

Seznam činností v rámci flexibilního rozsahu akreditace

Název subjektu: Fakultní nemocnice Ostrava

Název objektu: Ústav klinické a molekulární patologie a lékařské genetiky

Číslo akreditovaného objektu: 8194

Osvědčení o akreditaci č. 436/2025

Oblast akreditace: Zdravotnická laboratoř – ČSN EN ISO 15189 ed. 3:2023

Aktualizováno dne: 13.1.2026

1. Laboratoře Ústavu klinické a molekulární patologie a lékařské genetiky

17. Listopadu 1790/5, 708 52 Ostrava - Poruba

Vyšetření:

| Poř. číslo | Analyt / parametr/diagnostika | Princip vyšetření | Identifikace postupu/ přístrojové vybavení | Vyšetřovaný materiál | Stupně volnosti ¹ |
|--|---------------------------------------|-----------------------|---|--|------------------------------|
| 816 - Laboratoř lékařské genetiky | | | | | |
| 1. | Vyšetření variant germinálního genomu | Sangerovo sekvenování | SOP-LAB-ÚKMPLG-07 revize 04 SOP-LAB-ÚKMPLG-12 revize 02 ABI3130 ABI3130xl | Periferní krev, plodová voda, choriové klky, fetální tkáň, izolovaná NK | A, B, C, D |
| 2. | Vyšetření variant germinálního genomu | Fragmentační analýza | SOP-LAB-ÚKMPLG-07 revize 04 SOP-LAB-ÚKMPLG-14 revize 00 ABI3130, ABI3130xl | Periferní krev, plodová voda, choriové klky, fetální tkáň, kostní dřev, izolovaná NK | A, B, C, D |
| 3. | Vyšetření variant somatického genomu | Fragmentační analýza | SOP-LAB-ÚKMPLG-07 revize 04 SOP-LAB-ÚKMPLG-18 revize 00 ABI3130 ABI3130xl | Tkáň, buňky, periferní krev, kostní dřev, izolovaná NK | A, B, C, D |
| 4. | Stanovení konstitučního karyotypu | Mikroskopie | SOP-LAB-ÚKMPLG-05 revize 02 | Krev, plodová voda, choriová tkáň, tkáň | A, B, D |

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|------------|--|-------------------------|---|---|------------------------------|
| 5. | Vyšetření variant germinálního genomu | FISH | SOP-LAB-ÚKMPLG-06 revize 04 | Fixovaná buněčná suspenze, krev, plodová voda, choriová tkáň, tkáň | A, B, C, D |
| 6. | Vyšetření chromozomových aberací | FISH | SOP-LAB-ÚKMPLG-06 revize 04 | Fixovaná buněčná suspenze, krev, plodová voda, choriová tkáň, tkáň | A, B, C, D |
| 7. | Vyšetření získaných chromozomových aberací | Mikroskopie | SOP-LAB-ÚKMPLG-08 revize 01 | Lymfocyty z periferní krve | A, B |
| 8. | Vyšetření variant germinálního genomu | MPS - NGS | SOP-LAB-ÚKMPLG-07 revize 04 SOP-LAB-ÚKMPLG-11 revize 05 MiniSeq NextSeq 550 NovaSeq 6000Dx | Periferní krev, plodová voda, choriové klky, fetální tkáň, izolovaná NK | A, B, C, D |
| 9. | Vyšetření variant somatického genomu | MPS - NGS | SOP-LAB-ÚKMPLG-07 revize 04 SOP-LAB-ÚKMPLG-11 revize 05 MiniSeq NextSeq 550 NovaSeq 6000Dx | Tkáně, buňky, kostní dřeň, plazma, izolovaná NK | A, B, C, D |
| 10. | Vyšetření nebalancovaných chromozomových aberací | CGH array, SNP array | SOP-LAB-ÚKMPLG-07 revize 04 SOP-LAB-ÚKMPLG-09 revize 02 Agilent G5761A SureScan Dx NextSeq 550 | Periferní krev, plodová voda, choriové klky, fetální tkáň, izolovaná NK | A, B, C, D |

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|----------------------------------|--|-------------------|---|---|------------------------------|
| 11. | Vyšetření variant germinálního genomu | MLPA | SOP-LAB-ÚKMPLG-07 revize 04 SOP-LAB-ÚKMPLG-13 revize 03 ABI3130 ABI3130xl | Periferní krev, plodová voda, choriové klky, fetální tkáň, izolovaná NK | A, B, C, D |
| 12. | Vyšetření variant somatického genomu | MLPA | SOP-LAB-ÚKMPLG-07 revize 04 SOP-LAB-ÚKMPLG-13 revize 03 ABI3130 ABI3130xl | Tkáň, buňky, kostní dřeň, plazma, izolovaná NK | A, B, C, D |
| 13. | Vyšetření variant germinálního genomu | Real-time PCR | SOP-LAB-ÚKMPLG-07 revize 04 SOP-LAB-ÚKMPLG-10 revize 00 LightCycler 480II LightCycler v1.5 | Periferní krev, plodová voda, choriové klky, fetální tkáň, izolovaná NK | A, B, C, D |
| 14. | Vyšetření variant somatického genomu | Real-time PCR | SOP-LAB-ÚKMPLG-07 revize 04 SOP-LAB-ÚKMPLG-19 revize 00 EasyPGX Idylla™ | Tkáň, buňky, kostní dřeň, plazma, izolovaná NK | A, B, C, D |
| 823 - Laboratoř patologie | | | | | |
| 1. | Histologické vyšetření a diagnostika | Mikroskopie | SOP-LAB-ÚKMPLG-01 revize 02 tkáňový procesor | Tkáň | A, B |
| 2. | Peroperační histologické a cytologické vyšetření a diagnostika | Mikroskopie | SOP-LAB-ÚKMPLG-02 revize 02 kryotom | Tkáň, buňky | A, B |
| 3. | Stanovení základní cytologické diagnózy | Mikroskopie | SOP-LAB-ÚKMPLG-03 revize 02 cytocentrifuga | Buňky z punkce tkání, z tělních tekutin a obsahu patologických dutin | A, B |

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|------------|--|----------------------|--|---|------------------------------|
| 4. | Imunohistochemický/ imunocytochemický průkaz sledovaného markeru | Mikroskopie | SOP-LAB-ÚKMPLG-04 revize 02 imunostainer | Tkáně, buňky | A, B, C |
| 5. | Vyšetření chromozomových aberací | FISH | SOP-LAB-ÚKMPLG-06 revize 04 | Tkáně, buňky | A, B, C, D |
| 6. | Vyšetření variant somatického genomu | MPS - NGS | SOP-LAB-ÚKMPLG-07 revize 04 SOP-LAB-ÚKMPLG-11 revize 05 MiniSeq NextSeq 550 NovaSeq 6000Dx | Tkáně, buňky, kostní dřeň, plazma, izolovaná DNA/RNA | A, B, C, D |
| 7. | Vyšetření variant somatického genomu | Real-time PCR | SOP-LAB-ÚKMPLG-07 revize 04 SOP-LAB-ÚKMPLG-19 revize 00 EasyPGX Idylla™ | Tkáně, buňky, kostní dřeň, plazma, izolovaná NK DNA/RNA | A, B, C, D |
| 8. | Vyšetření variant somatického genomu | Fragmentační analýza | SOP-LAB-ÚKMPLG-07 revize 04 SOP-LAB-ÚKMPLG-18 revize 00 ABI3130 ABI3130xl | Tkáně, buňky, kostní dřeň, izolovaná DNA/RNA | A, B, C, D |

Upřesnění rozsahu akreditace:

| Odbornost / poř. číslo | Detailní informace k činnostem v rozsahu akreditace |
|------------------------|--|
| 816/1 | Geny: <i>GJB2</i> , <i>HFE</i> , <i>CYP2C9</i> , <i>VKORC1</i> |
| 816/2 | Aneuploidie chromozomů 13, 18, 21, X a Y Syndrom fragilního X (analýza počtu repetice CGG v 5'UTR oblasti genu <i>FMRI</i>) Chimerismus: vyšetření vybraných vysoce polymorfních mikrosatelitů (STR), delecí a inzercí za účelem typizace a monitoringu dárce a příjemce hematopoetických buněk |
| 816/3 | Mikrosatelitová instabilita: BAT-25, BAT-26, NR-21, NR-24, MONO-27, Penta C a Penta D |
| 816/5 | POSEIDON™ (f. KREATECH), AQUARIUS ^R (f. CYTOCELL), Human BAC FISH Probes (f. EmpireGenomics) |

| Odbornost / poř. číslo | Detailní informace k činnostem v rozsahu akreditace |
|---------------------------|---|
| 816/6 | <p>1p36/1q25, 19q13/19p13, 1p/12, ALK, ALK/EML4, ATM/CEN12, BCL2, BCL2/IGH, BCL6, BCOR, BIRC3/MAL1, CCND1, CCND1/IGH, CDKN2A/CEN9, CDK4/CEN12, CIC, COL1A1, DDIT3, EGFR/CEN7, ERG, ETV6, EWSR1, EWSR1/FLI1, EGFR1, FOXO1, FUS, HER2/CEN17, IGH, IRF4/DUSP22, JAZF1, MALT1, MAML2, MDM2/CEN12, MET/CEN7, MYC, MYC/CEN8, NR4A3, NTRK3, NUTM1, PDGFB, PDGFRA/FIP1L1, PHF1, PTEN/CEN10, RB1/13q34, RET, ROS1, SMARCB1/22q12, SS18, SS18/SSX1, TFE3, TP53/CEN17, USP6, VHL/CEN3, WT1, WWTR1</p> |
| 816/8 | <p>Neurodegenerativní onemocnění</p> <p><i>AARS1, AARS2, ABCA7, ABCD1, ACOX1, ACP33, ADCY5, AFG3L2, ALDH18A1, ALS2, ANG, ANO10, ANO3, ANXA11, AOPEP, AP4B1, AP4E1, AP4M1, AP5Z1, APOE, APP, APTX, ARSA, ATLI, ATM, ATP13A2, ATP1A3, ATP7B, ATXN2, ATXN3, AUH, B4GALNT1, BCAP31, BSCL2, C19orf12, C9orf72, CACNA1A, CACNA1B, CACNA1G, CACNB4, CAMTA1, CAPN1, CCNF, CIZ1, CLCN2, COASY, COL4A2, COL6A3, COLGALT1, CP, CPT1C, CRAT, CSF1R, CTC1, CTSF, CYP27A1, CYP2U1, CYP7B1, DAB1, DAO, DARS2, DCAF17, DCTN1, DDC, DDHD1, DNAJC12, DNAJC13, DNAJC5, DNAJC6, DNAJC7, DNMT1, DPGFB, DRD2, EEF2, EIF2AK2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF4G1, ELOVL4, ELOVL5, ENTPD1, EPHA4, ERBB4, ERLIN1, ERLIN2, FA2H, FARS2, FAT2, FBXO7, FGF14, FIG4, FTL, FUS, FXN, GBA, GBA2, GCDH, GCH1, GIGYF2, GJC2, GLE1, GLT8D1, GLUD2, GNAL, GNAO1, GNE, GPX3, GRM1, GRN, GTPBP2, HEXA, HEXB, HNRNPA1, HNRNPA2B1, HPCA, HPDL, HPRT1, HSPD1, HTRA1, HTRA2, HTT, CHCHD10, CHCHD2, CHMP2B, IBA57, ITM2B, ITPR1, JAKMIP3, JAM2, KCNC3, KCND3, KCNMA1, KCTD17, KIF1A, KIF1C, KIF5A, KMT2B, LMNB1, LRRK2, MAG, MAPT, MARS2, MATR3, MME, MMUT, MYORG, NDUFC2, NEFH, NEK1, NFUI, NIPAI, NKX2-1, NOTCH3, NRROS, NT5C2, NUP50, OPTN, PANK2, PANK3, PARK7/DJ1, PCCA, PCCB, PCNA, PDE10A, PDGFB, PDGFRB, PDYN, PFN1, PINK1, PLA2G6, PLD3, PLP1, PNKD, PNPLA6, POLG, PPT1, PRKCG, PRKN/PARK2, PRKRA, PRNP, PRPH, PRRT2, PSAP, PSEN1, PSEN2, PTS, PUM1, RAB39B, REEP1, REEP2, RELN, REPS1, RNF170, RNF216, RTN2, SACS, SCFD1, SCN2A, SCP2, SETX, SGCE, SIGMARI, SLC18A2, SLC1A3, SLC20A2, SLC2A1, SLC30A10, SLC33A1, SLC39A14, SLC6A3, SLC9A8, SNC1A, SNCA, SNCB, SNORD118, SOD1, SPART, SPAST, SPG11, SPG7, SPR, SPTBN2, SPTLC1, SQSTM1, SS18L1/KIAA0693, STUB1, SYNE1, SYNJ1, TAF1, TAF15, TARDBP, TBK1, TBP, TFG, TGM6, TH, THAP1, TIA1, TIMM8A, TMEM240, TMEM63A, TNIP1, TOR1A, TPP1, TREX1, TRPC3, TTBK2, TTPA, TTR, TUBA4A, TUBB4A, TWNK, TYROBP, UBAP1, UBQLN2, UCHL1, UNC13A, USP18, VAMPI1, VAPB, VCP, VPS13A, VPS13C, VPS13D, VPS16, VPS35, VPS37A, VPS41, WASHC5, WDR45, XK, XPR1, ZFYVE26, ZFYVE27 (v3)</i></p> <p>Hereditární nádorové syndromy a somatické varianty v nádorových tkáních</p> <p>Germinální testování: <i>ACD, AIP, AKT1, ALK, APC, AR, ATM, ATR, AXIN2, BAP1, BARD1, BLM, BMP1A, BRAF, BRCA1, BRCA2, BRF1, BRIP1, CARD11, CCND1, CDC73, CDH1, CDK4, CDKN1B, CDKN2A, CYLD, DICER1, DLST, DNMT3A, EGFR, ELP1, EMSY, EPCAM, ERBB2, ERBB3, ERCC2, ERCC3, ERCC5, ETV6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAT1, FBXW7, FH, FLCN, GATA2, GREM1, HNF1A, HOXB13, HRAS, CHEK1, CHEK2, IDH1, IDH2, KIF1B, KIT, KLLN, KMT2A, KRAS, MAP2K1, MAX, MC1R, MEN1, MET, MITF, MLH1, MLH3, MN1, MRE11, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2, NRAS, NRG1, NTHL1, PALB2, PALD, PARP1, PARP2, PARP3, PARP4, PBRM1, PDGFB, PDGFD, PDGFRA, PDGFRB, PHOX2B, PIK3CA, PMS2, POLD1, POLE, POT1, PPP2R2A, PRKARIA, PTEN, PTCH1, PTCH2, RABL3, RAD50, RAD51, RAD51B, RAD51C, RAD51D, RAD52, RAD54L, RB1, RECQL, RECQL4, RET, REXO2, RNF43, RPS20, SDHA, SDHAF2, SDHB, SDHC, SDHD, SH2D1A, SH2D2A, SLC25A11, SLX4, SMAD4, SMARCA4, SMARCB1, SMARCE1, SRGAP1, SRY, STK11, SUFU, TERF2IP, TERT, TET2, TMEM127, TP53, TSC1, TSC2, VHL, WRN, WT1, XPA, XRCC2, XRCC3 (hg38).</i> Somatické testování: <i>ABRAXAS1, ACVR1, ALK, ARAF, ARID1A, ARID1B, ATRX, BRK1, CCDC153, CCNE1, CD79B, CDADC1, CDK12, CDK6, CDKN2B, COX6C, CTNNA1, CTNNB1, DDR2, DPYD, EIF1AX, ENG, ENOX1, ERG, ESRI, EZHIP, FAM124A, FANCD2OS, FGFR1, FGFR2, FGFR3, FOXL2, GEN1, HDAC2, H3-C14 (H3), H3C2 (HIST1H3B), H3C3, H3-3A, IL18, INA, INCA1, KEAP1, KHDC3L, KIAA1549, MAD2L2, MAMLD1, MALT1, MTOR, MYB, MYBL1, MYC, MYCN, MYD88, MYH13, NAT1,</i></p> |

