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

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
~~Cystická fibróza: *CFTR* (3120+1G>A, 711+1G>T, 621+1G>T, 1717-1G>A, CFTRdele2,3(21kb), 3849+10kbC>T, 2789+5G>A, 1898+1G>A, Gly542X, Gly85Glu, Tyr1092X(C>A), Gly551Asp, Arg553X, 3659delC, Asn1303Lys, Arg560Thr, Arg117His, Arg1162X, Leu1077Pro, Arg117Cys, Arg1066Cys, Leu1065Pro, Trp1282X, Arg347His, Arg347Pro, Ile507del, Thr338Ile, Phe508del, Ile336Lys, 1677delTA, Arg334Trp, 3272-26A>G, 1078delT, 2183AA>G, 2184insA, 2143delT, 5T (9-13TG)~~
Mikrodelece na chromozomu Y (AZFa, AZFb, AZFc), SRY
Syndrom fragilního X (analýza počtu repetic CGG v 5'UTR oblasti genu FMR1)

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 (~~95~~185 genů: ~~(~~*ACD, AIP, AKT1, APC, AR, ATM, ATR, AXIN2, BAP1, BARD1~~–~~, BLM~~–~~, BMPR1A~~–~~, BRAF, BRCA1, BRCA2~~-~~, BRF1, BRIP1~~-~~, CARD11, CCND1, CDC73, CDH1, ~~CDK12,~~ CDK4, CDKN1B, CDKN2A, CYLD , DICER1, DLST, DNMT3A, EGFR, ~~ENG~~ EMSY, EPCAM~~–~~, ERBB2, ERBB3, ERCC2, ERCC3, FANCA , FANCB~~-~~, FANCC~~-~~, FANCD2~~-~~, FANCE~~-~~, FANCF~~-~~, FANCG~~-~~, FANCI~~-~~, FANCL, FANCM~~-~~, ~~FANCP~~ , FAT1, FH, FLCN, ~~FOXL2~~GATA2, GREM1~~-~~, HNF1A, HOXB13, HRAS, CHEK1, CHEK2, IDH1, IDH2, KIF1B, KIT, KLLN~~-~~, KMT2A, KRAS, ~~MAD2L2~~ 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, PDGFB, PDGFD, ~~PML,~~ PDGFRA, PDGFRB, PIK3CA, PMS2, POLD1~~–~~, POLE~~–~~, POT1, PPP2R2A, PTEN, PTCH1, RAD50~~–~~, RAD51, RAD51B, RAD51C~~-~~, RAD51D~~-~~, RAD52, RAD54L~~-~~, RB1, RECQL~~-~~, RECQL4, RET, REXO2, RNF43~~-~~, RPS20~~–~~, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, SLX4, SMAD4, SMARCA4, SMARCB1, SMARCE1, SRY~~–~~, STK11~~–~~, SUFU, TERF2IP, TERT, TET2, TMEM127, TP53, VHL, WRN~~),~~, WT1, XPA, XRCC2, XRCC3, ALK, ARAF, ARID1A, ARID1B, CCDC153, CCNE1, CD79B, CDADC1, CDK12, CDK6, CTNNB1, DDR2, ENG, ENOX1, ERG, ESR1, FAM124A, FGFR1, FGFR2, FGFR3, FOXL2, H3-3A, IL18, INCA1, KEAP1, MAD2L2, MALT1, MTOR, MYC, MYD88, NTRK1, NTRK2, NTRK3, NUTM1, PIK3R1, PML, PPP2R1A, RHOA, ROS1, RPAIN, S1PR1, TBX21, TMPRSS2, TOE1, TOP2A, WSCD1)*

NGS panel Mentální retardace a mikrocefalie (~~678~~701 genů: *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, DNM1L, DNMT3A, DONSON, DPAGT1, DPF2, DPM1, DPP6, DYNC1H1, DYNC1I2, DYRK1A, EBF3, 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, QRICH1, 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, TECR, TELO2, TGIF1, THOC2, THOC6, TIMM8A, TLK2, TMCO1, TMEM165, TMTC3, TMX2, TNIK, TOE1, TOP3A, TP53RK, TPRKB, TRAIP, 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 (359 genů: *AARS1, ABCC1, ABHD12, ACOX1, ACTB, ACTG1, ACVR1, ADCY1, ADGRV1 (GPR98), AFG3L2, AIFM1, AK2, ALG11, ALG12, ALMS1, AMMECR1, ANKH, AP1B1, AP1S1, ARSG, ATL1, ATOH1, ATP1A2, ATP1A3, ATP2B2, ATP6AP1, ATP6V0A4, ATP6V1B1, ATP6V1B2, ATRX, BCAP31, BCS1L, BDP1, BRAF, BSND, BTD, C2orf71, CABP2, CACNA1D, CATSPER2, CCDC50, CD151, CD164, CDC14A, CDC2L1, CDC42, CDC45L, CDH11, CDH23, CEACAM16, CEP250, CEP78, CIB2, CISD2, CLCNKA, CLCNKB, CLDN14, CLIC5, CLPP,*

CLRN1(USH3), CNBP, CNRIP1, COCH, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COLEC11, COQ2, COQ6, CREB3L1, CRYM, DCAF17, DCDC2, DDX11, DHX16, DCHS1, DIABLO, DIAPH1, DIAPH3, DLX5, DMXL2, DNAJC3, DNMT1, DSPP, EDN3, EDNRB, EFEMP1, EFTUD2, ECHS1, ELMOD1, ELMOD3, EPS8L2, ERAL1, ERCC3, ERCC6, ERCC8, ESPN, ESRP1, ESRRB, EXOSC2, EYA1, EYA4, FAT4, FGF3, FGFR3, FIT2, FKBP14, FLNA, FLNB, FOXC1, FOXI1, FTO, GAB1, GATA2, GATA3, GDF6, GGPS1, GIPC3, GJA1, GJB1, GJB2, GJB3, GJB6, GLA, GLIS3, GLYCTK, GMNN, GNAI3, GPC4, GPRASP2, GPSM2, GRAP, GRHL2, GRXCR1, GRXCR2, GSDME (DFNA5) , HARS1, HARS2, HGF, HOMER2, HOXA1, HOXA2, HOXB1, HSD17B4, HSPA9, HUWE1, CHD7, CHSY1, IARS2, , IGF1, IL17RD, ILDR1, INF2, IRX5, ITM2B, JAG1, KARS1, KCNE1, KCNJ10, KCNQ1, KCNQ4, KIT, KITLG, KMT2D, LARS2, LHFPL5, LHFPL5, , LHX3, LMX1A, LOXHD1, LRP2, LRTOMT, MAF, MAFB, MAN2B1, MAP3K7, MARVELD2, MASP1, MCM2, MCM3AP, MCM5, MED12, MET, MFN2, MGP, MICOS13, MITF, MN1, MPZ, MPZL2, MRPS7, MSRB3, MTO1, MYCN, MYH14, MYH9, MYO15A, MYO1A, MYO1C, MYO3A, MYO6, MYO7A, NARS2, NDP, NDRG1, NEBL, NLRP12, NLRP3, NOG, NOTCH2, OPA1, ORC1, ORC4, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX3, PBX1, PCDH15, PCGF2, PDE1C, PDSS1, PDZD7, PEX1, PEX12, PEX6, PEX7, PHYH, PIGL, PISD, PJVK, PLCG2, PLEK, PLOD3, PLS1, PMP22, PNPT1, POLD1, POLR1B, POLR1C, POLR1D, POU3F4, POU4F3, PPIP5K2, PRDM5, PRPS1, PSMC3, PSMD12, PTPN11, PTPRQ, PTRH2, RAI1, Rbm24, RDX, REST, RFT1, RIPOR2, RMND1, ROR1, RPGR, RPS6KA3, S1PR2, SALL1, SALL4, SCD5, SCN9A, SDHD, SEMA3E, SERAC1, SERPINB6, SGPL1, SIX1, SIX5, SLC12A1, SLC12A2, SLC17A8, SLC19A2, SLC26A4, SLC26A5, SLC29A3, SLC33A1, SLC44A4, SLC4A1, SLC4A11, SLC52A2, SLC52A3 (C20ORF54), SLC9A1, SLITRK6, SMAD4, SMPX, SNAI2, SNAP29, SOX10, SOX2, SOX6, SPATA5, SPNS2, SPTBN4, SPTLC1, SQSTM1, SRP72, SSBP1, STAG2, STRC, SUCLA2, SYNE4, TBC1D24, TBL1Y, TBX1, TCIRG1, TCOF1, TECTA, TFAM, TFAP2A, THRB, TIMM8A, TIMMDC1, TLK2, TMC1, TMEM132E, TMIE, TMPRSS3, TNC, TNFRSF11A, TPRN, TRIOBP, TRMT10C, TRPV3, TRRAP, TSPEAR, TSR2, TTR, TUBB4B, TWIST2, TWNK, TXNL4A, TYMP, TYR, UBR1, USH1C, USH1G, USH2A, VAC14, VCPKMT, VPS13B, VPS33B, WAC, WBP2, WFS1, WHRN)

