Příloha je nedílnou součástí
osvědčení o akreditaci č.: 645/2021 ze dne: 10. 12. 2021

Akreditovaný subjekt podle ČSN EN ISO 15189:2013:

Fakultní nemocnice Ostrava
Laboratoře Ústavu klinické a molekulární patologie a lékařské genetiky
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Laboratoř uplatňuje flexibilní přístup k rozsahu akreditace upřesněný v dodatku. Aktuální seznam činností prováděných v rámci flexibilního rozsahu má laboratoř k dispozici na webových stránkách laboratoře www.fno.cz a u vedoucího laboratoře.

Vyšetření:

Pořadové číslo	Přesný název postupu vyšetření	Identifikace postupu vyšetření	Předmět vyšetření
823 - Laboratoř patologie			
1.	Stanovení základní histologické diagnózy	SOP-LAB-ÚKMPLG-01	Tkáně
2.	Stanovení diagnózy u peroperačního vyšetření	SOP-LAB-ÚKMPLG-02	Tkáně, buňky
3.	Stanovení základní cytologické diagnózy	SOP-LAB-ÚKMPLG-03	Buňky z punkce tkání, z tělních tekutin a obsahu patologických dutin
4.	Imunohistochemický/ imunocytochemický průkaz sledovaného markeru [1]	SOP-LAB-ÚKMPLG-04	Tkáně, buňky
5.	Analýza histologických a cytologických vzorků metodou in situ hybridizace [2]	SOP-LAB-ÚKMPLG-06a	Tkáně, buňky
6.	Analýza lidského somatického genomu metodou masivně paralelního sekvenování [NGS panely] [3]	SOP-LAB-ÚKMPLG-11a	Tkáně, buňky
7.	Analýza sekvenčních variant lidského somatického genomu metodou real-time PCR [4]	SOP-LAB-ÚKMPLG-10a	Tkáně, buňky
8.	Analýza lidského somatického genomu metodou reverzní hybridizace [5]	SOP-LAB-ÚKMPLG-15a	Tkáně, buňky

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Pořadové číslo	Přesný název postupu vyšetření	Identifikace postupu vyšetření	Předmět vyšetření
816 - Laboratoř lékařské genetiky			
1.	Přímá sekvenace DNA lidského genomu dle Sangera [6]	SOP-LAB-ÚKMPLG-12	Biologický materiál obsahující lidskou DNA
2.	Fragmentační analýza lidského genomu pomocí kapilární elektroforézy [7]	SOP-LAB-ÚKMPLG-14	Biologický materiál obsahující lidskou DNA
3.	Vyšetření chromozomů klasickou cytogenetickou analýzou - stanovení karyotypu	SOP-LAB-ÚKMPLG-05	Krev, plodová voda, choriová tkáň, tkáň
4.	Vyšetření chromozomů metodou FISH	SOP-LAB-ÚKMPLG-06b	Fixovaná buněčná suspenze, krev, plodová voda, choriová tkáň, tkáň
5.	Cytogenetická analýza periferních lymfocytů [CAPL]	SOP-LAB-ÚKMPLG-08	Krev
6.	Analýza lidského genomu metodou masivně paralelního sekvenování [NGS panely] [8, 9, 10]	SOP-LAB-ÚKMPLG-11b	Biologický materiál obsahující lidskou DNA
7.	Analýza variant lidského genomu na biočipu [Microarray analýza]	SOP-LAB-ÚKMPLG-09	Biologický materiál obsahující lidskou DNA
8.	Analýza lidského genomu metodou MLPA [11]	SOP-LAB-ÚKMPLG-13	Biologický materiál obsahující lidskou DNA
9.	Analýza sekvenčních variant lidského genomu metodou real-time PCR [12]	SOP-LAB-ÚKMPLG-10b	Biologický materiál obsahující lidskou DNA
10.	Analýza lidského genomu metodou reverzní hybridizace [13]	SOP-LAB-ÚKMPLG-15b	Biologický materiál obsahující lidskou DNA

V závorkách […] jsou uvedeny názvy vyšetření dle názvu na výsledkových listech.

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Dodatek:
Flexibilní rozsah akreditace

Pořadová čísla postupů vyšetření
823: 4, 5, 6, 7, 8
816: 1, 2, 6, 8, 9, 10

Laboratoř může modifikovat v dodatku uvedené postupy vyšetření v dané oblasti akreditace při zachování principu měření.
U vyšetření v dodatku neuvedených nemůže laboratoř uplatňovat flexibilní přístup k rozsahu akreditace

Upřesnění rozsahu akreditace:

Vyšetřované geny / panely genů / sekvenční varianty / oblasti / markery
Markery pro prediktivní diagnostikou jsou uvedeny **tučným písmem**

[1] Seznam používaných protilátek

Název prokazovaného markeru
Aktin hladkosvalový
ACTH
ADAMTS 13
Alfa-1-fetoprotein
Anaplastic lymphoma
ALK - D5F3
AMACR
Amyloid A
Annexin1
Androgenový receptor
ATRX
Beta-Catenin
BCL 2 onkoprotein
BCL 6 protein
BerEP4 (epiteliální antigen)
BOB1
Carbonic Anhydrase IX
Calcitonin
Caldesmon
Calponin
Calretinin
c-erbB-2 onkoprotein
C-myc
CD1a
C1q
C4d

