

**Aktuální seznam činností prováděných v rámci flexibilního rozsahu akreditace  
Laboratoř Oddělení lékařské genetiky**

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

Geny: LDLR, ATP7B, GJB2

**Postup č. 2: 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 repetice CGG v 5'UTR oblasti genu FMR1)

**Postup č. 6: Analýza lidského genomu metodou masivně paralelního sekvenování (NGS panely)**

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

NGS panel Hereditární nádorové syndromy a somatické varianty v nádorových tkáních (84 genů: AKT, APC, ATM, ATR, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRAF, BRCA1, BRCA2, BRIP1, BRF1, CDH1, CDK4, CDKN2A, CYLD, DICER1, ENG, EPCAM, ERCC3, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCM, FANCP, FLCN, FOXL2, GPRC5A, GREM1, CHEK2, IDH1, IDH2, KLLN, KRAS, LKB1/STK11, MAD2L2, MET, MLH1, MLH3, MN1, MRE11, MSH2, MSH3, MSH6, MUTYH, NBN, NTHL1, NRAS, PALB2, PARP1, PARP2, PDGFB, PIK3CA, PMS2, PMS2CL, POLD1, POLE, POT1, PPM1D, PTCH1, PTEN, RAD50, RAD51C, RAD51D, RECQL, RECQL4, RET, RINT1, RNF43, RPS20, SCG5, SMAD4, SMARCA4, SRY, TEX15, TP53, WRN)

NGS panel Mikrocefalie (105 genů: ACTB, AGMO, ANKLE2, ANKRD11, AP3B1, ARFGF2, ARID1B, ASPM/MCPH5, ATR, AUTS2, BUB1B, CASC5, CASK, CDK5RAP2, CDK6, CENPE, CENPF, CENPJ, CEP135, CEP152, CKAP2L, CLP1, COX7B, CRIPT, CTNNB1, DIAPH1, DMRTA2, DNML1, DPP6, DYRK1A, EFTUD2, EP300, ERCC6, EXOSC3, FOXG1, FTO, GLI2, HDAC8, HMGB3, CHR1, IER3IP1, IQSEC2, KATNB1, KIF11, KIF1BP, MCPH1, MED17, MFSD2A, MSMD1, MYCN, NBN, NDE1, NHEJ1, NIPBL, OCLN, ORC1, PAFAH1B1, PCLO, PEX6, PHC1, PHGDH, PLEKHG2, PLK4, PNKP, POGZ, PPP1R15B, PQBP1, QARS, RAB3GAP1, RAD21, RAD50, RBBP8, RTTN, SASS6, SHH, SHOX2, SLC1A4, SLC25A19, SLC2A1, SLC9A6, SMC1A, SMC3, SOX11, SPATA5, STAMBP, STEAP3, STIL, TAF2, THOC6, TRAPPC9, TRMT10A, TSEN54, TUBA1A, TUBB, TUBB2B, TUBGCP4, TUBGCP6, VPS13B, WDR62, WDR73, XRCC4, ZBTB18, ZEB2, ZNF335)

NGS panel Porucha sluchu (174 genů: ABHD12, ACTG1, ADCY1, APOD, ATP1A2, ATP2B2, ATP6V1B1, BDP1, BSND, C19ORF53, CA14, CABP2, CATSPER2, CCDC50, CCS, CDH23, CEACAM16, CEP250, CIB2, CLDN14, CLIC5, CLPP, CLRN1, CLU, COCH, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, CRYM, DCDC2, DDR2, DFNA5, DGCR6, DIABLO, DIAPH1, DIAPH3, DNMT1, DSPP, EDN3, EDNRB, ELMOD3, EPS8, ESPN, ESRRB, EYA1, EYA4, FAM189A2, FAM65B, FBXO2, FGF3, FGFR3, FOXD1, FOXI1, GIPC3, GJA1, GJB2, GJB3, GJB6, GPR98, GPSM2, GRHL2, GRXCR1, GRXCR2, HARS2, HGF, HOMER2, HSD17B4, HYI, CHD7, IARS2, IGFBP6, ILDR1, INF2, ISLR, KARS, KCNE1, KCNJ10, KCNQ1, KCNQ4, KIAA1199, LARS2, LHFP, LHFPL5, LOXHD1, LRP1, LRTOMT, MARVELD2, MASP1, MET, MGP, MIA, MITF, MSRB3, MT3, MYH14, MYH9, MYO15A, MYO1A, MYO3A, MYO6, MYO7A, NARS2, NDP, OSBPL2, OTOA, OTOF, OTOG, OTOG, OTOR, P2RX2, PAX3, PCDH15, PCDHGC3, PDZD7, PJKV, PLEKHB1, PNPT1, POLR1D, POU3F4, POU4F3, PRPS1, PTGDS, PTPRQ, RBP1, RDX, S100A13, SELM, SEMA3E, SERPINB6, SIX1, SIX5, SLC12A1, SLC17A8, SLC26A4, SLC26A5, SLC33A1, SMPX, SNAI2, SOX10, STRC, SYNE4, TBC1D24, TCOF1, TECTA, TIMM8A, TJP2, TMC1, TMEM132E, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TSPEAR, UBA52, USH1C, USH1G, USH2A, USH3, USP11, UTY, VAMP5, WFS1, WHRN, WNK2, ZNF288, ZP2)

