



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, CTD1P1, 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,



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Formulář | F-KPDPM-DMP-S-16 | strana 2 z 3 | verze 1

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TAB2, TAZ, TBX20, TBX3, TBX5, TCAP, TECRL, TGFB2, TGFB3, TGFB1, TGFB2, 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, EFN1, 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, OSL1, 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, PTSS1, 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, SMARCA1, 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, TGFB1, TGFB2, 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 hypercholesterolemie (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



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Seznam genů analyzovaných v panelu pro dědičná onemocnění zadního segmentu oka (314 genů)

ABCA4, ABCC6, ABCD1, ABHD12, ACO2, ADAM9, ADAMTS18, ADGRV1, ADIPOR1, AGBL5, AHI1, AIPL1, ALMS1, AMACR, ARHGEF18, ARL13B, ARL2BP, ARL3, ARL6, ARMC9, ARR3, ARSG, ATF6, ATOH7, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C1QTNF5, CFAP410, PCARE, CPLANE1, CFAP418, CA4, CABP4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CDH23, CDH3, CDHR1, CEP104, CEP120, CEP164, CEP19, CEP250, CEP290, CEP41, CEP78, CEP83, CERKL, CHM, CIB2, CISD2, CLN3, CLN5, CLN6, CLN8, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL11A2, COL18A1, COL2A1, COL9A1, COL9A2, COL9A3, COQ2, CPE, CRB1, CRX, CSPP1, CTC1, CTNNA1, CTNNB1, CTSD, CWC27, CYP4V2, WHRN, DHDDS, DHX38, DNAJC5, DRAM2, DTHD1, DYNC2H1, EFEMP1, ELOVL4, EMC1, ESPN, EXOSC2, EYS, FAM161A, FDXR, FLVCR1, FRMD7, FZD4, GNAT1, GNAT2, GNB3, GNPTG, GPR143, GPR179, GRK1, GRM6, GUCA1A, GUCY2D, HARS, HGSNAT, HK1, HMX1, IDH3A, IDH3B, IFT140, IFT172, IFT27, IFT81, IMPDH1, IMPG1, IMPG2, INPP5E, INVS, IQCB1, CRPPA, JAG1, KCNJ13, KCNV2, KATNIP, KIAA0586, KIAA0753, KIAA1549, KIF11, KIF7, KIZ, KLHL7, LAMA1, LCA5, LRAT, LRIT3, LRP2, LRP5, LZTFL1, MAK, MERTK, MFN2, MFRP, MFSD8, MKKS, MKS1, MMACHC, MTPP, MVK, MYO7A, NAGLU, NDP, NEK2, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NR2F1, NRL, NYX, OAT, OCA2, OFD1, OPA1, OPA3, OPN1SW, OTX2, P3H2, PANK2, PAX2, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6D, PDE6G, PDE6H, PDSS1, PDSS2, PDZD7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, PISD, PITPNM3, PLA2G5, PLK4, PNPLA6, POC1B, POMGNT1, PPT1, PRCD, PRDM13, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, RAB28, RAX2, RBP3, RBP4, RCBTB1, RD3, RDH11, RDH12, RDH5, REEP6, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, ROM1, RP1, RP1L1, RP2, RPE65, RPGR, RPGRIP1, RPGRIP1L, RS1, RTN4IP1, SAG, SAMD11, SCAPER, SCLT1, SDCCAG8, SEMA4A, SGSH, SLC24A1, SLC25A46, SLC45A2, SLC7A14, SNRNP200, SPATA7, SPP2, SRD5A3, TCTN1, TCTN2, TCTN3, TEAD1, TIMM8A, TIMP3, TMEM107, TMEM126A, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TOPORS, TPP1, TRAF3IP1, TREX1, TRIM32, TRPM1, TSPAN12, TTC21B, TTC8, TLL5, TTPA, TUB, TUBB4B, TUBGCP4, TUBGCP6, TULP1, TYR, TYRP1, USH1C, USH1G, USH2A, VCAN, VPS13B, WDPCP, WDR19, WFS1, YME1L1, ZNF408, ZNF423, ZNF513

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