

Akreditovaný subjekt podle ČSN EN ISO 15189:2013:

GHC GENETICS, s.r.o.
Laboratoř GHC GENETICS
V Holešovičkách 1156/29, 182 00 Praha 8

Laboratoř uplatňuje flexibilní přístup k rozsahu akreditace upřesněný v dodatku. Aktuální seznam činností prováděných v rámci flexibilního rozsahu má laboratoř k dispozici u manažera kvality laboratoře.

Vyšetření:

Pořadové číslo	Přesný název postupu vyšetření	Identifikace postupu vyšetření	Předmět vyšetření
816 - Laboratoř lékařské genetiky			
1.	Molekulárně genetické vyšetření I/D mutace v genu PAI-1 metodou Melting Curve Genotyping [MCG]	SOP-316	Nesrážlivá venózní krev, bukální stěr
2.	Neobsazeno		
3.	Molekulárně genetické vyšetření mutací v genech ¹⁾ metodou Real-Time PCR	SOP-701	Nesrážlivá venózní krev, bukální stěr
4.	Molekulárně genetické vyšetření duplikací a delecí v genech ²⁾ metodou Multiplex Ligation-dependent Probe Amplification [MLPA]	SOP-702	Nesrážlivá venózní krev, bukální stěr
5.	Molekulárně genetické vyšetření genů ³⁾ metodou masivního paralelního sekvenování	SOP-703	Nesrážlivá venózní krev, bukální stěr
6.	Molekulárně genetické vyšetření pro detekci alel HLA ⁴⁾ pro predispozici k chorobám metodou strip assay	SOP-704	Nesrážlivá venózní krev, bukální stěr
7.	Molekulárně genetické vyšetření mutací ⁵⁾ genu CFTR metodou fragmentační analýzy	SOP-418	Nesrážlivá venózní krev, bukální stěr
8.	Molekulárně genetické vyšetření mutací v genech ⁶⁾ metodou fragmentační analýzy	SOP-705	Nesrážlivá venózní krev, bukální stěr

**Příloha je nedílnou součástí
osvědčení o akreditaci č.: 629/2021 ze dne: 02. 12. 2021**

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802 - Lékařská mikrobiologie			
1.	Detekce přítomnosti viru SARS-CoV-2 ⁷⁾ metodou RT-PCR	SOP-706	Nasopharyngeální a oropharyngeální stěry, sliny

Dodatek:

Flexibilní rozsah akreditace

Pořadová čísla postupů vyšetření
<i>odbornost 816:3 - 6, 8; odbornost 802:1</i>

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

Odběr primárních vzorků:

Pořadové číslo	Přesný název postupu odběru primárního vzorku	Identifikace postupu odběru primárního vzorku	Primární vzorek
1.	Odběr biologického materiálu	SOP-521	Periferní krev
2.	Odběr biologického materiálu	SOP-522	Bukální stěr

Vysvětlivky:

- 1) Detekované mutace pomocí metody Real-Time PCR: G1691A v genu pro Faktor V, G20210A v genu pro Faktor II, C677T v genu pro MTHFR, A1298C v genu pro MTHFR; c.181T>G, c.213-12A>G, c.5266dupC, c.68_69del2, c.1687C>T, c. 843_846del4, c. 4243delG v genu *BRCA1*, c.8537_8538del2, c.658_659del2, c.3847_3848del2, c.5645C>A v genu *BRCA2*
- 2) Vyšetření genů *BRCA1*, *BRCA2*, *PALB2*, *ATM*, *APC*, *BARD1*, *BRIP1*, *CDH1*, *CHEK2*, *EPCAM*, *MLH1*, *MSH2*, *MSH6*, *MUTYH*, *NBN*, *PMS2*, *PTEN*, *RAD50*, *RAD51C*, *RAD51D*, *STK11*, *TP53*
- 3) Vyšetření genů *BRCA1*, *BRCA2*, *PALB2*, *ATM*, *APC*, *BARD1*, *BRIP1*, *CDH1*, *CHEK2*, *EPCAM*, *MLH1*, *MSH2*, *MSH6*, *MUTYH*, *NBN*, *PMS2*, *PTEN*, *RAD50*, *RAD51C*, *RAD51D*, *STK11*, *TP53*, *A2M*, *A4GALT*, *A4GNT*, *AAAS*, *AADAC*, *AADA2L*, *AAGAB*, *AANAT*, *AARS*, *AARS2*, *AASS*, *ABAT*, *ABCA1*, *ABCA10*, *ABCA12*, *ABCA13*, *ABCA2*, *ABCA3*, *ABCA4*, *ABCB1*, *ABCB11*, *ABCB4*, *ABCB6*, *ABCB7*, *ABCC1*, *ABCC11*, *ABCC2*, *ABCC3*, *ABCC4*, *ABCC6*, *ABCC8*, *ABCC9*, *ABCD1*, *ABCD3*, *ABCD4*, *ABCG2*, *ABCG5*, *ABCG8*, *ABHD12*, *ABHD5*, *ABI3BP*, *ABL1*, *ABL2*, *ABO*, *ACACA*, *ACACB*, *ACAD10*, *ACAD11*, *ACAD8*, *ACAD9*, *ACADL*, *ACADM*, *ACADS*, *ACADSB*, *ACADVL*