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NGS panel Mentální retardace (141 genů: ABCD1, ACSL4, AFF2, AGTR2, AIFM1, AP1S2, ARHGEF6, ARHGEF9, ARX, ATP6AP2, ATP7A, ATRX, B3GALNT2, B3GNT1, BCOR, BRWD3, CASK, CCDC22, CCDC88C, CDK16, CDKL5, CLCN4, CLIC2, CNKSR2, COL4A1, CUL4B, DAG1, DCX, DKC1, DLG3, DMD, EBP, EIF2S3, FAAH2, FANCB, FGD1, FKR1, FKTN, FLNA, FMR1, FRMPD4, FTSJ1, GDI1, GK, GMPPB, GPC3, GRIA3, GSPT2, GTDC2, HCCS, HCFC1, HDAC6, HDAC8, HPRT1, HSD17B10, HUWE1, IDS, IGBP1, IKBKG, IL1RAPL1, IQSEC2, ISPD, JARID1C, KDM5C, KIAA2022, KLF8, L1CAM, LAMP2, LAS1L, MAGT1, MAOA, MBTPS2, MEC2, MED12, MID1, MPDZ, MTM1, NAA10, NDP, NDUFA1, NHS, NLGN3, NLGN4X, NSDHL, NXF5, OCRL, OFD1, OGT, OPHN1, OTC, PAK3, PCDH19, PDHA1, PGK1, PHF6, PHF8, PHGDH, PLP1, POMGNT1, POMT1, POMT2, PORCN, PQBP1, PRPS1, PSAT1, PSPH, PTEN, PTCH1, RAB39B, RAB40AL, RBM10, RIBC1, RPL10, RPS6KA3, SGK196, SHROOM4, SLC16A2, SLC6A8, SLC9A6, SMC1A, SMS, SOX3, SRPX2, SYN1, SYP, TAF1, THOC2, TIMM8A, TM4SF2, TMEM5, TSPAN7, UBE2A, UPF3B, USP9X, WDR13, ZC4H2, ZCCHC12, ZDHHC15, ZDHHC9, ZMYM3, ZNF41, ZNF674, ZNF711, ZNF81)