CD3
CD4
CD5
CD8
CD10
CD15
CD20cy,B cell
CD23
CD30
CD31
CD34
CD35
CD43
CD45RO,T cell
CD45LCA
CD56
CD61
CD68
CD79a
CD99
CD117
CD138
CD163
CDX-2
Carcinoembryonální antigen
CK AE1/AE3
CK 5/6

CK 7
CK 8
CK14
CK17
CK18
CK19
CK20
CK HMW
Cytomegalovirus
Cyclin D1
C3c
D 2-40
Desmin
DOG1
EBV
E-cadherin
Epiteliální membránový antigen
Estrogenový receptor
ERG
Fibrinogen
GATA3
GFAP
Glycophorin C
Glypican-3
Granzym B
Mesothelial Cell
HCG

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Hepatocyte
HER2neu
HMB45 (melanosom)
Helicobacter pylori
HPV16
Chromogranin A
Isocitrát dehydrogenáza 1
Ig A
Ig D
Ig G
Ig G4
Ig M
Inhibin, alpha
Kappa
Ki 67
Lambda
Lymfoid E.F.
MDM2
Melan A
Mitochondriální antigen
MLH1
Myeloperoxidase
MSH2
MSH6

MUC1
MUC2
MUC4
MUC5AC
MUC6
MUM1
MyoD1
Napsin A
NKX3.1
Neuron specifická enoláza
OCT-2
PD-L1
p16
p21
p27
p40
p53
p57
p63
P120
Pax-5
Pax-8
Perforin
Phosphohistone H3

Placentární alkalická fosfatáza
PMS2
Prealbumin
Progesteronový receptor
PSA (prostatic specific antigen)
Sérový amyloid P
Renal Cell Carcinoma
ROS1
SALL4
S100
SOX-10
SOX-11
STAT6
Synaptophysin
SV40 T Antigen
TAG72
TdT
Thrombomodulin
Thyroglobulin
Thyroid transcription factor
Vimentin
Von Willebrandův faktor (F VIII)
Wilms Tumor Protein

[2] ***HER2, ALK, ROS1***

[3] NGS panel Somatické varianty v nádorových tkáních (*AKT1, APC, ATM, ATR, AXIN2, BAP1, BARD1, BLM, BMPR1A,* ***BRAF, BRCA1, BRCA2****, BRF1 , BRIP1 , CDH1, CDK12, CDK4, CDKN2A, CYLD, DICER1,* ***EGFR****, ENG, EPCAM, ERCC3, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCM, FANCP, FLCN, FOXL2, GREM1, HRAS, CHEK1, CHEK2, IDH1, IDH2, KLLN,* ***KRAS****, MAD2L2, MEN1, MET, MLH, MLH3, MN1, MRE11, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2,* ***NRAS****, NTHL1, PALB2, PARP1, PARP2, PDGFD, PML, PMS2, POLD1, POLE, POT1, PTEN, PTCH1, RAD50, RAD51, RAD51B, RAD51C, RAD51D, RAD54L, RECQL, RECQL4, RET, RNF43, RPS20, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA4, SRY, STK11, SUFU, TP53, VHL, WRN*)

[4] ***KRAS, NRAS, BRAF, EGFR***

[5] ***KRAS, NRAS, BRAF, EGFR***

[6] *LDLR, ATP7B, GJB2*

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[7] C*FTR* (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)), aneuploidie chromozomů 13, 18, 21, X a Y, mikrodelece na chromozomu Y (AZFa, AZFb, AZFc), *SRY,* Syndrom fragilního X (analýza počtu repetic CGG v 5'UTR oblasti genu *FMR1)*

[8] NGS panely do 20 genů: NGS panel Alportův syndrom (*COL4A5, COL4A3, COL4A4, COL4A6, CFHR5, MYH9*)

[9] NGS panely 20 až 100 genů: NGS panel Hereditární nádorové syndromy a somatické varianty v nádorových tkáních (*AKT1, APC, ATM, ATR, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRAF, BRCA1, BRCA2, BRF1, BRIP1, CDH1, CDK12, CDK4, CDKN2A, CYLD, DICER1, EGFR, ENG, EPCAM , ERCC3 , FANCA , FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCM, FANCP, FLCN, FOXL2, GREM1 , HRAS, CHEK1, CHEK2, IDH1, IDH2, KLLN , KRAS, MAD2L2 , MEN1, MET, MLH, MLH3, MN1, MRE11, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2, NRAS, NTHL1, PALB2, PARP1, PARP2, PDGFD, PML, PMS2, POLD1, POLE, POT1, PTEN, PTCH1, RAD50, RAD51, RAD51B, RAD51C, RAD51D, RAD54L, RECQL, RECQL4, RET, RNF43, RPS20, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA4, SRY, STK11, SUFU, TP53, VHL, WRN*),
NGS panel ALL IN ONE (*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*)

[10] NGS panely nad 100 genů: NGS panel Vrozená porucha sluchu (*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,*

