

**Aktuální seznam činností prováděných v rámci flexibilního rozsahu akreditace  
Ústav klinické a molekulární patologie a lékařské genetiky  
Oddělení lékařské genetiky**

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**Postup č. 1/816: Přímá sekvence DNA lidského genomu dle Sangera**

Geny: *LDLR, ATP7B, GJB2*

**Postup č. 2/816: Fragmentační analýza lidského genomu pomocí kapilární elektroforézy**

Aneuploidie chromozomů 13, 18, 21, X a Y

Mikrodelece na chromozomu Y (*AZFa, AZFb, AZFc*), *SRY*

Syndrom fragilního X (analýza počtu repetice *CGG* v 5'UTR oblasti genu *FMR1*)

Chimerismus

Vyšetření mikrosatelitové instability (*MSI*)

**Postup č. 6/816: Analýza lidského genomu metodou masivně paralelního sekvenování**

NGS panel Alportův syndrom (*COL4A5, COL4A3, COL4A4, COL4A6, CFHR5, MYH9*)

NGS panel Hereditární nádorové syndromy a somatické varianty v nádorových tkáních (*ACD, AIP, AKT1, APC, AR, ATM, ATR, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRAF, BRCA1, BRCA2, BRF1, BRIP1, CARD11, CCND1, CDC73, CDH1, CDK4, CDKN1B, CDKN2A, CYLD, DICER1, DLST, DNMT3A, EGFR, EMSY, EPCAM, ERBB2, ERBB3, ERCC2, ERCC3, ERCC5, ETV6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAT1, FBXW7, FH, FLCN, GATA2, GREM1, HNF1A, HOXB13, HRAS, CHEK1, CHEK2, IDH1, IDH2, KIF1B, KIT, KLLN, KMT2A, KRAS, MAP2K1, MAX, MC1R, MEN1, MET, MITF, MLH1, MLH3, MN1, MRE11, MSH2, MSH3, MSH6, MSR1, MUTYH, NBN, NF1, NF2, NRAS, NRG1, NTHL1, PALB2, PARP1, PARP2, PARP3, PARP4, PBRM1, PDGFB, PDGFD, PDGFRA, PDGFRB, PIK3CA, PMS2, POLD1, POLE, POT1, PPP2R2A, PRKAR1A, PTEN, PTCH1, RAD50, RAD51, RAD51B, RAD51C, RAD51D, RAD52, RAD54L, RB1, RECQL, RECQL4, RET, REXO2, RNF43, RPS20, SDHA, SDHAF2, SDHB, SDHC, SDHD, SH2D1A, SH2D1A, SLC25A11, SLX4, SMAD4, SMARCA4, SMARCB1, SMARCE1, SRGAP1, SRY, STK11, SUFU, TERF2IP, TERT, TET2, TMEM127, TP53, TSC1, TSC2, VHL, WRN, WT1, XPA, XRCC2, XRCC3 (*hg38*)). Somatické varianty: *ACVR1, ALK, ARAF, ARID1A, ARID1B, ATRX, BRK1, CCDC153, CCNE1, CD79B, CDADC1, CDK12, CDK6, CDKN2B, COX6C, CTNNA1, CTNNB1, DDR2, ENG, ENOX1, ERG, ESR1, FAM124A, FANCD2OS, FGFR1, FGFR2, FGFR3, FOXL2, H3-C14 (H3), H3C2 (HIST1H3B), H3-3A, IL18, INA, INCA1, KEAP1, KIAA1549, MAD2L2, MAMLD1, MALT1, MTOR, MYB, MYBL1, MYC, MYCN, MYD88, MYH13, NTRK1, NTRK2, NTRK3, NUTM1, PABPC1, PABPC3, PAX8, PIK3R1, PML, PPARG, PPP2R1A, QKI, RAF1, RELA, RHOA, ROS1, RPAIN, SETD2, S1PR1, TACC1, TACC3, TBX21, THADA, TMPRSS2, TOE1, TOP2A, WSCD1, YAP1, ZFTA (C11orf95).**

NGS panel Mentální retardace a mikrocefalie (*AARS1, ABCA2, ACBD5, ACSL4, ACTB, ACTL6B, ADAR, ADARB1, ADAT3, ADNP, AFF2, AGA, AGTPBP1, AHDC1, AIFM1, AIMP1, AIMP2, AKT3, ALDH3A2, ALG1, ALG9, ALG11, ALG12, ALG13, ALG3, ALKBH8, AMPD2, ANK3, ANKLE2, ANKRD11, AP1S2, AP2M1, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, ARCN1, ARFGEF2, ARHGEF6, ARHGEF9, ARID1A, ARID1B, ARID2, ARNT2, ARSE, ARV1, ARX, ASH1L, ASNS, ASPM, ASXL1, ASXL3, ATAD3A, ATP6AP2, ATP6V1A, ATR, ATRIP, ATRX, AUTS2, B3GALNT2, B3GNT1, B4GAT1, BCAP31, BCL11A, BCL11B, BPTF, BRAT1, BRD4, BRF1, BRPF1, BRWD3, BUB1B, C12orf4, CA8, CACNA1B, CACNA1G, CACNG2, CAMK2A, CAMK2B, CAMK2G, CAMTA1, CARS1, CASK, CC2D1A, CCDC22, CCDC32, CCDC47, CCDC88C, CCND2, CD96, CDC45, CDC6, CDH15, CDK16, CDK5, CDK5RAP2, CDK6, CDKL5, CDON, CDT1, CENPE, CENPF, CENPJ, CENPT, CEP135, CEP152, CEP63, CEP85L, CFAP43, CHD7, CIC, CILK1, CIT, CKAP2L, CLCN4, CLIC2, CLP1, CLTC, CNKSR2, CNNM2, CNOT1, CNOT3, COASY, COG1, COG2, COG4, COG5, COG6, COG7, COL4A1, COL4A3BP, COPB2, COX7B, CRADD, CRBN, CREBBP, CRIPT, CSNK2A1, CTCF, CTNNA2, CTNNB1, CTU2, CUL4B, CUX1, CXorf56, CYFIP2, DAG1, DCPS, DCX, DDX11, DDX3X, DEAF1, DENND5A, DHCR24, DHCR7, DHX37, DIAPH1, DISP1, DLG3, DLG4, DLL1, DMXL2, DNA2, DNMT1L, DNMT3A, DONSON, DPAGT1, DPF2, DPM1, DPP6, DYNC1H1, DYNC1I2, DYRK1A, EBF3,*

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EBP, EDC3, EEF1A2, EFTUD2, EHMT1, EIF2S3, EIF3F, ELP2, EMC1, EML1, EMX2, EP300, EPB41L1, EPG5, ERCC1, ERCC2, ERCC3, ERLIN2, ETHE1, EXOC7, EXOSC3, EXOSC8, EXOSC9, EXT2, FAR1, FAM20C, FAM50A, FARSA, FARSB, FBXL3, FBXL4, FBXO31, FGD1, FGF8, FGFR1, FGFR2, FGFR3, FH, FKRP, FKTN, FLVCR2, FMN2, FMR1, FOXG1, FOXH1, FOXP1, FREM1, FRMD4A, FRMPD4, FTO, FTSJ1, FUT8, GABBR2, GABRB2, GAD1, GALNT2, GAS1, GATAD2B, GDI1, GEMIN4, GFAP, GLDC, GLI2, GLI3, GLYCTK, GMNN, GMPPA, GMPPB, GNAO1, GNB1, GOT2, GPC3, GPSM2, GPT2, GRIA3, GRIA4, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRM7, GSPT2, GTF2E2, GTF2H5, GTPBP2, HACE1, HCCS, HCFC1, HDAC6, HDAC8, HERC2, HHAT, HIVEP2, HMGB3, HNMT, HNRNPH2, HNRNPU, HPDL, HPRT1, HSD17B10, HS6ST2, HUWE1, HYLS1, CHAMP1, CHD2, CHD3, CHMP1A, IARS1, IER3IP1, IFIH1, IFT122, IGBP1, IGF1, IGF1R, IL1RAPL1, IMPA1, INPP5E, INTS8, IQSEC2, ISPD, KANSL1, KAT6A, KAT6B, KATNB1, KCNA4, KCNQ5, KCNT1, KDM5B, KDM5C, KIAA1109, KIF7, KIF11, KIF14, KIF1A, KIF2A, KIF4A, KIF5C, KIFBP, KIRREL3, KLF8, KLHL7, KLHL15, KMT2A, KMT2C, KMT2D, KMT2E, KMT5B, KNL1, KPTN, L1CAM, LAGE3, LAMB1, LARGE1, LARP7, LAS1L, LIG4, LINGO1, LINS1, LMAN2L, LMNB1, LMNB2, MAB21L1, MACF1, MAGT1, MAN1B1, MAN2B1, MAOA, MAP11, MAP1B, MAPRE2, MAST1, MBD5, MBOAT7, MCM5, MCPH1, MECP2, MED12, MED13, MED13L, MED17, MED23, MED25, MEF2C, MEIS2, METTL23, METTL5, MFF, MFSD2A, MID1, MID2, MIR17HG, MOCS1, MORC2, MPDZ, MPLKIP, MSL3, MSMO1, MTHFS, MTX2, MYCN, MYMK, MYO18B, MYT1L, NAA10, NAA15, NADK2, NALCN, NANS, NARS1, NBEA, NBN, NCAPD2, NCAPD3, NCAPH, NDE1, NDST1, NEXMIF, NGLY1, NHEJ1, NIN, NIPBL, NKAP, NLGN3, NLGN4X, NODAL, NONO, NRXN1, NSDHL, NSMCE2, NSUN2, NT5C2, NTNG2, NUP107, NUP133, NUP188, NUP37, NUS1, OCLN, OGT, OPHN1, ORC1, ORC4, ORC6, OSGEP, OTC, P4HB, PACS1, PAFAH1B1, PAK3, PARS2, PCDH12, PCGF2, PCLO, PCNT, PDE2A, PDHA1, PGAP1, PGAP2, PGAP3, PGK1, PHC1, PHF6, PHF8, PHGDH, PIGG, PIGN, PIGO, PIGV, PIGY, PIK3R2, PLAA, PLEKHG2, PLK4, PLP1, PMM2, PNKP, POC1A, POGZ, POLA1, POLR1A, POLR2A, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PORCN, PPP1R15B, PPP2CA, PPP2R1A, PPP2R5D, PQBP1, PRMT7, PRSS12, PRUNE1, PSAT1, PSMD12, PSPH, PTCH1, PTCHD1, PTPN23, PURA, PUS7, PYCR2, QARS, QRI1, RAB11B, RAB18, RAB39B, RAB3GAP1, RAB3GAP2, RAB40AL, RAC1, RAD21, RAD50, RARS1, RARS2, RBBP8, RBMX, RBM10, RELN, RFT1, RHOBTB2, RIC1, RLIM, RNASEH2A, RNASEH2B, RNASEH2C, RNF113A, RNF13, RNF125, RNU4ATAC, RPL10, RPS23, RPS6KA3, RSRC1, RTTN, RUSC2, RXYLT1, SAMHD1, SARS1, SASS6, SCN1A, SCN2A, SEC24D, SEPSECS, SCN3A, SEMA3E, SET, SETBP1, SETD5, SF3B4, SHANK3, SHH, SHROOM4, SIN3A, SIX3, SKI, SLC12A6, SLC16A2, SLC1A2, SLC1A4, SLC25A19, SLC2A1, SLC35A2, SLC35C1, SLC5A6, SLC6A17, SLC6A8, SLC9A6, SLC9A7, SMARCA2, SMARCA4, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SMC1A, SMC3, SMG9, SMPD4, SMS, SNAP29, SNRPB, SOBP, SON, SOX11, SOX3, SOX4, SOX5, SPATA5, SPOP, SPTAN1, SRCAP, SRPX2, SSR4, ST3GAL3, STAG1, STAG2, STAMBP, STIL, STRADA, STT3A, STT3B, STXBP1, SVBP, SYN1, SYNGAP1, SYP, TAF1, TAF13, TAF2, TAF6, TARS1, TASP1, TBC1D20, TBC1D23, TBCD, TBCK, TBL1XR1, TBR1, TBX1, TCF20, TCF4, TDP2, TECPR2, TECP, TELO2, TGIF1, THOC2, THOC6, TIMM8A, TLK2, TMCO1, TMEM165, TMTC3, TMX2, TNIK, TOE1, TOP3A, TP53RK, TPRKB, TRAP, TRAPPC12, TRAPPC4, TRAPPC6B, TRAPPC9, TREX1, TRIM71, TRIO, TRIP12, TRMT1, TRMT10A, TRRAP, TSEN15, TSEN2, TSEN34, TSEN54, TSPAN7, TTI2, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, TUBGCP2, TUBGCP4, TUBGCP6, TUSC3, UBA5, UBE2A, UBE3A, UBE3B, UBR1, UBR7, UBTF, UFC1, UFM1, UGP2, UNC80, UPB1, UPF3B, USP27X, USP9X, VARS1, VLDLR, VPS11, VPS13B, VPS51, VPS53, VRK1, WASHC4, WDFY3, WDR13, WDR37, WDR4, WDR45B, WDR62, WDR73, WDR81, WWOX, XRCC4, YIF1B, YY1, ZBTB11, ZBTB16, ZBTB18, ZBTB20, ZC3H14, ZC4H2, ZDHHC15, ZDHHC9, ZEB2, ZIC1, ZIC2, ZMYND11, ZNHIT3, ZNF148, ZNF292, ZNF335, ZNF711, ZNF81, ZSWIM6),