NGS panel Vrozené kostní anomálie (444 genů: ACAN, ACP5, ACVR1, ADAMTSL2, AGA, AGO3, AGPS, AKT1, ALDH1A2, ALPL, ALX1, ALX3, ALX4, AMER1, ANKH, ANKRD35, ANO5, ANTXR2, ARHGAP31, ARID1A, ARID1B, ARL6, ARSB, ARSE, ATP6V0A2, B4GALT7, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BLM, BMP1, BMP2, BMP4, BMP7, BMPER, BMPR1B, BRCA2, BRIP1, BTRC, CA2, CANT1, CASR, CC2D2A, CCDC8, CD96, CDC6, CDH3, CDKN1C, CDT1, CENPE, CEP120, CEP290, CLCN5, CLCN7, COG1, COL10A1, COL11A1, COL1A1, COL1A2, COL2A1, COL9A1, COL9A2, COL9A3, COMP, CREB3L1, CREBBP, CRTAP, CSPP1, CTNNA1, CTSA, CTSK, CUL7, CYP26B1, CYP27B1, CYP2R1, DDR2, DDX59, DHCR24, DHCR7, DHODH, DKK1, DLL3, DLX1, DLX2, DLX3, DLX5, DLX6, DMP1, DOCK6, DYM, DYNC2H1, EBP, EDN1, EFNB1, EIF2AK3, EMX2, EN1, ENPP1, EOGT, EP300, EPHA3, ESCO2, ETS1, ETS2, ETV4, ETV5, EVC, EVC2, EXT1, EXT2, EZH2, FAM111A, FAM20C, FAM58A, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FBLN1, FBN1, FBN2, FBXW4, FERMT3, FGF4, FGF8, FGF9, FGF10, FGF16, FGF17, FGF23, FGFR1, FGFR2, FGFR3, FIG4, FKBP10, FLNA, FLNB, FMN1, FOXC1, FREM1, FREM2, FREM3, FUCA1, GABPA, GALNS, GALNT3, GDF5, GDF6, GJA1, GLB1, GLI1, GLI2, GLI3, GNAS, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GPC3, GPC6, GREM1, GUSB, HAND2, HDAC4, HES7, HFE2, HOXA3, HOXA9, HOXA10, HOXA11, HOXA13, HOXB3, HOXB9, HOXC9, HOXC10, HOXD3, HOXD4, HOXD9, HOXD10, HOXD11, HOXD12, HOXD13, HPGD, HSGNAT, HSS, CHD7, CHST3, CHST14, CHSY1, CHUK, ICK, IDH1, IDH2, IDS, IDUA, IFITM5, IFT43, IFT80, IFT122, IFT140, IFT172, IHH, IKBKG, IL1RN, IMPAD1, INPP5E, INPPL1, IRF6, ISL1, ITGA10, KAT6B, KIF22, KIF7, KL, LBR, LBX1, LBX2, LEMD3, LEPRE1, LFNG, LIFR, LIX1L, LMBR1, LMNA, LMX1B, LOXL3, LPIN2, LRP4, LRP5, MAFB, MAN2B1, MANBA, MATN3, MEIS1, MEIS2, MEIS3, MEOX1, MEPE, MESP2, MGP, MIPOL1, MKKS, MKS1, MMP13, MMP2, MMP9, MNX1, MSX2, MYCN, NAGLU, NBN, NEK1, NEU1, NF1, NFIX, NFKB2, NIN, NIPBL, NKX3-2, NLRP3, NOG, NOTCH2, NPHP3, NPPC, NPR2, NSD1, NSDHL, NSMCE2, OBSL1, OFD1, OPG, ORC1, ORC4, ORC6, OSTM1, P4HB, PALB2, PAM16, PAPSS2, PAX1, PBX1, PBX2, PBX3, PBX4, PCNA, PCNT, PCYT1A, PDE4D, PEX11B, PEX7, PGAP2, PGAP3, PGM3, PHEX, PHF8, PIAS3, PIGO, PIGV, PITX1, PITX2, PLC, PLEKHM1, PLOD2, PLS3, POC1A, POLR1C, POLR1D, POLR3GL, POR, PORCN, PPIB, PRDM1, PRG4, PRKAR1A, PROM1, PRRX1, PRRX2, PTHLH, PTHR1, PTCH1, PTPN11, PYCR1, RAB23, RAD51C, RANK, RANKL, RARB, RASGRP2, RBM8A, RBPJ, RDH10, RECQL4, ROR2, RPRG1, RPL11, RPL5, RPS19, RUNX2, SALL1, SALL4, SAMD9, SBDS, SC5D, SEC24D, SEDL, SEMA3E, SERPINF1, SERPINH1, SETBP1, SF3B4, SH3BP2, SH3PXD2B, SHH, SHOX, SKI, SLC17A5, SLC26A2, SLC29A3, SLC34A1, SLC34A3, SLC35D1, SLC39A13, SLC9A3R1, SLCO2A1, SLCO5A1, SMAD1, SMAD4, SMAD5, SMAD9, SMARCA2, SMARCA4, SMARCAL1, SMARCB1, SMARCE1, SNX10, SOST, SOX9, SP7, SP8, SP9, SULF1, SUMF1, TBCE, TBX15, TBX3, TBX4, TBX5, TBXAS1, TCIRG1, TCOF1, TCTN3, TFAP2A, TFAP2B, TGFB1, TGFB1R1, TGFB1R2, THPO, TMEM216, TMEM38B, TMEM67, TP63, TREM2, TRIM32, TRIP11, TRPS1, TRPV4, TTC21B, TTC8, TWIST1, TWIST2, TXNIP, TYROBP, VC, WDR19, WDR34, WDR35, WDR60, WISP3, WNT1, WNT10A, WNT10B, WNT2B, WNT3, WNT5A, WNT7A, WNT8A, WNT8B, XRCC4, XYLT1, ZBTB16, ZMPSTE24)