| Odbornost / poř. číslo | Detailní informace k činnostem v rozsahu akreditace |
|---------------------------|---|
| | <p><i>MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY (v5)</i></p> <p>Vrozené kostní anomálie</p> <p><i>AAAS, ABCC9, ACAN, ACKR3, ACOX1, ACP5, ACTA1, ACTB, ACTG1, ACVR1, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTSL2, ADCY6, ADGRG6, AFF4, AGA, AGPS, AHDC1, AH11, AIFM1, AIRE, AKT1, AKT3, ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, ALPL, ALX1, ALX3, ALX4, AMER1, ANAPC1, ANKH, ANKRD11, ANO5, ANTXR1, ANTXR2, AP1S1, APC, ARCNI, ARHGAP31, ARID1A, ARID1B, ARID2, ARL6, ARMC9, ARSB, ARSL, ASCC1, ASXL1, ASXL2, ASXL3, ATAD1, ATLI, ATP1A2, ATP2B1, ATP2B3, ATP6AP1, ATP6AP2, ATP6V0A2, ATP7A, ATR, ATRIP, ATRX, B3GALT6, B3GAT3, B3GLCT, B4GALT1, B4GALT7, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCOR, BGN, BHLHA9, BICD2, BLM, BLTP1, BMP1, BMP2, BMP4, BMP7, BMPER, BMPR1B, BPNT2, BRAF, BRCA2, BRD4, BRIP1, BRWD3, C2CD3, CA2, CACNA1C, CANT1, CASR, CBFEB, CBL, CC2D2A, CCBE1, CCDC115, CCDC22, CCDC47, CCDC8, CCN6, CCND2, CCNK, CCNQ, CD96, CDAN1, CDC42, CDC45, CDC6, CDH1, CDH11, CDH3, CDIN1, CDKN1C, CDT1, CENPE, CENPF, CENPJ, CEP120, CEP152, CEP290, CEP41, CEP55, CEP63, CERT1, CFAP410, CFAP418, CFL2, CFTR, CIBAR1, 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, CRIPT, CRTAP, CSGALNACT1, CSPP1, CST6, CTNNB1, CTNS, CTSA, CTSC, CTSK, CTU2, CUL4B, CUL7, CWC27, CYP19A1, CYP26B1, CYP27B1, CYP2R1, DCPS, DDR2, DDRGK1, DDX11, DDX59, DEAF1, DHCR24, DHCR7, DHODH, DCHS1, DIS3L2, DLL3, DLL4, DLX3, DLX5, DMP1, DNA2, DNAJC21, DNMT2, DNMT3A, DOCK6, DOK7, DONSON, DPAGT1, DPF2, DPH1, DPM1, DPM2, DPM3, DPYS, DSE, DUOX2, DUOXA2, DVLI, DVL3, DYM, DYNC2H1, DYNC2L1I, DYNLT2B, EBP, ECEL1, EDARADD, EDN1, EED, EFEMP1, EFEMP2, EFL1, EFNA4, EFN1, EFTUD2, EIF2AK3, EIF4A3, ELMO2, ENPPI, 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, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FAT4, FBLN1, FBLN5, FBN1, FBN2, FBXL3, FBXO11, FBXW11, FCSK, FDFT1, FERMT1, FERMT3, FGD1, FGF10, FGF13, FGF16, FGF17, FGF23, FGF8, FGF9, FGFR1, FGFR2, FGFR3, FHL1, FIBP, FIG4, FKBP10, FLNA, FLNB, FLVCR2, FN1, FOXE1, FRAS1, FREM1, FREM2, FTO, FUCA1, FUT8, FUZ, FZD2, GALNS, GALNT2, GALNT3, 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, GRHL2, GRHL3, GRIPI, GSC, GTF2E2, GUSB, GZF1, H1-4, HAAO, HDAC4, HDAC6, HDAC8, HERC1, HES7, HESX1, HGSNAT, HMGA2, HMX2, HNRNPK, HNRNPR, HOXA11, HOXA13, HOXD10, HOXD13, HOXD3, HPGD, HRAS, HSD17B4, HSPA9, HSPG2, HUWE1, HYAL1, HYL1, CHAT, CHD4, CHD7, CHKA, CHRNG, CHST11, CHST14, CHST3, CHSY1, CHUK, IARS2, IBA57, 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, ITPR1, IYD, JAG1, KAT6A, KAT6B, KBTBD13, KCNH1, KCNJ1, KCNJ2, KCNJ8, KCNK4, KCNMA1, KCNN3, KCTD1, KDM1A, KDM6A, KDM6B, KIAA0586, KIAA0753, KIAA0825, KIAA1279, KIF14, KIF1A, KIF22, KIF3B, KIF5C, KIF7, KL, KLHL40, KLHL41, KMT2A, KMT2C, KMT2D, KPTN, KRAS, KYNU, LBR, LEMD3, LFNG, LGI4, LHX4, LIFR, LIG4, LMBR1, LMNA, LMNB1, LMOD3, LMX1B, LONP1, LPIN2, LRP2, LRP4, LRP5, LTBP2, LTBP3, LZTFL1, LZTR1, MAB21L1, MAB21L2, MAD2L2, MAF, MAFB, MAGEL2, MAGI2, MAGT1, MAN2B1, MANBA, MAP1B, MAP2K1, MAP2K2, MAP3K20, MAP3K7, MAPK1, MASP1, MATN3, MBTPS1, MBTPS2, MCM3AP, MCM5, Mecom, MED12, MEGF8, MEIS2, MEOX1, MESD, MESP2, MET, MGAT2, MGP, MKKS, MKS1, MMP13, MMP14, MMP2, MMP9, MN1, MNX1, MOGS, MPDU1, MPI, MPZ, MRAS,</i></p> |

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| | <p><i>MRPS16, MRPS28, MSX1, MSX2, MTAP, MTRFR, MTX2, MUSK, MYBPC1, MYCN, MYH3, MYH8, MYO18B, MYO9A, MYOD1, MYPN, NAA10, NAGLU, NANS, NBAS, NBN, NCAPG2, NEB, NECTIN1, NECTIN4, NEDD4L, NEK1, NEK9, NEPRO, NEU1, NEXMIF, NF1, NFIX, NFKB2, NGLY1, NHEJ1, NHERF1, NIN, NIPBL, NKX2-1, NKX2-5, NKX3-2, NLRP3, NOG, NOTCH1, NOTCH2, NPHP3, NPR2, NPR3, NRAS, NSD1, NSDHL, NSMCE2, NSUN2, NUP88, NUS1, NXN, OBSL1, OCRL1, OFD1, ORC1, ORC4, ORC6, OSTM1, P3H1, P4HB, PALB2, PAM16, PAPSS2, 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, PLCB3, PLEKHM1, PLK4, PLOD1, PLOD2, PLOD3, PLS3, PMM2, PMP22, POC1A, POLE, POLR1A, POLR1B, POLR1C, POLR1D, POLR3A, POMP, POP1, POR, PORCN, PPIB, PPM1D, PPP1CB, PPP1R12A, PPP3CA, PRDM5, PRG4, PRIM1, PRKARIA, PRMT7, PROM1, PRRX1, PSAT1, PSMD12, PSPH, PTDSS1, PTEN, PTH, PTH1R, PTHLH, PTCH1, PTCH2, PTK7, PTPN11, PUF60, PUM1, PYCRI, QRICHI, RAB23, RAB33B, RAB34, RAD21, RAD51, RAD51C, RAF1, RAI1, RALGAP1, RAPIA, RAPIB, RAPSN, RARB, RBBP8, RBM10, RBM8A, RBMX, RBPJ, RECQL4, RFT1, RFXD3, RHOA, RIN2, RIPK4, RIPPLY2, RNF125, ROR2, RPGRIP1L, RPL10, RPL11, RPL13, RPL15, RPL26, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS6KA3, RPS7, RRAS2, RSPO2, RSPRY1, RTTN, RUNX2, RYR1, SALL1, SALL4, SAMD9, SATB2, SBDS, SBF1, SC5D, SCARF2, SCYL2, SDCCAG8, SEC23A, SEC24D, SEM1, SEMA3E, SERPINF1, SERPINH1, SETBP1, SETD2, SF3B4, SFMBT1, 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, SLC35B2, SLC35C1, SLC35D1, SLC39A13, SLC39A8, SLC4A2, SLC5A5, SLC5A7, SLC6A9, SLCO2A1, SLURP1, SLX4, SMAD3, SMAD4, SMAD6, SMARCA2, SMARCA4, SMARCAL1, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SMC1A, SMC3, SMN1, SMN2, SMO, SMOC1, SMPD4, SMS, SNRPA, SNRPB, SNX10, SON, SOS1, SOS2, SOST, SOX11, SOX3, SOX4, SOX6, SOX9, SP7, SPARC, SPART, SPECC1L, SPRTN, SQSTM1, SRCAP, SRD5A3, SSR4, STAC3, STAG2, STAMPB, STAT3, STT3A, STT3B, STX16, SUFU, SUMF1, SUZ12, SYNE1, SYNE2, TAB2, TALDO1, TAPT1, TASP1, TBC1D24, TBC1D2B, TBCD, TBCE, TBL1X, TBL1XR1, TBX1, TBX15, TBX18, TBX2, TBX22, TBX3, TBX4, TBX5, TBX6, TBXAS1, TBXT, TCF12, TCF20, TCIRG1, TCOF1, TCTN2, TCTN3, TELO2, TENT5A, TERT, TFAP2A, TFAP2B, TG, TGDS, TGFBI, TGFBI2, TGFBI3, TGFBR1, TGFBR2, THRA, TLK2, TMC01, TMEM107, TMEM138, TMEM165, TMEM199, TMEM216, TMEM231, TMEM237, TMEM263, TMEM38B, TMEM43, TMEM67, TMEM94, TNFRSF11A, TNFRSF11B, TNFSF11, TNNT2, TNNT1, TNNT3, TONSL, TOR1A, TP63, TPM2, TPM3, TPO, TRAF3IP1, TRAI, TRAPPC2, TREM2, TRH, TRHR, TRIM32, TRIO, TRIP11, TRIP4, TRPS1, TRPV4, TRPV6, TRRAP, TSHB, TSHR, TTC21B, TTC8, TTN, TWIST1, TWIST2, TXNDC15, TXNL4A, TYROBP, UBA1, UBA2, UBE2T, UBE3B, UBR1, UFSP2, USP9X, VAC14, VDR, VIPAS39, VPS13B, VPS33A, VPS33B, VPS35L, VPS41, VPS4A, WBP4, WDPCP, WDR19, WDR34, WDR35, WDR37, WDR60, WNT1, WNT10A, WNT10B, WNT3, WNT5A, WNT7A, XRCC2, XRCC4, XYLT1, XYLT2, YY1, YY1AP1, ZBTB16, ZBTB42, ZBTB7A, ZC4H2, ZFH4, ZIC1, ZIC3, ZMIZ1, ZMPSTE24, ZNF141, ZNF335, ZNF462, ZNF469, ZSWIM6 (v5)</i></p> <p>Onemocnění ledvin a ciliopatie</p> <p><i>ABCA3, ABCB11, ABCB4, ABCC2, ABCC6, ACE, ACTG2, ACTN4, ACVR2B, ADAMTS13, ADAMTS9, AFF4, AGT, AGTR1, AGTR2, AGXT, AH11, AKR1D1, ALDOB, ALG5, ALG8, ALG9, ALMS1, AMACR, AMN, ANKFY1/KIAA1255, ANKS6, ANLN, ANOS1/KALI, AP2S1, APOA1, APOL1, APRT, AQP2, AR, ARHGAP24, ARHGAP24, ARHGAP24, ARL13B, ARL2BP, ARL3, ARL6, ARMC4, ARMC9, ATP1A1, ATP6AP2, ATP6V0A4, ATP6V1B1/ATP6B1, ATP8B1, AVIL, AVP, AVPR2, B2M, B9D1, B9D2, BAAT, BBIP1/BBS18, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9/PTHB1, BDNF, BICC1, BMP4, BMP7, BMPR1B, BNC2, BSND, C21orf59/CFAP298, C2CD3, C2orf71/PCARE, C3, C5orf42/CPLANE1, c8orf37/CFAP418, C8ORF38/NDUFAF6, CA12, CA2, CACNA1H, CAMKMT, CASR, CBY1, CC2D2A, CCDC103, CCDC114, CCDC141, CCDC28B, CCDC32, CCDC39, CCDC40, CCDC65, CCM2/C7orf22,</i></p> |