NGS panel Vrozená porucha sluchu (AARS1, ABCC1, ABHD5, ABHD12, ACOX1, ACTB, ACTG1, ACVR1, ADCY1, ADGRV1 (GPR98), AFG3L2, AIFM1, AK2, ALG9, ALG11, ALG12, ALMS1, AMMECR1, ANKH, AP1B1, AP1S1, ARSG, ASAH1, ATL1, ATOH1, ATP1A2, ATP1A3, ATP2B1, ATP2B2, ATP2B3, ATP6AP1, ATP6V0A4, ATP6V1B1, ATP6V1B2, ATP11A, ATRX, BCAP31, BCS1L, BDP1, BRAF, BSND, BTBD, C2orf71, CABP2, CACNA1D, CAPG, CASK, CATSPER2, CCDC50, CCNQ, CD151, CD164, CDC14A, CDC2L1, CDC42, CDC45L, CDH11, CDH23, CEACAM16, CEMIP,

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CENPP, CEP250, CEP78, CIB2, CISD2, CLCN7, CLCNKA, CLCNKB, CLDN9, CLDN14, CLIC5, CLPP, CLRN1 (USH3), CLRN2, CNBP, CNRIP1, COCH, COL1A1, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, COLEC11, COQ2, COQ6, CREB3L1, CRYM, DCAF17, DCDC2, DDX11, DCHS1, DHODH, DHX16, DIABLO, DIAPH1, DIAPH3, DLX5, DMXL2, DNAJC3, DNMT1, DSPP, EDN1, EDN3, EDNRB, EFEMP1, EFTUD2, ECHS1, ELMOD1, ELMOD3, EPG5, EPHA10, EPS8L2, ERAL1, ERCC3, ERCC6, ERCC8, ESPN, ESRP1, ESRRB, EXOSC2, EYA1, EYA4, FANCF, FAT4, FGF3, FGFR3, FIT2, FKBP14, FLNA, FLNB, FOXC1, FOXE1, FOXE3, FOXI1, FTO, GAB1, GATA2, GATA3, GBA1, GDF6, GGPS1, GIPC3, GJA1, GJB1, GJB2, GJB3, GJB6, GLA, GLI2, GLI3, GLIS3, GLYCTK, GMNN, GNAI3, GPC4, GPRASP2, GPSM2, GRAP, GREB1L, GRHL2, GRXCR1, GRXCR2, GSDME (DFNA5), HARS1, HARS2, HAS1, HGF, HOMER2, HOXA1, HOXA2, HOXB1, HSD17B4, HSPA9, HUWE1, CHD7, CHSY1, IARS1, IARS2, IDS, IDUA, IGF1, IL17RD, ILDR1, INF2, IRX5, ITGA6, ITM2B, JAG1, KARS1, KCNE1, KCNJ1, KCNJ10, KCNQ1, KCNQ4, KIT, KITLG, KMT2C, KMT2D, LARS2, LHFPL5, LHX3, LMX1A, LOXHD1, LRP2, LRTOMT, MAF, MAFB, MAN2B1, MAP1B, MAP3K7, MARVELD2, MASP1, MCM2, MCM3AP, MCM5, MED12, MET, MFN2, MGP, MICOS13, MINAR2, MITF, MN1, MPZ, MPZL2, MRPS7, MSRB3, MTO1, MYCN, MYH14, MYH9, MYO15A, MYO1A, MYO1C, MYO3A, MYO6, MYO7A, NARS2, NDP, NDRG1, NEBL, NEFL, NEUROG1, NLRP12, NLRP3, NMNAT1, NOG, NOTCH2, NR2F1, OPA1, ORC1, ORC4, OSBPL2, OTOA, OTOF, OTOG, OTOGL, OXR1, P2RX2, PAX3, PBX1, PCDH15, PCGF2, PDE1C, PDGFRB, PDSS1, PDZD7, PEX1, PEX12, PEX6, PEX7, PHEX, PHF9, PHYH, PIGL, PISD, PJVK, PLCB4, PLCG2, PLEK, PLOD3, PLS1, PMP22, PNPT1, POLD1, POLR1A, POLR1B, POLR1C, POLR1D, POU3F4, POU4F3, PPIP5K2, PRDM5, PROSER1, PRPS1, PRRX1, PSMC3, PSMD12, PTEN, PTPN11, PTPRQ, PTRH2, RABGAP1, RAI1, RAI2, Rbm24, RCBTB1, RDX, REST, RFT1, RIPOR2, RMND1, ROR1, RPGR, RPS26, RPS6KA3, RRM2B, S1PR2, SACS, SALL1, SALL4, SCD5, SCML1, SCN9A, SDHD, SEMA3E, SERAC1, SERPINB6, SF3B2, SGPL1, SIX1, SIX5, SLC12A1, SLC12A2, SLC17A8, SLC19A2, SLC26A4, SLC26A5, SLC29A3, SLC33A1, SLC44A4, SLC4A1, SLC4A11, SLC52A2, SLC52A3 (C200RF54), SLC6A20, SLC9A1, SLITRK6, SMAD4, SMPX, SNAI2, SNAP29, SOST, SOX10, SOX2, SOX6, SOX9, SPATA5, SPATA5L1, SPECC1L, SPNS2, SPTBN4, SPTLC1, SQSTM1, SRP72, SSBP1, STAG2, STRC, SUCLA2, SUCLG1, SYNE4, TBC1D24, TBL1X, TBL1Y, TBX1, TCIRG1, TCOF1, TECTA, TFAM, TFAP2A, TGDS, THRB, TIMM8A, TIMMDC1, TLK2, TMC1, TMEM132E, TMIE, TMPRSS3, TNC, TNFRSF11A, TNFRSF11B, TPRN, TRIOBP, TRMT10C, TRPV3, TRRAP, TSHZ1, TSPEAR, TSR2, TTR, TUBB4B, TWIST2, TWNK, TXNL4A, TYMP, TYR, UBE2A, UBR1, USH1C, USH1G, USH2A, USP48, VAC14, VCPKMT, VPS13B, VPS33B, WAC, WBP2, WFS1, WHRN, XYLT2, ZNF462. Geny pro mitochondriální DNA: MTF, MTRNR1, MTTV, MTRNR2, MTTL1, MTND1, MTTI, MTTQ, MTTM, MTND2, MTTW, MTTA, MTTN, MTTC, MTTY, MTCO1, MTTTS1, MTTD, MTCO2, MTTK, MTATP8, MTATP6, MTCO3, MTTG, MTND3, MTTT, MTND4L, MTND4, MTHH, MTTTS2, MTTL2, MTND5, MTND6, MTTE, MTCYB, MTTT, MTTT),