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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 Vrozené kostní anomálie (*AAAS, ABCC9, ABR, ACAN, ACOX1, ACP5, ACTA1, ACTB, ACTG1, ACVR1, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTSL2, ADCY6, ADGRG6, AFF4, AGA, AGPS, 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 , ARCN1, ARHGAP31, ARID1A, ARID1B, ARID2, ARL6 , ARMC9, ARSB, ARSL/ARSE, ASCC1, ASXL1, ASXL2, ASXL3, ATAD1, 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 , BTK, C12ORF65, C15ORForf41, C2CD3, C8ORF37, CA2, CACNA1C, CAD, CANT1, CASR, CBFB, CBL, CC2D2A, CCBE1, CCDC115, CCDC22, CCDC47, CCDC8, CCN6/WISP3 , CCND2 , CD96, CDAN1, CDC42, CDC45, CDC6, CDH1, CDH11, CDH3, CDKN1C, CDT1, CENPE, CENPF , CENPJ, CEP120, CEP152, CEP290, CEP41, CEP55, CEP63, CERT1/COL4A3BP , CFAP410/C21orf2 , CFL2, CIBAR1/FAM92A, CILK1, CKAP2L,*

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CLCF1, CLCN5, CLCN7, CNOT2, CNTNAP1, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL27A1, COL2A1, COL9A1, COL9A2, COL9A3, COLEC10, COLEC11, COMP, COX4I2, CPLANE1, CREB3L1, CREBBP, CRIM1, CRIPT, CRTAP, CSGALNACT1, CSPP1, CST6, CTNNB1, CTSA, CTSC, CTSK, CTU2, CUL4B, CUL7, CWC27, CYP26B1, CYP27B1, CYP2R1, DCPS, DDR2, DDRGK1, DDX11, DDX59, DEAF1, DHCR24, DHCR7, DHODH, DCHS1, DIS3L2, DLL3, DLL4, DLX1, DLX3, DLX5, DMP1, DNA2, DNAJC21, DNM2, DNMT3A, DOCK1, DOCK6, DOK7, DOLK, DONSON, DPAGT1, DPF2, DPH1, DPM1, DPM2, DPM3, DSE, DVL1, DVL3 , DYM , DYNC2H1, DYNC2LI1, DYT1/TOR1A, EBP, ECEL1, EDARADD, EDN1, EED, EFL1, EFNA4, EFNB1, EFTUD2, EIF2AK3, EIF4A3, ELMO2, ENPP1, EOGT, EP300, EPS15L1, ERBB3, ERCC1, ERCC4, ERCC5, ERCC6, 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, FBN1, FBN2, FBXL3, FBXO11, FBXW11, FCSK, FDFT1, FERMT1, FERMT3 , FGD1, FGF10, FGF16, FGF17 , FGF23, FGF8, FGF9, FGFR1, FGFR2, FGFR3, FHL1, FIG4, FKBP10, FLI1, FLNA, FLNB, FLVCR2, FN1, FRAS1, FREM1, FREM2, FTO, FUCA1, FUT8, FZD2, GALNS, GALNT2, GALNT3, GBA, GBE1, GDF3, GDF5, GDF6, GFM2, GH1, GHR, GHRHR, GHSR, GJA1, GLB1, GLDN, GLE1, GLI1, GLI2, GLI3, GMNN, GNAS, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GPC3, GPC4, GPC6, GPX4, GRHL2, GRHL3, GRIP1, GSC, GTF2E2, GUSB, GZF1 , HAAO, HAPLN1/CRTL1, HDAC6, HDAC8, 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, CHL1/CALL, CHRNA7, CHRNG, CHST11, CHST14, CHST3, CHSY1, CHUK , IARS2, IBA57, ICK/CILK1, IDS, IDUA, IFIH1 , IFITM5, IFT122, IFT140, IFT172, IFT27, IFT43, IFT52, IFT57, IFT74, IFT80, IFT81, IGF1 , IGF1R , IGF2, IHH, IKBKG , IL11RA, IL1RN, IL6ST, INPP5E, INPPL1, INTU, IQCE, IRF6, IRX5, ISCA2, ITPR1, JAG1, KAT6A, KAT6B, KBTBD13, KCNH1, KCNJ2, KCTD1, KDM1A, KDM6A, KDM6B, KIAA0586, KIAA0753 , KIAA0825, KIAA1109, KIAA1279, KIF14, KIF1A, KIF22, KIF3B, KIF5C, KIF7, KL , KLHL40, KLHL41, KMT2A, KMT2D, KPTN, KRAS, KYNU, LBR, LBX1, LEMD3, LFNG, LGI4, LIFR, LIG4, LMBR1, LMNA, LMOD3, LMX1B, LONP1, LPIN2, LRP2, LRP4, LRP5, LTBP2, LTBP3, LZTFL1, LZTR1, MAB21L1, MAB21L2, MACROH2A1, MAD2L2, MAF, MAFB, MAGEL2, MAGI2, MAGT1, MAN2B1 , MANBA, 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, NECTIN 4, NECTIN1, NEDD4L, NEK1, NEK9, NEPRO, NEU1, NEXMIF, NF1 , NFIX, NFIX , NFKB2, NGLY1, NHEJ1, NIN, NIPBL, NIT1, NKX3-2, NLRP3, NOG, NOTCH1, NOTCH2, NPHP3, NPR2, NRAS, NSD1, NSDHL, NSMCE2, NSUN2, NUP88, NUS1, NXN, OBSL1, OCRL1, OFD1, ORC1, ORC4, ORC6,