NGS panel Neurodegenerativní onemocnění (190 genů: *ABCA7, ADCY5, ADH1C, ALDH18A1, ALOX5, ALS2, ANG, ANO3, ANXA11, APOE, APP, APTX, ~~ARSA,~~ ATL1, ATM, ATP13A2, ATP1A3, ATP7B, ATXN2, ATXN3, ~~ATXN8OS,~~ AUH, BCAP31, BSCL2, C19orf12, C9orf72, CACNA1B, CACNA1G, CD36, CIZ1, CLU ~~(ApoJ),~~ COASY, COL6A3, CP, CPT1C, CRAT, CSF1R, DAB1, DAO, DCAF17, DCTN1, DDC, DNAJC12, DNAJC13, DNAJC6, DNMT1, ~~DYT1~~DPGFB, EEF2, EIF4G1, ELOVL4, ELOVL5, ERBB4, ERLIN1, FA2H, FAT2, FBXO7, FGF14, FIG4, FTL, FUS, GBA, GCDH, GCH1, GIGYF2, GJC2, GLE1, GLUD2, GNAL, GNAO1, GNE, GRM1, GRN, GTPBP2, HEXA, HEXB, HNRNPA1, HPCA, HPRT1, HSPD1, HTRA1, HTRA2, HTT, CHAT, CHCHD10, CHCHD2, CHMP2B, IDE, ITM2B, ITPR1, JAM2, KCNC3, KCND3, KCNMA1, KCTD17, KIF1A, KIF1C, KIF5A, KMT2B, LMNB1, LRRK2, MAPT, MATR3, MME, MMUT, MYORG, NEFH, NEK1, NIPA1, NOTCH3, OPTN, PANK2, PANK3, ~~PARK2,~~ PARK7, PCCA, PCCB, PCNA, ~~PDGFB,~~ PDGFRB, PDYN, PFN1, ~~PGKCG, PICALM~~PICAML, PINK1, PLA2G6, PLD3, PLP1, PNKD, POLG, PRKN/PARK2, PRKRA, PRNP, PRPH, PRRT2, PSEN1, PSEN2, PTS, PUM1, RAB39B, REEP1, RELN, REPS1, ~~SCA20, SCA32~~RTN2, SCP2, SETX, SGCE, SIGMAR1, SLC18A2, SLC20A2, SLC2A1, SLC30A10, SLC33A1, SLC39A14, SLC6A3, SNCA, SOD1, SPAST, SPG11, SPG7, SPR, SPTBN2, SQSTM1, SS18L1 ~~(~~/KIAA0693~~),~~ STUB1, SYNJ1, TAF1, TARDBP, TBK1, TBP, TGM6, TH, THAP1, TIA1, TIMM8A, TMEM240, TMEM63A, TNK1, TOMM40, TOR1A, TRPC3, TTBK2, TUBA4A, TUBB4A, TYROBP, UBAP1, UBQLN2, UCHL1, VAPB, VCP, VPS13C, VPS35, WASHC5, WDR45~~),~~ XPR1, ZFYVE27)*

NGS panel Vrozené kostní anomálie (~~920~~1029 genů: *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, C15ORForf41, C2CD3, C8ORF37, CA2, CACNA1C, CAD, CANT1, CASR, CBFB, 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/C21orf2, CFAP410, 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, CTNNB1, 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, DUOXA2, DVL1, DVL3, DYM, DYNC2H1, DYNC2LI1, DYT1/TOR1A, EBP, ECEL1, EDARADD, EDN1, EED, EFEMP2, EFL1, EFNA4, EFNB1, 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, FAM149B1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FAT4, FBLN1, FBLN5, FBN1, FBN2, FBXL3, FBXO11, FBXW11, FCSK, FDFT1, FERMT1, FERMT3, FGD1, FGF10, FGF16, FGF17, FGF23, FGF8, FGF9, FGF13, 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, 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, HYLS1, 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, KCNQ1OT1, KCTD1, KDM1A, KDM6A, KDM6B, KIAA0586, KIAA0753, KIAA0825, KIAA1109, KIAA1279, KIF14, KIF1A, KIF22, KIF3B, KIF5C, KIF7, KL, KLHL40, KLHL41, KMT2A, KMT2C, KMT2D, KPTN, KRAS, KYNU, LBR, LBX1, LEMD3, LFNG, LGI4, LHX4, LIFR, LIG4, LMBR1, LMNA, LMNB1, LMOD3, LMX1B, LONP1, LPIN2, LRP2, LRP4, LRP5, LTBP2, LTBP3, LZTFL1, LZTR1, MAB21L1, MAB21L2, MACROH2A1, MAD2L2, MAF, MAFB, MAGEL2, MAGI2, MAGT1, MAN2B1, MANBA, MAP1B, MAP2K1, MAP2K2, MAPK1, MAPK3, MAP3K7, MASP1, MATN3, MBTPS1, MBTPS2, MCM5, MED12, MEGF8, MEIS2, MEOX1, MESD, MESP2, MET, MGAT2, MGP, MKKS, MKS1, MMP13, MMP14, MMP2, MMP9, MN1, MNX1, MOGS, MPDU1, MPI, MPZ, MRAS, MRPS16, MRPS28, MSX1, MSX2, MTAP, MTX2, MUSK, MYBPC1, MYCN, MYH3, MYH8, MYO18B, MYO9A, MYOD1, MYPN, NAA10, NAGLU, NANS, NBAS, NBN, NCAPG2, NEB, NECTIN4, NECTIN1, NEDD4L, NEK1, NEK9, NEPRO, NEU1, NEXMIF, NF1, NFIX, NFIX, NFKB2, NGLY1, NHEJ1, NIN, NIPBL, NIT1, NKX2-1, NKX3-2, NKX2-5, NLRP3, NOG, NOTCH1, NOTCH2, NPEPPS, NPHP3, NPR2, NPR3, NRAS, NSD1, NSDHL, NSMCE2, NSUN2, NUP88, NUS1, NXN, OBSL1, OCRL1, OFD1, ORC1, ORC4, ORC6, OSTM1, P3H1/LEPRE1, P4HB, PALB2, PAM16, PAPSS2, PARP1, PARP2, PARP3, PARP4, PAX1, PAX3, PAX8, PBX1, PCDH12, PCNA, PCNT, PCYT1A, PDE3A, PDE4D, PDE6D, PDGFRB, PEX5, PEX7, PGAP2, PGAP3, PGK1, PGM1, PHEX, PHF8, PHGDH, PHIP, PI4KA, PIBF1, PIEZO1, PIEZO2, PIGA, PIGB, PIGK, PIGO, PIGS, PIGT, PIGV, PIGW, PIGY, PIK3C2A, PIK3CA, PIK3R1, PIK3R2, PIP5K1C, PISD, PITX1, PITX2, PLAA, PLAG1, PLC/HSPG2, PLCB3, PLEKHM1, PLK4, PLOD1, PLOD2, PLS3, PML, PPM1M, PMM2, POC1A, POLE, POLR1A, POLR1B, POLR1C, POLR1D, POLR3A, POMP, POP1, POP1, POR, PORCN, PPIB, PPM1D, PPP1CB, PPP1R12A, PPP3CA, PRDM5, PRG4, PRIM1, PRKAR1A, PRMT7, PROM1, PRRX1, PSAT1, PSMD12, PSPH,