**NGS panel Neurodegenerativní onemocnění**

(APOE, GBA, SOD1, OPTN, PFN1, FIG4, CSF1R, ABCA7, ANG, CHCHD10, DCTN1, ITM2B, ALS2, ATL1, HEXA, HEXB, SETX, WDR45, CRAT, GNAO1, REPS1, ATXN2, UBQLN2, SQSTM1, TUBA4A, SPAST, HNRNPA1, VAPB, NOTCH3, TYROBP, MATR3, SPG11, DCAF17, FTL, DNAJC12, VPS13C, CHCHD2, GIGYF2, FBXO7, RAB39B, SLC6A3, GLUD2, PRKRA, TAF1, TBP, GCH1, PNKD, SLC2A1, SPR, COL6A3, AUH, STUB1, GTPBP2, APTX, FA2H, KMT2B, TOR1A, THAP1, ATM, PRRT2, ANO3, TH, ATP1A3, ADCY5, DNMT1, PRNP, BCAP31, DPGFB, PDGFRB, SGCE, GNAL, TUBB4A, KIF1C, SLC30A10, SLC39A14, PCNA, PDYN, TMEM240, PUM1, DAB1, KCND3, ITPR1, MME, TRPC3, FAT2, ELOVL5, ELOVL4, GRM1, SPTBN2, FGF14, ATXN3, TTBK2, CACNA1G, EEF2, PLD3, KCNC3, TGM6, C19orf12, ATP7B, DDC, TIMM8A, HPRT1, KCTD17, PTS, KCNMA1, SLC18A2, COASY, PANK3, PANK2, ATP13A2, HPCA, CACNA1B, CIZ1, GCDH, MMUT, POLG, PCCB, PCCA, CP, RELN, HTT, ERBB4, ANXA11, DAO, SS18L1/KIAA0693, SIGMAR1, GLE1, PRPH, NEFH, LMNB1, NEK1, ERLIN1, KIF5A, TIA1, GNE, SNCA, LRRK2, PARK7/DJ1, PRKN/PARK2, VPS35, PINK1, SYNJ1, DNAJC13, DNAJC6, HTRA2, PLA2G6, EIF4G1, UCHL1, MAPT, GRN, VCP, FUS, CHMP2B, TARDBP, TBK1, C9orf72, PSEN1, PSEN2, APP, HTRA1, MYORG, SLC20A2, JAM2, XPR1, SCP2, REEP1, HSPD1, KIF1A, GJC2, TMEM63A, SLC33A1, WASHC5, UBAP1, ALDH18A1, ZFYVE27, BSCL2, NIPA1, SP

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G7, RTN2, CPT1C, PLP1, PSAP, CACNA1A, SNC1A, DRD2, VPS16, AFG3L2, AOPEP, EIF2AK2, VPS13A, CTC1, CYP2U1, NRROS, PDGFB, SNORD118, TREX1, USP18, RNF216, XK, PRKCG, SYN E1, TPP1, TTPA, TWNK, FXN, CACNB4, ANO10, CAMTA1, CAPN1, CYP27A1, DARS2, DNAJC5, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, MARS2, RNF170, SACS, SCN2A, SLC1A3, VAMP1, COL4A1, COLGALT1, CTSF, PPT1, AARS1, AARS2, ABCD1, ACOX1, ARSA, CLCN2, B4GALNT1, CYP7B1, HPDL, ZFYVE26, AP5Z1, VPS13D, VPS41, TTR, ERLIN2, SPART, ACP33, DDHD1, PNPLA6, GB A2, AP4B1, AP4M1, AP4E1, VPS37A, TFG, ENTPD1, NT5C2, MAG, FARS2, NFU1, IBA57, REEP2, NUP50, CCNF, SPTLC1, UNC13A, DNAJC7, GLT8D1, HNRNPA2B1, TAF15, EPHA4, SCFD1, GPX3, TNIP1, SLC9A8, JAKMIP3, NDUFC2, PDE10A, SNCB, NKX2-1),

NGS panel Vrozené kostní anomálie (AAAS, ABCC9, ABR, ACAN, ACOX1, ACP5, ACTA1, ACTB, ACTG1, ACVR1, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTSL2, ADCY6, ADGRE5, ADGRG6, AFF4, AGA, AGPS, AHDC1, AHI1, AIFM1, AIRE, AKT1, AKT3, ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, ALPL, ALX1, ALX3, ALX4, AMER1, ANAPC1, ANKH, ANKRD11, ANKRD35, ANO5, ANTXR1, ANTXR2, AP1S1, ARCN1, ARHGAP31, ARHGEF6, ARID1A, ARID1B, ARID2, ARL6, ARMC9, ARSB, ARSL/ARSE, ASCC1, ASXL1, ASXL2, ASXL3, ATAD1, ATP6AP1, ATP6AP2, ATP6V0A2, ATP7A, ATR, ATRIP, ATRX, B3GALT6, B3GAT3, B3GLCT, B4GALT1, B4GALT7, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9/PTHB1, BCOR, BGN, BHLHA9, BICD2, BLM/RECQL3, BMP1, BMP2, BMP4, BMP7, BMPER, BMPR1B, BPNT2/IMPAD1, BRAF, BRCA2, BRD4, BRIP1, BRWD3, BTK, C12ORF65, C15ORF41, C2CD3, C8ORF37, CA2, CACNA1C, CAD, CANT1, CASR, CBF, CBL, CC2D2A, CCBE1, CCDC115, CCDC22, CCDC47, CCDC8, CCN6/WISP3, CCND2, CCNK, CD40LG, CD96, CDAN1, CDC42, CDC45, CDC6, CDH1, CDH11, CDH3, CDKN1C, CDT1, CENPE, CENPF, CENPJ, CEP120, CEP152, CEP290, CEP41, CEP55, CEP63, CERT1/COL4A3BP, CFAP410, CFAP410/C21orf2, CFL2, CFTR, CIBAR1/FAM92A, CILK1, CKAP2L, CLCF1, CLCN5, CLCN7, CNOT2, CNTNAP1, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL27A1, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, COLEC10, COLEC11, COMP, COX4I2, COX5A, CPLANE1, CREB3L1, CREBBP, CRIM1, CRIPT, CRTAP, CSGALNACT1, CSPP1, CST6, CTNBN1, CTNS, CTSA, CTSC, CTSK, CTU2, CUL4B, CUL7, CWC27, CYP26B1, CYP27B1, CYP2R1, DCPS, DDR2, DDRGK1, DDX11, DDX59, DEAF1, DHCR24, DHCR7, DHODH, DCHS1, DICER1, DIS3L2, DLL3, DLL4, DLX1, DLX3, DLX5, DMP1, DNA2, DNAJC21, DNM2, DNMT3A, DOCK1, DOCK6, DOK7, DOLK, DONSON, DPAGT1, DPF2, DPH1, DPM1, DPM2, DPM3, DSE, DUOX2, DUOX2A, DVL1, DVL3, DYM, DYNC2H1, DYNC2LI1, DYT1/TOR1A, EBP, ECEL1, EDARADD, EDN1, EED, EFEMP2, EFL1, EFNA4, EFN1, EFTUD2, EIF2AK3, EIF4A3, ELMO2, ENPP1, EOGT, EP300, EPS15L1, ERBB3, ERCC1, ERCC4, ERCC5, ERCC6, ERCC8, ERF, ERGIC1, ESCO2, EVC, EVC2, EXOC6B, EXOSC2, EXOSC9, EXT1, EXT2, EXTL3, EZH2, FAM111A, FAM149B1, FAM20B, FAM20C, FAM58A/CCNQ, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FAT4, FBLN1, FBLN5, FBN1, FBN2, FBXL3, FBXO11, FBXW11, FCSK, FDFT1, FERMT1, FERMT3, FGD1, FGF10, FGF13, FGF16, FGF17, FGF23, FGF8, FGF9, FGFR1, FGFR2, FGFR3, FHL1, FIBP, FIG4, FKBP10, FLI1, FLNA, FLNB, FLVCR2, FN1, FOXE1, FRAS1, FREM1, FREM2, FTO, FUCA1, FUT8, FUZ, FZD2, GALNS, GALNT2, GALNT3, GATA6, GBA, GBE1, GDF3, GDF5, GDF6, GFM2, GH1, GHR, GHRHR, GHSR, GJA1, GJB2, GLB1, GLDN, GLE1, GLI1, GLI2, GLI3, GLIS3, GMNN, GNAS, GNB2, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GPC3, GPC4, GPC6, GPR101, GPX4, GPX7, GRHL2, GRHL3, GRIP1, GSC, GTF2E2, GUSB, GZF1, HAAO, HAPLN1/CRTL1, HDAC6, HDAC8, HERC1, HES7, HESX1, HGSNAT, HIST1H1E/H1-4, HMGA2, HMX2, HNRNPK, HOXA10, HOXA11, HOXA13, HOXD10, HOXD13, HOXD3, HPGD, HRAS, HSD17B4, HSPA9, HSPG2, HUWE1, HYAL1, HYL1, CHAT, CHD4, CHD7, CHKA, CHL1/CALL, CHRNA7, CHRNG, CHST11, CHST14, CHST3, CHSY1, CHUK, IARS2, IBA57, ICK/CILK1, IDH1, IDH2, IDS, IDUA, IFIH1, IFITM5, IFT122, IFT140, IFT172, IFT27, IFT43, IFT52, IFT57, IFT74, IFT80, IFT81, IGF1, IGF1R, IGF2, IGSF1, IHH, IKBKG, IL11RA, IL1RN, IL6ST, INPP5E, INPPL1, INSR, INTU, IQCE, IRF6, IRS4, IRX5, ISCA2, ITPR1, IYD, JAG1, KAT6A, KAT6B, KBTBD13, KCNH1, KCNJ2, KCNJ8, KCNK4, KCNMA1, KCNN3, KCNQ10T1, KCTD1, KDM1A, KDM6A, KDM6B, KIAA0586, KIAA0753, KIAA0825, KIAA1109, KIAA1279, KIF14, KIF1A, KIF22, KIF3B, KIF5C, KIF7, KL, KLHL40, KLHL41, KMT2A, KMT2C, KMT2D, KPTN, KRAS,