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OSTM1, P3H1/LEPRE1, P4HB, PALB2 , PAM16, PAPSS2, PAX1, PAX3, PBX1, PCDH12, PCNA, PCNT, PCYT1A, PDE3A, PDE4D, PDE6D, PDGFRB, PEX5, PEX7, PGAP2, PGAP3, PGK1, PGM1, PHEX, PHF8, PHGDH, PHIP, PI4KA, PIBF1, PIEZO2, PIGK, PIGO, PIGS, PIGT, PIGV, PIGW, PIGY, PIK3C2A, PIK3CA , PIK3R1, PIK3R2, PIP5K1C, PISD, PITX1, PITX2, PLAA, PLAG1, PLC/HSPG2, PLCB3, PLEKHM1, PLK4, PLOD2, PLS3, PML, PMM2, POC1A, POLE, POLR1A , POLR1B, POLR1C, POLR1D, POLR3A, POMP, POP1, POR, PORCN, PPIB, PPM1D, PPP1CB, PPP1R12A, PPP3CA, PRG4, PRKAR1A, PRMT7, PROM1 , PRRX1, PSAT1 , PSMD12, PSPH, PTDSS1, PTH, PTH1R, PTHLH, PTCH1, PTCH2, PTPN11, PUF60, PUM1, PYCR1, QRICH1, RAB23, RAB33B, RAD21, RAD51, RAD51C, RAF1, RAI1, RALGAPA1, RAP1A, RAP1B, RAPSN, RARB, RBBP8 , RBBPS, RBM10, RBM8A , RBPJ, RECQL4, RFT1 , RFWD3, RHOA , RIN2, RIPK4, RIPPLY2, ROR2, RPGRIP1L, RPL10, RPL11, RPL13, RPL15, RPL26, RPL5, RPS10 , RPS17, RPS19, RPS24, RPS26, RPS6KA3, RPS7 , RRAS2, RSPOR2, RSPRY1, RTTN, RUNX2, RYR1, SALL1, SALL4, SAMD9, SBDS, SBF1, SC5D/SC5DL , SCARF2, SCYL2, SDC2 , SDCCAG8, SEC23A, SEC24D, SEM1, SEMA3E, SERPINF1, SERPINH1, SETBP1, SETD2, SF3B4, SFRP4, SGMS2, SGSH, SH3BP2, SH3PXD2B, SHH, SHOC2, SHOX, SIK3, SIN3A, SIX2, SKI, SLC10A7, SLC17A5, SLC18A3, SLC25A24, SLC26A2, SLC29A3, SLC2A1, SLC2A2, SLC34A1, SLC34A3, SLC35A1, SLC35A3, SLC35C1 , SLC35D1, SLC39A13, SLC39A8, SLC5A7, SLC6A9, SLC9A3R1/NHERF1 , SLCO2A1, SLCO5A1, SLURP1, SLX4, SMAD3, SMAD4, SMARCA2, SMARCA4, SMARCAL1, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SMC1A, SMC3, SMO, SMOC1, SMPD4, 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, SYNE 2, SYNE1, TALDO1 , TAPT1, TASP1, TBC1D24, TBCD, TBCE, TBL1XR1, TBX1, TBX15, TBX2, TBX22, TBX3, TBX4, TBX5, TBX6, TBXAS1, TBXT, TCF12, TCIRG1, TCOF1, TCTEX1D2, TCTN2 , TCTN3, TELO2, TENT5A/FAM46A, TERT, TFAP2A, TFAP2B, TGDS, TGFB1, TGFB2, TGFB3, TGFBR1, TGFBR2, TLK2, TMCO1, TMEM107, TMEM138, TMEM165, TMEM199, TMEM216, TMEM231, TMEM237, TMEM38B, TMEM43, TMEM67, TNFRSF11A/RANK, TNFRSF11B/OPG, TNFSF11/RANKL, TNNI2, TNNT1, TNNT3, TONSL, TP63, TPM2, TPM3, TRAF3IP1, TRAIP, TRAPPC2/SEDL, TRAPPC6A, TREM2, TRIM32 , TRIO, TRIP11, TRIP4, TRPS1, TRPV4, TRPV6, TRRAP, TTC21B, TTC8 , TTN, TWIST1, TWIST2, TXNDC15 , TYROBP, UBA1, UBA2, UBE2T, UBE3B, UFSP2, USP9X, VAC14, VDR, VIPAS39, VPS33A, VPS33B, VPS35L/C16ORF62 , WDPCP, WDR19, WDR34, WDR35, WDR37, WDR60, WNT1, WNT10A, WNT10B, WNT3, WNT5A, WNT7A, XAGE1B, XRCC2, XRCC4, XYLT1, XYLT2, YY1, YY1AP1, ZBTB16, ZBTB42, ZC4H2, ZFHX4, ZIC1, ZIC3, ZMIZ1, ZMPSTE24, ZNF141, ZNF30, ZNF335, ZNF462, ZNF469, ZSWIM6),

NGS panel Onemocnění a vady očí (*AAAS, ABCA1, ABCA4, ABCB6, ABCC6, ABHD12, ABHD5, ACBD5, ACO2, ACTB, ACTG1, ADAM9, ADAMTS10, ADAMTS17, ADAMTS18, ADAMTSL4, ADGRV1 (GPR98, USH2B), AGBL1, AGBL5, AGK, AGPS, AHI1, AIPL1, ALDH18A1, ALDH1A3, ALMS1, ANOS1 (KAL1), AP3B1, AP3D1, APC, APTX, ARHGEF18,*