PTDSS1, PTEN, PTH, PTH1R, PTHLH, PTCH1, PTCH2, PTK7, PTPN11, PUF60, PUM1, PYCR1, QRICH1, RAB23, RAB33B, RAB34, RAD21, RAD51, RAD51C, RAF1, RAI1, RALGAPA1, RAP1A, RAP1B, RAPSN, RARB, RBBP8, RBBPS, RBM10, RBM8A, RBMX, RBPJ, , RECQL4, RFT1, RFWD3, RHOA, RIN2, RIPK4, RIPPLY2, RMRP, RNF125, RNU4ATAC, ROR2, RPGRIP1L, RPL10, RPL11, RPL13, RPL15, RPL26, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS6KA3, RPS7, RRAS2, RSPOR2, RSPRY1, RTTN, RUNX2, RYR1, SALL1, SALL4, SAMD9, SATB2 , SBDS, SBF1, SC5D/SC5DL, SCARF2, SCYL2, SDC2, SDCCAG8, SEC23A, SEC24D, SEM1, SEMA3E, SERPINF1, SERPINH1, SETBP1, SETD2, SF3B4, SFMBT1, SFN, SFRP4, SGMS2, SGSH, SH3BP2, SH3PXD2B, SHH, SHOC2, SHOX, SIK3, SIN3A, SIX2, SKI, SLC10A7, SLC17A5, SLC18A3, SLC25A24, SLC26A2, SLC26A4, SLC26A7, SLC29A3, SLC2A1, SLC2A2, SLC34A1, SLC34A3, SLC35A1, SLC35A3, ~~SLC35C1~~SLC35B2, LC35C1, SLC35D1, SLC39A13, SLC39A8, SLC4A2, SLC5A5, SLC5A7, SLC6A9, SLC9A3R1/NHERF1, SLCO2A1, SLCO5A1, SLURP1, SLX4, SMAD3, SMAD4, SMARCA2, SMARCA4, SMARCAL1, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SMC1A, SMC3, SMO, SMOC1, SMPD4, SMS, SNRPB, SNX10, SON, SOS1, SOS2, SOST, SOX11, SOX3, SOX4, SOX6, SOX9, SP7, SPARC, SPECC1L, SPG20, SPRTN, SRCAP, SRD5A3, SSR4, STAC3, STAG2, STAMBP, STAT3, STT3A, STT3B, STX16, SUFU, SUMF1, SUZ12, SYNE2, SYNE1, TAB2 , TALDO1, TAPT1, TASP1, TBC1D2B, TBC1D24, TBCD, TBCE, TBL1X, TBL1XR1, TBX1, TBX15, TBX2, TBX22, TBX3, TBX4, TBX5, TBX6, TBX18, TBXAS1, TBXT, TCF12, TCF20, TCIRG1, TCOF1, TCTEX1D2, TCTN2, TCTN3, TELO2, TENT5A/FAM46A, TERT, TFAP2A, TFAP2B, TG, TGDS, TGFB1, TGFB2, TGFB3, TGFBR1, TGFBR2, THRA, TLK2, TMCO1, TMEM38B, TMEM43, TMEM67, TMEM94, TMEM107, TMEM138, TMEM165, TMEM199, TMEM216, TMEM231, TMEM237, ~~TMEM38B, TMEM43, TMEM67,~~ TMEM263, TNFRSF11A/RANK, TNFRSF11B/OPG, TNFSF11/RANKL, TNNI2, TNNT1, TNNT3, TONSL, TP63, TPM2, TPM3, TPO, TRAF3IP1, TRAIP, TRAPPC2/SEDL, TRAPPC6A, TREM2, TRH, TRHR, TRIM32, TRIO, TRIP11, TRIP4, TRPS1, TRPV4, TRPV6, TRRAP, TSHB, TSHR, TTC21B, TTC8, TTN, TWIST1, TWIST2, TXNDC15, TYROBP, UBA1, UBA2, UBE2T, UBE3B, UBR1, UFSP2, USP9X, VAC14, VDR, VIPAS39, VPS13B, VPS33A, VPS33B, VPS35L/C16ORF62, VPS41, VPS4A, WDPCP, WDR19, WDR34, WDR35, WDR37, WDR60, WNT1, WNT10A, WNT10B, WNT3, WNT5A, WNT7A, XAGE1B, XRCC2, XRCC4, XYLT1, XYLT2, YY1, YY1AP1, ZBTB7A, ZBTB16, ZBTB42, ZC4H2, ZFHX4, ZIC1, ZIC3, ZMIZ1, ZMPSTE24, ZNF141, ZNF30, ZNF335, ZNF462, ZNF469, ZSWIM6)

~~NGS panel ALL IN ONE (57 genů: *APOB, APOE, LDLR, LDLRAP1, PCSK9, STAP1, PHEX, COL2A1, FGFR3, SHOX, RUNX2, ATP7B, CP, B3GALNT2, B4GAT1, CCDC88C, CRB2, DAG1, FKRP, FKTN, FLNA, FLVCR2, ISPD, L1CAM, LARGE1, MPDZ, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PTEN, WDR81, CFTR, CPA1, CTRC , SPINK1, PAH, BMP2, FTH1, HAMP, HFE, HJV (HFE2), SLC40A1, TFR2, BRAF (Exon 15), DCC (Exon 13), DCC (Exon 14), DCC (Exon 15a), DCC (Exon 15b), DCC (Exon 16), EGFR, IDH1 (Exon4), IDH2 (Exon4), KRAS, NRAS, CCL2, FUZ, TBXT, VANGL1, VANGL2*)~~

NGS panel Onemocnění a vady očí (~~625~~637 genů: *AAAS, ABCA1, ABCA4, ABCB6, ABCC6, ABHD12, ABHD5, ACBD5, ACO2, ACTB, ACTG1, ADAM9, ADAMTS10, ADAMTS17, ADAMTS18, ADAMTSL4, ADGRA3 (GPR125), ADGRV1 (GPR98, USH2B), ADIPOR1, AFG3L2 , AGBL1, AGBL5, AGK, AGPS, AHI1, AHR, AIPL1, ALDH18A1, ALDH1A3, ALMS1, ALX1, ANOS1 (KAL1), AP3B1, AP3D1, APC, APOE, APTX, ARHGEF18, ARL13B, ARL2BP, ARL3, ARL6, ARMC9, ARMS2, ARNT2, ARR3, ARSG (USH4), ASB10, ATF6, ATOH7, ATP1A2, ATP1A3, ATXN1, ATXN7, B3GALNT2, B3GALT1, B3GLCT, B4GAT1 (B3GNT1), B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCOR, BDNF, BEST1, BFSP1, BFSP2, BLOC1S3, BLOC1S6, BMP4, BMP7, BRD4, C12orf57, C12orf65, C1QTNF5, C2, C3, C8orf37, C9, CA4, CABP4, CACNA1F, CACNA2D4, CAPN5, CASK, CC2D2A, CDH23 (USH1D), CDH3, CDHR1, CENPF, CEP104, CEP164, CEP250, CEP290 (NPHP6), CEP41, CEP78, CERKL, CFAP410~~, ~~CFAP410~~ *(C21orf2), CFB, CFH, CFHR1, CFHR3, CFI, CIB2 (USH1J,KIP2), CISD2, CLCC1, CLDN19, CLN3, CLRN1 (USH3A), CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL11A2, COL17A1, COL18A1, COL25A1, COL2A1, COL4A1, COL4A3, COL8A2, COL9A1, COL9A2, COL9A3, COX7B, CPAMD8, CPLANE1 (C5orf42), CPSF1, CRB1, CRIM1, CRPPA (ISPD), CRX, CRYAA, CRYAB, CRYBA1, CRYBA2, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGB, CRYGC, CRYGD, CRYGS, CSPP1, CST3, CTC1, CTDP1, CTNNA1, CTNNB1, CTNS, CX3CR1,*