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NGS panel ALL IN ONE (54 genů: ATP7B, CCDC88C, CFTR, MPDZ, PTEN, POMT1, POMT2, POMGNT1, ISPD, FKTN, FKRP, DAG1, B3GALNT2, SGK196 (POMK), LDLR, APOB, APOE, PCSK9, LDLRAP1, STAP1, TBX5, SALL4, PHEX, CLCN5, L1CAM, CP, GNAS, FGFR2, FGFR3, DHCR7, CHD7, SEMA3E, SHOX, FGF23, PTPN14, GJC2, GATA2, PIK3CA, ADAMTS3, FAT4, VEGFC, RASA1, FLT4, GJA1, HGF, EPHB4, KIF11, AKT1, FOXC2, PIEZO1, CCBE1, SOX18, CELSR1, IKBKG)

NGS panel Onemocnění a vady očí (599 genů: AAAS, ABCA1, ABCA4, ABCB6, ABCC6, ABHD12, ABHD5, ACBD5, ACO2, ACTB, ACTG1, ADAM9, ADAMTS10, ADAMTS17, ADAMTS18, ADAMTSL4, ADGRV1, AGLB1, AGLB5, AGK, AGPS, AHI1, AIPL1, ALDH18A1, ALDH1A3, ALMS1, ANOS1, AP3B1, AP3D1, APC, APTX, ARHGEF18, ARL13B, ARL2BP, ARL6, ARMC9, ARNT2, ARR3, ARSG, ASB10, ATF6, ATOH7, ATP1A2, ATP1A3, ATXN1, ATXN7, B3GALNT2, B3GALT1, B3GLCT, B3GNT1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCOR, BDNF, BEST1, BFSP1, BFSP2, BLOC1S3, BLOC1S6, BMP4, BMP7, BRD4, C10ORF2, C12orf57, C12orf65, C1QTNF5, C5orf42, C8orf37, CA4, CABP4, CACNA1F, CACNA2D4, CAPN5, CASK, CC2D2A, CCDC111, CDH23, CDH3, CDHR1, CENPF, CEP104, CEP164, CEP250, CEP290, CEP41, CEP78, CERKL, CFAP410, CFB, CFH, CFI, CIB2, CISD2, CLDN19, CLN3, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL11A2, COL17A1, COL18A1, COL25A1, COL2A1, COL4A1, COL4A3, COL8A2, COL9A1, COL9A2, COL9A3, COX7B, CPAMD8, CRB1, CRX, CRYAA, CRYAB, CRYBA1, CRYBA2, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGB, CRYGC, CRYGD, CRYGS, CSPP1, CST3, CTC1, CTDSP1, CTNNA1, CTNNA2, CX3CR1, CYP11B1, CYP27A1, CYP4V2, CYP51A1, DAG1, DCN, DDX59, DHCR7, DHDDS, DHX38, DNMT1, DNMBP, DRAM2, DTHD1, DTNBP1, EDN3, EDNRB, EED, EFEMP1, ELOVL4, ELOVL5, ELP4, EMC1, EPG5, EPHA2, EYA1, EYS, FAM126A, FAM161A, FAR1, FBLN5, FBN1, FBN2, FGF10, FGF8, FGFR1, FKRP, FKTN, FLNA, 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, GMPPB, GNAT1, GNAT2, GNB3, GNPAT, GNPTG, GP1BA, GPR125, GPR143, GPR179, GRHL2, GRID2, GRIP1, GRK1, GRM6, GSN, GTF2H5, GUCA1A, GUCA1B, GUCY1A1, GUCY2D, GZF1, HARS, HCCS, HERC2, HESX1, HGSNAT, HK1, HMCN1, HMGB3, HMX1, HOXA1, HPS1, HPS3, HPS4, HPS5, HPS6, HS6ST2, HSF4, HTRA1, HTRA2, HYLS1, CHD7, CHM, CHMP4B, CHN1, CHRDL1, CHST6, IARS2, IDH3B, IFT140, IFT172, IFT27, IFT43, ELP1, IMPDH1, IMPG1, IMPG2, INPP5E, INPP5K, IQCB1, ISPD, ITM2B, ITPR1, JAG1, JAM3, KCNJ13, KCNV2, KERA, KIAA0586, KIAA1549, KIF11, KIF21A, KIF7, KIT, KITLG, KIZ, KLHL7, KMT2D, KRT12, KRT3, LAMA1, LAMB2, LARGE, LCA5, LCAT, LEMD2, LHX3, LHX4, LIM2, LMX1B, LONP1, LOXHD1, LOXL1, LOXL3, LRAT, LRIT3, LRMDA, LRP2, LRP5, LRPAP1, LSS, LTBP2, LYST, LZTFL1, MAB21L2, MAF, MAFB, MAK, MAN2B1, MC1R, MED25, MERTK, MFN2, MFRP, MIP, MIPEP, MITF, MKKS, MKS1, MLPH, MSMO1, MSTO1, MTPAP, MTPP, MUSK, MVK, MYH9, MYO5A, MYO7A, MYOC, NAA10, NDP, NDUFB11, NDUFS1, NEK2, NEUROD1, NF2, NHS, NLRP1, NMNAT1, NOTCH2, NPHP1, NPHP2, NPHP3, NPHP4, NR2E3, NR2F1, NRL, NTF4, NYX, OAT, OCA2, OCRL, OFD1, OPA1, OPA3, OPN1LW, OPN1MW, OPN1SW, OPTN, OR2W3, OTX2, OVOL2, P3H2, P4HA2, PABPN1, PANK2, PANK4, PAX2, PAX3, PAX6, PCARE, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6G, PDE6H, PDZD7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGK1, PHOX2A, PHYH, PIK3R1, PIKFYVE, PITPNM3, PITX1, PITX2, PITX3, PLA2G5, PLK4, PMM2, PNKP, PNPLA6, POC1B, POLA1, POLG, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PORCN, POU1F1, PRCD, PRDM13, PRDM5, PROKR2, PROM1, PROP1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, PRSS56, PTCH1, PUF60, PXDN, RAB18, RAB27A, RAB28, RAB3GAP1, RAB3GAP2, RAPSN, RARB, RAX, RAX2, RBP3, RBP4, RCBTB1, RD3, RDH11, RDH12, RDH5, REEP6, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, ROBO3, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, RPGRIP1, RPGRIP1L, RS1, RTN4IP1, RYR1, SACS, SAG, SALL2, SALL4, SC5D, SCAPER, SCLT1, SCO2, SDCCAG8, SEMA4A, SETX, SH3PXD2B, SHH, SIL1, SIPA1L3, SIX3, SIX6, SLC16A12, SLC24A1, SLC24A5, SLC25A3, SLC25A4, SLC2A1, SLC33A1, SLC38A8, SLC39A5, SLC45A2, SLC4A11, SLC7A14, SLITRK6, SMO, SMOC1, SNAI2, SNRNP200, SNX3, SOX1, SOX10, SOX2, SOX3, SPATA7, SRD5A3, STN1, STRA6, STS, STX3, TACSTD2, TAX1BP3, TBC1D20, TCF4, TDRD7, TEAD1, TEK, TENM3, TFAP2A, TGFBI, TIMM8A, TIMP3, TIN2, TLR4, TMEM126A, TMEM237, TMEM5, TMEM98, TOPORS, TP63, TPP1, TRAF3IP1, TREX1, TRIM32, TRIM37, TRIM44, TRNT1, TRPM1, TSPAN12, TTC8, TTLL5, TTPA, TUB, TUBB3, TUBGCP4, TUBGCP6, TULP1, TYR,

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TYRP1, UBE3B, UBIAD1, UNC119, UNC45B, USH1C, USH1G, USH2A, VAX1, VCAN, VIM, VPS13B, VSX1, VSX2, WDPCP, WDR19, WDR36, WFS1, WHRN, WRN, WT1, XPNPEP2, XYLT2, YAP1, YME1L1, ZBTB20, ZEB1, ZFH4, ZNF408, ZNF423, ZNF469, ZNF513, ZNF644)