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ARL13B, ARL2BP, ARL6, ARMC9, 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, TWNK (C10orf2), C12orf57, C12orf65, C1QTNF5, CFAP410 (C21orf2), PCARE (C2orf71), CPLANE1 (C5orf42), C8orf37, CA4, CABP4, CACNA1F, CACNA2D4, CAPN5, CASK, CC2D2A, PRIMPOL (CCDC111), CDH23 (USH1D), CDH3, CDHR1, CENPF, CEP104, CEP164, CEP250, CEP290 (NPHP6), CEP41, CEP78, CERKL, CFAP410, CFB, CFH, CFI, CIB2 (USH1J,KIP2), CISD2, CLDN19, CLN3, CLRN1 (USH3A), 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, CTDP1, CTNNA1, CTNNB1, CX3CR1, CYP1B1, CYP27A1, CYP4V2, CYP51A1, DAG1, DCN, DDX59, DHCR7, DHDDS, DHX38 (PRPF16), DNM1L, DNMBP, DRAM2, DTHD1, DTNBP1, EDN3, EDNRB, EED, EFEMP1, ELOVL4, ELOVL5, 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, GMPPB, GNAT1, GNAT2, GNB3, GNPAT, GNPTG, GP1BA, ADGRA3 (GPR125), GPR143, GPR179, GRHL2, GRID2, GRIP1, GRK1, GRM6, GSN, GTF2H5, GUCA1A, GUCA1B, GUCY1A1 (GUCY1A3), GUCY2D, GZF1, HARS1 (USH3B), HCCS, 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, ELP1 (IKBKAP), IMPDH1, IMPG1, IMPG2, INPP5E, INPP5K, IQCB1, CRPPA (ISPD), 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, NAA10, NDP, NDUFB11, NDUFS1, NEK2, NEUROD1, NF2, NHS, NLRP1, NMNAT1, NOTCH2, NPHP1, INVS (NPHP2), 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, 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, PIK3R1, PIKFYVE, PITPNM3, PITX1, PITX2, PITX3, PLA2G5, PLK4, PMM2, PNKP, PNPLA6, POC1B, POLA1, POLG, POMGNT1,

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POMGNT2, POMK, POMT1, POMT2, PORCN, POU1F1, PRCD, PRDM13, PRDM5, 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, 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 (MATP), SLC4A11, SLC7A14, SLITRK6, SMO, SMOC1, SNAI2, SNRNP200, SNX3 (MCOPS8), SOX1, SOX10, SOX2, SOX3, SPATA7, SRD5A3, STN1, STRA6, STS, STX3, TACSTD2, TAX1BP3, TBC1D20, TCF4, TDRD7, TEAD1, TEK, TENM3, TFAP2A, TGFBI, TIMM8A, TIMP3, TINF2, TLR4 (ARMD10), TMEM126A, TMEM237, RXYLT1 (TMEM5), TMEM98, TOPORS, TP63, TPP1, TRAF3IP1, TREX1, TRIM32, TRIM37, TRIM44, TRNT1, TRPM1, TSPAN12, TTC8, TTLL5, TTPA, TUB, TUBB3, TUBGCP4, TUBGCP6, TULP1, TYR, TYRP1, UBE3B, UBIAD1, UNC119, UNC45B, USH1C (PDZ73), USH1G (SANS), USH2A, VAX1, VCAN, VIM, VPS13B, VSX1, VSX2 (CHX10), WDPCP, WDR19 (KIAA1638, NPHP13), WDR36, WFS1, WHRN (USH2D,DFNB31), WRN, WT1, XPNPEP2, XYLT2, YAP1, YME1L1, ZBTB20, ZEB1, ZFHX4 (ZFH4), ZNF408, ZNF423, ZNF469, ZNF513, ZNF644, GMPPA, TRAPPC11, SBF2, DDX58, SLC4A4, CPSF1, CRIM1, SEMA3E, TBX22, PIGL, TMEM67, WASHC5 (KIAA0196), ALX1, HDAC6, IGBP1, CTNS, TAT, PLCB3, SAMD11, CLCC1, AHR, ADIPOR1, ARL3, SPP2, STAG2, PPP1CB),
NGS panel Onemocnění ledvin (*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, ABCC6, ACE, ACTN4, ACVR2B, ADAMTS13, ADAMTS9, AGT, AGTR1, AGTR2, AGXT, AHI1, ALG8, ALMS1, ANKFY1/KIAA1255, ANKS6, ANLN, ANOS1/KAL1, AP2S1, APOL1, AQP2, AR, ARHGAP24, ARHGDIA, ARL13B, ARL3, ARL6, ARMC4, ARMC9, ATP6AP2, ATP6V0A4, ATP6V1B1, AVIL, AVPR2, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BICC1, BMP4, BMP7, BMPR1B, BSND, C21orf59, C2orf71, C3, C5orf42/CPLANE1, c8orf37, CA2, CC2D2A, CCDC103, CCDC114, CCDC141, CCDC151, CCDC28B, CCDC39, CCDC40, CCDC65, CCM2/C7orf22, CCNO, CD2AP, CD46, MCP, CDC5L, CDC73, CENPF, CEP104, CEP120, CEP164, CEP290/NPHP6, CEP41, CEP55, CEP83, CFAP221/PCDP1, CFAP300, CFAP53/CCDC11, CFB, CFC1, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CFTR, CLCN5, CLCN5, CLCNKA, CLCNKB, CLDN14, CLDN16, CLDN19, CNNM2, COL4A1, COL4A3, COL4A4, COL4A5, COL4A6, COQ2, COQ8B, CRB2, CRELD1, CSF2RA , CSF2RB, CSPP1, CTNS, CUL3, CYP11B1, CYP11B2, CYP24A1, DACT1, DAW1, DCDC2, DGKE, DMP1, DNAAF1, DNAAF2, KTU, DNAAF3, DNAAF4, DYX1C1, DNAAF5, DNAH1, DNAH11, DNAH3, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2,*