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SLC25A46, SLC2A1, SLC33A1, SLC37A3, SLC38A8, SLC39A5, SLC45A2 (MATP), SLC4A11, SLC4A4, SLC52A2, SLC6A6, SLC7A14, SLITRK6, SMCHD1, SMO, SMOC1, SNAI2, SNRNP200, SNX10, SNX3 (MCOPS8), SOX1, SOX10, SOX2, SOX3, SPATA7, SPG7 (PGN), SPP2, SPTLC1, SRD5A3, SSBP1, STAG2, STN1, STRA6, STS, STX3, SUCLA2, SUCLG1, TACO1, TACSTD2, TAT, TAX1BP3, TBC1D20, TBX22, TCF4, TCOF1, TDRD7, TEAD1, TEK, TENM3, TFAP2A, TFG, TGFBI, TIMM8A, TIMP3, TINF2, TK2, TLCD3B (FAM57B), TLR4 (ARMD10), TMEM126A, TMEM216, TMEM218, TMEM237, TMEM67, TMEM70, TMEM98, TMX3, TOP3A, TOPORS, TP63, TPP1, TRAF3IP1, TRAPPC11, TREX1, TRIM32, TRIM37, TRIM44, TRNT1, TRPM1, TSFM, TSPAN12, TTC8, TTLL5, TTPA, TUB, TUBA3D, TUBA8, TUBB3, TUBB4B, TUBGCP4, TUBGCP6, TULP1, TWNK (C10orf2), TYR, TYRP1, UBE3B, UBIAD1, UCHL1, UNC119, UNC45B, USH1C (PDZ73), USH1G (SANS), USH2A, USP45, VAX1, VCAN, VIM, VPS13B, VPS35L (C16orf62), VSX1, VSX2 (CHX10), WASHC5 (KIAA0196), WDPCP, WDR19 (KIAA1638, NPHP13), WDR36, WDR87, WFS1, WHRN (USH2D, DFNB31), WRN, WT1, XPNPEP2, XYLT2, YAP1, YME1L1, ZBTB20, ZEB1, ZFH4 (ZFH4), ZFYVE26, ZNF408, ZNF423, ZNF469, ZNF513, ZNF644, ZNHIT3, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY).

NGS panel Onemocnění ledvin a ciliopatií (ABCA3, ABCB11, ABCB4, ABCC2, ABCC6, ACE, ACTG2, ACTN4, ACVR2B, ADAMTS13, ADAMTS9, AFF4, AGT, AGTR1, AGTR2, AGXT, AHI1, AKR1D1, ALDOB, ALG5, ALG8, ALG9, ALMS1, AMACR, AMN, ANKFY1/KIAA1255, ANKS6, ANLN, ANOS1/KAL1, AP2S1, APOA1, APOL1, APRT, AQP2, AR, ARHGAP24, ARHGDI, ARL13B, ARL2BP, ARL3, ARL6, ARMC4, ARMC9, ATP1A1, ATP6AP2, ATP6V0A4, ATP6V1B1/ATP6B1, ATP8B1, AVIL, AVP, AVPR2, B2M, B9D1, B9D2, BAAT, BBIP1/BBS18, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9/PTHB1, BDNF, BICC1, BMP4, BMP7, BMPR1B, BNC2, BSND, C21orf59/CFAP298, C2CD3, C2orf71/PCARE, C3, C5orf42/CPLANE1, c8orf37/CFAP418, C8ORF38/NDUF6, CA12, CA2, CACNA1H, CAMKMT, CASR, CBY1, CC2D2A, CCDC103, CCDC114, CCDC141, CCDC28B, CCDC32, CCDC39, CCDC40, CCDC65, CCM2/C7orf22, CCNO, CD19, CD2AP, CD46/MCP, CD81, CDC151/ODAD3, CDC5L, CDC73, CELSR2, CELSR3, CENPF, CEP104, CEP120, CEP164, CEP19, CEP290/NPHP6/BBS14, CEP41, CEP55, CEP83, CFAP221/PCDP1, CFAP300, CFAP52/WDR16, CFAP53/CCDC11, CFB, CFC1/CRYPTIC, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CFTR, CLCN2, CLCN5, CLCNKA, CLCNKB, CLDN10, CLDN14, CLDN16, CLDN19, CNNM2, COL4A1, COL4A3, COL4A4, COL4A5, COL4A6, COQ2, COQ6, COQ8B/ADCK4, CPE, CR2, CRB2, CRELD1, CRIM1, CRKL, CSF2RA, CSF2RB, CSPP1, CTNS, CTU2, CUBN, CUL3, CYP11B1, CYP11B2, CYP24A1, CYP27A1, DAAM2, DACT1, DAW1, DCDC2, DEAF1, DGKE, DLC1, DLG5, DMP1, DNAAF1, DNAAF2/KTU, DNAAF3, DNAAF4/DYX1C1, DNAAF5, DNAH1, DNAH11, DNAH3, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB11, DNAJB13, DNAL1, DRC1, DSTYK, DYNC2H1, DYNC2LI1, DYRK1B, DZIP1L, EGF, EHHADH, EIF2S3, EMP2, ENPP1, EVC, EVC2, EXOC3L2, EYA1, FAH, FAM111A, FAM20A, FAM58A/CCNQ, FAN1, FAT1, FGA, FGF20, FGF23, FGF8, FGF9, FLCN, FN1, FOXC1, FOXE1, FOXF1, FOXH1, FOXI1, FOXJ1, FRAS1, FREM1, FREM2, FXD2, G6PC, GALNT3, GANAB, GAPVD1, GAS2L2, GAS8/GAS11/DRC4, GATA3, GATA6, GATM, GBE1, GCM2, GDF1, GDNF, GFRA1, GJA1, GLA, GLI3, GLIS2/NPHP7, GLIS3, GNA11, GNAS, GON7, GREB1L/KIAA1772/C18ORF6, GREM1, GRHRP, GRIP1, GSN, HAAO, HNF1A, HNF1B, HNF4A, HOGA1, HOXA13, HPRT1, HPSE2, HS2ST1, HSD11B2, HSD3B7, HTRA1, HYLS1, CHD1L, CHRM3, CHRNA3, ICK/CILK1, ICOS, IFIH1, IFT122, IFT140, IFT172, IFT27/BBS19, IFT43, IFT52, IFT74/CCDC2, IFT80, IFT81, IKZF1, IL21, ILK, IMPDH1, INF2/C14ORF173, INPP5E, INTU/KIAA1284, INVS/NPHP2, IQCB1/NPHP5, IRF2BP2, IRS4, ITGA3, ITGA8, ITGB4, ITS1N1, KANK1, KANK2/KIAA1518, KANK4, KAT6B, KCNA1, KCNJ1, KCNJ10, KCNJ16, KCNJ5, KIAA0556/KATNIP, KIAA0586, KIAA0753, KIAA0974/FAM149B1, KIDINS220, KIF14, KIF3B, KIF7, KIRREL1, KL, KLHL3, KRIT1/CCM1, KSR2, KYNU, LAGE3, LAMA5, LAMB2, LAS1L, LCA5, LEFTY2, LEP, LEPR, LIFR, LIN7C, LMX1B, LRBA, LRIG2, LRP2BP, LRP4, LRP5, LRRC56, LRRC6/DNAAF11, LRRC1, LYZ, LZTFL1/BBS17, MAGED2, MAGEL2, MAGI2/KIAA0705/AIP1,



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MAPKBP1, MC3R, MC4R, MCIDAS, MEN1, MKKS, MKS1, MMACHC, MMP21, MNS1, MOCOS, MS4A1, MUC1, MXRA5, MYH9, MYO1E, MYO5B, MYOCD, MYOG, MYRF, MYT1L, NADSYN1, NAT1, NCF1, NDN, NEK1, NEK10, NEK4, NEK8/NPHP9, NFKB1, NFKB2, NFU1, NKX2-1, NKX2-5, NME8, NNT, NODAL, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2/PDCN, NPNT, NR1H4, NR3C1, NR3C2, NRIP1, NTRK2, NUP107, NUP133, NUP160/NPHS19, NUP205, NUP37, NUP85, NUP93, OCRL, OFD1, OSGEP, PAX2, PAX8, PBX1, PCBD1, PCDH15, PCSK1, PCSK5, PDCD10, PDE2A, PDE3A, PDE6D, PDSS2, PGM2L1, PHEX, PHF6, PHIP, PIBF1, PIGA, PIGT, PIH1D3, PKD1, PKD1L1, PKD2, PKHD1, PLCE1, PLVAP, PLXND1, PMM2, PNPLA6, PODXL, POMC, PRAG1, PREPL, PRKCD, PRKCSH, PTH, PTPN22, PTPRO, RAB23, REN, RET, ROBO2, ROR2, RPE65, RPGR, RPGRIP1, RPGRIP1L/NPHP8, RRAGD, RSPH1, RSPH3, RSPH4A, RSPH9, SALL1, SALL4, SARS2, SCAPER, SCARB2, SCLT1, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SEC61A1, SEC63, SEMA3E, SFTPA1, SFTPB, SFTPC, SFTPD, SGPL1, SH2B1, SHROOM3, SIM1, SIX1, SIX2, SIX5, SLC12A1, SLC12A3, SLC1A1, SLC20A1, SLC22A12/URAT1, SLC25A13, SLC26A1, SLC26A4, SLC2A2, SLC2A9, SLC34A1, SLC34A3, SLC36A2, SLC36A4, SLC37A4, SLC3A1, SLC4A1, SLC4A4, SLC51A, SLC5A1, SLC5A2, SLC6A19, SLC6A20, SLC7A9, SLC9A3R1, SLIT2, SMAD4, SMARCA1, SMO, SOX17, SPAG1, SPEF2, SPRY2, STK36, STRA6, SUFU, TAC3, TBC1D1, TBC1D32, TBC1D8B, TBL1X, TBX18, TCTEX1D2/DYNLT2B, TCTN1/TECT1, TCTN2, TCTN3, TFAP2A, TH, THBD, THRA, THRB, TJP2, TMEM107, TMEM127, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TMEM256, TMEM260, TMEM67, TNFRSF13B, TNFRSF13C, TNS2, TOGARAM1, TOPORS, TP53RK, TP63, TPO, TPP2, TPRKB, TRAF3IP1, TRAP1, TRAPPC9, TRHR, TRIM32, TRIM8, TRPC6, TRPM6, TRPV6, TSC1, TSC2, TSHB, TSHR, TTC12, TTC21B, TTC25, TTC8, TTF1, TUB, TXNDC15/C5orf14, ULK4, UMOD, UPK3A, USF2, VHL, VIPAS39, VPS33B, WBP11, WDPCP/C2ORF86, WDR19, WDR34/DYNC2I2, WDR35, WDR4, WDR60/DYNC2I1, WDR72, WDR73, WNK1, WNK4, WNT11, WNT4, WNT5A, WNT9B, WT1, XDH, XPNPEP3, XPO5, YRDC, ZIC3, ZMYM2, ZMYND10, ZNF423),

NGS panel Obezita s časným nástupem (BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9/PTHB1, ALMS1, ARL6, CEP19, GNAS, CPE, KIDINS220, LEP, LEPR, MC4R, INPP5E, CEP290/NPHP6/BBS14, MKKS, MKS1, MYT1L, NTRK2, PCSK1, PGM2L1, PHF6, PHIP, POMC, SDCCAG8, SIM1, TTC8, DYRK1B, KSR2, SH2B1, TUB, MC3R, AFF4, WT1, WDPCP/C2ORF86, TRIM32, TBC1D8B, LZTFL1/BBS17, IFT172, CNM2, C8orf37/CFAP418, BBIP1/BBS18, BDNF, DEAF1, EIF2S3, IFT174, LAS1L, MAGEL2, NDN, SCAPER, TRAPPC9).