NGS panel Onemocnění ledvin (357 genů: ABCC6, ACE, ACTN4, ADAMTS9, AGT, AGTR1, AGTR2, AGXT, AHI1, ALG8, ALMS1, ANKFY1, ANKS6, ANLN, ANOS1, AP2S1, APOL1, AQP2, ARHGAP24, ARHGDI, ARL13B, ARL3, ARL6, ARMC9, ATP6AP2, ATP6V0A4, ATP6V1B1, ATP8B1, AVIL, AVPR2, B9D1, B9D2, BAG6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BICC1, BMP4, BMP7, BMPR1B, BMPR2, BOLA3, BSND, C3, C5orf42, c8orf37, C9orf72, CA2, CASR, CC2D2A, CCDC28B, CD2AP, CD46, CDC5L, CDC73, CENPF, CEP104, CEP120, CEP164, CEP290, CEP41, CEP55, CEP83, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CLCN5, CLCNKA, CLCNKB, CLDN14, CLDN16, CLDN19, CNNM2, COL4A1, COL4A3, COL4A4, COL4A5, COL4A6, COQ2, COQ8B, CRB2, CSPP1, CTC1, CTNS, CUL3, CYP11B2, CYP17A1, CYP24A1, DACT1, DCDC2, DGKE, DMP1, DNAJB11, DSTYK, DYNC2H1, DYNC2LI1, DZIP1L, EGF, EHHADH, EIF2AK4, ELMOD2, EMP2, EMX2, ENG, ENPP1, EVC, EVC2, EYA1, FAM111A, FGF20, FGF23, FGF8, FGF9, FN1, FOXC1, FOXH1, FOXI1, FRAS1, FREM1, FREM2, FXYD2, G6PC, GALNT3, GANAB, GAPVD1, GATA3, GATA6, GBE1, GDNF, GLIS2, GLIS3, GNA11, GOSR2, GREB1L, GREM1, GRHPR, GRIP1, HNF1B, HNF4A, HOGA1, HSD11B2, HYLS1, CHD1L, CHRM3, IFIH1, IFT122, IFT140, IFT172, IFT27, IFT43, IFT52, IFT74, IFT80, IFT81, ILK, INF2, INPP5E, INTU, INVS, IQCB1, ITGA8, KANK2, KAT6B, KCNA1, KCNA5, KCNJ1, KCNJ10, KCNJ13, KCNJ5, KCNK3, KIAA0556, KIAA0586, KIAA0753, KIF14, KIF3B, KIF7, KL, KLHL3, LAMB2, LCA5, LIN7C, LRP4, LRP5, LRRCC1, LZTFL1, MAGED2, MAGI2, MAPKBP1, MEN1, MKKS, MKS1, MYH9, MYO1E, MYOG, NCF1, NEK1, NEK4, NEK8, NFU1, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NR1H4, NR3C2, NRIP1, NUP107, NUP133, NUP160, NUP205, NUP37, NUP85, NUP93, OCRL, PAX2, PBX1, PCBD1, PCDH15, PCNT, PCSK5, PDE2A, PDE6D, PHEX, PIBF1, PKD1, PKD2, PKHD1, PLCE1, PLK4, PLXND1, PMM2, PNPLA6, POC1A, PRKCD, PRKCSH, PTPN22, PTPRO, RDH12, REN, RET, ROBO2, RPE65, RPGRIP1, RPGRIP1L, SALL1, SALL4, SCARB2, SCN1A, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SEC61A1, SEC63, SGPL1, SIX1, SIX2, SIX5, SLC12A1, SLC12A3, SLC26A4, SLC2A2, SLC34A1, SLC34A3, SLC37A4, SLC3A1, SLC4A1, SLC4A4, SLC5A1, SLC5A2, SLC7A9, SLC9A3R1, SLIT2, SMAD4, SMAD9, SMARCA1, SOX17, SPRY2, SUFU, TBC1D8B, TBX18, TCTEX1D2, TCTN1, TCTN2, TCTN3, TH, THBD, TJP2, TMEM107, TMEM127, TMEM138, TMEM216, TMEM231, TMEM237, TMEM256, TMEM67, TRAF3IP1, TRIM32, TRPC6, TRPM6, TRPM8, TSC2, TTC21B, TTC8, TXNDC15, ULK4, UMOD, UPK3A, USF2, UTRN, VHL, WDPCP, WDR19, WDR34, WDR35, WDR60, WDR72, WDR73, WHRN, WNK1, WNK4, WNT4, WT1, XDH, XPNPEP3, ZNF423, KRIT1, CCM2, PDCD10, FOXE1, NKX2-1, NKX2-5, PAX8, SLC36A4, THRA, THRB, TSHR, ABCA3, CSF2RA, CSF2RB, SFTPA1, SFTPB, SFTPC, SFTPD, TTF1)