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| | <p>CCNO, CD19, CD2AP, CD46/MCP, CD81, CDC151/ODAD3, CDC5L, CDC73, CELSR2, CELSR3, CENPF, CEP104, CEP120, CEP164, CEP19, CEP290/NPHP6/BBS14, CEP41, CEP55, CEP83, CFAP221/PCDP1, CFAP300, CFAP52/WDR16, CFAP53/CCDC11, CFB, CFC1/CRYPTIC, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CFTR, CLCN2, CLCN5, CLCNKA, CLCNKB, CLDN10, CLDN14, CLDN16, CLDN19, CNNM2, COL4A1, COL4A3, COL4A4, COL4A5, COL4A6, COQ2, COQ6, COQ8B/ADCK4, CPE, CR2, CRB2, CRELD1, CRIM1, CRKL, CSF2RA, CSF2RB, CSPP1, CTNS, CTU2, CUBN, CUL3, CYP11B1, CYP11B2, CYP24A1, CYP27A1, DAAM2, DACT1, DAW1, DCDC2, DEAF1, DGKE, DLC1, DLG5, DMP1, DNAAF1, DNAAF2/KTU, DNAAF3, DNAAF4/DYX1C1, DNAAF5, DNAH1, DNAH11, DNAH3, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB11, DNAJB13, DNALI1, DRC1, DSTYK, DYNC2H1, DYNC2LI1, DYRK1B, DZIP1L, EGF, EHHADH, EIF2S3, EMP2, ENPP1, EVC, EVC2, EXOC3L2, EYA1, FAH, FAM111A, FAM20A, FAM58A/CCNQ, FAN1, FAT1, FGA, FGF20, FGF23, FGF8, FGF9, FLCN, FN1, FOXC1, FOXE1, FOXF1, FOXH1, FOXI1, FOXJ1, FRASI, FREM1, FREM2, FXYD2, G6PC, GALNT3, GANAB, GAPVD1, GAS2L2, GAS8/GAS11/DRC4, GATA3, GATA6, GATM, GBE1, GCM2, GDF1, GDNF, GFRA1, GJAI, GLA, GLI3, GLIS2/NPHP7, GLIS3, GNAI1, GNAS, GON7, GREB1L/KIAA1772/C18ORF6, GREM1, GRHRP, GRIP1, GSN, HAAO, HNF1A, HNF1B, HNF4A, HOGA1, HOXA13, HPRT1, HPSE2, HS2ST1, HSD11B2, HSD3B7, HTRA1, HYLS1, CHD1L, CHRM3, CHRNA3, ICK/CILK1, ICOS, IFIH1, IFT122, IFT140, IFT172, IFT27/BBS19, IFT43, IFT52, IFT74/CCDC2, IFT80, IFT81, IKZF1, IL21, ILK, IMPDH1, INF2/C14ORF173, INPP5E, INTU/KIAA1284, INVS/NPHP2, IQCB1/NPHP5, IRF2BP2, IRS4, ITGA3, ITGA8, ITGB4, ITSNI, KANK1, KANK2/KIAA1518, KANK4, KAT6B, KCNA1, KCNJ1, KCNJ10, KCNJ16, KCNJ5, KIAA0556/KATNIP, KIAA0586, KIAA0753, KIAA0974/FAM149B1, KIDINS220, KIF14, KIF3B, KIF7, KIRREL1, KL, KLHL3, KRIT1/CCM1, KSR2, KYNU, LAGE3, LAMA5, LAMB2, LAS1L, LCA5, LEFTY2, LEP, LEPR, LIFR, LIN7C, LMX1B, LRBA, LRIG2, LRP2BP, LRP4, LRP5, LRRC56, LRRC6/DNAAF11, LRRCC1, LYZ, LZTFL1/BBS17, MAGED2, MAGEL2, MAGI2/KIAA0705/AIP1, MAPKBP1, MC3R, MC4R, MCIDAS, MEN1, MKKS, MKS1, MMACHC, MMP21, MNS1, MOCOS, MS4A1, MUC1, MXRA5, MYH9, MYO1E, MYO5B, MYOCD, MYOG, MYRF, MYTIL, NADSYN1, NAT1, NCF1, NDN, NEK1, NEK10, NEK4, NEK8/NPHP9, NFKB1, NFKB2, NFU1, NKX2-1, NKX2-5, NME8, NNT, NODAL, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2/PDCN, NPNT, NR1H4, NR3C1, NR3C2, NRIP1, NTRK2, NUP107, NUP133, NUP160/NPHS19, NUP205, NUP37, NUP85, NUP93, OCRL, OFD1, OSGEP, PAX2, PAX8, PBX1, PCBD1, PCDH15, PCSK1, PCSK5, PDCD10, PDE2A, PDE3A, PDE6D, PDSS2, PGM2L1, PHEX, PHF6, PHIP, PIBF1, PIGA, PIGT, PIH1D3, PKD1, PKD1L1, PKD2, PKHD1, PLCE1, PLVAP, PLXND1, PMM2, PNPLA6, PODXL, POMC, PRAG1, PREPL, PRKCD, PRKCSH, PTH, PTPN22, PTPRO, RAB23, REN, RET, ROBO2, ROR2, RPE65, RPGR, RPGRIP1, RPGRIP1L/NPHP8, RRAGD, RSPH1, RSPH3, RSPH4A, RSPH9, SALL1, SALL4, SARS2, SCAPER, SCARB2, SCLT1, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SEC61A1, SEC63, SEMA3E, SFTPA1, SFTPB, SFTPC, SFTPD, SGPL1, SH2B1, SHROOM3, SIM1, SIX1, SIX2, SIX5, SLC12A1, SLC12A3, SLC1A1, SLC20A1, SLC22A12/URAT1, SLC25A13, SLC26A1, SLC26A4, SLC2A2, SLC2A9, SLC34A1, SLC34A3, SLC36A2, SLC36A4, SLC37A4, SLC3A1, SLC4A1, SLC4A4, SLC51A, SLC5A1, SLC5A2, SLC6A19, SLC6A20, SLC7A9, SLC9A3R1, SLIT2, SMAD4, SMARCAL1, SMO, SOX17, SPAG1, SPEF2, SPRY2, STK36, STRA6, SUFU, TAC3, TBC1D1, TBC1D32, TBC1D8B, TBLIX, TBX18, TCTEX1D2/DYNLT2B, TCTN1/TECT1, TCTN2, TCTN3, TFAP2A, TH, THBD, THRA, THRB, TJP2, TMEM107, TMEM127, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TMEM256, TMEM260, TMEM67, TNFRSF13B, TNFRSF13C, TNS2, TOGARAM1, TOPORS, TP53RK, TP63, TPO, TPP2, TPRKB, TRAF3IP1, TRAP1, TRAPPC9, TRHR, TRIM32, TRIM8, TRPC6, TRPM6, TRPV6, TSC1, TSC2, TSHB, TSHR, TTC12, TTC21B, TTC25, TTC8, TTF1, TUB, TXNDC15/C5orf14, ULK4, UMOD, UPK3A, USF2, VHL, VIPAS39, VPS33B, WBP11, WDPCP/C2ORF86, WDR19, WDR34/DYNC2I2, WDR35, WDR4, WDR60/DYNC2I1, WDR72, WDR73, WNK1, WNK4, WNT11, WNT4, WNT5A, WNT9B, WT1, XDH, XPNPEP3, XPO5, YRDC, ZIC3, ZMYM2, ZMYND10, ZNF423 (v4)</p> <p>Onemocnění a vady srdce</p> |

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| | <p>A2ML1, ABCA7, ABCC9, ABCG5, ABCG8, ACTA1, ACTA2, ACTC1, ACTN2, ACVR2B, ADAMTS2, ADAMTSL4, AEBP1, AKAP9, ALDH18A1, ALG10B, ALMS1, ALPK3, ANK2, ANKRD1, APOA1, APOA2, APOB, APOE, APP, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, B2M, B3GALT6, B4GALT7, BAG3, BGN, BMPR2, BRAF, CIR, CIS, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CALR3, CASQ2, CAV1, CAV3, CBL, CBS, CDH2, CFAP53/CCDC11, CFC1, CITED2, COL12A1, COL1A1, COL1A2, COL3A1, COL4A1, COL5A1, COL5A2, COX15, CRELD1, CRYAB, CSRP3, CST3, CTF1, CTNNA3, DES, DMD, DNAJC19, DOLK, DPP6, DSC2, DSE, DSG2, DSP, DTNA, EFEMP2/FBLN4, ELN, EMD, EMILIN1, EPG5, EVC, EVC2, EYA4, FBLN5, FBN1, FBN2, FGA, FHL1, FHL2, FHOD3, FKBP14, FKRP, FKTN, FLNA, FLNC, FOXE3, FOXF1, FOXH1, FREM1, GAA, GATA4, GATA5, GATA6, GATAD1, GDF1, GJA1, GJA5, GLA, GORAB, GPC3, GPD1L, GSN, GYG1, GYS1, HCN4, HFE, HRAS, CHST14, ILK, ITM2B, JAG1, JPH2, JUP, KAT6B, KCNA5, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, KLF10, KRAS, LAMA4, LAMP2, LDB3, LDLR, LDLRAP1, LEFTY2, LIPA, LMNA, LOX, LPL, LTBP1, LTBP2, LTBP3, LTBP4, LYZ, LZTR1, MAP2K1, MAP2K2, MAT2A, MED12, MFAP5, MIB1, MMP21, MNS1, MRAS, MURC, MYBPC3, MYH11, MYH6, MYH7, MYL2, MYL3, MYLK, MYLK2, MYO6, MYOM1, MYOZ2, MYPN, MYRF, NEBL, NEXN, NKX2-5, NKX2-6, NODAL, NOSIAP, NOTCH1, NPPA, NR2F2, NRAS, OSMR, P4HA1, PCSK9, PDLIM3, PGM1, PKD1L1, PKP2, PLN, PLOD1, PLXND1, PPA2, PPP1CB, PRDM16, PRDM5, PRDM6, PRKAG2, PRKARIA, PRKG1, PRNP, PSEN1, PSEN2, PTPN11, PYCR1, RAF1, RANGRF, RASA2, RBM20, RIN2, RIT1, ROBO4, RRAS, RRAS2, RSPH3, RYR2, SAA1, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCO2, SDHA, SGCD, SHOC2, SKI, SLC25A4, SLC2A10, SLC39A13, SLC8A1, SLMAP, SMAD2, SMAD3, SMAD4, SMAD6, SNTA1, SORL1, SOS1, SOS2, SPRED1, STAP1, STRN, SYNE2, TAB2, TAF1A, TAZ, TBX1, TBX20, TBX3, TBX5, TCAP, TECRL, TFAP2B, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD1, THSD4, TLL1, TMEM43, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TNXB, TOMM40, TPM1, TRDN, TREM2, TRPM4, TSPYL1, TTN, TTR, TXNRD2, VCL, ZFPM2, ZIC3, ZNF423, ZNF469 (v5)</p> <p>Mentální retardace a mikrocefalie</p> <p>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, ARCNI, ARFGEF2, ARHGEF6, ARHGEF9, ARID1A, ARID1B, ARID2, ARNT2, ARSE, ARVI, 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, DNMT3A, DONSON, DPAGT1, DPF2, DPM1, DPP6, DYNC1H1, DYNC1H2, 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, GDII, 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,</p> |