NGS panel Onemocnění a vady srdce (A2ML1, ABCA7, ABCC9, ABCG5, ABCG8, ACTA1, ACTA2, ACTC1, ACTN2, ACVR2B, ADAMTS2, ADAMTSL4, AEBP1, AKAP9, ALDH18A1, ALG10B, ALMS1, ALPK3, ANK2, ANKRD1, APOA1, APOA2, APOB, APOE, APP, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, B2M, B3GALT6, B4GALT7, BAG3, BGN, BMPR2, BRAF, C1R, C1S, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CALR3, CASQ2, CAV1, CAV3, CBL, CBS, CDH2, CFAP53/CCDC11, CFC1, CITED2, COL12A1, COL1A1, COL1A2, COL3A1, COL4A1, COL5A1, COL5A2, COX15, CRELD1, CRYAB, CSRP3, CST3, CTF1, CTNNA3, DES, DMD, DNAJC19, DOLK, DPP6, DSC2, DSE, DSG2, DSP, DTNA, EFEMP2/FBLN4, ELN, EMD, EMILIN1, EPG5, EVC, EVC2, EYA4, FBLN5, FBN1, FBN2, FGA, FHL1, FHL2, FHOD3, FKBP14, FKRP, FKTN, FLNA, FLNC, FOXE3, FOXF1, FOXH1, FREM1, GAA, GATA4, GATA5, GATA6, GATAD1, GDF1, GJA1, GJA5, GLA, GORAB, GPC3, GPD1L, GSN, GYG1, GYS1, HCN4, HFE, HRAS, CHST14, ILK, ITM2B, JAG1, JPH2, JUP, KAT6B, KCNA5, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, KLF10, KRAS, LAMA4, LAMP2, LDB3, LDLR, LDLRAP1, LEFTY2, LIPA, LMNA, LOX, LPL, LTBP1, LTBP2, LTBP3, LTBP4, LYZ, LZTR1, MAP2K1, MAP2K2, MAT2A, MED12, MFAP5, MIB1, MMP21, MNS1, MRAS, MURC, MYBPC3, MYH11, MYH6, MYH7, MYL2, MYL3, MYLK, MYLK2, MYO6, MYOM1, MYOZ2, MYPN, MYRF, NEBL, NEXN, NKX2-5, NKX2-6, NODAL, NOS1AP, NOTCH1, NPPA, NR2F2, NRAS, OSMR, P4HA1, PCSK9, PDLIM3, PGM1, PKD1L1, PKP2, PLN, PLOD1, PLXND1, PPA2, PPP1CB, PRDM16, PRDM5, PRDM6, PRKAG2, PRKAR1A, PRKG1, PRNP, PSEN1, PSEN2, PTPN11, PYCR1, RAF1, RANGRF, RASA2, RBM20, RIN2, RIT1, ROBO4, RRAS, RRAS2, RSPH3, RYR2, SAA1, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCO2, SDHA, SGCD, SHOC2, SKI, SLC25A4, SLC2A10, SLC39A13, SLC8A1, SLMAP, SMAD2,



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SMAD3, SMAD4, SMAD6, SNTA1, SORL1, SOS1, SOS2, SPRED1, STAP1, STRN, SYNE2, TAB2, TAF1A, TAZ, TBX1, TBX20, TBX3, TBX5, TCAP, TECRL, TFAP2B, TGFB2, TGFB3, TGFB3R1, TGFB3R2, THSD1, THSD4, TLL1, TMEM43, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TNXB, TOMM40, TPM1, TRDN, TREM2, TRPM4, TSPYL1, TTN, TTR, TXNRD2, VCL, ZFPM2, ZIC3, ZNF423, ZNF469),

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PAPP1521, PAPP1522, PAPP1523, PAPP1524, PAPP1525, P



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Holoprosencephaly (HPE), P196 TNNT2-BAG3, P197 KCNQ3, P201 CHARGE, P211 HSP region, P213 HSP mix-2, P214 COL2A1, P216 GHD, P219 PAX6, P221 LCA mix1, P222 LCA mix2, P225 PTEN, P229 OPA1, P232 FGD1, P233 MID1, P235 Retinitis, P241 MODY mix1, P259 RPS6KA3, P260 PALB2-RAD50-RAD51C-RAD51D, P266 CLCNKB, P272 COL1A2, P285 LRP5, P295 SPRED1, P298 BRAF-HRAS-KRAS-NRAS, P310 TCOF1, P311 Congenital Heart Disease, P314 ABCA3-SFTPC, P318 Hirschsprung-2, P319 Thyroid, P325 OCA2, P331 COL5A1 mix1, P332 COL5A1 mix2, P341 PKHD1 mix1, P342 PKHD1 mix2, P351 PKD1, P352 PKD1-PKD2, P354 KIT SNAI2, P359 PLOD1, P362 USH2A mix2, P366 CHM-RP2-RPGR, P367 BEST1-PRPH2, P389 MLL2, P398 CASK, P409 RASA1-EPHB4, P418 MYH7, P426 Cystinuria, P433 ARID1A-ARID1B, P445 KDM6A, P457 DHCR7, P472 SUFU, P488 RS1, P021 SMA, P034 DMD-1, P041 ATM1, P042 ATM2, P046 TSC2, P057 FANCD2-PALB2, P128 CYP450, P147 1p36, P163 GJB-WFS1-POU3F4, P179 Limb-1, P184 JAG1, P189 CDKL5/ARX/FOXG1, P236 CFH Region, P292 PCDH15, P294 Tumour Loss, P306 SPG11, P313 CREBBP, P321 VPS13B, P333 EP300, P347 Hemochromatosis, P388 AGS, P455 LZTR1, P460 SMA, P463 MRKH, P470 NCL, P473 CTNS, P480 WHS & Achondroplasia, ME012 MGMT-IDH1-IDH2, P013 ATRX, P017 MEN1, P027 Uveal melanoma, P029 WBS, P032 FANCA mix 2, P040 CLL, P049 SLC6A8-ABCD1, L1CAM, P055 PAH, P056 TP53, P071 LMNB1-PLP1-NOTCH3, P073-A1 IKBKG, P074 AR, P088 Oligodendroglioma, P093-C2 HHT/HPAH, P101 STK11, P113 FANCB, P116 SGC, P137 SCN1A, P158 JPS, P164 IDS, P183 EDA-EDAR-EDARADD, P188 22q13, P223 PHEX, P253-D1 NB mix3, P258 SMARCB1, P271 COL1A1, P275 MAPT-GRN, P305-B2 AGXT, P315 EGFR, P316-B4 Recessive Ataxias, P322 VPS13B mix2, P324 22q11, P328 EYS, P339 SHANK3, P340 EHMT1, P353 CMT4, P381 COL11A1 mix1, P382 COL11A1 mix2, P410 GRIN2A GRIN2B, P414-C1 MDS, P429 SDHA-MAX-TMEM127, P431 FOXF1, P436 ANO5, P451 Chromosome 16, P471 EOFAD, P475 FOXP1-FOXP2, P478 SMARCE1, P489 BARD1, P495 CYP11A1-CYP11B1-CYP11B2, ME011-D1 Mismatch Repair genes, ME033 TNDM, ME042-C1 CIMP. P035 DMD-2, P044 NF2, P120 PANK2/PLA2G6, P124 TSC1, P125 Mitochondrial DNA, P148 TGFBR1-TGFBR2, P215 EXT, P262 GHI, P308 MET, P329 CRLF2-CSF2RA-IL3RA, P432 MYH9, P437 Familial MDS-AML, P474 CD274-PDCD1LG2-JAK2, P483 HER gene family, P484 ALPL, P492 POLD1 -POLE.

