLC52A2, SLC52A3, TANGO2, TSEN54*

Peroxisomální onemocnění (34 genů): *ABCD1, ABCD3, ACBD5, ACOX1, AGPS, AGXT, AMACR, BAAT, CAT, DNM1L, FAR1, GDAP1, GNPAT, HSD17B4, MFF, PEX1, PEX2, PEX26, PEX3, PEX5, PEX5L, PEX6, PEX7, PEX10, PEX11A, PEX11B, PEX11G, PEX12, PEX13, PEX14, PEX16, PEX19, PHYH, SCP2*

Poruchy metabolismu neurotransmiterů (26 genů): *ABAT, ALDH5A1, ALDH7A1, AMT, DBH, DDC, DHFR, DNAJC12, FOLR1, GCH1, GCSH, GLDC, GLUL, MAOA, PCBD1, PHGDH, PNPO, PSAT1, PSPH, PTS, QDPR, SLC18A2, SLC46A1, SLC6A3, SPR, TH*

Neuronální ceroidlipofuscinózy (13 genů): *ATP13A2, CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, DNAJC5, GRN, KCTD7, MFSD8, PPT1, TPP1*

Poruchy cyklu močoviny, orotové acidurie (19 genů): *ARG1, ASL, ASS1, CAD, CPS1, DHODH, FTCD, NAGS, OTC, SHMT1, SHMT2, SLC25A13, SLC25A15, SLC25A2, SLC46A1, SLC7A7, TYMP, TYMS, UMPS*

Leucinózy (4 geny): *BCKDHA, BCKDHB, DBT, DLD*

Isovalerová acidurie (1 gen): *IVD*

Propionová acidurie (2 geny): *PCCA, PCCB*

Hyperhomocysteinémie (53 genů): *ABCD4, ADK, AHCY, ALDH7A1, AMN, CBS, CD320, CDO1, CTH, CUBN, DHFR, ETHE1, FOLH1, FOLR1, FOLR2, FOLR3, FTCD, FUT2, GIF, GNMT, GPHN, HCFC1, LMBRD1, LRP2, MAT1A, MAT2A, MAT2B, MCEE, MMAA, MMAB, MMACHC, MMADHC, MOCS1, MOCS2, MTHFD1, MTHFR, MTHFS, MTR, MTRR, MUT, PDXK, PDXP, PNPO, SLC19A1, SLC46A1, SQOR, SUCLA2, SUOX, TCN1, TCN2, THAP11, TST, ZNF143*

Cystinurie (2 geny): *SLC3A1, SLC7A9*

Hyperurikémie (5 genů a vybrané SNP):
ABCG2, HPRT1, PRPS1, REN, UMOD, GCKR – rs1260326, *ALDH2* – rs671, *ALDH16A1* – rs150414818, *ADH1B* – rs1229984

CADASIL (1 gen): *NOTCH3*

Seznam genů analyzovaných v kardiologickém panelu (180 genů)

ABCC9, ABCG5, ABCG8, ACTA1, ACTA2, ACTC1, ACTN2, AKAP9, ALMS1, ANK2, ANKRD1, APOA4, APOA5, APOB, APOC2, APOE, BAG3, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2 , CALM3, CALR3, CASQ2, CASZ1, CAV3, CBL, CETP, COL3A1, COL4A5, COL5A1, COL5A2, CREB3L3, CRELD1, CRYAB, CSRP3, CTF1, CTNNA3, DES, DMD, DNAJC19, DOLK, DPP6, DSC2, DSG2, DSP, DTNA, EFEMP2, ELN, EMD, EYA4, FBN1, FBN2, FHL1, FHL2, FKRP, FKTN, FLNC, FXN, GAA, GATA4, GATA5, GATAD1, GJA5, GLA, GPD1, GPD1L, GPIHBP1, HCN4, HFE, JAG1, JPH2, JUP, KCNA5, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, KLF10, LAMA2, LAMA4, LAMP2, LDB3, LDLR, LDLRAP1, LMF1, LMNA, LOX, LPL, LTBP2, MAP2K1, MAP2K2, MIB1, MT-TL1 - rs199474657, MURC, MYBPC3, MYH11, MYH6, MYH7, MYL2, MYL3, MYLK, MYLK2, MYOM1, MYOZ2, MYPN, NEBL, NEXN, NKX2-5, NODAL, NOS1AP, NOTCH1, NPPA, OBSCN, PCSK9, PDLIM3, PKP2, PLN, PRDM16, PRKAG2, PRKG1, PTPN11, RAF1, RANGRF, RBM20, RIT1, ROBO4, RYR2, SALL4, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SDHA, SGCD, SGCG, SHOC2, SLC2A10, SLMAP, SMAD3, SMAD4, SNTA1, SOS1, SYNE1, SYNE2,

NGS PANELY - SEZNAM VYŠETŘOVANÝCH GENŮ

TAB2, TAZ, TBX20, TBX3, TBX5, TCAP, TECRL, TGFB2, TGFB3, TGFBR1, TGFBR2, TMEM43, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRDN, TRIM63, TRPM4, TTN, TTR, TXNRD2, VCL, ZIC3

Seznam genů analyzovaných v panelu pro onemocnění skeletu (337 genů)