CYP1B1, CYP27A1, CYP4A2 (CYP4A11), CYP4V2, CYP51A1, DAG1, DCN, DCT, DDX58, DDX59, DHCR7, DHDDS, DHX38 (PRPF16), DNM1L, DNMBP, DRAM2, DTHD1, DTNBP1, EDN3, EDNRB, EED, EFEMP1, ELOVL4, ELOVL5, ELP1 (IKBKAP), ELP4, EMC1, EPG5, EPHA2 (ARCC2,CTPP), EYA1, EYS, FAM126A, FAM161A, FAR1, FBLN5, FBN1, FBN2, FGF10, FGF8, FGFR1, FKRP, FKTN, FLNA (FLN1), FLVCR1, FOXC1, FOXD3, FOXE3, FOXL2, FRAS1, FREM1, FREM2, FRMD7, FSCN2, FTL, FXN, FYCO1, FZD4, FZD5, GALK1, GALT, GCNT2, GDF3, GDF6, GFER, GJA1, GJA3, GJA8, GLI2, GLI3, GLIS3, GMPPA, GMPPB, GNAT1, GNAT2, GNB3, GNPAT, GNPTG, GP1BA, GPR143, GPR179, GRHL2, GRID2, GRIP1, GRK1, GRM6, GSN, GTF2H5, GUCA1A, GUCA1B, GUCY1A1 (GUCY1A3), GUCY2D, GZF1, HARS1 (USH3B), HCCS, HDAC6, HERC2, HESX1, HGSNAT, HK1, HMCN1, HMGB3, HMX1 (NKX5-3), HOXA1, HPS1, HPS3, HPS4, HPS5, HPS6, HS6ST2, HSF4, HTRA1, HTRA2, HYLS1, CHD7, CHM, CHMP4B, CHN1, CHRDL1, CHST6, IARS2, IDH3B, IFT140, IFT172, IFT27, IFT43, IGBP1, IMPDH1, IMPG1, IMPG2, INPP5E, INPP5K, INVS (NPHP2), IQCB1, ITM2B, ITPR1, JAG1, JAM3, KCNJ13, KCNV2, KERA, KIAA0586, KIAA1549, KIF11, KIF21A, KIF7, KIT, KITLG, KIZ, KLHL7, KMT2D, KRT12, KRT3, LAMA1, LAMB2, LARGE1, LCA5 (C6orf152), LCAT, LEMD2, LHX3, LHX4, LIM2, LMX1B, LONP1, LOXHD1, LOXL1, LOXL3, LRAT, LRIT3, LRMDA (C10orf11), LRP2, LRP5, LRPAP1, LSS, LTBP2, LYST, LZTFL1, MAB21L2, MAF, MAFB, MAK, MAN2B1, MC1R, MED25, MERTK, MFN2, MFRP, MIP (AQP0), MIPEP, MITF, MKKS, MKS1, MLPH, MSMO1, MSTO1, MTPAP, MTTP, MUSK, MVK, MYH9, MYO5A, MYO7A (USH1B), MYOC, MYRF, NAA10, NDP, NDUFB11, NDUFS1, NEK2, NEUROD1, NF2, NHS, NLRP1, NMNAT1, NOTCH2, NPHP1, NPHP3, NPHP4, NR2E3, NR2F1, NRL, NTF4, NYX, OAT, OCA2, OCRL, OFD1, OPA1, OPA3, OPN1LW, OPN1MW, OPN1SW, OPTN, OR2W3, OTX2, OVOL2, P3H2 (LEPREL1), P4HA2, PABPN1, PANK2, PANK4, PAX2, PAX3, PAX6, PCARE (C2orf71), PCDH15 (USH1F), PCYT1A, PDE6A, PDE6B, PDE6C, PDE6G, PDE6H, PDZD7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGK1, PHOX2A, PHYH, PIGL, PIK3R1, PIKFYVE, PITPNM3, PITX1, PITX2, PITX3, PLA2G5, PLCB3, PLK4, PMM2, PNKP, PNPLA6, POC1B, POLA1, POLG, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PORCN, POU1F1, PPP1CB, PRCD, PRDM13, PRDM5, PRIMPOL (CCDC111), PROKR2, PROM1, PROP1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2 (RDS), PRPS1, PRSS56, PTCH1, PUF60, PXDN, RAB18, RAB27A, RAB28, RAB3GAP1, RAB3GAP2, RAPSN, RARB, RAX, RAX2 (RAXL1), RBP3, RBP4, RCBTB1, RD3, RDH11, RDH12, RDH5, REEP6, RGR, RGS9, RGS9BP, RHO (OPN2), RIMS1, RLBP1 (CRALBP), ROBO3, ROM1, RP1, RP1L1, RP2, RP9 (PAP1), RPE65, RPGR, RPGRIP1, RPGRIP1L, RS1, RTN4IP1, RXYLT1 (TMEM5), RYR1, SACS, SAG, SALL2, SALL4, SAMD11, SBF2, SC5D, SCAPER, SCLT1, SCO2, SDCCAG8, SEMA3E, SEMA4A, SETX, SH3PXD2B, SHH, SIL1, SIPA1L3, SIX3, SIX6, SLC16A12, SLC24A1, SLC24A5, SLC25A3, SLC25A4, SLC2A1, SLC33A1, SLC38A8, SLC39A5, SLC45A2 (MATP), SLC4A11, SLC4A4, SLC7A14, SLITRK6, SMO, SMOC1, SNAI2, SNRNP200, SNX3 (MCOPS8), SOX1, SOX10, SOX2, SOX3, SPATA7, SPP2, SRD5A3, SSBP1, STAG2, STN1, STRA6, STS, STX3, TACSTD2, TAT, TAX1BP3, TBC1D20, TBX22, TCF4, TDRD7, TEAD1, TEK, TENM3, TFAP2A, TGFBI, TIMM8A, TIMP3, TINF2, TLR4 (ARMD10), TMEM126A, TMEM237, TMEM67, TMEM98, TOPORS, TP63, TPP1, TRAF3IP1, TRAPPC11, TREX1, TRIM32, TRIM37, TRIM44, TRNT1, TRPM1, TSPAN12, TTC8, TTLL5, TTPA, TUB, TUBB3, TUBGCP4, TUBGCP6, TULP1, TWNK (C10orf2), TYR, TYRP1, UBE3B, UBIAD1, UNC119, UNC45B, USH1C (PDZ73), USH1G (SANS), USH2A, USP45, VAX1, VCAN, VIM, VPS13B, VSX1, VSX2 (CHX10), WASHC5 (KIAA0196), WDPCP, WDR19 (KIAA1638, NPHP13), WDR36, WFS1, WHRN (USH2D,DFNB31), WRN, WT1, XPNPEP2, XYLT2, YAP1, YME1L1, ZBTB20, ZEB1, ZFHX4 (ZFH4), ZNF408, ZNF423, ZNF469, ZNF513, ZNF644)