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| | <p><i>HUWE1, HYLS1, CHAMP1, CHD2, CHD3, CHMP1A, IARS1, IER3IP1, IFIH1, IFT122, IGBP1, IGF1, IGF1R, ILIRAPL1, 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, KNLI, KPTN, LICAM, 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, QRICHI 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, STXPB1, 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, TMC01, TMEM165, TMTC3, TMX2, TNIK, TOE1, TOP3A, TP53RK, TPRKB, TRAI, TRAPPC12, TRAPPC4, TRAPPC6B, TRAPPC9, TREX1, TRIM71, TRIO, TRIP12, TRMT1, TRMT10A, TRRAP, TSEN15, TSEN2, TSEN34, TSEN54, TSPAN7, TT12, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, TUBGCP2, TUBGCP4, TUBGCP6, TUSC3, UBA5, UBE2A, UBE3A, UBE3B, UBR1, UBR7, UBTF, UFC1, UFMI, 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 (v2)</i></p> <p>Onemocnění a vady očí</p> <p><i>AAAS, ABCA1, ABCA3, ABCA4, ABCB6, ABCC6, ABCD1, ABHD12, ABHD5, ACBD5, ACO2, ACTB, ACTG1, ADAM9, ADAMTS10, ADAMTS17, ADAMTS18, ADAMTSL4, ADGRA3/GPR125, ADGRV1/GPR98/USH2B, ADIPOR1, AFG3L2, AGBL1, AGBL5, AGK, AGPS, AH11, AHR, AIFM1, AIPL1, AKR1E2, ALDH18A1, ALDH1A3, ALDH3A2, ALG3, ALMS1, ALPK1, ALX1, AMACR, ANTXR1, AP3B1, AP3D1, APOE, APTX, ARHGEF18, ARL13B, ARL2BP, ARL3, ARL6, ARMC9, ARMS2, ARNT2, ARR3, ARSG/USH4, ASB10, ASRGL1, ATAD3A, ATF6, ATG7, ATOH7, ATP1A2, ATP1A3, ATP5F1A/ATP5A1, ATXN1, ATXN7, AUH, B3GALNT2, B3GLCT, B4GAT1/B3GNT1, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9/PTHB1, BCOR, BCS1L, BEST1, BFSP1, BFSP2, BLOC1S3, BLOC1S5, BLOC1S6, BMP4, BMP7, BRD4, BTB, C12orf57, C19ORF12, C1QBP, C1QTNF5, C2, C3, C9, CA4, CABP4, CACNA1F, CACNA2D4, CAPN15, CAPN5, CASK, CC2D2A, CCT2, CDH2, CDH23/USH1D, CDH3, CDHR1, CENPF, CEP104, CEP164, CEP250, CEP290/NPHP6, CEP41, CEP78, CERKL, CFAP410/C21orf2, CFAP418/C8orf37, CFB, CFH, CFHR1, CFHR3, CFI, CIB2/USH1J/KIP2, CISD2, CLCC1, CLDN19, CLN3, CLN5, CLN6, CLN8, CLPB, CLRN1/USH3A,</i></p> |

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| | <p>SOX1, SOX10, SOX2, SOX3, SPATA7, SPG7, SPP2, SPTLC1, SRD5A3, SSBP1, STAG2, STN1, STRA6, STS, STX3, SUCLA2, SUCLG1, SURF1, TACO1, TACSTD2, TAT, TAX1BP3, TBC1D20, TBX22, TCF4, TCOF1, TDRD7, TEAD1, TEK, TENM3, TFAP2A, TFG, TGFBI, TIMM8A, TIMP3, TINF2, TK2, TLCD3B/FAM57B, TLR4/ARMD10, TMEM126A, TMEM216, TMEM218, TMEM237, TMEM67, TMEM70, TMEM98, TMX3, TOP3A, TOPORS, TP63, TPK1, TPP1, TRAF3IP1, TRAPPC11, TREX1, TRIM32, TRIM37, TRIM44, TRMU, TRNT1, TRPM1, TSFM, TSPAN12, TTC19, TTC8, TTLL5, TTPA, TUB, TUBA3D, TUBA8, TUBB3, TUBB4B, TUBGCP4, TUBGCP6, TULP1, TWNK/C10orf2, TYMP, TYR, TYRP1, UBE3B, UBIAD1, UCHL1, UNC119, UNC45B, UQCRCQ, USH1C/PDZ73, USH1G/SANS, USH2A, USP45, VAX1, VCAN, VIM, VPS13B, VPS35L/C16orf62, VSX1, VSX2/CHX10, WASHC5/KIAA0196, WDPCP, WDR19/KIAA1638/NPHP13, WDR36, WDR45, WDR87, WFS1, WHRN/USH2D/DFNB31, WT1, XYLT2, YAP1, YME1L1, ZBTB20, ZEB1, ZFH4/ZFH4, ZFYVE26, ZNF408, ZNF423, ZNF469, ZNF513, ZNF644, ZNHIT3, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY (v6)</p> <p>Vzácná onemocnění</p> <p>A2ML1, AAAS, ABL1, ACAN, ACSL4, ACTB, ACTG1, ACVR1, ACVRL1, ADAMTS2, ADAMTS3, ADAMTS10, ADAMTS17, ADAMTSL2, ADAT3, ADGRG2, ADNP, AEBP1, AFF4, AGGF1, AIRE, AKR1C2, AKR1C4, AKT1, AKT2, AKT3, ALMS1, ALX1, ALX3, ALX4, AMER1, AMT, ANAPC1, ANGPT2, ANK1, ANKRD11, ANOS1, ANTXR2, AP1S1, AP1S2, APC2, AR, ARAP3, ARCNI, ARHGAP29, ARHGAP31, ARID1A, ARID1B, ARID2, ARSL, ARX, ASCC1, ASCL1, ASXL1, ASXL2, ASXL3, ATN1, ATP7B, ATR, ATRX, ATRIP, B3GALT/B3GLCT, B3GALT6, B4GALT7, B3GAT3, BANF1, BCOR, BHLHA9, BICD2, BICRA, BMP1, BMP2, BMP4, BMPR1B, BMPR2, BPTF, BRAF, BRF1, BRPF1, BRD4, BRWD3, BTB, BTK, BTRC, CIR, C1S, C2CD3, CALCRL, CANT1, CASR, CBL, CBX2, CC2D2A, CCBE1, CCDC8, CCDC22, CCDC32, CCDC47, CCDC88C, CCDC134, CCDC141, CCM2, CCND2, CCL2, CD96, CDC45, CDH1, CDH2, CDH3, CDH11, CDK13, CDK5, CDKL5, CDKN1C, CDON, CDT1, CDC6, CELSRI, CENPJ, CENPE, CENPF, CEP120, CEP152, CEP63, CEP85L, CFTR, CIBAR1, CIC, CILK1/ICK, CIT, CKAP2L, CLCN4, CLTC, CNOT1, CNOT3, CNTNAP2, COG4, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL10A1, COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, COL12A1, COLEC10, COLEC11, COMP, COQ6, CP, CPA1, CPE, CPLANE1/C5orf42, CREB3L1, CREBBP, CRIPT, CRTAP, CSNK2A1, CTNND1, CTCF, CTCRC, CTU2, CUL4B, CUL7, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP26B1, DCAF17, DCC, DCPS, DCX, DCHS1, DDR2, DDX11, DDX58/RIGI, DDX59, DHCR7, DHCR24, DHH, DHODH, DHX37, DIS3L2, DISP1, DLG4, DLL1, DLL4, DLX3, DLX4, DLX5, DLX6, DNA2, DNM2, DNMT3A, DOCK6, DOK7, DONSON, DPF2, DPYSL5, DSE, DUSP6, DVLI, DVL3, DYNC1H1, DYNC2H1, DYNC2LI1, DYRK1A, EBF3, EBP, ECE1, ECEL1, EDN1, EDN3, EDNRA, EDNRB, EED, EEF1A2, EFNBI, EFTUD2, EHMT1, EIF2AK4, EIF2S3, EIF4A3, EML1, ENG, EOGT, EP300, EPB42, EPG5, EPHB4, EPS15L1, ERCC1, ERCC2, ERCC5, ERCC6, ERCC8, ERF, ESCO2, EVC, EVC2, EXOC6B, EXT1, EXT2, EYA1, EZH2, FAM20C, FAM50A, FAT4, FBLN1, FBN1, FBN2, FEZF1, FGD1, FGF10, FGF16, FGF17, FGF8, FGF9, FGFRI, FGFR2, FGFR3, FH, FIG4, FKBP10, FKBP14, FLNA, FLNB, FLRT3, FLT4, FLVCR2, FOXC2, FOXE1, FOXG1, FOXH1, FOXPI, FRAS1, FREM1, FREM2, FRMD4A, FSHB, FTH1, FUZ, FZD2, GABBR2, GAS1, GATA2, GATAD2B, GATA4, GDF11, GDF2, GDF3, GDF5, GDF6, GDII, GDNF, GH1, GHR, GHRH, GHRHR, GHSR, GJA1, GJB2, GJC2, GLDC, GLE1, GLII, GLI2, GLI3, GLMN, GLYCTK, GMNN, GNAI3, GNAS, GNB1, GNRH1, GNRHR, GPC3, GPC4, GPC6, GPT2, GON7, GRB10, GRHL3, GRIA4, GRIPI, GSX2, GZF1, HAMP, HDAC4, HDAC6, HDAC8, HECW2, HELLS/SMARCA6, HESX1, HFE, HHAT, HIST1H1E, HIVEP2, HJV, HMGA2, HOXA13, HOXD13, HPRT1, HRAS, HS6ST1, HSD17B3, HSD3B2, HSPA9, HYAL2, HYL1, CHAMP1, CHD3, CHD4, CHD7, CHD8, CHNG, CHST3, CHST14, CHSY1, IARS1, IFIH1, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT52, IFT57, IFT80, IGF1, IGF1R, IGF2, IGFALS, IHH, IL17RD, IMPAD1/BPNT2, INSR, INTU, IQCE, IQSEC2, IRF6, JAG1, JAM2, KANSL1, KAT6A, KAT6B, KATNB1, KCNH1, KCNJ2, KCTD1,</p> |