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ACAN, ACAT1, ACAT2, ACBD5, ACBD6, ACCS, ACE, ACHE, ACLY, ACO2, ACOX1, ACP1, ACP2, ACP5, ACSF3, ACSL4, ACSL5, ACSL6, ACSM3, ACTA1, ACTA2, ACTB, ACTC1, ACTG1, ACTN2, ACTN4, ACVR1, ACVR1B, ACVR1C, ACVR2A, ACVR2B, ACVRL1, ACY1, ADA, ADAM10, ADAM12, ADAM17, ADAM19, ADAM22, ADAM23, ADAM33, ADAM7, ADAM9, ADAMTS1, ADAMTS10, ADAMTS13, ADAMTS17, ADAMTS18, ADAMTS19, ADAMTS2, ADAMTSL2, ADAMTSL3, ADAMTSL4, ADAR, ADARB1, ADC, ADCK3, ADCY10, ADCY3, ADCY5, ADCY6, ADCY9, ADD1, ADD2, ADH1B, ADH1C, ADH4, ADH7, ADIPOQ, ADIPOR1, ADK, ADM, ADORA2A, ADRA1A, ADRA2A, ADRA2B, ADRA2C, ADRB1, ADRB2, ADRB3, ADRBK2, ADSL, ADTRP, AFF2, AFF3, AFG3L2, AFP, AGA, AGBL1, AGGF1, AGK, AGL, AGMO, AGO1, AGPAT2, AGPS, AGRN, AGRP, AGT, AGTPBP1, AGTR1, AGTR2, AGXT, AGXT2, AHCY, AH1, AHR, AHRR, AHSG, AHSP, AICDA, AIFM1, AIMP1, AIP, AIPL1, AIRE, AK1, AK2, AK7, AK8, AKAP10, AKAP2, AKAP9, AKR1B1, AKR1C2, AKR1C3, AKR1C4, AKR1D1, AKR7A2, AKT1, AKT2, AKT3, ALAD, ALAS2, ALB, ALDH16A1, LDH18A1, ALDH1A1, ALDH1A2, ALDH2, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG12, ALG13, ALG14, ALG2, ALG3, ALG5, ALG6, ALG8, ALG9, ALK, ALMS1, ALOX12, ALOX12B, ALOX15, ALOX5, ALOX5AP, ALOXE3, ALPL, ALS2, ALS2CL, ALX1, ALX3, ALX4, AMACR, AMBN, AMELX, AMELY, AMER1, AMH, AMHR2, AMN, AMPD1, AMPD3, AMT, ANG, ANGPTL3, ANGPTL4, ANGPTL5, ANGPTL6, ANK1, ANK2, ANK3, ANKH, ANKK1, ANKRD1, ANKRD11, ANKRD26, ANKS1A, ANKS6, ANO10, ANO3, ANO5, ANO6, ANO7, ANTXR1, ANTXR2, ANXA5, AOA, AP1S1, AP1S2, AP3B1, AP3B2, AP3D1, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, PAF1, APBA2, APBB1IP, APBB2, APCDD1, APEX1, APOA1, APOA2, APOA4, APOA5, APOB, APOBEC1, APOBEC3H, APOC4_APOC2, APOD, APOE, APOH, APOL1, APOL3, APP, APRT, APTX, AQP1, AQP2, AQP3, AQP4, AQP5, AQP7, AR, ARF4, ARFGEF2, ARG1, ARHGAP18, ARHGAP24, ARHGAP26, ARHGAP31, ARHGAP6, ARHGAP9, ARHGADIA, ARHGEF10, ARHGEF12, ARHGEF6, ARHGEF7, ARHGEF9, ARIDIA, ARID1B, ARID4A, ARID4B, ARL13B, ARL14EP, ARL3, ARL6, ARMS2, ARSA, ARSB, ARSE, ART4, ARX, AS3MT, ASAH1, ASB10, ASCC1, ASCC3, ASCL1, ASIC3, 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DGCR2, DGKD, DGKE, DGUOK, DHCR24, DHCR7, DHDDS, DHH, DHODH, DHRS4L1, DHTKD1, DHX37, DIABLO, DIAPH1, DIAPH2, DIAPH3, DICER1, DIP2A, DIP2B, DIP2C, DIRC2, DIS3L2, DISC1, DISP1, DKC1, DKK3, DLAT, DLC1, DLD, DLEC1, DLG2, DLG3, DLG4, DLGAP3, DLL1, DLL3, DLX3, DLX5, DLX6, DMC1, DMD, DMGDH, DMP1, DMPK, DMRT1, DMXL1, DNAAF1, DNAAF2, DNAAF3, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAJB2, DNAJB6, DNAJC19, DNAJC30, DNAJC5, DNAJC6, DNAL1, DNASE1, DNASE1L3, DNASE2, DNMI1, DNMI1L, DNMI2, DNMT1, DNMT3A, DNMT3B, DNMT3L, DOC2A, DOCK3, DOCK4, DOCK6, DOCK7, DOCK8, DOCK9, DOK1, DOK2, DOK5, DOK7, DOLK, DOLPP1, DOT1L, DPAGT1, DPCD, DPM1, DPM2, DPM3, DPP6, DPY19L2, DPYD, DPYS, DRD1, DRD2, DRD3, DRD4, DRD5, DSC2, DSC3, DSCAM, DSG1, DSG2, DSG3, DSG4, DSP, DSPP, DST, DTNA, DTNBP1, DUOX2, DUOX2A, DUX4, DUX4L3, DVL2, DYM, DYNAP, DYNC1H1, DYNC2H1, DYRK1A, DYSF, DYX1C1, E2F1, E2F4, E2F5, EARS2, EBAG9, EBF4, EBP, ECE1, ECE2, EC11, ECM1, ECM2, ECSIT, EDA, EDA2R, EDAR, EDARADD, EDN1, EDN3, EDNRA, EDNRB, EFCAB5, EFEMP1, EFEMP2, EFHC1, EFHC2, EFNA5, EFN1, EFR3A, EFTUD2, EGF, EGFR, EGLN1, EGR2, EHP1, EHD2, EHHADH, EHMT1, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2D, EIF2S3, EIF4E, EIF4G1, EIF4H, ELAC2, ELANE, ELF4, ELK1, ELK3, ELMOD2, ELN, ELOVL4, ELP2, ELP4, EMC4, EMD, EME1, EMG1, EMX1, EMX2, EN2, ENAM, ENG, ENOX1, ENPP1, ENTPD1, EOMES, EP300, EPAS1, EPB41, EPB41L1, EPB41L2, EPB42, EPC2, EPHA2, EPHA3, EPHA5, EPHA7, EPHB2, EPHB6, EPHX1, EPHX2, EPM2A, EPO, EPOR, EPX, ERAP1, ERAP2, ERBB2, ERBB3, ERBB4, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, ERLIN2, ERMAP, ERFF1, ESCO2, ESPN, ESR1, ESR2, ESRRB, ETFA, ETFB, ETFDH, ETHE1, ETV4, ETV6, EVC, EVC2, EVI5, EWSR1, EXO1, EXO5, EXOC4, EXOSC3, EXPH5, EXT1, EXT2, EXTL1, EXTL2, EXTL3, EYA1, EYA4, EYS, EZH2, F10, F11, F12, F13A1, F13B, F2, F2RL1, F5, F7, F8, F9, FA2H, FAAH, FAAH2, FABP1, FABP2, FADD, FAH, FAM120A, FAM126A, FAM134B, FAM161A, FAM175A, FAM205A, FAM20A, FAM20C, FAM47B, FAM58A, FAM83H, FAM8A1, FAM91A1, FAN1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FARS2, FAS, FASLG, FASN, FASTKD2, FBLN1, FBLN5, FBN1, FBN2, FBP1, FBXL6, FBXO10, FBXO11, FBXO18, FBXO7, FBXW11, FBXW4, FBXW7, FCER1G, FCGR2A, FCGR2B, FCN2, FCN3, FDFT1, FECH, FEM1A, FEM1B, FERMT1, FERMT3, FEZF2, FFAR1, FFAR4, FGA, FGB, FGD1, FGD3, FGD4, FGF10, FGF14, FGF2, FGF23, FGF3, FGF8, FGF9, FGFR1, FGFR2, FGFR3, FGFR4, FGFR1L, FGG, FH, FHIT, FHL1, FHL2, FIG4, FIGLA, FIP1L1, FKBP10, FKBP14, FKBP1A, FKBP4, FKBP5, FKBP6, FKBP8, FKRP, FKTN, FLCN, FLG, FLI1, FLNA, FLNB, FLNC, FLOT1, FLT1, FLT3, FLT4, FLVCR1, FLVCR2, FMN1, FMN2, FMO1, FMO2, FMO3, FMO4, FMO5, FMO6, FMOD, FMRI, FMRI_AS1, FN1, FN3K, FOLH1, FOLR1, FOXA1, FOXA2, FOXA3, FOXC1, FOXC2, FOXD3, FOXE1, FOXE3, FOXF1, FOXF2, FOXG1, FOXH1, FOXI1, FOXJ1, FOXK1, FOXL2, FOXM1, FOXN1, FOXO1, FOXP1, FOXP2, FOXP3, FOXRED1, FPGS, FPR1, FRAS1, FREM1, FREM2, FREM3, FRK, FRMD5, FRMD7, FRMPD4, FRY, FRZB, FSCB, FSCN2, FSHB, FSHR, FST, FSTL1, FTCD, FTHL17, FTO, FTSJ1, FUCAL, FUS, FUT1, FUT2, FUT3, FUT6, FUT7, FUT8, FUZ, FXN, FXN2, FYCO1, FZD3, FZD4, FZD6, FZD9, G6PC, G6PC2, G6PC3, G6PD, GAA, GABARAPL1, GABBR1, GABRA1, GABRA2, GABRA6, GABRB3, GABRD, GABRG1, GABRG2, GABRR2, GAD1, GAD2, GADD45A, GAK, GAL3ST1, GAL3ST2, GAL3ST3, GAL3ST4, GALT, GALE, GALK1, GALNS, GALNT11, GALNT12, GALNT13, GALNT14, GALNT18, GALNT2, GALNT3, GALNT5, GALNT6, GALNT7, GALNT8, GALNT9, GALNTL6, GALT, GAMT, GAN, GAP43, GARS, GAS1, GAS2L2, GATA1, GATA2, GATA3, GATA4, GATA5, GATA6, GATAD1, GATM, GBA, GBE1, GBGT1, GC, GCDH, GCGR, GCH1, GCK, GCKR, GCLC, GCLM, GCM2, GCNT1, GCNT2, GDAP1, GDF1, GDF3, GDF5, GDF6, GDF9, GDI1, GDNF, GFAP, GFII, GFII1B, GFM1, GFPT1, GFRA1, GFRA2, GGCX, GGH,