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DNAJB11, DNAJB13, DNAL1, DRC1, DSTYK, DYNC2H1, DYNC2LI1, DZIP1L, EGF, EHHADH, EMP2, ENPP1, EVC, EVC2, EYA1, FAM111A, FGF20, FGF23, FGF8, FGF9, FN1, FOXC1, FOXE1, FOXH1, FOXI1, FOXJ1, FRAS1, FREM1, FREM2, FXYD2, G6PC, GALNT3, GANAB, GAPVD1, GAS2L2, GAS8/GAS11, DRC4, GATA3, GATA6, GBE1, GDF1, GDNF, GJA1, GLIS2/NPHP7, GLIS3, GNA11, GREB1L, KIAA1772/C18ORF6, GREM1, GRHPR, GRIP1, HNF1B, HNF4A, HOGA1, HSD11B2, HTRA1, HYLS1, CHD1L, CHRM3, IFIH1, IFT122, IFT140, IFT172, IFT27, IFT43, IFT52, IFT74/CCDC2, IFT80, IFT81, ILK, IMPDH1, INF2/C14ORF173, INPP5E, INTU/KIAA1284, INVS/NPHP2, IQCB1/NPHP5, ITGA8, KANK2/KIAA1518, KAT6B, KCNA1, KCNJ1, KCNJ10, KCNJ5, KIAA0556/KATNIP, KIAA0586, KIAA0753, KIF14, KIF3B, KIF7, KL, KLHL3, KRIT1, LAMB2, LCA5, LEFTY2, LIN7C, LRP4, LRP5, LRRC56, LRRC6, LRRCC1, LZTFL1/BBS17, MAGED2, MAGI2/KIAA0705, AIP1, MAPKBP1, MCIDAS, MEN1, MKKS, MKS1, MMP21, MMP21, MUC1, MYH9, MYO1E, MYOG, NAT1, NCF1, NEK1, NEK4, NEK8/NPHP9, NFU1, NKX2-1, NKX2-5, NME8, NODAL, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NR3C2, NRIP1, NUP107, NUP133, NUP160/NPHS19, NUP205, NUP37, NUP85, NUP93, OCRL, OFD1, PAX2, PAX8, PBX1, PCBD1, PCDH15, PCSK5, PDCD10, PDE2A, PDE3A, PDE6D, PHEX, PIBF1, PIH1D3, PKD1, PKD1L1, PKD2, PKHD1, PLCE1, PLXND1, PMM2, PNPLA6, PRKCD, PRKCSH, PTPN22, PTPRO, RAB23, REN, RET, ROBO2, RPE65, RPGR, RPGRIP1, RPGRIP1L/NPHP8, RSPH1, RSPH3, RSPH4A, RSPH9, SALL1, SALL4, SCARB2, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SEC61A1, SEC63, SFTPA1, SFTPB, SFTPC, SFTPD, SGPL1, SHROOM3, SIX1, SIX2, SIX5, SLC12A1, SLC12A3, SLC26A4, SLC2A2, SLC34A1, SLC34A3, SLC37A4, SLC3A1, SLC4A1, SLC4A4, SLC5A1, SLC5A2, SLC7A9, SLC9A3R1, SLIT2, SMAD4, SMARCAL1, SOX17, SPAG1, SPEF2, SPRY2, STK36, SUFU, TAC3, TBC1D8B, TBX18, TCTEX1D2, TCTN1/TECT1, TCTN2, TCTN3, TH, THBD, THRA, THRB, TMEM107, TMEM127, TMEM138, TMEM216, TMEM231, TMEM237, TMEM256, TMEM67, TRAF3IP1, TRIM32, TRPC6, TRPM6, TSC2, TSHR, TTC21B, TTC25, TTC8, TTF1, TXNDC15/C5orf14, ULK4, UMOD, UPK3A, USF2, VHL, WDPCP/C2ORF86, WDR19, WDR34, WDR35, WDR60, WDR72, WDR73, WNK1, WNK4, WNT4, WT1, XDH, XPNPEP3, ZIC3, ZMYND10, ZNF423),

NGS panel Onemocnění a vady srdce (*A2ML1, ABCA7, ABCC9, ACTA1, ACTA2, ACTC1, ACTN2, ACVR2B, ADAMTS2, ADAMTSL4, AEBP1, AKAP9, ALG10B, ALMS1, ALPK3, ANK2, ANKRD1, APOA1, APOA2, APOB, APOE, APP, B2M, B3GALT6, B4GALT7, BAG3, BGN, BRAF, C1R, C1S, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CALR3, CASQ2, CAV3, CBL, CFC1, CITED2, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, COX15, CRELD1, CRYAB, CSRP3, CST3, CTF1, CTNNA3, DES, DMD, DNAJC19, DOLK, DPP6, DSC2, DSG2, DSE, DSP, DTNA, EFEMP2, ELN, EMD, EPG5, EYA4, FBN1, FBN2, FGA, FHL1, FHL2, FHOD3, FKBP14, FKRP, FKTN, FLNA, FLNC, FOXE3, GAA, GATA4, GATA5, GATA6, GATAD1, GDF1, GJA1, GJA5, GLA, 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, LMNA, LOX, LTBP2,*