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| | <p>CHEK2, IDH1, IDH2, KIF1B, KIT, KLLN, KMT2A, KRAS, MAP2K1, MAX, MC1R, MEN1, MET, MITF, MLH1, MLH3, MN1, MRE11, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2, NRAS, NRG1, NTHL1, PALB2, PALLD, PARP1, PARP2, PARP3, PARP4, PBRM1, PDGFB, PDGFD, PDGFRA, PDGFRB, PHOX2B, PIK3CA, PMS2, POLD1, POLE, POT1, PPP2R2A, PRKARIA, PTEN, PTCH1, PTCH2, RABL3, RAD50, RAD51, RAD51B, RAD51C, RAD51D, RAD52, RAD54L, RB1, RECQL, RECQL4, RET, REXO2, RNF43, RPS20, SDHA, SDHAF2, SDHB, SDHC, SDHD, SH2D1A, SH2D2A, SLC25A11, SLX4, SMAD4, SMARCA4, SMARCB1, SMARCE1, SRGAP1, SRY, STK11, SUFU, TERF2IP, TERT, TET2, TMEM127, TP53, TSC1, TSC2, VHL, WRN, WT1, XPA, XRCC2, XRCC3 (hg38). Somatické testování: ABRAXAS1, ACVR1, ALK, ARAF, ARID1A, ARID1B, ATRX, BRK1, CCDC153, CCNE1, CD79B, CDADC1, CDK12, CDK6, CDKN2B, COX6C, CTNNA1, CTNNB1, DDR2, DPYD, EIF1AX, ENG, ENOX1, ERG, ESR1, EZHIP, FAM124A, FANCD2OS, FGFR1, FGFR2, FGFR3, FOXL2, GEN1, HDAC2, H3-C14 (H3), H3C2 (HIST1H3B), H3C3, H3-3A, IL18, INA, INCA1, KEAP1, KHDC3L, KIAA1549, MAD2L2, MAMLD1, MALT1, MTOR, MYB, MYBL1, MYC, MYCN, MYD88, MYH13, NAT1, NLRP7, NTRK1, NTRK2, NTRK3, NUTM1, PABPC1, PABPC3, PATZ1, PAX8, PIK3R1, PML, PPARG, PPM1D, PPP2R1A, QKI, RAB18, RAF1, RELA, RHOA, ROS1, RPAIN, SETD2, SIPR1, TACCI1, TACC3, TBX21, TCEB1 (ELOC), TFEB, TFE3, THADA, TMPRSS2, TOE1, TOP2A, TOP6BL (C11orf80), WSCD1, YAP1, ZFTA (C11orf95) (hg38). Hereditární pankreatitida: CFTR, CTRC, CPA1, SPINK1, PRSS1 (hg38). Lymfoproliferace: AICDA, AKT3, ASXL1, B2M, BAX, BCL2, BCL3, BCL6, BCL10, BIRC3, BLNK, BTG1, BTK, CFBF, CCDC50, CCND2, CCND3, CCR4, CCR7, CD28, CD44, CD58, CD70, CD79A, CD274, CDC25A, CIITA, CREBBP, CTLA4, CXCR4, DAPK1, DDX3X, DEK, DUSP22, E2F2, EBF1, EGR2, EP300, EZH2, FLT3, FOXO1, FOXP1, FYN, GNA13, H1-2, H1-4, CHD2, CHIC2, ID3, IKKB, IKZF1, IL16, INO80, IRF4, IRF8, ITPKB, JAK1, JAK3, KDM6A, KLF2, KLHL6, KMT2C, KMT2D, LMO2, MAP3K14, MCL1, MEF2B, MGA, MLF1, MLLT10, MME, NFKB2, NFKBIE, NOTCH1, NOTCH2, NSD2, P2RY8, PAX5, PDCD1, PDCD1LG2, PIK3CD, PIM1, PIM2, PLCG1, PLCG2, PRDM1, PRDM16, PRKCB, PTPN1, PTPN11, PTPRD, RAG1, RAG2, REL, RELN, ROCK1, RPS15, SIPR2, SAMHD1, SERPINA9, SF3B1, SGK1, SH2B3, SOCS1, STAT3, STAT5B, STAT6, TBC1D9, TCF3, TLN2, TNFAIP3, TNFRSF13B, TNFRSF14, TNFSF4, TP63, TRAF2, UBE2A, UBR5, XPO1, ZMYM3 (hg38) (v12)</p> <p>Somatické varianty v nádorových tkáních II</p> <p>A2M, ABCC9, ABL1, ABL2, ABRAXAS1, ACBD5, ACD, ACP5, ACTB, ACVR1, ACVR1B, ACVR2A, ADAMTS14, ADGRA2, AFAP1L2, AFF1, AFF3, AGTRAP, AHRR, AICDA, AIP, AKAP9, AKT1, AKT2, AKT3, ALK, ALOX12B, AMER1, AMN, ANKRD11, ANKRD26, ANTXR1, ANTXR2, ANXA11, APC, AR, ARAF, ARFRP1, ARHGEF2, ARID1A, ARID1B, ARID2, ARID5B, ARMC10, ASB13, ASPSCR1, ASXL1, ASXL2, ATF1, ATF5, ATIC, ATM, ATP1B1, ATR, ATRX, AURKA, AURKB, AXINI, AXIN2, AXL, B2M, BAG4, BAP1, BARD1, BATF3, BAX, BBC3, BCAM, BCL2, BCL2L1, BCL2L2, BCL2L11, BCL3, BCL6, BCL10, BCOR, BCORL1, BCR, BEST3, BIRC3, BIRC6, BLM, BLNK, BMF, BMP4, BMP7, BMPR1A, BRAF, BRCA1, BRCA2, BRD4, BRD8, BRF1, BRIP1, BTG1, BTK, BUB1, BUB1B, CAD, CALR, CAMK2A, CAMTA1, CARD11, CARS1, CASP8, CASZ1, CATSPERZ, CFBF, CBL, CC2D1A, CCDC6, CCDC50, CCN6, CCNA2, CCNB1, CCNB2, CCNB3, CCND1, CCND2, CCND3, CCNE1, CCR4, CCR7, CD28, CD34, CD44, CD58, CD70, CD74, CD79A, CD79B, CD274, CD276, CDADC1, CDC6, CDC25A, CDC73, CDH1, CDH11, CDK1, CDK4, CDK6, CDK8, CDK12, CDKNIA, CDKN1B, CDKN1C, CDKN2A, CDKN2B, CDKN2C, CDKN2D, CDX1, CDX2, CENPA, CEP43, CEP85L, CIC, CIITA, CITED2, CLCA2, CLCN6, CLIP1, CLTC, CNBP, CNTN1, COG8, COL1A1, COL1A2, COL25A1, COL3A1, COL17A1, COP1, CPM, CREB1, CREB3L1, CREB3L2, CREB3L3, CREBBP, CREM, CRKL, CRLF2, CRTCI1, CRTCI3, CSF1R, CSF3R, CSGALNACT2, CSNK1A1, CTCF, CTLA4, CTNNA1, CTNNB1, CUBN, CUL3, CUX1, CXCR4, CYB5R2, CYLD, CYP1A1, CYP3A4, CYSLTR2, DAPK1, DAXX, DCTN1, DCUN1D1, DDIT3, DDR2, DDX3X, DDX5, DDX41, DEK, DENND3, DERA, DFFA, DHX15, DHX57, DICER1, DIS3, DLL1, DLST, DNAJB1, DNMT1, DNMT3A, DNMT3B, DNMT3C, DNMT3L, DNMT3D, DNMT3E, DNMT3F, DNMT3G, DNMT3H, DNMT3I, DNMT3J, DNMT3K, DNMT3L, DNMT3M, DNMT3N, DNMT3O, DNMT3P, DNMT3Q, DNMT3R, DNMT3S, DNMT3T, DNMT3U, DNMT3V, DNMT3W, DNMT3X, DNMT3Y, DNMT3Z, DNMT3AA, DNMT3AB, DNMT3AC, DNMT3AD, DNMT3AE, DNMT3AF, DNMT3AG, DNMT3AH, DNMT3AI, DNMT3AJ, DNMT3AK, DNMT3AL, DNMT3AM, DNMT3AN, DNMT3AO, DNMT3AP, DNMT3AQ, DNMT3AR, DNMT3AS, DNMT3AT, DNMT3AU, DNMT3AV, DNMT3AW, DNMT3AX, DNMT3AY, DNMT3AZ, DNMT3BA, DNMT3BB, DNMT3BC, DNMT3BD, DNMT3BE, DNMT3BF, DNMT3BG, DNMT3BH, DNMT3BI, DNMT3BJ, DNMT3BK, DNMT3BL, DNMT3BM, DNMT3BN, 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DNMT3FV, DNMT3FW, DNMT3FX, DNMT3FY, DNMT3FZ, DNMT3GA, DNMT3GB, DNMT3GC, DNMT3GD, DNMT3GE, DNMT3GF, DNMT3GG, DNMT3GH, DNMT3GI, DNMT3GJ, DNMT3GK, DNMT3GL, DNMT3GM, DNMT3GN, DNMT3GO, DNMT3GP, DNMT3GQ, DNMT3GR, DNMT3GS, DNMT3GT, DNMT3GU, DNMT3GV, DNMT3GW, DNMT3GX, DNMT3GY, DNMT3GZ, DNMT3HA, DNMT3HB, DNMT3HC, DNMT3HD, DNMT3HE, DNMT3HF, DNMT3HG, DNMT3HH, DNMT3HI, DNMT3HJ, DNMT3HK, DNMT3HL, DNMT3HM, DNMT3HN, DNMT3HO, DNMT3HP, DNMT3HQ, DNMT3HR, DNMT3HS, DNMT3HT, DNMT3HU, DNMT3HV, DNMT3HW, DNMT3HX, DNMT3HY, DNMT3HZ, DNMT3IA, DNMT3IB, DNMT3IC, DNMT3ID, DNMT3IE, DNMT3IF, DNMT3IG, DNMT3IH, DNMT3II, DNMT3IJ, DNMT3IK, DNMT3IL, DNMT3IM, DNMT3IN, DNMT3IO, DNMT3IP, DNMT3IQ, DNMT3IR, DNMT3IS, DNMT3IT, DNMT3IU, DNMT3IV, DNMT3IW, DNMT3IX, DNMT3IY, DNMT3IZ, DNMT3JA, DNMT3JB, DNMT3JC, DNMT3JD, DNMT3JE, DNMT3JF, DNMT3JG, DNMT3JH, DNMT3JI, DNMT3JJ, DNMT3JK, DNMT3JL, DNMT3JM, DNMT3JN, DNMT3JO, DNMT3JP, DNMT3JQ, DNMT3JR, DNMT3JS, DNMT3JT, DNMT3JU, DNMT3JV, DNMT3JW, DNMT3JX, DNMT3JY, DNMT3JZ, DNMT3KA, DNMT3KB, 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DNMT3WZ, DNMT3XA, DNMT3XB, DNMT3XC, DNMT3XD, DNMT3XE, DNMT3XF, DNMT3XG, DNMT3XH, DNMT3XI, DNMT3XJ, DNMT3XK, DNMT3XL, DNMT3XM, DNMT3XN, DNMT3XO, DNMT3XP, DNMT3XQ, DNMT3XR, DNMT3XS, DNMT3XT, DNMT3XU, DNMT3XV, DNMT3XW, DNMT3XX, DNMT3XY, DNMT3XZ, DNMT3YA, DNMT3YB, DNMT3YC, DNMT3YD, DNMT3YE, DNMT3YF, DNMT3YG, DNMT3YH, DNMT3YI, DNMT3YJ, DNMT3YK, DNMT3YL, DNMT3YM, DNMT3YN, DNMT3YO, DNMT3YP, DNMT3YQ, DNMT3YR, DNMT3YS, DNMT3YT, DNMT3YU, DNMT3YV, DNMT3YW, DNMT3YX, DNMT3YY, DNMT3YZ, DNMT3ZA, DNMT3ZB, DNMT3ZC, DNMT3ZD, DNMT3ZE, DNMT3ZF, DNMT3ZG, DNMT3ZH, DNMT3ZI, DNMT3ZJ, DNMT3ZK, DNMT3ZL, DNMT3ZM, DNMT3ZN, DNMT3ZO, DNMT3ZP, DNMT3ZQ, DNMT3ZR, DNMT3ZS, DNMT3ZT, DNMT3ZU, DNMT3ZV, DNMT3ZW, DNMT3ZX, DNMT3ZY, DNMT3ZZ</p> |