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GH1, GH2, GHR, GHRH, GHRHR, GHRL, GHSR, GIF, GIGYF2, GIMAP8, GIP, GIPC3, GIPR, GIT1, GJA3, GJA5, GJA8, GJB1, GJB2, GJB3, GJB4, GJB6, GJC2, GJC3, GK, GLA, GLB1, GLCC11, GLDC, GLE1, GLI2, GLI3, GLIS2, GLIS3, GLMN, GLO1, GLP1R, GLRA1, GLRB, GLRX5, GLUL, GLYCTK, GM2A, GMDS, GMPS, GNAI2, GNAI3, GNAQ, GNAS, GNAT1, GNAT2, GNB1L, GNB3, GNE, GNG7, GNMT, GNPAT, GNPTAB, GNPTG, GNRH1, GNRHR, GNS, GOLGA3, GOLGA5, GON4L, GOPC, GORAB, GOSR2, GOT1, GP1BA, GP6, GP9, GPAM, GPATCH8, GPBAR1, GPC3, GPC4, GPC6, GPD1, GPD1L, GPD2, GPHN, GPI, GPIHBP1, GPNMB, GPR1, GPR12, GPR132, GPR139, GPR143, GPR179, GPR37, GPR56, GPR98, GPS1, GPSM2, GPT, GRB10, GREM1, GRHL1, GRHL2, GRHPR, GRIA1, GRIA3, GRID1, GRID2, GRIK1, GRIK2, GRIK4, GRIN1, GRIN2A, GRIN2B, GRIN3A, GRIP1, GRK1, GRK4, GRM1, GRM5, GRM6, GRM7, GRN, GRPR, GRXCR1, GSC, GSE1, GSN, GSPT2, GSR, GSS, GSTA2, GSTA3, GSTM1, GSTM3, GSTO1, GSTP1, GSTT1, GSTZ1, POMGNT2(GTDC2), GTF2H5, GTF2I, GTF2IRD1, GUCA1A, GUCA1B, GUCY2C, GUCY2D, GUCY2F, GUSB, GYG1, GYG2, GYLTL1B, GYPA, GYPC, GYPE, GYS1, GYS2, H6PD, HABP2, HACE1, HADH, HADHA, HADHB, HAL, HAMP, HAND1, HAND2, HAPLN1, HARS, HARS2, HAVCR1, HAX1, HBB, HBD, HBE1, HBM, HCCS, HCFC1, HCN1, HCN2, HCN3, HCN4, HCRT, HCRTR2, HDAC4, HDAC6, HDAC8, HDAC9, HDC, HDGFRP2, HDLBP, HDX, HEATR2, HEPACAM, HEPH, HERC1, HES7, HESX1, HEXA, HEXB, HEY1, HEY2, HFE, HFE2, HGD, HGF, HGSNAT, HIBCH, HIF1A, HIGD2A, HINT1, HIP1, HIST1H2AE, HIST3H3, HK1, HK2, HLA_A, HLA_B, HLA_C, HLA_DQA1, HLA_DQB1, HLA_DRB5, HLA_G, HLCS, HLX, HMBS, HMCN1, HMGA2, HMGCL, HMGCR, HMGCS2, HMHA1, HMMR, HMOX1, HMX1, HMX2, HNF1A, HNF1B, HNF4A, HNMT, HNRNPH3, HNRNPU, HOGA1, HOXA1, HOXA10, HOXA11, HOXA13, HOXA2, HOXA3, HOXA4, HOXB1, HOXB13, HOXB6, HOXB8, HOXC13, HOXD10, HOXD13, HOXD4, HP, HPCAL4, HPD, HPGD, HPRT1, HPS1, HPS3, HPS4, HPS5, HPS6, HPSE2, HR, HRAS, HRG, HSD11B1, HSD11B2, HSD17B10, HSD17B3, HSD17B4, HSD3B1, HSD3B2, HSD3B7, HSF1, HSF4, HSP90AA1, HSPA9, HSPB1, HSPB3, HSPB6, HSPB7, HSPB8, HSPG2, HTN3, HTR1A, HTR1B, HTR2A, HTR2B, HTR2C, HTR3A, HTR3B, HTR3C, HTR7, HTRA1, HTRA2, HTT, HUWE1, HYAL1, HYDIN, HYLS1, IAPP, ICAM1, ICAM4, ICK, ICOS, ID3, ID4, IDH1, IDH2, IDH3B, IDO1, IDS, IDUA, IER3IP1, IFIH1, IFITM3, IFITM5, IFNAR1, IFNAR2, IFNG, IFNGR1, IFNGR2, IFNL4, IFRD1, IFT122, IFT140, IFT172, IFT43, IFT80, IFT88, IGBP1, IGF1, IGF1R, IGF2, IGF2BP2, IGF2R, IGFALS, IGFBP7, IGHMBP2, IGLL1, IGSF1, IHH, IKBIP, IKBKAP, IKBKB, IKBKG, IKZF1, IKZF3, IL10, IL10RA, IL10RB, IL11, IL11RA, IL12B, IL12RB1, IL12RB2, IL13, IL17F, IL17RA, IL18RAP, IL1B, IL1R1, IL1RAPL1, IL1RN, IL20RB, IL21R, IL23R, IL2RA, IL2RG, IL3, IL31RA, IL36RN, IL4, IL4R, IL6, IL6R, IL6ST, IL7R, IL9R, ILDR1, ILK, IMMMP2L, IMPAD1, IMPDH1, IMPDH2, IMPG2, INF2, INGI, INHA, INMT, INPP5B, INPP5D, INPP5E, INS, INSL3, INSL6, INSR, INVS, IQCB1, IQGAP2, IQGAP3, IQSEC2, IRAK3, IRAK4, IRF1, IRF4, IRF5, IRF6, IRF8, IRGM, IRS1, IRS2, IRS4, IRX4, IRX5, ISCU, ISL1, ISPD, ITCH, ITGA1, ITGA2, ITGA2B, ITGA3, ITGA6, ITGA7, ITGA9, ITGAM, ITGB1, ITGB2, ITGB3, ITGB4, ITGB6, ITIH4, ITIH6, ITK, ITM2B, ITPA, ITPKC, ITPR1, ITS2N2, IVD, IYD, JAG1, JAG2, JAK2, JAK3, JAM3, JMJD1C, JPH2, JPH3, JRK, JUN, JUNB, JUP, KALI, KALRN, KANK1, KARS, KAT2A, KAT6A, KAT6B, KATNAL2, KBTBD13, KCNA1, KCNA4, KCNA5, KCNA6, KCNAB1, KCNAB2, KCNC3, KCND2, KCND3, KCNE1, KCNE1L, KCNE2, KCNE3, KCNE4, KCNH2, KCNH3, KCNIP4, KCNJ1, KCNJ10, KCNJ11, KCNJ12, KCNJ13, KCNJ2, KCNJ5, KCNJ6, KCNJ8, KCNK18, KCNK3, KCNK9, KCNMA1, KCNMB1, KCNMB4, KCNN2, KCNN3, KCNQ1, KCNQ2, KCNQ3, KCNQ4, KCNQ5, KCNS1, KCNT1, KCNV1, KCNV2, KCTD13, KCTD7, KDM3A, KDM5A, KDM5C, KDM6A, KDM6B, KDR, KEL, KERA, KHDC3L, KHK, KIAA0100, KIAA0196, KIAA0226, KIAA0232, KIAA0319, KIAA1033, KIAA1199, KIAA1279, KIAA1432, KIAA1462, KIAA2022, KIF11, KIF17, KIF1A, KIF1B, KIF21A, KIF22, KIF27, KIF3C,