ACAN, ACP5, ACTB, ACTG1, ACVR1, ADAMTS10, ADAMTS17, ADAMTSL2, AGPS, AIFM1, AKT1, ALPL, ALX3, ALX4, AMER1, ANKH, ANKRD11, ANO5, ARHGAP31, ARSB, ARSE, ATP6V0A2, ATR, B3GALT6, B3GAT3, B4GALT7, BCS1L, BGN, BHLHA9, BMP1, BMP2, BMPER, BMPR1B, BRAF, BRCA2, BRIP1, CA2, CANT1, CASR, CBL, CCDC8, CDC6, CDC45, CDKN1C, CDT1, CENPJ, CEP63, CEP152, CHST3, CHST14, CHSY1, CKAP2L, CLCN5, CLCN7, COL1A1, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, COL10A1, COL11A1, COL11A2, COMP, CREB3L1, CREBBP, CRTAP, CSPP1, CTSK, CUL7, CYP24A1, CYP27B1, DDR2, DHCR7, DHCR24, DHODH, DLL3, DLL4, DLX3, DLX5, DMP1, DOCK6, DVL1, DYM, DYNC2H1, EBP, EFNB1, EFTUD2, EIF2AK3, ENAM, ENPP1, EOGT, EP300, ERCC4, ESCO2, EVC, EVC2, EXT1, EXT2, EXTL3, EZH2, FAM20A, FAM20C, FAM58A, FAM83H, FAM111A, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FBN1, FBN2, FGD1, FGF10, FGF23, FGFR1, FGFR2, FGFR3, FKBP10, FLNA, FLNB, GALNT3, GDF5, GH1, GHR, GHRHR, GJA1, GLI2, GLI3, GNAS, GNPAT, GNS, GPC6, HDAC8, HESX1, HOXA13, HOXD13, HRAS, HSPG2, IFITM5, IFT43, IFT80, IFT122, IFT140, IFT172, IGF1, IGF1R, IGF2, IGFALS, IHH, IMPAD1, INPPL1, INSR, IRS1, KAT6B, KIF7, KIF22, KMT2A, KRAS, LARP7, LBR, LEMD3, LHX3, LHX4, LIFR, LMNA, LMX1B, LONP1, LRP4, LRP5, LTBP2, LTBP3, LZTR1, MAFB, MAP2K1, MAP2K2, MATN3, MBTPS2, MESP2, MGP, MMP2, MMP9, MMP13, MSX2, MYCN, NAGLU, NANS, NEK1, NF1, NFIX, NIPBL, NKX3-2, NOG, NOTCH1, NOTCH2, NPR2, NRAS, NSD1, NSDHL, OBSL1, ORC1, ORC4, ORC6, OSTM1, OTX2, P3H1, PALB2, PAPSS2, PCNT, PCYT1A, PDE4D, PEX7, PEX14, PEX19, PGM3, PHEX, PIK3CA, PITX2, PLOD2, PLS3, POC1A, POLR1C, POLR1D, POR, POU1F1, PPIB, PRKAR1A, PROP1, PTDSS1, PTH1R, PTHLH, PTPN11, PYCR1, RAB33B, RAD21, RAD51C, RAF1, RASA2, RBBP8, RBM8A, RBPJ, RECQL4, RIT1, RMRP, RNU4ATAC, ROR2, RRAS, RTTN, RUNX2, SALL1, SALL4, SBDS, SEC24D, SERPINF1, SERPINH1, SETBP1, SF3B4, SH3BP2, SH3PXD2B, SHOC2, SHOX, SKI, SLC26A2, SLC29A3, SLC34A3, SLC35D1, SLC39A13, SLCO2A1, SLX4, SMAD3, SMAD4, SMAD6, SMARCAL1, SMC1A, SMC3, SNX10, SOS1, SOST, SOX2, SOX3, SOX9, SP7, SPARC, SRCAP, STAMBP, STAT5B, TBX3, TBX4, TBX5, TBX6, TBX15, TBX19, TCF12, TCIRG1, TCOF1, TCTN3, TGFB1, TGFB2, TGFB3, TGFBR1, TGFBR2, TMEM38B, TNFRSF11A, TNFRSF11B, TNFSF11, TP63, TRAPPC2, TRIM37, TRIP11, TRPC3, TRPS1, TRPV4, TTC21B, TWIST1, TYROBP, VDR, VIPAS39, WDR19, WDR34, WDR35, WDR60, WISP3, WNT1, WNT5A, WNT7A, XRCC2, XRCC4, XYLT1

Seznam genů analyzovaných v panelu pro familiární hypercholesterolémie (11 genů a vybrané SNP)

Seznam vyšetřovaných genů:
ABCG5, *ABCG8*, *APOB*, *APOE*, *LDLR*, *LDLRAP1*, *LIPA*, *LIPC*, *LIPG*, *PCSK9*, *STAP1*
Seznam vyšetřovaných polymorfismů k vypočtení LDL-C skóre:
ABCG8 – rs4299376, *APOB* – rs1367117, *APOE* – rs429358, *APOE* – rs7412, *CELSR2* – rs629301, *HFE* – rs1800562, *LDLR* – rs6511720, *MYLIP* – rs3757354, *NYNRIN* – rs8017377, *PCSK9* – rs2479409, *SLC22A1* – rs1564348, *ST3GAL4* – rs11220462
Seznam vyšetřovaných polymorfismů asociovaných se statinovou léčbou:
LPA – rs10455872, *SLCO1B1* – rs4149056