NGS panel Onemocnění ledvin (~~358~~484 genů: ~~*C8ORF38/NDUFAF6, CASR, CD19, CD81, CFAP298, CFH, COQ6, CR2, GATM, GCM2, GNAS, GSN, ICOS, IKZF1, IL21, IRF2BP2, IRS4, ITGA3, KIAA0974/FAM149B1, LAGE3, LRBA, MNS1, MS4A1, NEK10, NFKB1, NFKB2, OSGEP, PDSS2, PTH, SLC36A4, TBL1X, TNFRSF13B, TNFRSF13C, TP53RK, TPP2, TPRKB, TRHR, TRPV6, TSC1, TSHB, TTC12, WDR4, WNT11, ABCA3*~~*ABCA3, ABCB11, ABCB4, ABCC2, ABCC6, ACE, ACTN4, ACVR2B, ADAMTS13, ADAMTS9, AGT, AGTR1, AGTR2, AGXT, AHI1, AKR1D1, ALDOB, ALG8, ALMS1, AMACR, ANKFY1/KIAA1255, ANKS6, ANLN, ANOS1/KAL1, AP2S1, APOA1, APOL1, AQP2, AR, ARHGAP24, ARHGDIA, ARL13B, ARL2BP, ARL3, ARL6, ARMC4, ARMC9, ATP6AP2, ATP6V0A4,*

ATP6V1B1, ATP8B1, AVIL, AVP, AVPR2, B2M, B9D1, B9D2, BAAT, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BICC1, BMP4, BMP7, BMPR1B, BSND, ~~C21orf59~~C2CD3, C2orf71, C3, ~~C5orf42/CPLANE1,~~ c8orf37, C8ORF38/NDUFAF6, CA2, CACNA1H, CAMKMT, CASR, CC2D2A, CCDC103, CCDC114, CCDC141, CCDC151, CCDC28B, CCDC39, CCDC40, CCDC65, CCM2/C7orf22, CCNO, CD19, CD2AP, CD46/MCP, CD81, CDC5L, CDC73, CELSR2, CENPF, CEP104, CEP120, CEP164, CEP290/NPHP6, CEP41, CEP55, CEP83, CFAP221/PCDP1, CFAP298/C21orf59, CFAP300, CFAP52, CFAP53/CCDC11, CFB, CFC1, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, ~~CFI,~~ CFTR, ~~CLCN5~~CLCN2, CLCN5, CLCNKA, CLCNKB, CLDN14, CLDN16, CLDN19, CNNM2, COL4A1, COL4A3, COL4A4, COL4A5, COL4A6, COQ2, COQ6, COQ8B, CR2, CRB2, CRELD1, CRIM1, CSF2RA~~,~~ CSF2RB, CSPP1, CTNS, CUL3, CYP11B1, CYP11B2, CYP24A1, CYP27A1, DACT1, DAW1, DCDC2, DGKE, DMP1, DNAAF1, DNAAF2/KTU, DNAAF3, DNAAF4~~-~~/DYX1C1, DNAAF5, DNAH1, DNAH11, DNAH3, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB11, DNAJB13, DNAL1, DRC1, DSTYK, DYNC2H1, DYNC2LI1, DZIP1L, EGF, EHHADH, EMP2, ENPP1, EVC, EVC2, EYA1, FAH, FAM111A, FGA, FGF20, FGF23, FGF8, FGF9, FN1, FOXC1, FOXE1, FOXH1, FOXI1, FOXJ1, FRAS1, FREM1, FREM2, FXYD2, G6PC, GALNT3, GANAB, GAPVD1, GAS2L2, GAS8/GAS11/DRC4, GATA3, GATA6, GATM, GBE1, GCM2, GDF1, GDNF, GJA1, GLI3, GLIS2/NPHP7, GLIS3, GNA11, GNAS, GREB1L~~–~~/KIAA1772/C18ORF6, GREM1, GRHPR, GRIP1, GSN, HNF1A, HNF1B, HNF4A, HOGA1, HSD11B2, HSD3B7, HTRA1, HYLS1, CHD1L, CHRM3, ICOS, IFIH1, IFT122, IFT140, IFT172, IFT27, IFT43, IFT52, IFT74/CCDC2, IFT80, IFT81, IKZF1, IL21, ILK, IMPDH1, INF2/C14ORF173, INPP5E, INTU/KIAA1284, INVS/NPHP2, IQCB1/NPHP5, IRF2BP2, IRS4, ITGA3, ITGA8, KANK2/KIAA1518, KAT6B, KCNA1, KCNJ1, KCNJ10, KCNJ5, KIAA0556/KATNIP, KIAA0586, KIAA0753, KIAA0974/FAM149B1, KIF14, KIF3B, KIF7, KL, KLHL3, KRIT1, LAGE3, LAMB2, LCA5, LEFTY2, LIFR, LIN7C, LMX1B, LRBA, LRP2BP, LRP4, LRP5, LRRC56, LRRC6, LRRCC1, LYZ, LZTFL1/BBS17, MAGED2, MAGI2/KIAA0705~~, /~~AIP1, MAPKBP1, MCIDAS, MEN1, MKKS, MKS1, MMACHC, MMP21, ~~MMP21~~MNS1, MS4A1, MUC1, MYH9, MYO1E, MYO5B, MYOG, NAT1, NCF1, NEK1, NEK10, NEK4, NEK8/NPHP9, NFKB1, NFKB2, NFU1, NKX2-1, NKX2-5, NME8, NODAL, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NR1H4, NR3C2, NRIP1, NUP107, NUP133, NUP160/NPHS19, NUP205, NUP37, NUP85, NUP93, OCRL, OFD1, OSGEP, PAX2, PAX8, PBX1, PCBD1, PCDH15, PCSK5, PDCD10, PDE2A, PDE3A, PDE6D, PDSS2, PHEX, PIBF1, PIH1D3, PKD1, PKD1L1, PKD2, PKHD1, PLCE1, PLXND1, PMM2, PNPLA6, PREPL, PRKCD, PRKCSH, PTH, PTPN22, PTPRO, RAB23, REN, RET, ROBO2, RPE65, RPGR, RPGRIP1, RPGRIP1L/NPHP8, RSPH1, RSPH3, RSPH4A, RSPH9, SALL1, SALL4, SCARB2, SCLT1, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SEC61A1, SEC63, SEMA3E, SFTPA1, SFTPB, SFTPC, SFTPD, SGPL1, SHROOM3, SIX1, SIX2, SIX5, SLC12A1, SLC12A3, SLC25A13, SLC26A4, SLC2A2, SLC34A1, SLC34A3, SLC36A4, SLC37A4, SLC3A1, SLC4A1, SLC4A4, SLC51A, SLC5A1, SLC5A2, SLC7A9, SLC9A3R1, SLIT2, SMAD4, SMARCAL1, SMO, SOX17, SPAG1, SPEF2, SPRY2, STK36, SUFU, TAC3, TBC1D8B, TBL1X, TBX18, TCTEX1D2, TCTN1/TECT1, TCTN2, TCTN3, TFAP2A, TH, THBD, THRA, THRB, TJP2, TMEM107, TMEM127, TMEM138, TMEM216, TMEM231, TMEM237, TMEM256, TMEM67, TNFRSF13B, TNFRSF13C, TOPORS, TP53RK, TPP2, TPRKB, TRAF3IP1, TRHR, TRIM32, TRPC6, TRPM6, TRPV6, TSC1, TSC2, TSHB, TSHR, TTC12, TTC21B, TTC25, TTC8, TTF1, TXNDC15/C5orf14, ULK4, UMOD, UPK3A, USF2, VHL, WDPCP/C2ORF86, WDR19, WDR34, WDR35, WDR4, WDR60, WDR72, WDR73, WNK1, WNK4, WNT11, WNT4, WT1, XDH, XPNPEP3, ZIC3, ZMYND10, ZNF423)