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KIF5A, KIF7, KIR2DL3, KIRREL3, KISS1, KISS1R, KIT, KITLG, KL, KLC1, KLF1, KLF10, KLF11, KLF6, KLF8, KLHDC8B, KLHL10, KLHL3, KLHL7, KLHL9, KLK1, KLK12, KLK4, KLKB1, KMT2D, KNG1, KPNA1, KRAS, KRIT1, KRT1, KRT10, KRT12, KRT13, KRT14, KRT16, KRT17, KRT18, KRT2, KRT3, KRT37, KRT38, KRT4, KRT5, KRT6A, KRT74, KRT75, KRT8, KRT81, KRT83, KRT85, KRT86, KRT9, KRTAPI_1, KYNU, LICAM, L2HGDH, L3MBTL1, LAMA1, LAMA2, LAMA3, LAMA4, LAMB1, LAMB2, LAMB3, LAMB4, LAMC2, LAMC3, LAMP2, LAMTOR2, LARGE, LASIL, LBR, LCA5, LCAT, LCE3B, LCE5A, LCK, LCN10, LCT, LDB1, LDB3, LDHA, LDHB, LDLR, LDLRAP1, LECT2, LEF1, LEFTY2, LEMD3, LEP, LEPR, LEPRE1, LEPRE1, LETM1, LFNG, LGALS13, LGALS2, LGI1, LGI4, LGR5, LHB, LHCGR, LHFPL5, LHX1, LHX3, LHX4, LHX8, LIAS, LIF, LIFR, LIG1, LIG3, LIG4, LILRA3, LIM2, LIMK1, LINC00470, LINC00523, LINS, LIPA, LIPC, LIPG, LIPH, LIPI, LIPN, LITAF, LLGL1, LMAN1, LMBR1, LMBRD1, LMF1, LMNA, LMNB1, LMNB2, LMO2, LMO4, LMTK3, LMX1B, LOC100271832, LOC100996485, LOC338817, LOR, LOX, LOXHD1, LOXL1, LPA, LPAR6, LPIN1, LPIN2, LPIN3, LPL, LPP, LRAT, LRBA, LRFN5, LRIT3, LRP1, LRP1B, LRP2, LRP4, LRP5, LRP6, LRP8, LRPPRC, LRRC4, LRRC6, LRRC8A, LRRFIP2, LRR1Q1, LRRK2, LRRTM1, LRSAM1, LRTOMT, LTBP1, LTBP2, LTBP3, LTBP4, LTBR, LTC4S, LTNI, LUM, LY96, LYN, LYST, LYZ, LZTFL1, LZTS1, MACROD2, MAD1L1, MADD, MAEA, MAF, MAFA, MAFB, MAGEE2, MAGEL2, MAGI2, MAGT1, MAK, MALT1, MAML2, MAMLD1, MANIA2, MAN1B1, MAN2A1, MAN2B1, MANBA, MAOA, MAOB, MAP2, MAP2K1, MAP2K2, MAP2K3, MAP2K4, MAP2K7, MAP3K1, MAP3K14, MAP3K15, MAP3K8, MAP6, MAP7D1, MAPK1, MAPK10, MAPK8IP1, MAPT, MARS2, MARVELD2, MASP1, MASP2, MASTL, MAT1A, MATN3, MATR3, MAX, MBD1, MBD3, MBD4, MBD5, MBL2, MBNL1, MBOAT7, MBTPS2, MC1R, MC2R, MC3R, MC4R, MCC, MCCC1, MCCC2, MCEE, MCFD2, MCHR1, MCM4, MCM5, MCM6, MCM9, MCOLN1, MCPH1, MDGA2, MDK, MDM2, MDN1, ME2, MECOM, MECP2, MED12, MED13L, MED17, MED23, MED25, MEF2A, MEF2C, MEFV, MEGF10, MEGF11, MEGF8, MEIS1, MEN1, MEPE, MERTK, MESDC2, MESP2, MEST, MET, MFAP4, MFF, MFGES, MFN2, MFRP, MFSD8, MGAT1, MGAT2, MGAT3, MGAT4A, MGAT4B, MGAT4C, MGAT5, MGAT5B, MGLL, MGMT, MGP, MGST2, MGST3, MICA, MID1, MIF, MINPP1, MIP, MIPOL1, MIR182, MIR183, MIR96, MITF, MKKS, MKL1, MKS1, MLC1, MLH3, MLLT10, MLLT11, MLLT3, MLPH, MLXIPL, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MME, MMEL1, MMP1, MMP13, MMP14, MMP2, MMP20, MMP3, MMP8, MMP9, MNI, MNX1, MOCOS, MOCS1, MOCS2, MOG, MOGS, MOK, MPC1, MPDU1, MPDZ, MPG, MPHOSPH8, MPI, MPL, MPLKIP, MPO, MPP6, MPST, MPV17, MPZ, MR1, MRAP, MRC1, MRE11A, MREG, MRPL3, MRPL48, MRPS16, MRPS22, MRRF, MS4A1, MS4A12, MS4A2, MSH3, MSMB, MSMO1, MSRI, MSRA, MSRB3, MST1, MST1R, MSTN, MSX1, MSX2, MTA1, MTA2, MTAP, MTFMT, MTHFD1, MTHFR, MTHFS, MTM1, MTMR14, MTMR2, MTMR9, MTNR1A, MTNR1B, MTO1, MTPAP, MTR, MTRR, MTSS1, MTPP, MUC5B, MUC7, MURC, MUS81, MUSK, MUT, MVK, MXI1, MYB, MYBPC1, MYBPC3, MYC, MYCN, MYD88, MYF5, MYF6, MYH11, MYH14, MYH2, MYH3, MYH6, MYH7, MYH8, MYH9, MYL1, MYL2, MYL3, MYLK, MYLK2, MYO15A, MYO1A, MYO1C, MYO1D, MYO1E, MYO1F, MYO3A, MYO5A, MYO5B, MYO5C, MYO6, MYO7A, MYO7B, MYO9B, MYOC, MYOCD, MYOD1, MYOM1, MYOT, MYOZ2, MYPN, MYT1, MYT1L, NAA10, NAALADL2, NAGA, NAGLU, NAGPA, NAGS, NAIP, NAPRT1, NARS2, NAT1, NAT2, NAT8L, NAV2, NBAS, NBEA, NBEAL2, NBPF1, NCALD, NCAPD2, NCF2, NCF4, NCKAP1, NCOA1, NCOA4, NCR3, NCS1, NCSTN, NDE1, NDN, NDOR1, NDP, NDRG1, NDST1, NDST2, NDST3, NDST4, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA4, NDUFA6, NDUFA8, NDUFA9, NDUFAF1, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF7, NDUFB1, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS5, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NDUFV3, NEB, NEBL, NEDD4,

