

Vyšetřované panely genů (uplatnění FRA)**NGS panely 20 až 100 genů:**

- NGS panel Hereditární nádorové syndromy (*APC, ATM, BAP1, BARD1, BLM, BMP1A, BRCA1, BRCA2, BRIP1, CDHI, CDK4, CDKN2A, CTNNA1, DICER1, EPCAM, ERCC2, ERCC3, FANCC, FANCM, FH, FLCN, GREM1, HOXB13, CHEK2, KIT, MAX, MEN1, MET, MITF, MLH1, MLH3, MRE11A, MSH2, MSH6, MUTYH, NBN, NF1, NF2, PALB2, PMS2, POLD1, POLE, PRKAR1A, PTEN, PTCH1, RAD50, RAD51C, RAD51D, RAD52, RB1, RECQL, RECQL4, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLX4, SMAD4, SMARCA4, STK11, SUFU, TMMEM127, TP53, TSC1, TSC2, VHL, WRN, WT1*)

NGS panely nad 100 genů:

- NGS panel Metabolické poruchy (*ACADM, ACADS, ACADSB, ACADVL, AKAP9, AKR1C3, ANK2, APOA1, APOA2, APOA4, APOA5, APOB, APOC1, APOC2, APOC3, APOC4, APOD, APOE, ARG1, ARSB, ASS1, ATP7B, BCKDHA, BCKDHB, BTBD9, C3, C3P1, CACNA1C, CALM1, CALM2, CAV3, CD46, CF46P1, CFB, CFH, CFHR5, CFI, CFTR, CPT1A, CPT2, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP19A1, CYP21A1P, CYP21A2, DBT, DHCR7 - SLOsy, DUOX2, DUOX2A, EPHX2, GALNS, GCDH, GCHI - HPABH4B, GLBI, GNS, GUSB, HADHA, HGSNAT, HSD17B3, HSD3B2, HYAL1, IDS, IDUA, IVD, IYD, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, LDLR, LDLRAP1, LPA, LPAL2, LPL, LRPAP1, MMACHC, MTHFR, NAGLU, NKX2-5, OTC, P4HA2, PAH - PKU, PAX8, PCBD1 - HPABH4D, PCSK9, POR, PRIMPOL, CCDC111, PTS - HPABH4A, QDPR - HPABH4C, SCN4B, SCN5A, SCO2, SERPINA1, SGSH, SLC25A20, SLC26A4, SLC39A5, SLC5A5, SNTA1, SRD5A2, TG, THBD, THRA, TPO, TSHB, TSHR, UGT1A1, USF1, USF1, ZNF644*),
- NGS panel Dědičné poruchy sluchu a Dědičné poruchy zraku (*ABCA4, ABCB6, ACO2, ACTG1, ADCY1, ADGRV1, AGBL1, AGBL5, AIFM1, AIPL1, ALDH1A3, ARHGAP18, ARL6, ASB10, ATF6, ATP2B2, BBOX1, BBS2, BDNF, BEST1, BEST2, BEST3, BEST4, BFSP2, BSND, C10orf11, C10orf2, C2orf71, C8orf37, CA4, CABP2, CACNA1F, CANT1, CCDC50, CD164, CDC14A, CDH23, CDHR1, CEACAM16, CEP290, CERKL, CIB2, CLDN14, CLIC5, CLPP, CLRNI, CNGA1, CNGA3, CNGB1, CNGB3, COCH, COL11A2, COL4A3, COL4A4, COL4A5, COL4A6, COL8A2, CPAMD8, CRB1, CRX, CRYAA, CRYAB, CRYBB2, CRYGD, CRYM, CYP1B1, CYP4V2, DCDC2, DFNA5, DFN59, DHDDS, DIABLO, DIAPH1, DIAPH3, DNMI1, EDN3, EDNRB, ELMOD3, ELOVL4, EPS8, ESPN, ESRRB, EYA1, EYA4, EYS, FAM161A, FAM65B, FOXC1, FOXE3, FOXI1, FSCN2, GDF3, GDF6, GIPC3, GJA3, GJA8, GJB2, GJB3, GJB6, GNAT2, GPR143, GRHL2, GRXCR1, GRXCR2, GUCA1B, GUCY2D, HARS, HARS2, HERC2, HGF, HGSNAT, HK1, HOMER2, HSD17B4, CHD7, CHN1, IDH3B, IFT140, IFT172, IFT43, ILDR1, IMPDH1, IMPG1, IMPG2, ITPR1, KARS, KCNE1, KCNJ10, KCNJ13, KCNQ1, KCNQ4, KITLG, KIZ, KLHL7, LARS2, LCA5, LHFPL5, LMX1B, LOXHD1, LOXL1, LRAT, LRTOMT*),

Vyšetřované panely genů (uplatnění FRA)

LTBP2, MAF, MAFB, MAK, MARVELD2, MC1R, MCM2, MERTK, MET, MFRP, MIR122, MIR124-1, MIR147A, MIR147B, MIR16-1, MIR181A2, MIR181B1, MIR181D, MIR195, MIR204, MIR211, MIR24-1, MIR24-2, MIR26A1, MIR26B, MIR29B1, MIR29B2, MIR301A, MIR301B, MIR3074, MIR323A, MIR323B, MIR346, MIR3591, MIR372, MIR373, MIR378A, MIR378C, MIR378G, MIR383, MIR4499, MIR450A1, MIR450A2, MIR450B, MIR499A, MIR499B, MIR520A, MIR520D, MIR520G, MIR544A, MIR877, MIR892C, MIR92B, MIR96, MITF, MPPED2, MSRB3, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, MYOC, NDP, NEK2, NHS, NMNAT1, NR2E3, NRL, NTF4, OCA2, OFD1, OPA1, OPA3, OPN1LW, OPN1MW, OPN1SW, OPTN, OSBPL2, OTOA, OTOF, OTOG, OTOGL, OTX2, P2RX2, PAUPAR, PAX3, PAX6, PCDH15, PDE6A, PDE6B, PDE6C, PDE6G, PDE6H, PDZD7, PITX2, PITX3, PNPT1, POMGNT1, POU3F4, POU4F3, PRCD, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, PRRG4, PRSS56, PTPRQ, PXDN, RAX, RBP3, RBP4, RD3, RDH12, RDH5, RDX, REEP6, RGR, RHO, RLBP1, ROM1, RP1, RP2, RP9, RPE65, RPGR, RPGRIP1, RTN4IP1, SIPR2, SAG, SANS, SBF2, SEMA3E, SEMA4A, SERPINB6, SHH, SIX1, SIX5, SLC17A8, SLC24A5, SLC26A4, SLC26A5, SLC45A2, SLC4A11, SLC7A14, SMPX, SNAI2, SNRNP200, SOX10, SPATA7, STRA6, STRC, SYNE4, TBC1D24, TBK1, TCF4, TECTA, TEK, TENM3, TGFB1, TMC1, TMEM126A, TMIE, TMPRSS3, TNC, TOPORS, TPRN, TRIM44, TRIOBP, TSPEAR, TTC8, TULP1, TYR, TYRP1, USH1C, USH2A, VSX2, WDR36, WFS1, WHRN, YME1L1, ZEB1, ZNF408, ZNF513)

Vypracoval: Mgr. Václav Palata

Podpis: 

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