**Postup č. 9/816: Analýza sekvenčních variant lidského genomu metodou real-time PCR**

Hemochromatóza (sekvenční varianty H63D, S65C a C282Y v genu *HFE*)

Faktor V Leiden (sekvenční varianta 1691G>A v genu *F5*)

Faktor II Prothrombin (sekvenční varianta 20210G>A v genu *F2*)

Thiopurin S-metyltransferáza (sekvenční varianty 238G>C, 460G>A a 719 A>G v genu *TPMT*)

Deficit alfa 1-antitrypsinu (sekvenční varianty E264V a E342K v genu *SERPINA1*)

Sekvenční varianty genů *KRAS*, *NRAS*, *BRAF* a *EGFR*:

Idylla: *KRAS* (sekvenční varianty: c.34G>T, c.34G>C, c.34G>A, c.35G>C, c.35G>A, c.35G>T, c.38G>A, c.176C>A, c.176C>G, c.175G>A, c.181C>A, c.180\_181delinsAA, c.182A>T, c.182A>G, c.183A>C, c.183A>T, c.351A>C, c.351A>T, c.436G>C, c.436G>A, c.437C>T) *NRAS* (sekvenční varianty: c.34G>T, c.34G>A, c.35G>A, c.35G>C, c.35G>T, c.38G>A, c.38G>T, c.37G>C, c.175G>A, c.181C>A, c.182A>T, c.182A>G, c.183A>C, c.183A>T, c.351G>C, c.351G>T, c.436G>A, c.437C>T) *BRAF* (sekvenční varianty: c.1799T>A, c.1799\_1800delinsAA, c.1799\_1800delinsAC, c.1799\_1800delinsAT, c.1798\_1799delinsAA, c.1798\_1799delinsAG, c.1798G>A) *EGFR* (sekvenční varianty: c.2156G>C, c.2155G>T, c.2154\_2155delinsTT, c.2155G>A, c.2238\_2248delinsGC, c.2239\_2248delinsC, c.2240\_2248del, c.2239\_2247del, c.2239\_2251delinsC, c.2240\_2251del, c.2235\_2249del, c.2236\_2250del, c.2239\_2253del, c.2240\_2254del, c.2238\_2252del, c.2237\_2251del, c.2235\_2252delinsAAT, c.2237\_2252delinsT, c.2234\_2248del, c.2236\_2253delinsCTA, c.2237\_2253delinsTA, c.2235\_2251delinsAG, c.2236\_2253delinsCAA, c.2230\_2249delinsGTCAA, c.2240\_2257del, c.2237\_2255delinsT, c.2239\_2256del, c.2236\_2253del, c.2239\_2258delinsCA, c.2237\_2254del, c.2238\_2255del, c.2237\_2257delinsTCT, c.2236\_2255delinsAT, c.2236\_2256delinsATC, c.2237\_2256delinsTT, c.2237\_2256delinsTC, c.2235\_2255delinsGGT, c.2238\_2258del, c.2236\_2256del, c.2253\_2276del, c.2369C>T, c.2303G>T, c.2310\_2311insGGT,

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c.2308\_2309insGCCAGCGTG, c.2308\_2311delinsCCAGCGTGGAT, c.2311\_2312insGCGTGGACA, c. 2319\_2320insCAC, c.2573T>G, c.2573\_2574delinsGT, c.2573\_2574delinsGA, c.2582T>A).  
PGX: *KRAS* (sekvenční varianty: c.34G>T, c.34G>C, c.34G>A, c.35G>C, c.35G>A, c.35G>T, c.38G>A, c.176C>A, c.176C>G, c.175G>A, c.181C>A, c.182A>T, c.182A>G, c.183A>C, c.183A>T, c.349A>G, c.350A>G, c.351A>C, c.351A>T, c.436G>C, c.436G>A, c.437C>T) *NRAS* (sekvenční varianty: c.34G>T, c.34G>A, c.35G>A, c.35G>C, c.35G>T, c.38G>A, c.38G>T, c.37G>C, c.175G>A, c.176C>T, c.181C>A, c.182A>T, c.182A>G, c.183A>C, c.183A>T, c.350A>G, c.351G>C, c.351G>T, c.436G>A, c.437C>T) *BRAF* (sekvenční varianty: c.1799T>A, c.1799\_1800TG>AA, c.1799\_1800TG>AT, c.1798\_1799GT>AA, c.1798\_1799GT>AG).  
Dihydropyrimidin dehydrogenáza (sekvenční varianty: c.1905+1G>A, c.1679T>G, c.1236G>A, c.2846A>T v genu *DPYD*).  
Gilbertův syndrom (vyšetření alelických variant 5TA, 6TA, 7TA, 8TA v promotoru genu *UGT1A1*).  
Chimerismus - vybraných vysoce polymorfních delecí a inzercí za účelem typizace a monitoringu dárce a příjemce hematopoetických buněk.

**Postup č. 10/816: Analýza lidského genomu metodou reverzní hybridizace**

*KRAS* (sekvenční varianty: c.35G>C, c.34G>C, c.35G>A, c.34G>T, c.34\_35delGGinsAT, c.34\_35delGGinsCT, c.34G>A, c.35G>T, c.38G>C, c.37G>C, c.38G>A, c.37G>T, c.37G>A, c.38G>T, c.176C>A, c.176C>G, c.175G>A, c.179G>T, c.182A>G, c.183A>C, c.183A>T, c.182A>T, c.181C>A, c.351A>C, c.351A>T, c.349A>G, c.436G>C, c.436G>A, c.437C>T)  
*NRAS* (sekvenční varianty: c.35G>C, c.34G>C, c.35G>A, c.34G>T, c.34G>A, c.35G>T, c.37G>C, c.38G>A, c.37G>T, c.38G>T, c.176C>A, c.175G>A, c.178G>C, c.179G>A, c.182A>G, c.181C>G), c.183A>C, c.183A>T, c.182A>T, c.181C>A, c.182A>C, c.436G>A)  
*BRAF* (sekvenční varianty: c.1799T>C, c.1799\_1800TG>AT, c.1799T>A, c.1799\_1800TG>AA, c.1799T>G, c.1798\_1799GT>AA, c.1798G>A, c.1798\_1799GT>AG, c.1801A>G),  
*EGFR* (sekvenční varianty: c.2156G>C, c.2155G>T, c.2155G>A, c.2233\_2247del, c.2235\_2249del, c.2235\_2248delinsAATTC, c.2236\_2250del, c.2235\_2251delinsAATTC, c.2236\_2253del, c.2237\_2251del, c.2237\_2252delinsT, c.2237\_2253delinsTTGCT, c.2235\_2255delinsAAT, c.2237\_2254del, c.2237\_2255delinsT, c.2238\_2255del, c.2237\_2257delinsTCT, c.2239\_2247del, c.2238\_2248delinsGC, c.2239\_2248delinsC, c.2239\_2251delinsC, c.2240\_2251del, c.2240\_2254del, c.2239\_2256del, c.2239\_2256delinsCAA, c.2239\_2258delinsCA), c.2240\_2257del, c.2369C>T, c.2573T>G, c.2582T>A).  
*VKORC1* (sekvenční varianta 1639G>A)  
*CYP2C9* (sekvenční varianty: 1075A>C, 430C>T)

**Postup č. 5/823: Analýza histologických a cytologických vzorků metodou *in situ* hybridizace**

Sondy: *HER2*, *ALK*, *ROS1*, *ALK/EML4*, *ATM/CEN12*, *BCL2*, *BCL6*, *BCOR*, *BCL2/IGH*, *BIRC3/MALT1*, *CCND1/IGH*, *CCND1*, *CDK4/CEN12*, *CDKN2A/CEN9*, *CIC*, *COL1A1*, *DDIT3(CHOP)*, *EGFR/CEN7*, *ETV6*, *EWSR*, *EWSR/FLI1*, *FGFR1*, *FOXO1*, *FUS*, *JAZF1*, *MAML2*, *MALT1*, *MYC/CEN8*, *MYC*, *MET/CEN7*, *MDM2/CEN12*, *NTRK3*, *NR4A3*, *NUTM1*, *PTEN/CEN10*, *PDGFB*, *PHF1*, *PDGFRA/FIP1L1*, *RET*, *RB1/13q12*, *SMARCB1/22q12*, *SS18*, *SS18/SSX1*, *TP53/CEN17*, *TFE3*, *USP6*, *VHL/CEN3*, *WWTR1*, *WT1*, 1p12 , 1p/19q, *IGH*

**Postup č. 6/823: Analýza lidského somatického genomu metodou masivně paralelního sekvenování**

NGS panel Hereditární nádorové syndromy a somatické varianty v nádorových tkáních (*ACD*, *AIP*, *AKT1*, *APC*, *AR*, *ATM*, *ATR*, *AXIN2*, *BAP1*, *BARD1*, *BLM*, *BMPR1A*, *BRAF*, *BRCA1*, *BRCA2*, *BRF1*, *BRIP1*, *CARD11*, *CCND1*, *CDC73*, *CDH1*, *CDK4*, *CDKN1B*, *CDKN2A*, *CYLD*, *DICER1*, *DLST*, *DNMT3A*, *EGFR*, *EMSY*, *EPCAM*, *ERBB2*, *ERBB3*, *ERCC2*, *ERCC3*, *ERCC5*, *ETV6*, *FANCA*, *FANCB*, *FANCC*, *FANCD2*, *FANCE*, *FANCF*, *FANCG*, *FANCI*, *FANCL*, *FANCM*, *FAT1*, *FBXW7*, *FH*,



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FLCN, GATA2, GREM1, HNF1A, HOXB13, HRAS, CHEK1, CHEK2, IDH1, IDH2, KIF1B, KIT, KLLN, KMT2A, KRAS, MAP2K1, MAX, MC1R, MEN1, MET, MITF, MLH1, MLH3, MN1, MRE11, MSH2, MSH3, MSH6, MSR1, MUTYH, NBN, NF1, NF2, NRAS, NRG1, NTHL1, PALB2, PARP1, PARP2, PARP3, PARP4, PBRM1, PDGFB, PDGFD, PDGFRA, PDGFRB, PIK3CA, PMS2, POLD1, POLE, POT1, PPP2R2A, PRKAR1A, PTEN, PTCH1, RAD50, RAD51, RAD51B, RAD51C, RAD51D, RAD52, RAD54L, RB1, RECQL, RECQL4, RET, REXO2, RNF43, RPS20, SDHA, SDHAF2, SDHB, SDHC, SDHD, SH2D1A, SH2D1A, SLC25A11, SLX4, SMAD4, SMARCA4, SMARCB1, SMARCE1, SRGAP1, SRY, STK11, SUFU, TERF2IP, TERT, TET2, TMEM127, TP53, TSC1, TSC2, VHL, WRN, WT1, XPA, XRCC2, XRCC3 (hg38). Somatické varianty: ACVR1, ALK, ARAF, ARID1A, ARID1B, ATRX, BRK1, CCDC153, CCNE1, CD79B, CDADC1, CDK12, CDK6, CDKN2B, COX6C, CTNNA1, CTNNB1, DDR2, ENG, ENOX1, ERG, ESR1, FAM124A, FANCD2OS, FGFR1, FGFR2, FGFR3, FOXL2, H3-C14 (H3), H3C2 (HIST1H3B), H3-3A, IL18, INA, INCA1, KEAP1, KIAA1549, MAD2L2, MAMLD1, MALT1, MTOR, MYB, MYBL1, MYC, MYCN, MYD88, MYH13, NTRK1, NTRK2, NTRK3, NUTM1, PABPC1, PABPC3, PAX8, PIK3R1, PML, PPARG, PPP2R1A, QKI, RAF1, RELA, RHOA, ROS1, RPAIN, SETD2, S1PR1, TACC1, TACC3, TBX21, THADA, TMPRSS2, TOE1, TOP2A, WSCD1, YAP1, ZFTA (C11orf95).