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NEDD4L, NEFH, NEFL, NEFM, NEGR1, NEIL1, NEIL2, NEK1, NEK8, NEU2, NEUROD1, NEUROG3, NEXN, NF1, NF2, NFATC2, NFATC3, NFATC4, NFE2L1, NFIA, NFIB, NFIX, NFKB2, NFKBIA, NFKBIZ, NFU1, NGF, NGLY1, NHEJ1, NHLRC1, NHS, NICN1, NIN, NINJ1, NIPAI, NIPAL4, NIPBL, NIPSNAP1, NIPSNAP3A, NKAIN2, NKX2_1, NKX2_5, NKX2_6, NKX3_1, NKX3_2, NLGN1, NLGN2, NLGN3, NLGN4X, NLGN4Y, NLRP1, NLRP12, NLRP14, NLRP2, NLRP3, NLRP7, NLRX1, NME1, NME5, NME7, NME8, NMNAT1, NMT2, NMU, NNT, NOBOX, NOD1, NOD2, NODAL, NOG, NOL3, NOP10, NOP56, NOS1, NOSIAP, NOS2, NOS3, NOTCH1, NOTCH2, NOTCH3, NOX3, NPAS2, NPAS3, NPAT, NPC1, NPC1L1, NPC2, NPFFR2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NPL, NPM1, NPPA, NPPC, NPR2, NPSR1, NPY, NPY1R, NPY2R, NQO1, NR0B1, NR0B2, NR1H2, NR1H3, NR1H4, NR1I2, NR1I3, NR2E1, NR2E3, NR2F1, NR2F2, NR3C1, NR3C2, NR4A1, NR4A2, NR4A3, NR5A1, NRAS, NRCAM, NRG1, NRL, NRP2, NRTN, NRXN1, NRXN2, NRXN3, NSD1, NSDHL, NSMF, NSUN2, NSUN7, NT5C3A, NT5E, NTF3, NTF4, NTHL1, NTNG1, NTRK1, NTRK2, NTRK3, NUA1, NUBPL, NUDC, NUDT1, NUMA1, NUP155, NUP214, NUP62, NXF5, NXNL1, NYX, OAS1, OAS2, OAT, OBSCN, OBSL1, OCA2, OCLN, OCRL, ODC1, OFD1, OGG1, OGT, OLFM2, OLIG2, OLR1, OMG, OPA1, OPA3, OPCML, OPHN1, OPLAH, OPN1SW, OPRL1, OPRM1, OPTC, OPTN, OR10X1, OR13G1, OR1B1, OR51G1, OR52H1, OR52N4, OR5AC2, OR5H6, OR8K3, ORAI1, ORC1, ORC4, ORC6, OSMR, OSTM1, OTC, OTOA, OTOF, OTOG, OTOGL, OTX2, OVCH2, OXCT1, P2RX1, P2RX5, P2RX7, P2RY12, P2RY4, PABPC4L, PACRG, PACS1, PADI4, PAFAH1B1, PAFAH1B3, PAH, PAK3, PALLD, PAMR1, PANK2, PAPSS2, PARD3B, PARK2, PARK7, PARL, PARP1, PASK, PAX1, PAX2, PAX3, PAX4, PAX5, PAX6, PAX7, PAX8, PAX9, PBX1, PC, PCBD1, PCBP3, PCCA, PCCB, PCDH11X, PCDH15, PCDH18, PCDH19, PCDHA1, PCDHA10, PCDHA13, PCDHB4, PCK2, PCLO, PCM1, PCMT1, PCNT, PCOLCE, PCP4, PCSK1, PCSK5, PCSK9, PDCD1, PDCD10, PDE11A, PDE4B, PDE4D, PDE6A, PDE6B, PDE6C, PDE6G, PDE6H, PDE8B, PDGFB, PDGFRA, PDGFRB, PDGFRL, PDHA1, PDHB, PDHX, PDK1, PDLIM3, PDLIM4, PDP1, PDPK1, PDSS1, PDSS2, PDX1, PDYN, PDZD7, PECCR, PEPD, PER1, PER2, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PFN1, PGAM2, PGAM5, PGK1, PGM1, PGR, PGRMC1, PHB, PHEX, PHF11, PHF2, PHF3, PHF6, PHF8, PHGDH, PHIP, PHKA1, PHKA2, PHKB, PHKG2, PHLPP2, PHOX2A, PHOX2B, PHYH, PICALM, PICK1, PIF1, PIGA, PIGL, PIGM, PIGN, PIGO, PIGR, PIGV, PIGZ, PIK3CA, PIK3CB, PIK3CD, PIK3R1, PIK3R2, PIK3R5, PIKFYVE, PIM1, PIN1, PINK1, PIP5K1C, PITPNA, PITPNM3, PITX1, PITX2, PITX3, PKD1, PKD2, PKHD1, PKLR, PKM, PKN3, PKP1, PKP2, PLA2G10, PLA2G2A, PLA2G4A, PLA2G5, PLA2G6, PLA2G7, PLAG1, PLAGL1, PLAU, PLCB1, PLCB4, PLCD1, PLCE1, PLCG2, PLCZ1, PLD2, PLEC, PLEKHG4, PLEKHG5, PLG, PLIN1, PLOD1, PLOD2, PLOD3, PLP1, PLP2, PLSCR3, PLTP, PML, PMM2, PMP22, PMS1, PNKD, PNKP, PNLIP, PNMT, PNP, PNPLA1, PNPLA2, PNPLA6, PNPO, PNPT1, POC1A, POF1B, POFUT2, POGZ, POLB, POLD1, POLE2, POLG, POLG2, POLH, POLL, POLR1C, POLR1D, POLR3A, POLR3B, POLRMT, POMC, POMGNT1, POMP, POMT1, POMT2, PON1, PON2, PON3, POP1, POR, PORCN, POSTN, POU1F1, POU3F4, POU4F3, POU5F1, POU6F2, PPARA, PPARG, PPARGC1B, PPIB, PPM1B, PPM1D, PPM1G, PPM1K, PPOX, PPP1R12B, PPP1R17, PPP1R1A, PPP1R3A, PPP1R3C, PPP2R1B, PPP2R2A, PPP2R2B, PPP2R2C, PPP3CA, PPP3R1, PPT1, PQBP1, PRAF2, PRB1, PRB3, PRB4, PRCC, PRCD, PRCP, PRDM16, PRDM5, PRDM9, PREPL, PRF1, PRG4, PRICKLE1, PRICKLE2, PRKACA, PRKAG2, PRKAG3, PRKAR1A, PRKCA, PRKCG, PRKCH, PRKCSH, PRKDC, PRKG2, PRLH, PRLR, PRM1, PRM2, PRMT10, PRMT3, PRMT7, PRND, PRNP, PROC, PROCR, PRODH, PROK1, PROK2, PROKR1, PROKR2, PROM1, PROPI, PROS1, PROXI, PROZ, PRPF3, PRPF31, PRPF6, PRPF8, PRPH, PRPH2, PRPS1, PRRT2, PRRX1, PRSS1, PRSS12, PRSS3P2, PRSS56, PRSS8, PRTG, PRX, PSAP, PSAT1, PSEN1,