NGS panel Onemocnění a vady srdce (~~241~~263 genů: *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, BRAF, C1R, C1S, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CALR3, CASQ2, CAV3, CBL, CBS, CFC1, CITED2, COL12A1, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, ~~COL12A1,~~ COX15, CRELD1, CRYAB, CSRP3, CST3, CTF1, CTNNA3, DES, DMD, DNAJC19, DOLK, DPP6, DSC2, DSE, DSG2, ~~DSE,~~ DSP, DTNA, EFEMP2~~,~~ (FBLN4), ELN, EMD, EPG5, EYA4, FBLN5, FBN1, FBN2, FGA, FHL1, FHL2, FHOD3, FKBP14, FKRP, FKTN, FLNA, FLNC, FOXE3, GAA, GATA4, GATA5, GATA6, GATAD1, GDF1, GJA1, GJA5, GLA, GORAB, GPD1L, GSN, HCN4, HFE, HRAS, CHST14, ILK, ITM2B, JAG1, JPH2, JUP,*

KAT6B, KCNA5, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5 (KCNE1L), KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, KLF10, KRAS, LAMA4, LAMP2, LDB3, LDLR, LDLRAP1, LEFTY2, LIPA, LMNA, LOX, LPL, LTBP2, LTBP3, LTBP4, LYZ, LZTR1, MAP2K1, MAP2K2, MED12, MFAP5, MIB1, MMP21, MNS1, MRAS, MURC, MYBPC3, MYH11, MYH6, MYH7, MYL2, MYL3, MYLK, MYLK2, MYO6, MYOM1, MYOZ2, MYPN, NEBL, NEXN, NKX2-5, NKX2-6, NODAL, NOS1AP, NOTCH1, NPPA, NR2F2, NRAS, OSMR, P4HA1, PCSK9, PDLIM3, PKD1L1, PKP2, PLN, PLOD1, PPA2, PPP1CB, ~~PRDM5,~~ PRDM16, PRDM5, PRKAG2, ~~PRKG1,~~ PRKAR1A, PRKG1, PRNP, PSEN1, PSEN2, PTPN11, PYCR1, RAF1, RANGRF, RASA2, RBM20, RIN2, RIT1, ROBO4, RRAS, RRAS2, RYR2, SAA1, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, ~~SCN10A,~~ SCO2, SDHA, SGCD, SHOC2, SKI, SLC25A4, SLC2A10, SLC39A13, SLMAP, SMAD2, SMAD3, SMAD4, SMAD6, ~~SLMAP,~~ SNTA1, SORL1, SOS1, SOS2, SPRED1, STAP1, SYNE2, TAF1A, TAZ, TBX1, TBX20, TBX3, TBX5~~, TBX20~~, TCAP, TECRL, TGFB2, TGFB3, TGFBR1, TGFBR2, TLL1, TMEM43, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TNXB, TOMM40, TPM1, TRDN, TREM2, TRPM4, TSPYL1, TTN, TTR, TXNRD2, VCL, ZFPM2, ZIC3, ZNF469)

NGS panel Vzácná onemocnění (~~525~~636 genů: *A2ML1, AAAS, ABL1, ACAN, ACVR1, ACVRL1, ACTB, ACTG1, ADAMTS2, ADAMTS10, ADAMTS17, ADAMTSL2, ADGRG2, AEBP1, AFF4, AGGF1, AIRE, AKR1C2, AKR1C4, AKT1, AKT2, ALX1, ALX3, ALX4, ANAPC1, ANGPT2, ANKRD11, ANOS1, ANTXR2, AP1S1, APC2, AR, ARHGAP29, ARHGAP31, ARID1A, ARID1B, ARID2, ARSL, ARX, ASCC1, ASCL1, ASXL1, ASXL2, ASXL3, ATN1, ATP7B, ATR, ATRIP, B3GALTL, B3GALT6, B4GALT7, B3GAT3, BHLHA9, BICD2, BICRA, BMP2, BMP4, BMPR1B, BRAF, BRD4, BRWD3, BTK, BTRC, C1R, C1S, C2CD3, CASR, CANT1, CALCRL, CBL, CBX2, CCDC8, CCDC22, CCDC141, CCND2, CCL2, CD96, CDC45, CDH1, CDH3, CDK5, CDKL5, CDKN1C, CDON, CDT1, CDC6, CELSR1, CENPJ, CENPE, CENPF, CEP120, CEP152, CEP63, CFTR, CIBAR1, CKAP2L, CNOT1, CNTNAP2, COG4, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL10A1, COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, COL12A1, COLEC10, COLEC11, COMP, COQ6, CP, CPA1, CPE, CPLANE1, CREBBP, CTRC, CUL4B, CUL7, CYP11A1, CYP11B1, CYP17A1, CYP26B1, DCAF17, DCC, DCX, DDR2, DDX59, DHCR7, DHCR24, DHH, DHX37, DIS3L2, DISP1, DLL1, DLL4, DLX3, DLX4, DLX5, DLX6, DNA2, DNM2, DNMT3A, DOCK6, DOK7, DPF2, DPYSL5, DSE, DUSP6, DYNC1H1, DYNC2H1, DYNC2LI1, DVL1, DVL3, EBF3, EBP, ECE1, ECEL1, EDN3, EDNRB, EED, EFNB1, EFTUD2, ENG, EOGT, EP300, EPHB4, EPS15L1, ERF, ESCO2, EVC, EVC2, EXOC6B, EXT1, EXT2, EZH2, FAM20C, FBLN1, FBN1, FBN2, FEZF1, FGD1, FGF10, FGF16, FGF17, FGF8, FGF9, FGFR1, FGFR2, FGFR3, FIG4, FKBP14, FLNA, FLNB, FLRT3, FLT4, FOXC2, FOXE1, FOXG1, FOXH1, FRAS1, FREM1, FREM2, FSHB, FTH1, FUZ, FZD2, GABBR2, GAS1, GDF2, GDF3, GDF5, GDF6, GDNF, GH1, GHR, GHRH, GHRHR, GHSR, GJA1, GJB2, GJC2, GLE1, GLI1, GLI2, GLI3, GMNN, GNAS, GNRH1, GNRHR~~-~~, GPC3, GPC4, GPC6, GRHL3, GRIP1, GZF1, HAMP, HDAC4, HDAC8, HECW2, HESX1, HFE, HIST1H1E, HJV, HMGA2, HOXA13, HOXD13, HRAS, HS6ST1, HSD3B2, HSPA9, HYAL2, HYLS1, CHD7, CHD8, CHST3, CHST14, CHSY1, IARS1, IDH1, IDH2, IFT122, IFT140, IFT172, IFT43, IFT52, IFT57, IFT80, IGF1, IGF1R, IGF2, IHH, IL17RD~~-~~, INTU, IQCE, IQSEC2, IRF6, JAG1, JAM2, KAT6A, KAT6B, KATNB1, KCNH1, KCNQ1OT1, KCTD1, KDM6A, KIAA0586, KIAA0753, KIAA0825, KIFBP, KIF22, KISS1~~-~~, KISS1R~~-~~, KITLG, KLB, KLHL7, KMT2A, KMT2D, KRAS, KYNU, LAMB1, L1CAM, LBR, LGI4, LHB~~-~~, LHCGR, LHX3, LHX4, LIFR, LIG4, LMBR1, LMX1B, LOXL3, LRP2, LRP4, LTBP2, LTBP3, LZTR1, MACF1, MAGEL2, MAMLD1, MAP2K1, MAP2K2, MAP3K1, MATN3, MAU2, MASP1, MC2R, MCM5, MECP2, MED12, MEFV, MEGF8, MEIS2, MEOX1, MID1, MMP13, MNX1, MRAP, MRAS, MSX1, MSX2, MTOR, MUSK, MVK, MYBPC1, MYCN, MYH3, MYH8, MYLPF, MYO18B, MYOD1, MYORG, MYT1, NALCN, NBN, NDE1, NDNF, NECTIN1, NEK1, NEK9, NF1, NF1, NF2, NFIB, NFIX, NHLH2, NIN, NIPBL, NNT, NODAL, NOG, NOTCH1, NOTCH2, NR0B1, NR5A1, NRAS, NRTN, NRXN1, NSD1, NSMCE2, NSMF, NSUN2, NTNG1, NUP88, NXN, OBSL1, OFD1, ORC1, ORC4, ORC6, OTX2, P4HA1, PAFAH1B1, PAH, PAPSS2, PARP1, PAX3, PCNT, PDE3A, PDGFB, PDGFRB, PGAP2, PGAP3, PHF6, PHOX2B, PI4KA, PIEZO1, PIEZO2, PIGA, PIGL, PIGN, PIGO, PIGQ, PIGT, PIGV, PIGW, PIGY, PIK3CA, PIK3R1, PIK3R2, PITX1, PITX2, PLAG1, PLK4, PLOD1, PLXND1, POLA1, POLR1B, POLR1C, POLR1D, POMT1, POMT2, POR, PORCN, POU1F1, PPP1CB, PRKACB, PRKAR1A, PRMT7, PROK2, PROKR2, PROP1, PTEN, PTH1R, PTHLH, PTCH1, PTCH2, PTPN11, PTPN14, RAB23, RAD21, RAF1, RAI1, RAP1A, RAP1B, RAPSN, RASA1, RASA2, RASA3, RBBP8,*