NGS panel Onemocnění a vady srdce (214 genů: A2ML1, ABCA7, ABCC9, ACTA1, ACTA2, ACTC1, ACTN2, ACVR2B, AKAP9, ALG10B, ALMS1, ALPK3, ANK2, ANKRD1, APOA1, APOA2, APOB, APOE, APP, B2M, BAG3, BRAF, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CALR3, CASQ2, CAV3, CBL, CFC1, CITED2, COL3A1, COL5A1, COL5A2, COX15, CRELD1, CRYAB, CSRP3, CST3, CTF1, CTNNA3, DES, DMD, DNAJC19, DOLK, DPP6, DSC2, DSG2, DSP, DTNA, EFEMP2, ELN, EMD, EPG5, EYA4, FBN1, FBN2, FGA, FHL1, FHL2, FHOD3, FKRP, FKTN, FLNA, FLNC, GAA, GATA4, GATA5, GATA6, GATAD1, GDF1, GJA1, GJA5, GLA, GPD1L, GSN, HCN4, HFE, HRAS, ILK, ITM2B, JAG1, JPH2, JUP, KCNA5, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, KLF10, KRAS, LAMA4, LAMP2, LDB3, LMNA, LOX, LTBP2, LYZ, LZTR1, MAP2K1, MAP2K2, MFAP5, MIB1, MMP21, MRAS, MURC, MYBPC3, MYH11, MYH6, MYH7, MYL2, MYL3, MYLK, MYLK2, MYO6, MYOM1, MYOZ2, MYPN, NEBL, NEXN, NKX2-5, NKX2-6, NODAL, NOS1AP, NOTCH1, NPPA, NRAS, OSMR, PDLIM3, PKD1L1, PKP2, PLN, PPA2, PPP1CB, PRDM16, PRKAG2, PRKG1, PRKAR1A, PRNP, PSEN1, PSEN2, PTPN11, RAF1, RANGRF, RASA2, RBM20, RIT1, ROBO4, RRAS, RRAS2, RYR2, SAA1, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCN10A, SCO2, SDHA, SGCD, SHOC2, SLC25A4, SLC2A10, SMAD3, SMAD4, SMAD6, SLMAP, SNTA1, SORL1, SOS1, SOS2, SYNE2, TAF1A, TAZ, TBX1, TBX3, TBX5, TBX20, TCAP, TECRL, TGFB2, TGFB3, TGFB1, TGFB2, TLL1, TMEM43, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TOMM40, TPM1, TRDN, TREM2, TRPM4, TSPYL1, TTN, TTR, TXNRD2, VCL, ZFPM2, ZIC3)