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PSEN2, PSENEEN, PSMA4, PSMA6, PSMB8, PSMC3IP, PSMD7, PSPH, PSPN, PSTPIP1, PSTPIP2, PTAFR, PTC1, PTCH1, PTCH2, PTCHD1, PTCHD3, PTF1A, PTGDR, PTGER2, PTGER4, PTGIR, PTGIS, PTGS1, PTGS2, PTH, PTH1R, PTHLH, PTK7, PTPN1, PTPN11, PTPN12, PTPN14, PTPN22, PTPN6, PTPRC, PTPRJ, PTPRK, PTPRN2, PTPRO, PTPRQ, PTPRT, PTRF, PTS, PUS1, PVRL1, PVRL4, PXDN, PYCRI, PYGL, PYGM, PYY, PZP, QDPR, QKI, RAB11FIP5, RAB18, RAB23, RAB25, RAB27A, RAB27B, RAB2A, RAB39B, RAB3GAP1, RAB3GAP2, RAB7A, RAB7L1, RABGGTA, RABL6, RAC1, RAC2, RAD21, RAD21L1, RAD51, RAD51B, RAD52, RAD54B, RAD54L, RAF1, RAG1, RAG2, RAI1, RALGAP1, RALGDS, RANBP2, RANGRF, RAPIGDS1, RAPSN, RARA, RARS2, RASA1, RASGEF1B, RASGRP1, RASGRP2, RASSF5, RAX, RAX2, RB1, RB1CC1, RBBP8, RBOX1, RBL1, RBL2, RBM10, RBM15, RBM20, RBM28, RBMXL2, RBP3, RBP4, RBPJ, RC3H1, RCBTB1, RCHY1, RD3, RDH12, RDH5, RDH8, RDX, RECQL4, REEP1, REL, RELN, REN, REPS2, RET, RETN, RFC2, RFT1, RFX2, RFX5, RFX6, RFX8, RFXANK, RFXAP, RGL1, RGMA, RGR, RGS2, RGS5, RGS7, RGS9, RGS9BP, RHAG, RHBDF2, RHCE, RHD, RHO, RHOG, RHOH, RHPN2, RIMS1, RIMS2, RIMS3, RIN2, RIOK2, RIPK4, RIT2, RLBP1, RMDN2, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASEL, RNASET2, RNF113A, RNF135, RNF139, RNF168, RNF170, RNF212, RNF213, RNF219_AS1, RNF6, ROBO1, ROBO2, ROBO3, ROCK1, ROGDI, ROM1, ROR2, RORA, ROS1, RP1, RP1L1, RP2, RP9, RPA1, RPA4, RPE65, RPGR, RPGRIP1, RPGRIP1L, RPIA, RPL10, RPL11, RPL24, RPL35A, RPL36, RPL38, RPL5, RPL6, RPN2, RPS15, RPS17L, RPS19, RPS24, RPS26, RPS5, RPS6KA3, RPS6KL1, RPSAP52, RPTOR, RRAS2, RRH, RRM2B, RS1, RSC1A1, RSPH4A, RSPH9, RSPO1, RSPO4, RSRC1, RTN2, RTN4R, RTTN, RUNX1, RUNX2, RUNX3, RUVBL1, RXFP2, RXRA, RYK, RYR1, RYR2, SAA1, SACS, SAG, SAGE1, SALL1, SALL4, SAMD9, SAMHD1, SAR1B, SARDH, SARS2, SART3, SAT1, SATB2, SATL1, SBF2, SC5D, SCARB1, SCARB2, SCARF2, SCGB1A1, SCGB3A2, SCLT1, SCN10A, SCN11A, SCN1A, SCN1B, SCN2A, SCN2B, SCN3A, SCN3B, SCN4A, SCN4B, SCN5A, SCN7A, SCN8A, SCN9A, SCNN1A, SCNN1B, SCNN1D, SCNN1G, SCO1, SCO2, SCP2, SCRIB, SCUBE2, SDC3, SDCCAG8, SDHA, SDHAF1, SDHAF2, SDHB, SDHC, SDHD, SEC23A, SEC23B, SEC63, SECISBP2, SELE, SELL, SELP, SELPLG, SEMA3A, SEMA3E, SEMA4A, SEMA4C, SEMA4G, SEMA6D, SEMA7A, SEMG1, SEPNI1, SEPSECS, SEPT12., SEPT5_GP1BB, SEPT7, SERAC1, SERPINA1, SERPINA10, SERPINA3, SERPINA6, SERPINA7, SERPINB11, SERPINB5, SERPINB6, SERPINC1, SERPIND1, SERPINE1, SERPINF1, SERPINF2, SERPING1, SERPINH1, SERPINI1, SERPINI2, SESN2, SETBP1, SETD2, SETD8, SETX, SEZ6, SEZ6L, SEZ6L2, SF3B1, SF3B4, SFRP1, SFTPA1, SFTPA2, SFTPB, SFTPC, SFTPD, SGCA, SGCB, SGCD, SGCE, SGCG, POMK(SGK196), SGSH, SH2B1, SH2B3, SH2D1A, SH3BP2, SH3GL1, SH3PXD2B, SH3TC2, SHANK2, SHANK3, SHARPIN, SHBG, SHFM1, SHH, SHMT1, SHOC2, SHOX, SHOX2, SHROOM3, SHROOM4, SI, SIAE, SIGLEC12, SIGLEC14, SIGMAR1, SIK3, SIL1, SIM1, SIM2, SIPA1, SIPA1L1, SIRT1, SIRT3, SIX1, SIX2, SIX3, SIX5, SIX6, SKI, SLA2, SLC10A1, SLC10A2, SLC11A1, SLC11A2, SLC12A1, SLC12A3, SLC12A6, SLC13A2, SLC14A1, SLC15A1, SLC16A1, SLC16A12, SLC16A2, SLC17A3, SLC17A5, SLC17A8, SLC19A2, SLC19A3, SLC1A1, SLC1A3, SLC1A5, SLC20A2, SLC22A1, SLC22A12, SLC22A18, SLC22A2, SLC22A3, SLC22A4, SLC22A5, SLC22A6, SLC22A9, SLC24A1, SLC24A2, SLC24A5, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A3, SLC25A38, SLC25A39, SLC25A4, SLC26A1, SLC26A10, SLC26A2, SLC26A3, SLC26A4, SLC26A5, SLC26A6, SLC26A9, SLC27A1, SLC27A4, SLC27A5, SLC28A1, SLC28A2, SLC28A3, SLC29A1, SLC29A2, SLC29A3, SLC2A1, SLC2A10, SLC2A2, SLC2A4, SLC2A9, SLC30A10, SLC30A2, SLC30A5, SLC30A8, SLC31A1, SLC33A1, SLC34A1, SLC34A2, SLC34A3, SLC35A1, SLC35C1, SLC35D1, SLC35F3, SLC35G2, SLC36A2, SLC37A4, SLC39A12, SLC39A13, SLC39A4, SLC3A1, SLC40A1, SLC41A1, SLC44A2, SLC45A2, SLC46A1, SLC47A1, SLC47A2, SLC4A1, SLC4A10, SLC4A11, SLC4A3, SLC4A4, SLC4A7,