RBM10, RBM8A, RBPJ, RECQL4, RELN, RET, REV3L, RIT1, RLIM, RNF125, RNF135, RNF216, RNPC3, ROR2, RPS6KA3, RRAS, RRAS2, RTEL1, RUNX2, SALL1, SALL4, SAMD9, SATB2, SCARF2, SC5D, SEM1, SEMA3A, SEMA3C, SEMA3D, SEMA3E, SEMA7A, SETD2, SETD5, SF3B2, SF3B4, SGPL1, SHH, SHOC2, SHOX, SIX3, SKI, SLC18A3, SLC20A2, SLC26A2, SLC29A3, SLC39A13, SLC40A1, SMAD3, SMAD4, SMAD6, SMARCA2, SMARCA4, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SMC1A, SMC3, SMO, SMPD4, SMS, SNAP29, SON, SOS1, SOS2, SOST, SOX2, SOX3, SOX4, SOX9, SOX10, SOX11, SOX18, SPECC1L, SPRED1, SPRED2, SPINK1, SPINT2, SPRY4, SRCAP, SRY, STAMBP, STAR, STAG2, STIL, STXBP1, SUFU, SUMO1, SUZ12, TAC3, TACR3, TANC2, TBC1D24, TBCK, TBX1, TBX2, TBX5, TBX15, TBX19, TBX22, TBXT, TCF12, TCF4, TCOF1, TCTEX1D2, TCTN3, TDGF1, TEK, TELO2, TFAP2A, TFAP2B, TFR2, TGDS, TGFB2, TGFB3, TGFBR1, TGFBR2, TGIF1, TIE1, TMCO1, TMEM107, TMTC3, TNNI2, TNNT3, TNXB, TOR1A, TP63, TPM2, TRAIP, TRIP11, TRPV4, TRPS1, TTC7A, TTC21B, TUBA1A, TWIST1, TXNL4A, UBE3A, UGT1A1, UNC80, USP7, USP9X, VANGL1, VANGL2, VEGFC, VIPAS39, VPS13B, VPS33B, VPS35L, WASHC5, WDPCP, WDR11-, WDR19, WDR34, WDR35, WDR60, WNT10B, WNT4, WNT5A, WNT7A, WT1, XPR1, YWHAE, ZEB2, ZFPM2, ZIC1, ZIC2, ZIC3, ZNF141, ZSWIM6)

Postup č. 8/816: **Analýza lidského genomu metodou MLPA**

Probemixy: P092 ABCC6 (*ABCC6, ABCC1*), P343 Autism-1 (15q11, 15q12, 15q13, 16p11.2, *SHANK3*), P191 Alport mix 1 (*COL4A5*), P192 Alport mix 2 (*COL4A5, COL4A6*), P180-B1 Limb malformation-2/Heart (*SALL1, SALL4, TBX5*), P002 BRCA1 , P045 BRCA2/CHEK2 , P439 COL4A3, P444 COL4A4, P250 DiGeorge (22q11.2), P461 DIS (*STRC, CATSPER2, CKMT1B, OTOA*), P018 SHOX, P060 SMA (*SMN1, SMN2*), P061 Lissencephaly (*PAFAH1B1, DCX, POMT1, POMGNT1, FLNA*), P062 LDLR, P297 Microdeletion syndromes-2 (1q21.1, 1q21.1, 3q29, 7q36.1, 12p11.23, 15q13, 15q24.1 16p11, 17q12, 18q21.2, 20p12.2), P356 Chromosome 22q probemix (22q11, 22q13), ME031 GNAS, P379 NRXN1, P245-Microdeletion syndromes-1A, ME030 BWS/RSS, ME028 Prader-Willi/Angelman, P326 LARGE1, P309 MTM1, P220 Obesity, P003 MLH1/MSH2, P008 PMS2, P043 APC, P052 Parkinson mix 2, P063 FHIT-WWOX, P070 Subtelomeres Mix 2B, P072 MSH6-MUTYH probemix, P133 Kallmann-2, P152 ABCA4 mix-2 probemix, P190 CHEK2 probemix, P240 BRIP-CHEK1, P291 Human Telomere-12, P355 Microcephaly probemix, P361 USH2A mix, P365 Human Telomere-14, P385 DOCK8, P387 NPHP1, P396 SHANK2, P405 CMT1, P443 KANSL1, ME032 UPD7-UPD14, P010 POLG, P015 MECP2, P016 VHL, P026 Sotos syndrome, P031 FANCA mix 1, P048 LMNA/MYOT/ZMPSTE24, P050 CAH, P051 Parkinson mix 1, P054 FOXL2-TWIST1, P065 Marfan Syndrome, P066 Marfan Syndrome, P067 PTCH1, P080 Craniofacial, P081 NF1 mix 1, P082 NF1 mix 2, P091 CFTR, P100 MYBPC3, P106 MRX, P108 SCN5A, P114 Long-QT, P118 WT1, P130 CCM mix-1, P131 CCM mix-2, P132 Kallmann-1, P136 Gitelman syndrome, P141 NIPBL-1 mix1, P142 NIPBL-2 mix2, P151 ABCA4 mix-1, P153 EYA1, P155 EDS, P159 GLA, P160 STS, P168 ARVC-PKP2, P169 Hirschsprung-1, P177 CASR, P185 Intersex, P186 PAX3-MITF-SOX10, P187 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.