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NGS panel Vzácná onemocnění (525 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, ANKRD11, ANOS1, ANTXR2, APC2, AR, ARHGAP31, ARID1A, ARID1B, ARID2, ARSL, ARX, ASCC1, ASCL1, ASXL1, ASXL2, ASXL3, ATP7B, ATR, ATRIP, B3GALT, B3GALT6, B4GALT7, B3GAT3, BHLHA9, BICD2, BMP2, BMP4, BMPR1B, BRAF, BRD4, BRWD3, BTK, BTRC, C1R, C1S, CASR, CANT1, CBL, CBX2, CCDC8, CCDC141, CD96, CDC45, CDH3, CDK5, CDKL5, CDKN1C, CDON, CDT1, CDC6, CENPJ, CENPE, CEP120, CEP152, CEP63, CFTR, CKAP2L, CNOT1, CNTNAP2, COG4, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL10A1, COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, COL12A1, COLEC10, COLEC11, COMP, CREBBP, CUL4B, CUL7, CYP11A1, CYP11B1, CYP17A1, CYP26B1, DCAF17, DCX, DDR2, DHCR7, DHCR24, DHH, DIS3L2, DISP1, DLL1, DLL4, DLX3, DLX4, DLX5, DLX6, DNA2, DNM2, DNMT3A, DOCK6, DOK7, DPF2, DSE, DUSP6, DYNC1H1, DYNC2H1, DYNC2LI1, DVL1, DVL3, ECE1, ECEL1, EDN3, EDNRB, EED, EFN1, ENG, EOGT, EP300, EPHB4, EPS15L1, ERF, ESCO2, EVC, EVC2, EXOC6B, EXT1, EXT2, EZH2, FBLN1, FBN1, FBN2, FEZF1, FGD1, FGF10, FGF16, FGF17, FGF8, FGF9, FGFR1, FGFR2, FGFR3, FKBP14, FLNA, FLNB, FLRT3, FLT4, FOXC2, FOXC1, FOXH1, FREM1, FSHB, FTH1, 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, GZF1, HAMP, HDAC4, HDAC8, HESX1, HFE, HIST1H1E, HJV, HMGA2, HOXD13, HRAS, HS6ST1, HSD3B2, CHD7, CHD8, CHST3, CHST14, CHSY1, IDH1, IDH2, IFT122, IFT140, IFT172, IFT43, IFT52, IFT80, IGF1, IGF1R, IGF2, IHH, IL17RD, IQCE, IQSEC2, IRF6, JAG1, KAT6B, KATNB1, KCTD1, KDM6A, KIAA0586, KIFBP, KIF22, KISS1, KISS1R, KITLG, KLB, KLHL7, KMT2A, KMT2D, KRAS, LAMB1, L1CAM, 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, MRAP, MRAS, MSX1, MSX2, MTOR, MUSK, MVK, MYBPC1, MYH3, MYH8, MYO18B, MYOD1, MYT1, NALCN, NBN, NDE1, NECTIN1, NEK1, NEK9, NF1, NF2, NFIB, NFIX, 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, PCNT, PDE3A, PDGFRB, PGAP2, PGAP3, PHF6, PHOX2B, PIEZO1, PIEZO2, PIGL, PIGO, PIGV, PIGW, PIGY, PIK3CA, PIK3R1, PITX1, PITX2, PLAG1, PLK4, PLOD1, POLR1C, POLR1D, POMT1, POMT2, POR, POU1F1, PPP1CB, PRKAR1A, PRMT7, PROK2, PROKR2, PROP1, PTEN, PTH1R, PTHLH, PTCH1, PTCH2, PTPN11, RAB23, RAD21, RAF1, RAI1, RAP1A, RAP1B, RAPSN, RASA1, RASA2, RASA3, RBBP8, RBM8A, RBPJ, RECQL4, RELN, RET, RIT1, RLIM, RNF125, RNF135, RNPC3, ROR2, RPS6KA3, RRAS, RRAS2, RTEL1, RUNX2, SALL4, SAMD9, SC5D, SEM1, SEMA3A, SEMA3C, SEMA3D, SEMA3E, SEMA7A, SETD2, SETD5, SGPL1, SHH, SHOC2, SHOX, SIX3, SKI, SLC18A3, SLC26A2, SLC29A3, SLC39A13, SLC40A1, SMAD3, SMAD4, SMAD6, SMARCA2, SMARCA4, SMARCB1, SMARCC2, SMARCE1, SMC1A, SMC3, SMO, SMPD4, SOS1, SOS2, SOST, SOX2, SOX3, SOX4, SOX9, SOX10, SOX11, SOX18, SPECC1L, SPRED1, SPRY4, SRCAP, SRY, STAR, STAG2, STXBP1, SUFU, SUMO1, TAC3, TACR3, TBC1D24, TBCK, TBX1, TBX2, TBX5, TBX15, TBX19, TCF12, TCF4, TCOF1, TCTEX1D2, TCTN3, TDGF1, TFAP2A, TFR2, TGFB2, TGFB3, TGFB1, TGFB2, TGIF1, TMTC3, TNNT2, TNNT3, TNXB, TOR1A, TP63, TPM2, TRAP1, TRIP11, TRPV4, TRPS1, TTC21B, TUBA1A, TWIST1, UBE3A, UGT1A1, UNC80, VEGFC, VIPAS39, VPS13B, VPS33B, WDPCP, WDR11, WDR19, WDR34, WDR35, WDR60, WNT10B, WNT4, WNT5A, WNT7A, WT1, YWHAE, ZEB2, ZFPM2, ZIC1, ZIC2, ZNF141, ZSWIM6)

**Postup č. 8: 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

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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 MEC2, 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

**Postup č. 9: 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)

**Postup č. 10: 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)