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TSHB, TSHR, TSHZ1, TSLP, TSPAN12, TSPAN17, TSPAN7, TSPEAR, TSPO, TSPYL1, TSSC4, TSSK4, TST, TTBK2, TTC19, TTC21B, TTC37, TTC8, TTI2, TLL1, TTN, TTPA, TTR, TUB, TUBA1A, TUBA8, TUBB1, TUBB2B, TUBB3, TUBGCP5, TUBGCP6, TUFM, TULP1, TUSC3, TWIST1, TWIST2, TXNIP, TXNRD2, TYK2, TYMP, TYMS, TYR, TYRO3, TYROBP, TYRP1, UACA, UBA1, UBAC2, UBE2A, UBE2B, UBE2NL, UBE3A, UBE3C, UBIAD1, UBQLN2, UBR1, UBR3, UBR7, UCHL1, UCN, UCP1, UCP2, UCP3, UFD1L, UGCG, UGGT1, UGGT2, UGT1A1, UGT1A10, UGT1A3, UGT1A4, UGT1A5, UGT1A6, UGT1A7, UGT1A8, UGT1A9, UGT2A1, UGT2B10, UGT2B17, UGT2B4, UGT2B7, UGT8, UIMC1, ULK4, UMOD, UMPS, UNC119, UNC13D, UNC5C, UNC5CL, UNC80, UNC93A, UNC93B1, UNG, UNKL, UPB1, UPF3B, UPK3A, UQCRB, UQCRFS1, UQCRQ, UROC1, UROD, UROS, USB1, USF1, USH1C, USH1G, USH2A, USP1, USP15, USP26, USP3, USP46, USP9X, USP9Y, UST, UTF1, UTP14C, UTRN, UVSSA, VAMP7, VANGL1, VANGL2, VAPB, VAX1, VCAM1, VCAN, VCL, VCP, VCX3A, VDR, VEGFA, VEGFC, VHL, VIM, VIPAS39, VKORC1, VLDLR, VMA21, VNN1, VPS13A, VPS13B, VPS33B, VPS35, VPS37A, VPS54, VRK1, VSIG4, VSX1, VSX2, VWF, WAS, WASF3, WBSCR17, WDFY3, WDCP, WDR11, WDR13, WDR19, WDR35, WDR36, WDR4, WDR45, WDR45B, WDR62, WDR65, WDR72, WDR81, WFS1, WHSC1, WIPF1, WISP3, WNK1, WNK4, WNT10A, WNT10B, WNT3, WNT4, WNT5A, WNT7A, WRAP53, WRN, WT1, WWC1, WWOX, WWTR1, XBP1, XDH, XG, XIAP, XK, XKR6, XPA, XPC, XPNPEP2, XPNPEP3, XRCC1, XRCC2, XRCC3, XRCC4, XRCC5, XRCC6, XYLT1, XYLT2, YARS, YARS2, YBX2, YWHAE, YY1, ZAN, ZAP70, ZBTB16, ZBTB18, ZBTB24, ZBTB40, ZBTB41, ZC3H14, ZCCHC12, ZCCHC13, ZCCHC8, ZDHHC15, ZDHHC17, ZDHHC2, ZDHHC9, ZEB1, ZEB2, ZFAT, ZFH3, ZFP36, ZFP36L1, ZFP36L2, ZFP69, ZFP90, ZFPM2, ZFYVE26, ZFYVE27, ZHX3, ZIC2, ZIC3, ZIC4, ZKSCAN5, ZMPSTE24, ZMYM3, ZNF175, ZNF213, ZNF335, ZNF365, ZNF41, ZNF419, ZNF420, ZNF423, ZNF433, ZNF469, ZNF480, ZNF513, ZNF526, ZNF592, ZNF644, ZNF674, ZNF711, ZNF750, ZNF75D, ZNF80, ZNF81, ZBPB, ZBPB2.