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| | <p><i>EPC1, EPCAM, EPHA3, EPHA5, EPHA7, EPHB1, ERBB2, ERBB3, ERBB4, ERC1, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERG, ERRF11, ESR1, ESRP1, ESRRA, ETS1, ETV1, ETV4, ETV5, ETV6, EWSR1, EXOC2, EXT1, EXT2, EZH2, EZHIP, EZR, FAM124A, FAM131B, FAM216A, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FAT1, FBXO11, FBXW7, FEN1, FEV, FGF1, FGF2, FGF3, FGF4, FGF5, FGF6, FGF7, FGF8, FGF9, FGF10, FGF14, FGF19, FGF23, FGFR1, FGFR2, FGFR3, FGFR4, FH, FLCN, FLI1, FLT1, FLT3, FLT4, FN1, FOS, FOSB, FOXA1, FOXB2, FOXE1, FOXL2, FOXO1, FOXO4, FOXO1, FRS2, FUBP1, FUS, FUT8, FYN, GABRA6, GAPDH, GATA1, GATA2, GATA3, GATA4, GATA6, GCC2, GCGR, GEN1, GFPT1, GID4, GLI1, GLI2, GLMN, GLRX5, GLYR1, GNA11, GNA13, GNAI1, GNAQ, GNAS, GOLGA5, GOPC, GPS2, GREB1, GREM1, GRIN2A, GRM3, GSK3B, GTF2E1, GTF3C2, H1-2, H1-4, H2BC5, H3-3A, H3-3B, H3-4, H3-5, H3C1, H3C2, H3C3, H3C4, H3C6, H3C7, H3C8, H3C10, H3C11, H3C12, H3C13, H4C9, HAS2, HDAC2, HDAC9, HEY1, HGF, HIP1, HLA-A, HMGA2, HMGN2P46, HNF1A, HNRNPA1, HNRNPK, HOOK3, HOXB13, HRAS, HSD3B1, HSD17B2, HSP90AA1, CHD2, CHD4, CHEK1, CHEK2, CHIC2, CHN1, CHTOP, ICOSLG, ID3, IDH1, IDH2, IFNGR1, IFNL3, IGF1, IGF1R, IGF2, IGFBP5, IGSF11, IKBKB, IKBKE, IKZF1, IL1R1, IL7R, IL10, IL12RB1, IL16, IL18, IL21R, INCA1, ING1, INHA, INHBA, INO80, INPP4A, INPP4B, INSL3, INSM1, INSR, IRAG2, IRF2, IRF2BP2, IRF4, IRF8, IRS1, IRS2, IRX2, ITPKB, ITPR2, JAK1, JAK2, JAK3, JAZF1, JUN, KANSL1, KAT6A, KAT6B, KCNMB4, KCNQ1, KDM2A, KDM4D, KDM5A, KDM5B, KDM5C, KDM6A, KDM6B, KDR, KEAP1, KEL, KHDC3L, KIAA1217, KIAA1549, KIF1B, KIF5B, KIF13B, KIF15, KIT, KLC1, KLF2, KLF4, KLF17, KLHL6, KLK2, KLLN, KMT2A, KMT2B, KMT2C, KMT2D, KRAS, KRT8, KRT19, KTN1, L2HGDH, LAMC1, LAMP1, LATS1, LATS2, LCPI, LEMD3, LEUTX, LIMD1, LIN28B, LMNA, LMO1, LMO2, LMO7, LPP, LRIG3, LRP1, LRP1B, LYN, LZTR1, LZTS1, MAD2L1, MAD2L2, MAGEC2, MAGI2, MAL, MALT1, MAML2, MAML3, MAP2K1, MAP2K2, MAP2K4, MAP3K1, MAP3K4, MAP3K5, MAP3K13, MAP3K14, MAPK1, MAPK3, MAX, MBTD1, MC1R, MCL1, MDC1, MDH2, MDM2, MDM4, MEAF6, MED12, MEF2B, MEN1, MET, MGA, MID1, MINK1, MTF, MKRN1, MLF1, MLH1, MLH3, MLLT3, MLLT10, MME, MN1, MPL, MPRIP, MRAS, MRE11, MRTFA, MSH2, MSH3, MSH6, MSMB, MSN, MSR1, MST1, MST1R, MSX1, MTAP, MTOR, MUC1, MUTYH, MYB, MYBL1, MYC, MYCL, MYCN, MYD88, MYH9, MYH13, MYO5A, MYOD1, N4BP1, NAB2, NACC2, NAT1, NAV3, NBN, NCOA1, NCOA2, NCOA3, NCOA4, NCOR1, NEGR1, NEK6, NF1, NF2, NFATC2, NFE2L2, NFIB, NFKB1, NFKB2, NFKBIA, NFKBIE, NKAP, NKX2-1, NKX3-1, NLRP6, NLRP7, NME1, NOTCH1, NOTCH2, NOTCH3, NOTCH4, NPM1, NR4A2, NR4A3, NRAS, NRG1, NSD1, NSD2, NTHL1, NTRK1, NTRK2, NTRK3, NUP93, NUP160, NUTM1, NUTM2A, NUTM2B, OMD, OR51E2, P2RY8, PAFAH1B1, PAICS, PAK1, PAK3, PAK5, PALB2, PARP1, PARP2, PARP3, PARP4, PATZ1, PAX3, PAX5, PAX7, PAX8, PBRM1, PBX1, PBX3, PCLAF, PCMI, PCNA, PDCD1, PDCD1LG2, PDE11A, PDGFB, PDGFD, PDGFRA, PDGFRB, PDK1, PDPK1, PGR, PHF1, PHF6, PHLDA1, PHLDB1, PHOX2B, PIK3C2B, PIK3C2G, PIK3C3, PIK3CA, PIK3CB, PIK3CD, PIK3CG, PIK3R1, PIK3R2, PIK3R3, PIM1, PIM2, PINK1, PLA2R1, PLAG1, PLCB4, PLCG1, PLCG2, PLK2, PLXNA2, PMAIP1, PML, PMS1, PMS2, PNRC1, POLD1, POLE, POLE2, POLH, PORCN, POT1, POU2AF1, POU5F1, PPAR, PPARG, PPAT, PPFIBP1, PPFIBP2, PPL, PPM1D, PPP1CB, PPP1R15A, PPP2R1A, PPP2R1B, PPP2R2A, PPP6C, PRCC, PRDM1, PRDM10, PRDM16, PREX2, PRKARIA, PRKAR2B, PRKCB, PRKCI, PRKCZ, PRKDC, PRKG2, PRKN, PRRT2, PRSS8, PTEN, PTH1R, PTCH1, PTPN1, PTPN3, PTPN11, PTPN12, PTPRD, PTPRJ, PTPRK, PTPRS, PTPRT, PWWP2A, PYCRI, QKI, RAB2A, RAB18, RAB29, RAB35, RAC1, RAD21, RAD50, RAD51, RAD51B, RAD51C, RAD51D, RAD52, RAD54L, RAF1, RAG1, RAG2, RANBP1, RANBP2, RARA, RASA1, RBL1, RBM10, RCSD1, RECQL, RECQL4, REL, RELB, RELN, RET, REXO2, RHBDF2, RHEB, RHOA, RICTOR, RIMS2, RIT1, RNF43, RNF130, RNF213, ROCK1, ROS1, RPAIN, RPS6KA4, RPS6KB1, RPS6KB2, RPS15, RPS20, RPTOR, RRAS2, RRBP1, RSPO2, RSPO3, RTEL1, RUNX1, RUNX1T1, RUNX2, RYBP, S1PR1, S1PR2, SALL4, SAMHD1, SDC4, SDHA, SDHAF2, SDHB, SDHC, SDHD, SEC31A, SEPTIN7, SERPINA9, SERPINE1, SETBP1, SETD2, SEZ6L2, SF3B1, SGK1, SH2B3, SH2D1A, SH3BP2, SH3BP5, SHOC2, SHPRH, SHQ1, SHTN1, SLC3A2, SLC5A7, SLC9A8, SLC25A11, SLC34A2, SLC43A3, SLC45A3, SLC01C1, SLFN11, SLIT2, SLX4, SMAD2, SMAD3, SMAD4, SMARCA2, SMARCA4, SMARCA5, SMARCB1, SMARCC1, SMARCC2, SMARCD1,</i></p> |

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| | <p><i>SMARCE1, SMC1A, SMC3, SMO, SNCAIP, SNRNP25, SOCS1, SOS1, SOS2, SOX2, SOX4, SOX9, SOX10, SOX17, SP3, SP140, SPEN, SPOP, SPP1, SPRED1, SPTA1, SPTBN1, SQSTM1, SRC, SRF, SRGAP3, SRSF2, SRY, SS18, SS18L1, SSX1, SSX2, SSX2B, SSX4, STAG1, STAG2, STAR, STARD13, STAT3, STAT4, STAT5B, STAT6, STIL, STIP1, STK11, STK40, STRBP, STRN, SUFU, SUZ12, SYK, TACC3, TAF1, TAF4, TAF15, TANK, TBC1D9, TBX3, TBX21, TBXT, TCF3, TCF7L2, TCF12, TEK, TENT5C, TERC, TERF2IP, TERT, TET1, TET2, TFE3, TFEB, TFG, TERC, TGFBR1, TGFBR2, THBS1, THRAP3, TLN2, TLR2, TMC6, TMC8, TMED6, TMEM106B, TMEM127, TMEM178A, TMPRSS2, TNFAIP3, TNFRSF11A, TNFRSF13B, TNFRSF14, TNFSF4, TNNI2, TOE1, TOGARAM2, TOP1, TOP2A, TOP6BL, TP53, TP63, TPD52L1, TPM3, TPM4, TPR, TPRA1, TRAF1, TRAF2, TRAF3, TRAF7, TRIM11, TRIM24, TRIM27, TRIM33, TSC1, TSC2, TSHR, TSPAN31, TTC23, U2AF1, UBE2A, UBR5, UBTF, UEVLD, USP6, USP15, VAMP2, VCL, VEGFA, VEGFB, VGLL2, VHL, VMP1, VTCN1, WDCP, WDFY2, WRN, WSCD1, WT1, WWTR1, XIAP, XPA, XPO1, XRCC2, XRCC3, YAP1, YEATS4, YES1, YWHAE, ZBTB2, ZBTB7A, ZBTB16, ZC3H7A, ZC3H7B, ZCCHC8, ZFHX3, ZFP36, ZFTA, ZMYM3, ZNF217, ZNF444, ZNF703, ZRSR2 (v2)</i></p> |
| 816/10 | <p>Čipy: Sure Print G3 CGH ISCA v2, 8x60K, HumanCytoSNP-850K v1.3 DNA Analysis Beadchip Kit</p> |
| 816/11 | <p>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 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, 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, P013 ATRX, P017 MEN1, P027 Uveal melanoma, P029 WBS, P032 FANCA mix 2,</p> |

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| | <p>P040 CLL, P049 SLC6A8-ABCD1, L1CAM, P055 PAH, P056 TP53, P071 LMNB1-PLP1-NOTCH3, P073 IKBKG, P074 AR, P088 Oligodendroglioma, P093 HHT/HPAH, P101 STK11, P113 FANCB, P116 SGC, P137 SCN1A, P158 JPS, P164 IDS, P183 EDA-EDAR-EDARADD, P188 22q13, P223 PHEX, P253 NB mix3, P258 SMARCB1, P271 COL1A1, P275 MAPT-GRN, P305 AGXT, P315 EGFR, P316 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 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, ME033 TNDM, ME042 CIMP, P035 DMD-2, P044 NF2, P120 PANK2/PLA2G6, P124 TSC1, P125 Mitochondrial DNA, P148 TGFBR1-TGFBR2, P215 EXT, P262 GHI, P329 CRLF2-CSF2RA-IL3RA, P432 MYH9, P437 Familiar MDS-AML, P474 CD274-PDCD1LG2-JAK2, P484 ALPL, P492 POLD1 -POLE, P011 VWF mix 1, P242 Pancreatitis, P257 TERT-DKC1, P280 SLC26A4</p> <p>Digitální MLPA: D001 Hereditary Cancer Panel 1</p> |
| 816/12 | <p>Probemixy: ME011 Mismatch Repair genes, P056 TP53, ME012 MGMT-IDH-TERT, P002 BRCA1, P045 BRCA2/CHEK2, P003 MLH1/MSH2, P008 PMS2, P043 APC, P072 MSH6-MUTYH probemix, P190 CHEK2 probemix, P240 BRIP-CHEK1, P260 PALB2-RAD50-RAD51C-RAD51D, P298 BRAF-HRAS-KRAS-NRAS, P445 KDM6A, P483 HER gene family, P315 EGFR, P294 Tumour Loss, P258 SMARCB1, P308 MET, P377 Hematologic Malignancies, P419 CDKN2A/2B-CDK4</p> <p>Digitální MLPA: D001 Hereditary Cancer Panel 1</p> |
| 816/13 | <p><i>HFE</i> (sekvenční varianty c.187C>G, c.193A>T, c.845G>A) Faktor V Leiden (sekvenční varianta c.1691G>A v genu <i>F5</i>) Faktor II Prothrombin (sekvenční varianta c.20210G>A v genu <i>F2</i>) <i>TPMT</i> (sekvenční varianty c.238G>C, c.460G>A, c.719A>G) Deficit alfa 1-antitrypsinu (sekvenční varianty c.863A>T, c.1096G>A v genu <i>SERPINA1</i>) <i>DPYD</i> (sekvenční varianty: c.1905+1G>A, c.1679T>G, c.1236G>A, c.2846A>T) Gilbertův syndrom (vyšetření alelických variant 5TA, 6TA, 7TA, 8TA v promotoru genu <i>UGT1A1</i>) Chimerismus: vysoce polymorfní delece a inzerce za účelem typizace a monitoringu dárce a příjemce hematopoetických buněk <i>CYP2C19</i> (sekvenční varianty c.681G>A, c.636G>A, c.-806C>T)</p> |
| 816/14 | <p>Sekvenční varianty genů <i>KRAS</i>, <i>NRAS</i>, <i>BRAF</i> a <i>EGFR</i>: Idylla™: <i>KRAS</i> (sekvenční varianty: c.34G>T, c.34G>C, c.34G>A, c.35G>C, c.35G>A, c.35G>T, c.38G>A, c.176C>A, c.176C>G, c.175G>A, c.181C>A, c.180_181delinsAA, c.182A>T, c.182A>G, c.183A>C, c.183A>T, c.351A>C, c.351A>T, c.436G>C, c.436G>A, c.437C>T) <i>NRAS</i> (sekvenční varianty: c.34G>T, c.34G>A, c.35G>A, c.35G>C, c.35G>T, c.38G>A, c.38G>T, c.37G>C, c.175G>A, c.181C>A, c.182A>T, c.182A>G, c.183A>C, c.183A>T, c.351G>C, c.351G>T, c.436G>A, c.437C>T) <i>BRAF</i> (sekvenční varianty: c.1799T>A, c.1799_1800delinsAA, c.1799_1800delinsAC, c.1799_1800delinsAT, c.1798_1799delinsAA, c.1798_1799delinsAG, c.1798G>A) <i>EGFR</i> (sekvenční varianty: c.2156G>C, c.2155G>T, c.2154_2155delinsTT, c.2155G>A, c.2238_2248delinsGC, c.2239_2248delinsC, c.2240_2248del, c.2239_2247del, c.2239_2251delinsC, c.2240_2251del, c.2235_2249del, c.2236_2250del, c.2239_2253del, c.2240_2254del, c.2238_2252del, c.2237_2251del, c.2235_2252delinsAAT, c.2237_2252delinsT, c.2234_2248del, c.2236_2253delinsCTA, c.2237_2253delinsTA, c.2235_2251delinsAG, c.2236_2253delinsCAA, c.2230_2249delinsGTCAA, c.2240_2257del, c.2237_2255delinsT, c.2239_2256del, c.2236_2253del, c.2239_2258delinsCA, c.2237_2254del, c.2238_2255del, c.2237_2257delinsTCT, c.2236_2255delinsAT, c.2236_2256delinsATC, c.2237_2256delinsTT, c.2237_2256delinsTC, c.2235_2255delinsGGT, c.2238_2258del, c.2236_2256del, c.2253_2276del, c.2369C>T, c.2303G>T, c.2310_2311insGGT,</p> |