**NGS panel Somatické varianty v nádorových tkáních II** (A2M, ABCC9, ABL1, ABL2, ACBD5, ACD, ACP5, ACTB, ACVR1, ACVR2A, ACVR1B, ADAMTS14, ADGRA2, AFAP1L2, AFF1, AFF3, AGTRAP, AHRR, AKAP9, AIP, AKT1, AKT2, AKT3, ALDH1L2, ALK, ALL1, ALOX12B, AMER1, AMN, ANKRD11, ANKRD26, ANTXR1, ANTXR2, ANXA11, APC, AR, ARAF, ARFRP1, ARHGEF2, ARID1A, ARID1B, ARID5B, ARID2, ARMC10, ASPSCR1, ASXL1, ASXL2, ATF1, ATF5, ATIC, ATM, ATP1B1, ATR, ATRX, AURKA, AURKB, AXIN1, AXIN2, AXL, BAG4, BAP1, BARD1, BAX, BBC3, BCAM, BCL2, BCL6, BCL10, BCL2L2, BCL2L1, BCL2L11, BCOR, BCORL1, BCR, BEST3, BIRC3, BIRC6, BLIMP1, BLM, BMP4, BMPR1A, BRAF, BRCA1, BRCA2, BRD4, BRD8, BRF1, BRIP1, BTK, BUB1, BUB1B, BTG1, B2M, C11ORF30, C11orf95, C2orf44, CAD, CALR, CAMK2A, CAMTA1, CARD11, CARS1, CASP8, CASZ1, CATSPERZ, CBFEB, CBL, CCND1, CCND2, CCND3, CC2D1A, CCDC153, CCDC6, CCNA2, CCNB1, CCNB2, CCNB3, CCND1, CCND3, CCNE1, CD34, CD74, CD79B, CD274, CD276, CDADC1, CDC6, CDC73, CD79A, CD79B, CDH1, CDH11, CDK1, CDK8, CDK12, CDK17, CDK4, CDK6, CDKN1A, CDKN1B, CDKN1C, CDKN2C, CDKN2A, CDKN2B, CDKN2D, CDX1, CDX2, CEBPD, CEBPE, CEBPG, CENPA, CEP43, CEP85L, CIC, CIITA, CITED2, CLCA2, CLCN6, CLIP1, CLTC, CNBP, CNTN1, COG8, COL17A1, COL1A1, COL1A2, COL25A1, COL3A1, COP1, CPM, CREB1, CREB3L1, CREB3L2, CREB3L3, CREBBP, CREM, CRKL, CRTCL1, CRTCL3, CSF1R, CSF3R, CSGALNACT2, CTLA4, CSNK1A1, CTNNA1, CTNNB1, CUBN, CUL3, CUX1, CXCR4, CYLD, CYP1A1, CYP3A4, CYSLTR2, DAXX, DCTN1, DCUN1D1, DDIT3, DDR2, DDX5, DDX41, DEK, DERA, DFFA, DHX15, DHX57, DICER1, DIS3, DLL1, DLST, DNAJB1, DNMT1, DNMT3A, DNMT3B, DOT1L, CRLF2, DUSP22, CTCF, DUX4, E2F2, EBF1, EED, EGFL7, FGF2, EGFR, EGLN1, EGLN2, EHMT1, EI24, EIF1AX, EIF3E, EIF4E, EIF4A1, EIF4A2, ELF4, ELL, EML4, EMSY, ENG, ENOX1, ENTPD1, EP300, EPAS1, EPC1, EPCAM, EPHA3, EPHA5, EPHA7, EPHB1, ERBB2, ERBB3, ERBB4, ERC1, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERG, ERFF1, ESR1, ESRP1, ESRRA, ETS1, ETV1, ETV4, ETV5, ETV6, EVER1, TMC8, EWSR1, EXOC1, EXOC2, EXT1, EXT2, EZH2, EZHIP, EZR, FAM124A, FAM131B, FAM175A, FAM179A, FAM216A, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FANCP, FAS, FAT1, FBXO11, FBXW7, FEN1, FEV, FGF1, FGF3, FGF4, FGF5, FGF6, FGF7, FGF8, FGF9, FGF10, FGF14, FGF19, FGF23, FGFR1, FGFR1OP, FGFR2, FGFR3, FGFR4, FH, FLCN, FLI1, FLT1, FLT3, FLT4, FN1, FOS, FOSB, FOXA1, FOXB2, FOXE1, FOXL2, FOXO1, FOXO4, FOXP1, FRS2, FUBP1, FUS, FUT8, FYN, GABRA6, GAPDH, GATA1, GATA2, GATA3, GATA4, GATA6, GCC2, GCGR, GEN1, GFPT1, GID4, GLI1, GLI2, GLMN, GLRX5, GLYR1, GNA11, GNA13, GNAI1, GNAQ, GNAS, GOLGA5, GOPC, GPR124, GPS2, GREB1, GREM1, GRIN2A, GRM3, GSK3B, GTF2E1, GTF3C2, H1, H2BC5, H3, H3-3A, H3F3C, H3C1, H3C10, H3C11, H3C12, H3C13, H3C3, H3-4, H3C4, H3C6, H3C7, H3C8, H3-3A, H3-3B, H3K27, H4C9, HAS2, HDAC2, HDAC9, HEY1, HGF, HIP1, HIST1H3B, HLA-A, HMGA2, HMGN2P46, HNF1A, HNRNPA1, HNRNPK, HOOK3, HOXB13, HRAS, HSD17B2, HSD3B1, HSP90AA1, CHD2, CHD4, CHEK1, CHEK2, CHIC2, CHN, CHN1, CHTOP, ICOSLG, ID3, IDH1, IDH2, IFNGR1, IGF1, IGF2, IGF1R, IGFBP5, IGSF11, IKBKE, IKZF1, IL10, IL12RB1, IL16,