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

Hemochromatóza (sekvenční varianty H63D, S65C a C282Y v genu *HFE*)
~~Familiární hypercholesterolemie typu B (sekvenční varianta R3500Q v genu *APOB*)~~
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*: *KRAS* (kodon 12- c.34G>A, c.34G>C, c.34G>T, c.35G>A, c.35G>C, c.35G>T, kodon 13- c.37G>A, c.37G>C, c.37G>T, c.38G>A, c.38G>C, c.38G>T, kodon 59- c.175G>T, c.175G>A, c.176C>A, c.176C>G, c.176_178delCAG, kodon 60, 61- c.179G>A, c.179G>C, c.179G>T, c.180T>A, c.180T>C, c.181C>G, c.181C>A, c.182A>T, c.182A>G, c.182A>C, c.183A>T, c.183A>C, kodon 117- c.349A>G, c.350A>G, c.351A>C, c.351A>T, kodon 146- c.436G>C, c.436G>A, c.437C>G, c.437C>T, c.438A>G, c.438A>C, c.438A>T), *NRAS* (sekvenční varianty: kodon 12- c.34G>A, c.34G>C, c.34G>T, c.34_35GG>AA, c.34_35GG>CC, c.34_35GG>TA, c.35G>A, c.35G>C, c.35G>T, kodon 13- c.37G>A, c.37G>C, c.37G>T, c. 37_38GG>AA, c. 37_38GG>TA, c.38G>A, c.38G>C, c.38G>T, c. 38_39GT>TC, c.39T>C, kodon 59- c.175G>A, c.176C>A, c.176C>G, c.181C>A, c.181C>G, c.181_182CA>AG, c.181_182CA>TT, c.182A>C, c.182A>G, c.182A>T, c.182_183AA>GG, c.182_183AA>TG, c.183A>C, c.183A>G, c.183A>T, kodon 117- c.349A>G, c.350A>G, c.351G>T, c.351G>C, kodon 146- c.436G>A, c.436G>C, c.436G>T, c.437C>T, c.437C>G), *BRAF* (sekvenční varianty: kodon 600- c.1799T>A, c.1799_1800TG>AT, c.1798_1799GT>AA, c.1798_1799GT>AG, c.1799T>C), *EGFR* (sekvenční varianty: kodon 719- c.2156G>C, c.2155G>A, c.2155G>T, kodon 746-753- c.2235_2249 del 15, c.2235_2252>AAT, c.2236_2253 del 18, c.2237_2251 del 15, c.2237_2254 del 18, c.2237_2255>T, c.2236_2250 del 15, c.2238_2255 del 18, c.2238_2248 >GC, c.2238_2252 >GCA, c.2239_2247 del 9, c.2239_2253 del 15, c.2239_2256 del 18, c.2239_2248TTAAGAGAAG>C, c.2239_2258>CA, c.2240_2251 del 12, c.2240_2257 del 18, c.2240_2254 del 15, c.2239_2251>C, c.2235_2248>AATTC, c.2237_2252>T, c.2235_2251>AATTC, c.2235_2255>AAT, c.2237_2257>TCT, c.2238_2252 del 15, c.2239_2256>CAA, kodon 790-c.2369C>T, kodon 768- c.2303G>T, kodon 772-775- c.2319_2320 insCAC, c.2315_2316 insGACAACCCC, c.2315_2316 insGGGCAACCC, c.2318A>T, c.2319_2320 insCCCCAC, c.2321_2322 insCCACGT, kodon 858- c.2573T>G, c.2573_2574TG>GT, kodon 861- c.2582T>A).

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>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)
~~*HFE* (sekvenční varianty: V53M, V59M, H63D, H63H, S65C, Q127H, P160delC, E168Q, E168X, W169X, C282Y, Q293P)~~
~~*TFR2* (sekvenční varianty: E60X, M172K, Y250X, AVAQ594-597del), *FPN1* (sekvenční varianty: N144H, V162del)~~

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 a WT1.

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

NGS panel Hereditární nádorové syndromy a somatické varianty v nádorových tkáních (185 genů: *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, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAT1, 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, PDGFB, PDGFD, PDGFRA, PDGFRB, PIK3CA, PMS2, POLD1, POLE, POT1, PPP2R2A, PTEN, PTCH1, RAD50, RAD51, RAD51B, RAD51C, RAD51D, RAD52, RAD54L, RB1, RECQL, RECQL4, RET, REXO2, RNF43, RPS20, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, SLX4, SMAD4, SMARCA4, SMARCB1, SMARCE1, SRY, STK11, SUFU, TERF2IP, TERT, TET2, TMEM127, TP53, VHL, WRN, WT1, XPA, XRCC2, XRCC3, ALK, ARAF, ARID1A, ARID1B, CCDC153, CCNE1, CD79B, CDADC1, CDK12, CDK6, CTNNB1, DDR2, ENG, ENOX1, ERG, ESR1, FAM124A, FGFR1, FGFR2, FGFR3, FOXL2, H3-3A, IL18, INCA1, KEAP1, MAD2L2, MALT1, MTOR, MYC, MYD88, NTRK1, NTRK2, NTRK3, NUTM1, PIK3R1, PML, PPP2R1A, RHOA, ROS1, RPAIN, S1PR1, TBX21, TMPRSS2, TOE1, TOP2A, WSCD1*)

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

Sekvenční varianty genů *KRAS, NRAS, BRAF, EGFR*: *KRAS* (kodon 12- c.34G>A, c.34G>C, c.34G>T, c.35G>A, c.35G>C, c.35G>T, kodon 13- c.37G>A, c.37G>C, c.37G>T, c.38G>A, c.38G>C, c.38G>T, kodon 59- c.175G>T, c.175G>A, c.176C>A, c.176C>G, c.176_178delCAG, kodon 60, 61- c.179G>A, c.179G>C, c.179G>T, c.180T>A, c.180T>C, c.181C>G, c.181C>A, c.182A>T, c.182A>G, c.182A>C, c.183A>T, c.183A>C, kodon 117- c.349A>G, c.350A>G, c.351A>C, c.351A>T, kodon 146- c.436G>C, c.436G>A, c.437C>G, c.437C>T, c.438A>G, c.438A>C, c.438A>T), *NRAS* (sekvenční varianty: kodon 12- c.34G>A, c.34G>C, c.34G>T, c.34_35GG>AA, c.34_35GG>CC, c.34_35GG>TA, c.35G>A, c.35G>C, c.35G>T, kodon 13- c.37G>A, c.37G>C, c.37G>T, c. 37_38GG>AA, c. 37_38GG>TA, c.38G>A, c.38G>C, c.38G>T, c. 38_39GT>TC, c.39T>C, kodon 59- c.175G>A, c.176C>A, c.176C>G,

c.181C>A, c.181C>G, c.181_182CA>AG, c.181_182CA>TT, c.182A>C, c.182A>G, c.182A>T, c.182_183AA>GG, c.182_183AA>TG, c.183A>C, c.183A>G, c.183A>T, kodon 117- c.349A>G, c.350A>G, c.351G>T, c.351G>C, kodon 146- c.436G>A, c.436G>C, c.436G>T, c.437C>T, c.437C>G), *BRAF* (sekvenční varianty: kodon 600- c.1799T>A, c.1799_1800TG>AT, c.1798_1799GT>AA, c.1798_1799GT>AG, c.1799T>C), *EGFR* (sekvenční varianty: kodon 719- c.2156G>C, c.2155G>A, c.2155G>T, kodon 746-753- c.2235_2249 del 15, c.2235_2252>AAT, c.2236_2253 del 18, c.2237_2251 del 15, c.2237_2254 del 18, c.2237_2255>T, c.2236_2250 del 15, c.2238_2255 del 18, c.2238_2248 >GC, c.2238_2252 >GCA, c.2239_2247 del 9, c.2239_2253 del 15, c.2239_2256 del 18, c.2239_2248TTAAGAGAAG>C, c.2239_2258>CA, c.2240_2251 del 12, c.2240_2257 del 18, c.2240_2254 del 15, c.2239_2251>C, c.2235_2248>AATTC, c.2237_2252>T, c.2235_2251>AATTC, c.2235_2255>AAT, c.2237_2257>TCT, c.2238_2252 del 15, c.2239_2256>CAA, kodon 790- c.2369C>T, kodon 768- c.2303G>T, kodon 772-775- c.2319_2320 insCAC, c.2315_2316 insGACAACCCC, c.2315_2316 insGGGCAACCC, c.2318A>T, c.2319_2320 insCCCCAC, c.2321_2322 insCCACGT, kodon 858- c.2573T>G, c.2573_2574TG>GT, kodon 861- c.2582T>A).

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>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).