

Metodika – panel genů:

Cílená masivní paralelní sekvenace (KAPA kit, Roche; MGI) exonů a přilehlých intronových oblastí následujících genů:

AARS1, ABAT, ABCA2, ACTL6B, ADAM22, ADAR, ADARBI, ADGRG1, ADGRV1, ADNP, ADPRS, ADSL, AFF3, AFG2A, AGO1, AIMP1, AIMP2, AKT3, ALDH4A1, ALDH5A1, ALDH7A1, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALKBH8, ALPL, AMPD2, AMT, ANKRD11, APIG1, AP2M1, AP3B2, AP4S1, APC2, ARF1, ARFGEF1, ARFGEF2, ARG1, ARHGEF9, ARID1B, ARV1, ARX, ASAH1, ASH1L, ASNS, ASPA, ASXL3, ATAD1, ATN1, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP2B1, ATP6AP2, ATP6V0A1, ATP6V0A2, ATP6V1A, ATP7A, ATRX, BCORL1, BCS1L, BOLA3, BRAT1, BSCL2, BTB, C12orf57, CACNA1A, CACNA1B, CACNA1C, CACNA1D, CACNA1E, CACNA1G, CACNA1H, CACNA1I, CACNA2D1, CACNA2D2, CACNB4, CAD, CAMK2B, CARS2, CASK, CASR, CC2D2A, CCDC88A, CDC42BPB, CDK19, CDKL5, CELF2, CEP85L, CERS1, CIC, CLCN2, CLCN3, CLCN4, CLCN6, CLN3, CLN5, CLN6, CLN8, CLTC, CNKSR2, CNNM2, CNPY3, CNTN2, CNTN4, CNTNAP2, COG4, COG6, COG7, COG8, COL18A1, COL4A1, COL4A2, COQ2, COQ4, COQ8A, COQ9, CPA6, CPLX1, CPSF3, CSNK2A1, CSNK2B, CSTB, CTNNA2, CTNND2, CTSD, CTSF, CUL3, CUL4B, CUX2, CYFIP2, D2HGDH, DCX, DDC, DDX3X, DEAF1, DEGS1, DENND5A, DEPDC5, DHDDS, DHPS, DHX30, DIAPH1, DLG1, DLL1, DMXL2, DNAJC5, DNMI, DNMI1, DOCK7, DOLK, DPAGT1, DPM1, DPM2, DPYD, DTYMK, DYNC1H1, DYRK1A, EARS2, EEF1A2, EFHC1, EFTUD2, EHMT1, EIF2AK2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, EIF3F, EMC10, EML1, EMX2, EPG5, EPM2A, ETHE1, EXOC7, EXT2, FAM50A, FARS2, FBXL4, FBXO11, FBXO28, FCSK, FGF12, FGF13, FLNA, FOLR1, FOXG1, FRRS1L, FUCA1, FUT8, GABBR2, GABRA1, GABRA2, GABRA5, GABRA6, GABRB1, GABRB2, GABRB3, GABRD, GABRG2, GAD1, GAL, GALT, GALNT2, GAMT, GATAD2B, GBA1, GCSH, GFAP, GLB1, GLDC, GLRA1, GLRA2, GLRB, GLS, GLUD1, GLUL, GM2A, GNAO1, GNB1, GNB2, GNB5, GOSR2, GOT2, GPAAL, GPHN, GRIA2, GRIA3, GRIA4, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRM7, GRN, GTPBP2, GUF1, HACE1, HCN1, HCN2, HECTD4, HECW2, HEPACAM, HERC2, HEXA, HEXB, HID1, HNRNP2, HNRNPR, HNRNPU, HOXA1, HPDL, HSPD1, HTRA1, CHD2, CHD5, CHD8, CHKA, CHRNA2, CHRNA4, CHRNB2, IER3IP1, IFIH1, IQSEC2, IRF2BPL, ITPA, KANSL1, KARS1, KAT5, KAT8, KATNB1, KCNA1, KCNA2, KCNB1, KCNC1, KCNC2, KCNH1, KCNH5, KCNJ10, KCNJ11, KCNK4, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, KCTD3, KCTD7, KDM5C, KIF1A, KIF2A, KIF5C, KMT2E, KMT5B, KPTN, LGI1, LIAS, LIPT2, LMAN2L, LMBRD2, LMNB1, LMNB2, LNPBK, MACF1, MADD, MAF, MAST1, MBD5, MBOAT7, MDH2, MECP2, MED12, MED13L, MED17, MED27, MEF2C, MFF, MFSD8, MINPP1, MLC1, MOCS1, MOCS2, MOGS, MPDU1, MTHFS, MTOR, MYH1, MYT1L, NAA15, NAC1, NAPB, NARS1, NBEA, NCDN, NDE1, NDUFA1, NDUFA10, NDUFAF2, NDUFAF5, NDUFS4, NDUFS8, NDUFV1, NECAP1, NEDD4L, NEUROD2, NEXMIF, NGLY1, NHLRC1, NLGN3, NOTCH3, NPRL2, NPRL3, NR4A2, NRROS, NRXN1, NSD1, NSDHL, NSRP1, NTRK2, NUP214, NUS1, OCLN, OGDHL, OPHN1, OTUD6B, OXR1, P4HTM, PACS1, PACS2, PAFAH1B1, PAK1, PAK3, PARS2, PCDH12, PCDH19, PCDHGC4, PCLO, PDHA1, PDHX, PGM2L1, PHACTR1, PHF6, PHGDH, PIDD1, PIGA, PIGB, PIGC, PIGG, PIGH, PIGK, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGU, PIGV, PIGW, PIK3R2, PLA2G6, PLAA, PLCB1, PLPBP, PMM2, PNKD, PNKP, PNPO, POGZ, POLG, PPFIBP1, PPIL1, PPP2CA, PPP2R5D, PPP3CA, PPT1, PRDM8, PRICKLE1, PRICKLE2, PRIMA1, PRMT7, PRODH, PRRT2, PSAP, PTCH1, PTPN23, PTS, PURA, QARS1, RAB11A, RAB11B, RAB18, RAB39B, RAB3GAP1, RAB3GAP2, RALA, RALGAP1, RANBP2, RARS1, RARS2, RELN, RFT1, RHOTB2, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF13, RNF2, ROGDI, RORA, RORB, RUSC2, SAMD12, SAMHD1, SARS1, SATB1, SATB2, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCN9A, SEMA6B, SEPSECS, SERPINI1, SETBP1, SETD1A, SETD1B, SETD2, SETD5, SGCE, SGSH, SHANK3, SHH, SIK1, SIX3, SLC12A5, SLC13A5, SLC16A2, SLC19A3, SLC1A2, SLC1A4, SLC25A12, SLC25A22, SLC2A1, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC38A3, SLC39A8, SLC45A1, SLC46A1, SLC6A1, SLC6A5, SLC6A8, SLC7A6OS, SLC9A6, SMARCA2, SMARCC2, SMC1A, SMS, SNAP25, SNIP1, SPTAN1, SPTBN1, SPTBN4, SRPX2, ST3GAL3, ST3GAL5, STAG1, STAMBP, STARD7, STIL, STRADA, STX1B, STXBP1, SUCLA2, SUOX, SURF1, SYN1, SYNGAP1, SYNJI, SYP, SZT2, TAF8, TANC2, TANGO2, TBC1D20, TBC1D24, TBC1D2B, TBCD, TBCK, TBLIXR1, TBR1, TCF4, TET3, TFE3, TGIF1, TIAM1, TMEM106B, TMEM222, TMTC3, TMX2, TNPO2, TPBK, TPPI1, TRAK1, TRAPPC12, TRAPPC4, TRAPPC6B, TREX1, TRIM8, TRIP12, TRPM3, TRPM6, TRRAP, TSC1, TSC2, TSEN15, TSEN2, TSEN34, TSEN54, TUBA1A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP2, TWNK, UBA5, UBE2A, UBE3A, UBR7, UFM1, UFSP2, UGDH, UGP2, UNC80, USP18,

USP7, VAMP2, VARS1, VLDLR, VPS11, VPS50, VPS53, WARS2, WASF1, WDR37, WDR45, WDR45B, WDR62, WDR73, WWOX, YIF1B, YIPF5, YWHAG, ZBTB18, ZDHHC9, ZEB2, ZIC2, ZNF142, ZNF335