- 4) HLA-B*27, HLA-DQA1, HLA-DQB1
- 5) Detekované mutace v genu *CFTR*: CFTRdele2,3, E60X, P67L, G85E, 394delTT, 444delA, R117C, R117H, Y122X, 621+1G>T, 711+1G>T, L206W, 1078delT, R334W, R347P, R347H, A455E, I507del, F508del, 1677delTA, V520F, 1717-1G>A, G542X, S549R(T>G), S549N, G551D, R553X, R560T, 1811+1,6kbA>G, 1898+1G>A, 2143delT, 2184delA, 2347delG, W846X, 2789+5G>A, Q890X, 3120+1G>A, 3272-26A>G, R1066C, Y1092X(C>A), M1101K, D1152H, R1158X, R1162X, 3659delC, 3849+10kbC>T, S1251N, 3905insT, W1282X, N1303K
- 6) Detekované mutace pomocí metody fragmentační analýzy v genu *BRCA1*: c.1881_1884del4, c.1938_1947del10, c.1953_1956del4, c.1961delA, c.2068delA, c.2411_2412del2, c.2488_2497dupAAGTATCCAT, c.3018_3021del4, c.3642_3643del2, c.3700_3704delGTAAA, c.3756_3759del4, c.4065_4068del4; v genu *BRCA2*: c.7913_7917del5, c.9403delC, c.9435_9436del2
- 7) Vyšetřované geny viru SARS-CoV-2: *Endornase, Spike*