

METODIKA – PANEL GENŮ:

Cílená masivní paralelní sekvenace (KAPA HyperCap, Roche; Illumina) exonů a přilehlých intronových oblastí genů spojených s následujícími skupinami onemocnění:
AARS1, ABAT, ACTL6B, ADAM22, ADAR, ADGRV1, ADNP, ADSL, ALDH4A1, ALDH5A1, ALDH7A1, ALG13, AMT, AP3B2, AP4S1, ARHGEF9, ARID1B, ARV1, ARX, ASAHI, ASH1L, ASXL3, ATAD1, ATN1, ATP1A1, ATP1A2, ATP1A3, ATP1A2, ATP6AP2, ATP6VIA, ATP13A2, BRAT1, C10orf2, C12orf57, CACNA1A, CACNA1B, CACNA1D, CACNA1E, CACNA1H, CACNA2D2, CACNB4, CAD, CASK, CASR, CDKL5, CERS1, CLCN2, CLCN4, CLN3, CLN5, CLN6, CLN8, CLTC, CNKS2, CNPY3, CNTN2, CNTN4, CNTNAP2, COQ8A, CPA6, CSNK2B, CSTB, CTNND2, CTSD, CTSF, CYFIP2, DCX, DENND5A, DEPDC5, DHDDS, DLG1, DNAJC5, DNAJC6, DNM1, DNM1L, DOCK7, DYRK1A, EEF1A2, EFHC1, EHMT1, EPM2A, FARS2, FBXO7, FGF12, FOLR1, FOXG1, FRRS1L, GABBR2, GABRA1, GABRA6, GABRB2, GABRB3, GABRD, GABRG2, GAL, GAMT, GATM, GBA, GCSH, GIGYF2, GLDC, GLRA1, GLRB, GNAO1, GOSR2, GPA1, GPHN, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRN, HCN1, HCN2, HECW2, HNRNPU, CHD2, CHD8, CHCHD2, CHRNA2, CHRNA4, CHRN2, IER3IP1, IQSEC2, IRF2BPL, ITPA, KANSL1, KCNA1, KCNA2, KCNB1, KCNC1, KCNJ10, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, KCND7, LGI1, LIAS, LMNB2, LRRK2, MBD5, MDH2, MECP2, MED13L, MEF2C, MFSD8, MOCS1, MOCS2, MTOR, MYH1, MYT1L, NAA15, NBEA, NECAP1, NEUROD2, NEXMIF, NGLY1, NHLRC1, NLGN3, NOTCH3, NPRL2, NPRL3, NRXN1, NTRK2, NUS1, OPHN1, PANK2, PARK7, PARS2, PCDH19, PHACTR1, PHF6, PIGA, PIGB, PIGC, PIGG, PIGK, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGW, PINK1, PLA2G6, PLCB1, PLPBP, PNKD, PNKP, PNPO, POGZ, POLG, PPP3CA, PPT1, PRDM8, PRICKLE1, PRICKLE2, PRIMA1, PRKN, PRODH, PRRT2, PTRHD1, PURA, QARS, RAB11A, RAB39B, RANBP2, RELN, RHOBTB2, RNASEH2A, RNASEH2B, RNASEH2C, ROGDI, RORB, SAMHD1, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCN9A, SERPINI1, SETD2, SETD5, SGCE, SHANK2, SHANK3, SIK1, SLC12A5, SLC13A5, SLC19A3, SLC25A12, SLC25A22, SLC2A1, SLC35A2, SLC46A1, SLC6A1, SLC6A5, SLC6A8, SLC9A6, SMC1A, SNAP25, SNCA, SPATA5, SPTAN1, SRPX2, ST3GAL3, ST3GAL5, STRADA, STX1B, STXBP1, SYN1, SYNGAP1, SYNJ1, SZT2, TBC1D24, TBCK, TBLIXR1, TBR1, TCF4, TPK1, TPP1, TRAK1, TREX1, TRIM8, TRIP12, TRPM6, TSC1, TSC2, UBA5, UBE3A, USP7, VPS13C, VPS35, WDR45, WWOX, ZEB2