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LTBP3, LYZ, LZTR1, MAP2K1, MAP2K2, MED12, 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, P4HA1, PDLIM3, PKD1L1, PKP2, PLN, PLOD1, PPA2, PPP1CB, PRDM5, 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, SKI, SLC25A4, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, SMAD6, SLMAP, SNTA1, SORL1, SOS1, SOS2, SPRED1, SYNE2, TAF1A, TAZ, TBX1, 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 (*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, B3GALTL, 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, EFNB1, 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, FOXG1, 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,*

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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, TGFBR1, TGFBR2, TGIF1, TMTC3, TNNI2, TNNT3, TNXB, TOR1A, TP63, TPM2, TRAIP, 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),

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

NGS panel Mentální retardace a mikrocefalie (*ACSL4, ACTB, AFF2, AIFM1, ANKLE2, ANKRD11, AP1S2, ARFGEF2, ARHGEF6, ARHGEF9, ARID1B, ARX, ATP6AP2, ATR,*

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osvědčení o akreditaci č.: 645/2021 ze dne: 10. 12. 2021

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ATRX, AUTS2, B3GALNT2, BRWD3, BUB1B, CASK, CCDC22, CCDC88C, CDK16, CDK5RAP2, CDK6, CDKL5, CENPE, CENPF, CENPJ, CEP135, CEP152, CKAP2L, CLCN4, CLIC2, CLP1, CNKSR2, COL4A1, COX7B, CRIPT, CTNNB1, CUL4B, DAG1, DCX, DIAPH1, DLG3, DNM1L, DPP6, DYRK1A, EBP, EFTUD2, EIF2S3, EP300, EXOSC3, FGD1, FKRP, FKTN, FMR1, FOXG1, FRMPD4, FTO, FTSJ1, GDI1, GLI2, GMPPB, GPC3, GRIA3, GSPT2, HCCS, HCFC1, HDAC6, HMGB3, HPRT1, HSD17B10, HUWE1, IER3IP1, IGBP1, IL1RAPL1, ISPD, KATNB1, KIF11, KLF8, L1CAM, LAS1L, MAGT1, MAOA, MCPH1, MECP2, MED12, MED17, MFSD2A, MID1, MPDZ, MSMO1, MYCN, NAA10, NBN, NDE1, NHEJ1, NIPBL, NLGN3, NLGN4X, NSDHL, OCLN, OGT, OPHN1, ORC1, OTC, PAFAH1B1, PAK3, PCLO, PDHA1, PGK1, PHC1, PHF6, PHF8, PLEKHG2, PLK4, PLP1, PNKP, POGZ, POMGNT1, POMT1, POMT2, PORCN, PPP1R15B, PSAT1, PSPH, PTCHD1, QARS, RAB39B, RAB3GAP1, RAB40AL, RAD21, RAD50, RBBP8, RBM10, RPL10, RPS6KA3, RTTN, SASS6, SHH, SHROOM4, SLC16A2, SLC1A4, SLC25A19, SLC2A1, SLC6A8, SMC3, SMS, SOX11, SOX3, SPATA5, SRPX2, STAMBP, STIL, SYN1, SYP, TAF1, TAF2, THOC2, THOC6, TIMM8A, TRAIP, TRAPPC9, TRMT10A, TSEN54, TUBA1A, TUBB, TUBB2B, TUBGCP4, TUBGCP6, UBE2A, UPF3B, USP9X, VPS13B, WDR13, WDR62, WDR73, XRCC4, ZBTB18, ZC4H2, ZDHHC15, ZDHHC9, ZEB2, ZNF335, ZNF711, ZNF81, AARS1, ABCA2, ACBD5, ACTL6B, ADAR, ADARB1, ADAT3, ADNP, AGA, AGTPBP1, AHDC1, AIMP1, AIMP2, AKT3, ALDH3A2, ALG1, ALG11, ALG12, ALG13, ALG3, ALG9, ALKBH8, AMPD2, ANK3, AP2M1, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, ARCN1, ARID1A, ARID2, ARNT2, ARSE, ARV1, ASH1L, ASNS, ASPM, ASXL1, ASXL3, ATAD3A, ATP6V1A, ATRIP, B3GNT1 , BCAP31, BCL11A, BCL11B, BPTF, BRAT1, BRD4, BRF1, BRPF1, C12orf4, CA8, CACNA1B, CACNA1G, CACNG2, CAMK2A, CAMK2B, CAMK2G, CAMTA1, CARS1, CC2D1A, CCDC32, CCDC47, CCND2, CD96, CDC45, CDC6, CDH15, CDK5, CDON, CDT1, CENPT, CEP63, CEP85L, CIC, CIT, CLTC, CNNM2, CNOT1, CNOT3, COASY, COG1, COG2, COG4, COG5, COG6, COG7, COL4A3BP, COPB2, CRADD, CRBN, CREBBP, CSNK2A1, CTCF, CTNNA2, CTU2, CUX1, CXorf56, CYFIP2, DCPS, DDX11, DDX3X, DEAF1, DENND5A, DHCR7, DHX37, DISP1, DLG4, DLL1, DMXL2, DNA2, DNMT3A, DONSON, DPAGT1, DPF2, DPM1, DYNC1H1, DYNC1I2, EBF3, EDC3, EEF1A2, EHMT1, EIF3F, ELP2, EMC1, EMX2, EPB41L1, EPG5, ERCC1, ERCC2, ERCC3, ERLIN2, ETHE1, EXOC7, EXOSC8, EXOSC9, EXT2, FAM50A, FAR1, FARSA, FARSB, FBXL3, FBXL4, FBXO31, FGF8, FGFR1, FH, FLVCR2, FMN2, FOXH1, FOXP1, FREM1, FRMD4A, FUT8, GABBR2, GABRB2, GAD1, GALNT2, GAS1, GATAD2B, GEMIN4, GLDC, GLYCTK, GMNN, GMPPA, GNAO1, GNB1, GOT2, GPT2, GRIA4, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRM7, GTF2E2, GTF2H5, GTPBP2, HACE1, HDAC8, HERC2, HHAT, HIVEP2, HNMT, HNRNPH2, HNRNPU, HPDL, HS6ST2, CHAMP1, CHD2, CHD3, CHD7, CHMP1A, IARS1, IFIH1, IFT122, IGF1, IGF1R, IMPA1, INPP5E, INTS8, IQSEC2, KANSL1, KAT6A, KAT6B, KCNA4, KCNQ5, KCNT1, KDM5B, KDM5C, KIAA1109, KIF14, KIF1A, KIF2A, KIF4A, KIF5C, KIFBP, KIRREL3, KLHL15, KLHL7, KMT2A, KMT2C, KMT2D, KMT2E, KMT5B, KNL1, KPTN, LAGE3, LAMB1, LARGE1, LARP7, LIG4, LINGO1, LINS1, LMAN2L, LMNB1, LMNB2, MAB21L1, MACF1,