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IL18, IL1R1, IL21R, IFNL3, IL7R, INCA1, ING1, INHA, INHBA, INPP4B, INSL3, INSM1, INSR, IRF2, IRF2BP2, IRF4, IRF8, IRS1, IRS2, IRX2, ITPKB, INPP4A, ITPR2, JAK1, JAK2, JAK3, JAZF1, JUN, KANSL1, KAT6A, KAT6B, KCNMB4, KCNQ1, KDM2A, KDM4D, KDM5A, KDM5B, KDM5C, KDM6A, KDM6B, KDR, KEAP1, KEL, KIAA0101, KIAA1217, KIAA1549, KIF13B, KIF15, KIF1B, KIF5B, KIT, KLC1, KLF4, KLF17, KLHL6, KLK2, KLLN, KMT2A, KMT2B, KMT2C, KMT2D, KRAS, KRT8, KRT19, KTN1, L2HGDH, LAMC1, LAMP1, LATS1, LATS2, LCP1, LEMD3, LEUTX, LIMD1, LIN28B, LMNA, LMO1, LMO2, LMO7, TP53, LPP, LRIG3, IRAG2, LRMP, LRP1, LRP1B, LYN, LZTR1, LZTS1, MAD2L1, MAD2L2, MAGEC2, MAGI2, MAL, MALT1, MAML2, MAML3, MAPK3, MAP2K1, MAP2K2, MAP3K1, MAP3K4, MAP3K13, MAP3K14, MAP3K5, MAP4K3, MAP2K4, MAPK1, MAX, MBTD1, MC1R, MCL1, MDC1, MDH2, MDM2, MDM4, MEAF6, MED12, MEF2B, MEN1, MET, MGA, MID1, MINK1, MITF, MRTFA, MKRN1, MLF1, MLH1, MLH3, MLL2, MLLT3, MLLT10, MME, MN1, MPL, MPRIIP, MRAS, MRE11, MRE11A, MSH2, MSH3, MSH6, MSMB, MSN, MSR1, MST1, MST1R, MSX1, MTAP, MTOR, MUC1, MUTYH, MYB, MYBL1, MYC, MYCL, MYCL1, MYCN, MYD88, MYH13, MYH9, MYO5A, MYOD1, N4BP1, NAB2, NACCC, NAV3, NBN, NCOA1, NCOA2, NCOA3, NCOA4, NCOR1, NEGR1, NEK6, NF1, NF2, NFATC2, NFE2L2, NFIB, NFKB1, NFKB2, NFKBIA, NKAP, NKX3, NKX2-1, NKX3-1, NLRP6, NME1, NOTCH, NOTCH1, NOTCH2, NOTCH3, NOTCH4, NPM1, NR4A2, NR4A3, NRAS, NRG1, NSD1, NSD2, NTHL1, NTRK1, NTRK2, NTRK3, NUP160, NUP37, NUP93, NUTM1, NUTM2A, NUTM2B, OMD, OR51E2, ORL, P2RY8, PAFAH1B1, PAICS, PAK1, PAK3, PAK7, PALB2, PARK2, PARP1, PARP2, PARP3, PARP4, PATZ1, PAX3, PAX5, PAX7, PAX8, PBRM1, PBX1, PBX3, PCLAF, PCM1, PCNA, PDCD1, PDCD1LG2, PDE11A, PDGFB, PDGFD, PDGFRA, PDGFRB, PDK1, PDPK1, PGR, PHF1, PHF6, PHLDA1, PHLDB1, PHOX2B, PIK3C3, PIK3CA, PIK3CB, PIK3CD, PIK3C2B, PIK3CG, PIK3C2G, PIK3R1, PIK3R2, PIK3R3, PIM1, PIM2, PINK1, PLA2R1, PLAG1, PLCB4, PLCG1, PLCG2, PLK2, PLXNA2, PMAIP1, PML, PMS1, PMS2, PMS2CL, PMS2P1, PNRC1, POLD1, POLE, POLE2, POLH, PORCN, POT1, POU2AF1, POU5F1, PPARD, PPARG, PPAT, PPFIBP1, PPFIBP2, PPL, PPM1D, PPP6C, PPP1CB, PPP1R15A, PPP2R1A, PPP2R1B, PPP2R2A, PRCC, PRDM1, PRDM10, PRDM16, PREX2, PRKAR1A, PRKAR2B, PRKCI, PRKCZ, PRKDC, PRKG2, PRRT2, PRSS8, PTEN, PTH1R, PTCH1, PTPN1, PTPN11, PTPN12, PTPN3, PTPRD, PTPRJ, PTPRK, PTPRS, PTPRT, PWWP2A, PYCR1, QKI, RAB29, RAB35, RAB2A, RAC1, RAD21, RAD50, RAD51, RAD52, RAD51B, RAD51C, RAD51D, RAD54L, RAF1, RAG1, RAG2, RANBP1, RANBP2, RARA, RASA1, RB1, RBM10, RCSD1, RECQL, RECQL4, REL, RELA, RELB, REST, RET, REXO2, RHBDF2, RHEB, RHOA, RICTOR, RIMS2, RIT1, RNF130, RNF213, RNF43, ROS1, RPAIN, RPTOR, RPS20, RPS6KA4, RPS6KB1, RPS6KB2, RRAS2, RRBP1, RSPO2, RSPO3, RTEL1, RUNX1, RUNX2, RUNX1T1, RYBP, S1PR1, S1PR2, SALL4, SDC4, SDHA, SDHAF2, SDHB, SDHC, SDHD, SEC31A, SEPT7, SEPTIN9, SERPINE1, SETBP1, SETD2, SEZ6L2, SF3B1, SH2B3, SH3BP2, SH3BP5, SH2D1A, SHOC2, SHTN1, SHQ1, SLC25A11, SLC34A2, SLC3A2, SLC43A3, SLC45A3, SLC5A7, SLC9A8, SLC01C1, SLFN11, SLIT2, SLX4, SMAD2, SMAD3, SMAD4, SMARCA2, SMARCA4, SMARCA5, SMARCB1, SMARCC1, SMARCC2, SMARCD1, SMARCE1, SMC1A, SMC3, SMO, SNCAIP, SNRNP25, SOCS1, SOS1, SOS2, SOX2, SOX4, SOX9, SOX10, SOX17, SP140, SP3, SPEN, SPOP, SPP1, SPRED1, SPTA1, SPTBN1, SQSTM1, SRC, SRF, SRSF2, SRGAP3, SRY, SS18, SS18L1, SSSX1, SSSX2, SSSX2B, SSSX4, STAG1, STAG2, STAR, STARD13, STAT3, STAT4, STAT5B, STAT6, STIL, STIP1, STK11, STK40, STRBP, STRN, SUFU, SUZ12, SYK, SYT8, TACC3, TAF1, TAF15, TAF4, TANK, TBX3, TBX21, TBXT, TCEB1, TCF12, TCF3, TCF7L2, TEC, TEK, TENT5C, TERC, TERF2IP, TERT, TET1, TET2, TFE3, TFG, TFRC, TGFB1, TGFB2, THBS1, THRAP3, TLR2, TMC6, TMC8, TMED6, TMEM106B, TMEM127, TMEM178A, TMPRSS2, TNFAIP3, TNFRSF11A, TNFRSF13B, TNFRSF14, TNFSF4, TNNI2, TOE1, TOGARAM2, TOP1, TOP2A, TP53, TP63, TPD52L1, TPM3, TPM4, TPR, TPRA1, TRAF1, TRAF2, TRAF3, TRAF7, TRIM11, TRIM24, TRIM27, TRIM33, TSC1, TSC2, TSHR, TSPAN31, TTC23, U2AF1, UBTf, UEVLD, USP15, USP6, VAMP2, VCL, VDACC2, VEGFA, VEGFB, VGLL2, VHL, VMP1, VTCN1, WDCP, WDFY2, WISP3, WRN, RECQL2, WSCD1, WT1, WWTR1, XPA, XIAP, XPO1, XRCC2, XRCC3, YAP1, YEATS4, YES1, YWHAE, TST500C, ZBTB7A, ZBTB2, ZBTB16, ZC3H7A, ZC3H7B, ZCCHC8, ZFH3, ZFP36, ZNF217, ZNF444, ZNF703, ZRSR2).

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**Postup č. 7/823: Analýza sekvenčních variant lidského somatického genomu metodou real-time PCR**

Idylla: *KRAS* (sekvenční varianty: c.34G>T, c.34G>C, c.34G>A, c.35G>C, c.35G>A, c.35G>T, c.38G>A, c.176C>A, c.176C>G, c.175G>A, c.181C>A, c.180\_181delinsAA, c.182A>T, c.182A>G, c.183A>C, c.183A>T, c.351A>C, c.351A>T, c.436G>C, c.436G>A, c.437C>T) *NRAS* (sekvenční varianty: c.34G>T, c.34G>A, c.35G>A, c.35G>C, c.35G>T, c.38G>A, c.38G>T, c.37G>C, c.175G>A, c.181C>A, c.182A>T, c.182A>G, c.183A>C, c.183A>T, c.351G>C, c.351G>T, c.436G>A, c.437C>T) *BRAF* (sekvenční varianty: c.1799T>A, c.1799\_1800delinsAA, c.1799\_1800delinsAC, c.1799\_1800delinsAT, c.1798\_1799delinsAA, c.1798\_1799delinsAG, c.1798G>A) *EFGR* (sekvenční varianty: c.2156G>C, c.2155G>T, c.2154\_2155delinsTT, c.2155G>A, c.2238\_2248delinsGC, c.2239\_2248delinsC, c.2240\_2248del, c.2239\_2247del, c.2239\_2251delinsC, c.2240\_2251del, c.2235\_2249del, c.2236\_2250del, c.2239\_2253del, c.2240\_2254del, c.2238\_2252del, c.2237\_2251del, c.2235\_2252delinsAAT, c.2237\_2252delinsT, c.2234\_2248del, c.2236\_2253delinsCTA, c.2237\_2253delinsTA, c.2235\_2251delinsAG, c.2236\_2253delinsCAA, c.2230\_2249delinsGTCAA, c.2240\_2257del, c.2237\_2255delinsT, c.2239\_2256del, c.2236\_2253del, c.2239\_2258delinsCA, c.2237\_2254del, c.2238\_2255del, c.2237\_2257delinsTCT, c.2236\_2255delinsAT, c.2236\_2256delinsATC, c.2237\_2256delinsTT, c.2237\_2256delinsTC, c.2235\_2255delinsGGT, c.2238\_2258del, c.2236\_2256del, c.2253\_2276del, c.2369C>T, c.2303G>T, c.2310\_2311insGGT, c.2308\_2309insGCCAGCGTG, c.2308\_2311delinsCCAGCGTGGAT, c.2311\_2312insGCGTGGACA, c.2319\_2320insCAC, c.2573T>G, c.2573\_2574delinsGT, c.2573\_2574delinsGA, c.2582T>A).  
PGX: *KRAS* (sekvenční varianty: c.34G>T, c.34G>C, c.34G>A, c.35G>C, c.35G>A, c.35G>T, c.38G>A, c.176C>A, c.176C>G, c.175G>A, c.181C>A, c.182A>T, c.182A>G, c.183A>C, c.183A>T, c.349A>G, c.350A>G, c.351A>C, c.351A>T, c.436G>C, c.436G>A, c.437C>T) *NRAS* (sekvenční varianty: c.34G>T, c.34G>A, c.35G>A, c.35G>C, c.35G>T, c.38G>A, c.38G>T, c.37G>C, c.175G>A, c.176C>T, c.181C>A, c.182A>T, c.182A>G, c.183A>C, c.183A>T, c.350A>G, c.351G>C, c.351G>T, c.436G>A, c.437C>T) *BRAF* (sekvenční varianty: c.1799T>A, c.1799\_1800TG>AA, c.1799\_1800TG>AT, c.1798\_1799GT>AA, c.1798\_1799GT>AG).

**Postup č. 8/823: Analýza lidského somatického genomu metodou reverzní hybridizace**

*KRAS* (sekvenční varianty: c.35G>C, c.34G>C, c.35G>A, c.34G>T, c.34\_35delGGinsAT, c.34\_35delGGinsCT), c.34G>A, c.35G>T, c.38G>C, c.37G>C, c.38G>A, c.37G>T, c.37G>A, c.38G>T, c.176C>A, c.176C>G, c.175G>A, c.179G>T, c.182A>G, c.183A>C, c.183A>T, c.182A>T, c.181C>A, c.351A>C, c.351A>T, c.349A>G, c.436G>C, c.436G>A, c.437C>T)  
*NRAS* (sekvenční varianty: c.35G>C, c.34G>C, c.35G>A, c.34G>T, c.34G>A, c.35G>T, c.37G>C, c.38G>A, c.37G>T, c.38G>T, c.176C>A, c.175G>A, c.178G>C, c.179G>A, c.182A>G, c.181C>G), c.183A>C, c.183A>T, c.182A>T, c.181C>A, c.182A>C, c.436G>A)  
*BRAF* (sekvenční varianty: c.1799T>C, c.1799\_1800TG>AT, c.1799T>A, c.1799\_1800TG>AA, c.1799T>G, c.1798\_1799GT>AA, c.1798G>A, c.1798\_1799GT>AG, c.1801A>G),  
*EGFR* (sekvenční varianty: c.2156G>C, c.2155G>T, c.2155G>A, c.2233\_2247del, c.2235\_2249del, c.2235\_2248delinsAATTC, c.2236\_2250del, c.2235\_2251delinsAATTC, c.2236\_2253del, c.2237\_2251del, c.2237\_2252delinsT, c.2237\_2253delinsTTGCT, c.2235\_2255delinsAAT, c.2237\_2254del, c.2237\_2255delinsT, c.2238\_2255del, c.2237\_2257delinsTCT, c.2239\_2247del, c.2238\_2248delinsGC, c.2239\_2248delinsC, c.2239\_2251delinsC, c.2240\_2251del, c.2240\_2254del, c.2239\_2256del, c.2239\_2256delinsCAA, c.2239\_2258delinsCA, c.2240\_2257del, c.2369C>T, c.2573T>G, c.2582T>A).