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| | <p>c.2308_2309insGCCAGCGTG, c.2308_2311delinsCCAGCGTGGAT, c.2311_2312insGCGTGGACA, c. 2319_2320insCAC, c.2573T>G, c.2573_2574delinsGT, c.2573_2574delinsGA, c.2582T>A)</p> <p>EasyPGX: <i>KRAS</i> (sekvenční varianty: c.34G>T, c.34G>C, c.34G>A, c.35G>C, c.35G>A, c.35G>T, c.38G>A, c.176C>A, c.176C>G, c.175G>A, c.181C>A, c.182A>T, c.182A>G, c.183A>C, c.183A>T, c.349A>G, c.350A>G, c.351A>C, c.351A>T, c.436G>C, c.436G>A, c.437C>T) <i>NRAS</i> (sekvenční varianty: c.34G>T, c.34G>A, c.35G>A, c.35G>C, c.35G>T, c.38G>A, c.38G>T, c.37G>C, c.175G>A, c.176C>T, c.181C>A, c.182A>T, c.182A>G, c.183A>C, c.183A>T, c.350A>G, c.351G>C, c.351G>T, c.436G>A, c.437C>T) <i>BRAF</i> (sekvenční varianty: c.1799T>A, c.1799_1800TG>AA, c.1799_1800TG>AT, c.1798_1799GT>AA, c.1798_1799GT>AG)</p> |
| 823/4 | <p>HSA, AFP, ALK, ALK-D5F3, AMACR, AMYLOID-A, ANNEX, AR, ARG, ATRX, BAP1, B-CAT, BCL2, BCL6, BER, BOB1, BRACHY, C1q, C3, C3c, C4d, CA9, CAL, CALDE, CALP, CALRE, CD10, CD19, CD117, CD138, CD15, CD163, CD1a, CD20, CD21, CD23, CD3, CD30, CD31, CD34, CD4, CD43, CD45LCA, CD45RO, CD5, CD56, CD61, CD68, CD71, CD79a, CD8, CD99, CDX2, CEA, CK, CKHMW, CD14, CK17, CK18, CK19, CK20, CK5/6, CK7, CK8, c-MYC, CY-D1, D2, DES, DOG1, E-CAD, EMA, ER, ERG, Fb, FOLR1, GATA3, GFAP, GLY-C, GPC3, GRAN-B, HBME1, HCG, HEP, HER2neu, HHV8, HMB45, HP, H3K27me3, H3K27M, CHROM, IDH, IgA, IgG, IgG4, IgM, INH, INI-1, INSM1, KAPPA, Ki67, LAMBDA, LANG, LEF, LYS, MELAN, MIA, MLH1, MPO, MSH2, MSH6, MUC1, MUC2, MUC4, MUC5, MUC6, MUM1, MyoD1, NAP, NKX3.1, NSE, OCT2, OCT3/4, OLIG2, p120, p16, p21, p40, p53, p57, p63, PanTRK, PAX5, PAX8, PD-L1(22C3), PER, PHH3, PALP, PMS2, PR, PRAME, PREALBUMIN, PSA, P-SAP, RCC, ROS1, SALL4, SATB2, S100, SOX-2, SOX10, SOX11, STAT6, SV40, SYN, SSTR2, TDT, THYR, TTF1, VIM, WT-1</p> |
| 823/5 | <p>1p36/1q25, 19q13/19p13, 1p/12, ALK, ALK/EML4, ATM/CEN12, BCL2, BCL2/IGH, BCL6, BCOR, BIRC3/MAL1, CCND1, CCND1/IGH, CDKN2A/CEN9, CDK4/CEN12, CIC, COL1A1, DDIT3, EGFR/CEN7, ERG, ETV6, EWSR1, EWSR1/FLI1, EGFR1, FOXO1, FUS, HER2/CEN17, IGH, IRF4/DUSP22, JAZF1, MALT1, MAML2, MDM2/CEN12, MET/CEN7, MYC, MYC/CEN8, NR4A3, NTRK3, NUTM1, PDGFB, PDGFRA/FIP1L1, PHF1, PTEN/CEN10, RB1/13q34, RET, ROS1, SMARCB1/22q12, SS18, SS18/SSX1, TFE3, TP53/CEN17, USP6, VHL/CEN3, WT1, WWTR1</p> |
| 823/6 | <p>Hereditární nádorové syndromy a somatické varianty v nádorových tkáních</p> <p>Germinální testování: <i>ACD, AIP, AKT1, ALK, APC, AR, ATM, ATR, AXIN2, BAP1, BARD1, BLM, BMP1A, BRAF, BRCA1, BRCA2, BRF1, BRIP1, CARD11, CCND1, CDC73, CDH1, CDK4, CDKN1B, CDKN2A, CYLD, DICER1, DLST, DNMT3A, EGFR, ELP1, EMSY, EPCAM, ERBB2, ERBB3, ERCC2, ERCC3, ERCC5, ETV6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAT1, FBXW7, FH, FLCN, GATA2, GREM1, HNF1A, HOXB13, HRAS, CHEK1, CHEK2, IDH1, IDH2, KIF1B, KIT, KLLN, KMT2A, KRAS, MAP2K1, MAX, MC1R, MEN1, MET, MITF, MLH1, MLH3, MN1, MRE11, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2, NRAS, NRG1, NTHL1, PALB2, PALLD, PARP1, PARP2, PARP3, PARP4, PBRM1, PDGFB, PDGFD, PDGFRA, PDGFRB, PHOX2B, PIK3CA, PMS2, POLD1, POLE, POT1, PPP2R2A, PRKARIA, PTEN, PTCH1, PTCH2, RABL3, RAD50, RAD51, RAD51B, RAD51C, RAD51D, RAD52, RAD54L, RB1, RECQL, RECQL4, RET, REXO2, RNF43, RPS20, SDHA, SDHAF2, SDHB, SDHC, SDHD, SH2D1A, SH2D2A, SLC25A11, SLX4, SMAD4, SMARCA4, SMARCB1, SMARCE1, SRGAP1, SRY, STK11, SUFU, TERF2IP, TERT, TET2, TMEM127, TP53, TSC1, TSC2, VHL, WRN, WT1, XPA, XRCC2, XRCC3 (hg38).</i></p> <p>Somatické testování: <i>ABRAXAS1, ACVR1, ALK, ARAF, ARID1A, ARID1B, ATRX, BRK1, CCDC153, CCNE1, CD79B, CDADC1, CDK12, CDK6, CDKN2B, COX6C, CTNNA1, CTNNB1, DDR2, DPYD, EIF1AX, ENG, ENOX1, ERG, ESRI, EZHIP, FAM124A, FANCD2OS, FGFR1, FGFR2, FGFR3, FOXL2, GEN1, HDAC2, H3-C14 (H3), H3C2 (HIST1H3B), H3C3, H3-3A, IL18, INA, INCA1, KEAP1, KHDC3L, KIAA1549, MAD2L2, MAMLD1, MALT1, MTOR, MYB, MYBL1, MYC, MYCN, MYD88, MYH13, NAT1, NLRP7, NTRK1, NTRK2, NTRK3, NUTM1, PABPC1, PABPC3, PATZ1, PAX8, PIK3R1, PML, PPARG, PPM1D, PPP2R1A, QKI, RAB18, RAF1, RELA, RHOA, ROS1, RPAIN, SETD2, SIPR1, TAC1, TAC13, TBX21, TCEB1 (ELOC), TFEB, TFE3, THADA, TMPRSS2, TOE1, TOP2A, TOP6BL (C11orf80),</i></p> |