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MAN1B1, MAP11, MAP1B, MAPRE2, MAST1, MBD5, MBOAT7, MCM5, MED13, MED13L, MED23, MED25, MEF2C, MEIS2, METTL23, METTL5, MFF, MID2, MIR17HG, MOCS1, MORC2, MPLKIP, MSL3, MTHFS, MTX2, MYMK, MYO18B, MYT1L, NAA15, NADK2, NALCN, NANS, NARS1, NBEA, NCAPD2, NCAPD3, NCAPH, NDST1, NEXMIF, NGLY1, NIN, NKAP, NODAL, NONO, NRXN1, NSMCE2, NSUN2, NT5C2, NTNG2, NUP107, NUP133, NUP188, NUP37, NUS1, ORC4, ORC6, OSGEP, PACS1, PARS2, PCDH12, PCGF2, PCNT, PDE2A, PGAP1, PGAP2, PGAP3, PHGDH, PIGG, PIGN, PIGO, PIGV, PIGY, PIK3R2, PLAA, PMM2, POC1A, POLA1, POLR1A, POLR2A, POMK, PPP2CA, PPP2R1A, PPP2R5D, PQBP1, PRMT7, PRSS12, PRUNE1, PSMD12, PTCH1, PTPN23, PURA, PUS7, PYCR2, QRICH1, RAB11B, RAB18, RAB3GAP2, RAC1, RARS1, RARS2, RBMX, RELN, RFT1, RHOBTB2, RIC1, RLIM, RNASEH2A, RNASEH2B, RNASEH2C, RNF113A, RNF13, RNU4ATAC, RPS23, RSRC1, RUSC2, RXYLT1, SAMHD1, SARS1, SCN1A, SCN2A, SCN3A, SEMA3E, SEPSECS, SET, SETBP1, SETD5, SF3B4, SHANK3, SIN3A, SIX3, SLC12A6, SLC1A2, SLC35A2, SLC35C1, SLC5A6, SLC6A17, SLC9A6, SLC9A7, SMARCA2, SMARCA4, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SMC1A, SMG9, SMPD4, SNAP29, SNRPB, SOBP, SON, SOX4, SOX5, SPOP, SPTAN1, SRCAP, SSR4, ST3GAL3, STAG1, STAG2, STT3A, STT3B, STXBP1, SVBP, SYNGAP1, TAF13, TAF6, TARS1, TASP1, TBC1D20, TBC1D23, TBCD, TBCK, TBL1XR1, TBR1, TBX1, TCF20, TCF4, TDP2, TECPR2, TECR, TELO2, TGIF1, TLK2, TMCO1, TMEM165, TMTC3, TMX2, TNIK, TOE1, TOP3A, TP53RK, TPRKB, TRAPPC12, TRAPPC4, TRAPPC6B, TREX1, TRIO, TRIP12, TRMT1, TRRAP, TSEN15, TSEN2, TSEN34, TSPAN7, TTI2, TUBA8, TUBB2A, TUBB3, TUBG1, TUBGCP2, TUSC3, UBA5, UBE3A, UBE3B, UBR1, UBR7, UBTF, UFC1, UFM1, UGP2, UNC80, UPB1, USP27X, VARS1, VLDLR, VPS11, VPS51, VPS53, VRK1, WASHC4, WDFY3, WDR37, WDR4, WDR45B, WWOX, YIF1B, YY1, ZBTB11, ZBTB16, ZC3H14, ZIC1, ZIC2, ZMYND11, ZNF148, ZNF292, ZNHIT3, ZSWIM6).

[11] SALSA 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,

Akreditovaný subjekt podle ČSN EN ISO 15189:2013:

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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.

[12] Hemochromatóza (sekvenční varianty: H63D, S65C, 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, 719 A>G v genu *TPMT*), Deficit alfa 1-antitrypsinu (sekvenční varianty: E264V, E342K v genu *SERPINA1*), *KRAS* (sekvenční varianty: 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)

[13] *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)