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| | <p>WSCD1, YAP1, ZFTA (<i>C11orf95</i>) (<i>hg38</i>). Hereditární pankreatitida: <i>CFTR, CTRC, CPA1, SPINK1, PRSS1</i> (<i>hg38</i>). Lymfoproliferace: <i>AICDA, AKT3, ASXL1, B2M, BAX, BCL2, BCL3, BCL6, BCL10, BIRC3, BLNK, BTG1, BTK, CFBF, CCDC50, CCND2, CCND3, CCR4, CCR7, CD28, CD44, CD58, CD70, CD79A, CD274, CDC25A, CIITA, CREBBP, CTLA4, CXCR4, DAPK1, DDX3X, DEK, DUSP22, E2F2, EBF1, EGR2, EP300, EZH2, FLT3, FOXO1, FOXP1, FYN, GNA13, H1-2, H1-4, CHD2, CHIC2, ID3, IKBKB, IKZF1, IL16, INO80, IRF4, IRF8, ITPKB, JAK1, JAK3, KDM6A, KLF2, KLHL6, KMT2C, KMT2D, LMO2, MAP3K14, MCL1, MEF2B, MGA, MLF1, MLLT10, MME, NFKB2, NFKBIE, NOTCH1, NOTCH2, NSD2, P2RY8, PAX5, PDCD1, PDCD1LG2, PIK3CD, PIM1, PIM2, PLCG1, PLCG2, PRDM1, PRDM16, PRKCB, PTPN1, PTPN11, PTPRD, RAG1, RAG2, REL, RELN, ROCK1, RPS15, SIPR2, SAMHD1, SERPINA9, SF3B1, SGK1, SH2B3, SOCS1, STAT3, STAT5B, STAT6, TBC1D9, TCF3, TLN2, TNFAIP3, TNFRSF13B, TNFRSF14, TNFSF4, TP63, TRAF2, UBE2A, UBR5, XPO1, ZMYM3</i> (<i>hg38</i>) (<i>v12</i>)</p> <p>Somatické varianty v nádorových tkáních II</p> <p><i>A2M, ABCC9, ABL1, ABL2, ABRAXAS1, ACBD5, ACD, ACP5, ACTB, ACVR1, ACVR1B, ACVR2A, ADAMTS14, ADGRA2, AFAP1L2, AFF1, AFF3, AGTRAP, AHRR, AICDA, AIP, AKAP9, AKT1, AKT2, AKT3, ALK, ALOX12B, AMER1, AMN, ANKRD11, ANKRD26, ANTXR1, ANTXR2, ANXA11, APC, AR, ARAF, ARFRP1, ARHGGEF2, ARID1A, ARID1B, ARID2, ARID5B, ARMC10, ASB13, ASPSCR1, ASXL1, ASXL2, ATF1, ATF5, ATIC, ATM, ATP1B1, ATR, ATRX, AURKA, AURKB, AXIN1, AXIN2, AXL, B2M, BAG4, BAP1, BARD1, BATF3, BAX, BBC3, BCAM, BCL2, BCL2L1, BCL2L2, BCL2L11, BCL3, BCL6, BCL10, BCOR, BCORL1, BCR, BEST3, BIRC3, BIRC6, BLM, BLNK, BMF, BMP4, BMP7, BMPR1A, BRAF, BRCA1, BRCA2, BRD4, BRD8, BRF1, BRIP1, BTG1, BTK, BUB1, BUB1B, CAD, CALR, CAMK2A, CAMTA1, CARD11, CARS1, CASP8, CASZ1, CATSPERZ, CFBF, CBL, CC2D1A, CCDC6, CCDC50, CCN6, CCNA2, CCNB1, CCNB2, CCNB3, CCND1, CCND2, CCND3, CCNE1, CCR4, CCR7, CD28, CD34, CD44, CD58, CD70, CD74, CD79A, CD79B, CD274, CD276, CDADC1, CDC6, CDC25A, CDC73, CDH1, CDH11, CDK1, CDK4, CDK6, CDK8, CDK12, CDKN1A, CDKN1B, CDKN1C, CDKN2A, CDKN2B, CDKN2C, CDKN2D, CDX1, CDX2, CENPA, CEP43, CEP85L, CIC, CIITA, CITED2, CLCA2, CLCN6, CLIP1, CLTC, CNBP, CNTN1, COG8, COL1A1, COL1A2, COL25A1, COL3A1, COL17A1, COP1, CPM, CREB1, CREB3L1, CREB3L2, CREB3L3, CREBBP, CREM, CRKL, CRLF2, CRTCI, CRTCC3, CSF1R, CSF3R, CSGALNACT2, CSNK1A1, CTCF, CTLA4, CTNNA1, CTNNA1, CUBN, CUL3, CUX1, CXCR4, CYB5R2, CYLD, CYP1A1, CYP3A4, CYSLTR2, DAPK1, DAXX, DCTN1, DCUN1D1, DDIT3, DDR2, DDX3X, DDX5, DDX41, DEK, DENND3, DERA, DFFA, DHX15, DHX57, DICER1, DIS3, DLL1, DLST, DNAJB1, DNMT1, DNMT3A, DNMT3B, DNMT3C, DNMT3L, DNMT3D, DNMT3E, DNMT3F, DNMT3G, DNMT3H, DNMT3I, DNMT3J, DNMT3K, DNMT3L, DNMT3M, DNMT3N, DNMT3O, DNMT3P, DNMT3Q, DNMT3R, DNMT3S, DNMT3T, DNMT3U, DNMT3V, DNMT3W, DNMT3X, DNMT3Y, DNMT3Z, DNMT3AA, DNMT3AB, DNMT3AC, DNMT3AD, DNMT3AE, DNMT3AF, DNMT3AG, DNMT3AH, DNMT3AI, DNMT3AJ, DNMT3AK, DNMT3AL, DNMT3AM, DNMT3AN, DNMT3AO, DNMT3AP, DNMT3AQ, DNMT3AR, DNMT3AS, DNMT3AT, DNMT3AU, DNMT3AV, DNMT3AW, DNMT3AX, DNMT3AY, DNMT3AZ, DNMT3BA, DNMT3BB, DNMT3BC, DNMT3BD, DNMT3BE, DNMT3BF, DNMT3BG, DNMT3BH, DNMT3BI, DNMT3BJ, DNMT3BK, DNMT3BL, DNMT3BM, DNMT3BN, DNMT3BO, DNMT3BP, DNMT3BQ, DNMT3BR, DNMT3BS, DNMT3BT, DNMT3BU, DNMT3BV, DNMT3BW, DNMT3BX, DNMT3BY, DNMT3BZ, DNMT3CA, DNMT3CB, DNMT3CC, DNMT3CD, DNMT3CE, DNMT3CF, DNMT3CG, DNMT3CH, DNMT3CI, DNMT3CJ, DNMT3CK, DNMT3CL, DNMT3CM, DNMT3CN, DNMT3CO, DNMT3CP, DNMT3CQ, DNMT3CR, DNMT3CS, DNMT3CT, DNMT3CU, DNMT3CV, DNMT3CW, DNMT3CX, DNMT3CY, DNMT3CZ, DNMT3DA, DNMT3DB, DNMT3DC, DNMT3DD, DNMT3DE, DNMT3DF, DNMT3DG, DNMT3DH, DNMT3DI, DNMT3DJ, DNMT3DK, DNMT3DL, DNMT3DM, DNMT3DN, DNMT3DO, DNMT3DP, DNMT3DQ, DNMT3DR, DNMT3DS, DNMT3DT, DNMT3DU, DNMT3DV, DNMT3DW, DNMT3DX, DNMT3DY, DNMT3DZ, DNMT3EA, DNMT3EB, DNMT3EC, DNMT3ED, DNMT3EE, DNMT3EF, DNMT3EG, DNMT3EH, DNMT3EI, DNMT3EJ, DNMT3EK, DNMT3EL, DNMT3EM, DNMT3EN, DNMT3EO, DNMT3EP, DNMT3EQ, DNMT3ER, DNMT3ES, DNMT3ET, DNMT3EU, DNMT3EV, DNMT3EW, DNMT3EX, DNMT3EY, DNMT3EZ, DNMT3FA, DNMT3FB, DNMT3FC, DNMT3FD, DNMT3FE, DNMT3FF, DNMT3FG, DNMT3FH, DNMT3FI, DNMT3FJ, DNMT3FK, DNMT3FL, DNMT3FM, DNMT3FN, DNMT3FO, DNMT3FP, DNMT3FQ, 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DNMT3SO, DNMT3SP, DNMT3SQ, DNMT3SR, DNMT3SS, DNMT3ST, DNMT3SU, DNMT3SV, DNMT3SW, DNMT3SX, DNMT3SY, DNMT3SZ, DNMT3TA, DNMT3TB, DNMT3TC, DNMT3TD, DNMT3TE, DNMT3TF, DNMT3TG, DNMT3TH, DNMT3TI, DNMT3TJ, DNMT3TK, DNMT3TL, DNMT3TM, DNMT3TN, DNMT3TO, DNMT3TP, DNMT3TQ, DNMT3TR, DNMT3TS, DNMT3TT, DNMT3TU, DNMT3TV, DNMT3TW, DNMT3TX, DNMT3TY, DNMT3TZ, DNMT3UA, DNMT3UB, DNMT3UC, DNMT3UD, DNMT3UE, DNMT3UF, DNMT3UG, DNMT3UH, DNMT3UI, DNMT3UJ, DNMT3UK, DNMT3UL, DNMT3UM, DNMT3UN, DNMT3UO, DNMT3UP, DNMT3UQ, DNMT3UR, DNMT3US, DNMT3UT, DNMT3UU, DNMT3UV, DNMT3UW, DNMT3UX, DNMT3UY, DNMT3UZ, DNMT3VA, DNMT3VB, DNMT3VC, DNMT3VD, DNMT3VE, DNMT3VF, DNMT3VG, DNMT3VH, DNMT3VI, DNMT3VJ, DNMT3VK, DNMT3VL, DNMT3VM, DNMT3VN, DNMT3VO, DNMT3VP, DNMT3VQ, DNMT3VR, DNMT3VS, DNMT3VT, DNMT3VU, DNMT3VV, DNMT3VW, DNMT3VX, DNMT3VY, DNMT3VZ, DNMT3WA, DNMT3WB, DNMT3WC, DNMT3WD, DNMT3WE, DNMT3WF, DNMT3WG, DNMT3WH, DNMT3WI, DNMT3WJ, DNMT3WK, DNMT3WL, DNMT3WM, DNMT3WN, DNMT3WO, DNMT3WP, DNMT3WQ, DNMT3WR, DNMT3WS, DNMT3WT, DNMT3WU, DNMT3WV, DNMT3WW, DNMT3WX, DNMT3WY, DNMT3WZ, DNMT3XA, DNMT3XB, DNMT3XC, DNMT3XD, DNMT3XE, DNMT3XF, DNMT3XG, DNMT3XH, DNMT3XI, DNMT3XJ, DNMT3XK, DNMT3XL, DNMT3XM, DNMT3XN, DNMT3XO, DNMT3XP, DNMT3XQ, DNMT3XR, DNMT3XS, DNMT3XT, DNMT3XU, DNMT3XV, DNMT3XW, DNMT3XX, DNMT3XY, DNMT3XZ, DNMT3YA, DNMT3YB, DNMT3YC, DNMT3YD, DNMT3YE, DNMT3YF, DNMT3YG, DNMT3YH, DNMT3YI, DNMT3YJ, DNMT3YK, DNMT3YL, DNMT3YM, DNMT3YN, DNMT3YO, DNMT3YP, DNMT3YQ, DNMT3YR, DNMT3YS, DNMT3YT, DNMT3YU, DNMT3YV, DNMT3YW, DNMT3YX, DNMT3YY, DNMT3YZ, DNMT3ZA, DNMT3ZB, DNMT3ZC, DNMT3ZD, DNMT3ZE, DNMT3ZF, DNMT3ZG, DNMT3ZH, DNMT3ZI, DNMT3ZJ, DNMT3ZK, DNMT3ZL, DNMT3ZM, DNMT3ZN, DNMT3ZO, DNMT3ZP, DNMT3ZQ, DNMT3ZR, DNMT3ZS, DNMT3ZT, DNMT3ZU, DNMT3ZV, DNMT3ZW, DNMT3ZX, DNMT3ZY, DNMT3ZZ</i></p> |

| Odbornost / poř. číslo | Detailní informace k činnostem v rozsahu akreditace |
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| | <p>c.183A>C, c.183A>T, c.351A>C, c.351A>T, c.436G>C, c.436G>A, c.437C>T) <i>NRAS</i> (sekvenční varianty: c.34G>T, c.34G>A, c.35G>A, c.35G>C, c.35G>T, c.38G>A, c.38G>T, c.37G>C, c.175G>A, c.181C>A, c.182A>T, c.182A>G, c.183A>C, c.183A>T, c.351G>C, c.351G>T, c.436G>A, c.437C>T) <i>BRAF</i> (sekvenční varianty: c.1799T>A, c.1799_1800delinsAA, c.1799_1800delinsAC, c.1799_1800delinsAT, c.1798_1799delinsAA, c.1798_1799delinsAG, c.1798G>A) <i>EFGR</i> (sekvenční varianty: c.2156G>C, c.2155G>T, c.2154_2155delinsTT, c.2155G>A, c.2238_2248delinsGC, c.2239_2248delinsC, c.2240_2248del, c.2239_2247del, c.2239_2251delinsC, c.2240_2251del, c.2235_2249del, c.2236_2250del, c.2239_2253del, c.2240_2254del, c.2238_2252del, c.2237_2251del, c.2235_2252delinsAAT, c.2237_2252delinsT, c.2234_2248del, c.2236_2253delinsCTA, c.2237_2253delinsTA, c.2235_2251delinsAG, c.2236_2253delinsCAA, c.2230_2249delinsGTCAA, c.2240_2257del, c.2237_2255delinsT, c.2239_2256del, c.2236_2253del, c.2239_2258delinsCA, c.2237_2254del, c.2238_2255del, c.2237_2257delinsTCT, c.2236_2255delinsAT, c.2236_2256delinsATC, c.2237_2256delinsTT, c.2237_2256delinsTC, c.2235_2255delinsGGT, c.2238_2258del, c.2236_2256del, c.2253_2276del, c.2369C>T, c.2303G>T, c.2310_2311insGGT, c.2308_2309insGCCAGCGTG, c.2308_2311delinsCCAGCGTGGAT, c.2311_2312insGCGTGGACA, c.2319_2320insCAC, c.2573T>G, c.2573_2574delinsGT, c.2573_2574delinsGA, c.2582T>A)</p> <p>EasyPGX: <i>KRAS</i> (sekvenční varianty: c.34G>T, c.34G>C, c.34G>A, c.35G>C, c.35G>A, c.35G>T, c.38G>A, c.176C>A, c.176C>G, c.175G>A, c.181C>A, c.182A>T, c.182A>G, c.183A>C, c.183A>T, c.349A>G, c.350A>G, c.351A>C, c.351A>T, c.436G>C, c.436G>A, c.437C>T) <i>NRAS</i> (sekvenční varianty: c.34G>T, c.34G>A, c.35G>A, c.35G>C, c.35G>T, c.38G>A, c.38G>T, c.37G>C, c.175G>A, c.176C>T, c.181C>A, c.182A>T, c.182A>G, c.183A>C, c.183A>T, c.350A>G, c.351G>C, c.351G>T, c.436G>A, c.437C>T) <i>BRAF</i> (sekvenční varianty: c.1799T>A, c.1799_1800TG>AA, c.1799_1800TG>AT, c.1798_1799GT>AA, c.1798_1799GT>AG)</p> |
| 823/8 | Mikrosatelitová instabilita: BAT-25, BAT-26, NR-21, NR-24, MONO-27, Penta C a Penta D |

Vysvětlivky:

¹ Zavedené stupně volnosti podle MPA 00-09-23:

A - Flexibilita týkající se dokumentovaného postupu vyšetření / odběru

B - Flexibilita týkající se techniky

C - Flexibilita týkající se analytů/parametrů

D - Flexibilita týkající se vyšetřovaného materiálu

Není-li uveden žádný stupeň volnosti, nemůže laboratoř pro dané vyšetření uplatňovat flexibilní přístup k rozsahu akreditace.

CGH array – comparative genome hybridization array

SNP array – single nucleotide polymorphism

FISH – fluorescenční in situ hybridizace

PCR – polymerázová řetězová reakce

MPS – masivně paralelní sekvenování

NGS – next generation sequencing

MLPA – multiplex ligation dependent probe amplifikation

Real-time PCR – polymerázová řetězová reakce v reálném čase

NK – nukleová kyselina