# List of activities within the flexible scope of accreditation

**Accredited Body**: GENvia, s.r.o.

**CAB Name**: Laboratory of Medical Genetics

**CAB Number**: 8006

**Certificate of Accreditation No.**: 118/2024

**Field of Accreditation**: Medical Laboratory - ČSN EN ISO 15189 ed. 3:2023

**Updated**: 12.07. 2024

**Examinations:**

| Ordinal Number | Analyte/ parameter/diagnostics | Principle of examination | Identification of method procedure/ equipment | Examined material | Degrees of freedom[1] |
|---|---|---|---|---|---|
| **802 – Medical Microbiology** | | | | | |
| 1. | Detection of nucleic acids of infectious agents | Real-Time PCR | SOP26 – version 3;<br>BPP65 – version 2;<br>BPP66 – version 2;<br>BPP67 – version 2;<br>BPP68 – version 2;<br>BPP69 – version 2;<br>RotorGeneQ;<br>QuantStudio 7 Pro | Nasopharyngeal, oropharyngeal and nasal swab, saliva, sputum, oral cavity lavage, tracheal aspirate, BAL | A, B |

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|---|---|---|---|---|---|
| **816 – Medical Genetics Laboratory** | | | | | |
| 1. | Examination of constitutional karyotype | Conventional cytogenetic analysis | SOP01– version 3;<br>BPP01 – version 3;<br>BPP02 – version 3;<br>BPP07 – version 3;<br>BPP10 – version 3;<br>BPP19 – version 3;<br>BPP37 – version 3;<br>BPP38 – version 3;<br>SOP02 – version 3;<br>BPP03 – version 3;<br>BPP04 – version 3;<br>BPP07 – version 3;<br>BPP10 – version 3;<br>BPP37 – version 3;<br>BPP38 – version 3;<br>SOP03 – version 3;<br>BPP12 – version 3;<br>BPP13 – version 3;<br>BPP14 – version 3;<br>BPP07 – version 3;<br>BPP10 – version 3;<br>BPP37 – version 3;<br>BPP38 – version 3 | Amniotic fluid, fetal blood, peripheral blood, chorion, aborted tissue | A, B |
| 2. | Examination of constitutional chromosomal aberrations | FISH | SOP04 – version 3;<br>BPP05 – version 3;<br>BPP06 – version 3;<br>BPP07 – version 3;<br>BPP37 – version 3;<br>BPP38 – version 3 | Amniotic fluid, fetal blood, peripheral blood, chorion, aborted tissue, blastomere | A, B |
| 3. | Examination of germline genome variants | PCR with fragment analysis | SOP11 – version 7;<br>BPP34 – version 7;<br>SOP12 – version 8;<br>BPP35 – version 7;<br>SOP16 – version 3;<br>BPP28 – version 4;<br>SOP22 – version 3;<br>BPP56 – version 4;<br>ABI 3130 | Biological material containing genomic DNA | A, B, C |
| 4. | Examination of germline genome variants | QF PCR | SOP14 – version 8;<br>BPP44 – version 4;<br>BPP64 – version 4;<br>ABI3130 | Biological material containing genomic DNA | A, B, C |

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|---|---|---|---|---|---|
| 5. | Examination of germline genome variants | aCGH | SOP20 – version 4;<br>BPP47 – version 3;<br>SurePrint G3 Unrestricted CGH ISCA v2, 8x60K;<br>Microarray scanner Surescan | Biological material containing genomic DNA | A, B |
| 6. | Examination of germline genome variants | MLPA | SOP21 – version 9;<br>BPP51 – version 6;<br>ABI3130 | Biological material containing genomic DNA | A, B, C |
| 7. | Examination of germline genome variants | NGS-MPS | SOP24 – version 6;<br>BPP72-01-version 2;<br>BPP72-03-version 2;<br>BPP72-04-version 2;<br>BPP72-05-version 1;<br>BPP72-06-version 1;<br>MiSeq – Illumina | Biological material containing genomic DNA | A, B, C |
| 8. | Examination of germline genome variants | Real-Time PCR | SOP13 – version 5;<br>BPP36 – version 8;<br>BPP76 – version 1;<br>BPP77 – version 1;<br>BPP78 – version 1;<br>RotorGeneQ;<br>QuantStudio 7 Pro | Biological material containing genomic DNA | A, B, C |
| 9. | Examination of germline genome variants | Sanger sequencing | SOP23 – version 8;<br>BPP41 – version 7;<br>BPP60 – version 5;<br>BPP74 – version 3;<br>BPP75 – version 2;<br>ABI3130 | Biological material containing genomic DNA | A, B, C |
| 10. | Examination of germline genome variants | PCR with electrophoretic detection | SOP25 – version 5;<br>BPP62 – version 4 | Biological material containing genomic DNA | A, B, C |

## Specification of the scope of accreditation:

| Field Nr. / Ordinal Number | Detailed information on activities within the scope of accreditation |
|---|---|
| 802/1 | RNA SARS-CoV-2 |

# List of activities within the flexible scope of accreditation

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| 816/3 | CFTR gene mutations: R347H, R347P, 2789+5G>A, 3120+1G>A, 711+1G>T, R334W, I507del, F508del, 3849+10kbC>T, 1677delTA, 1078delT, V520F, L206W, W1282X, R560T, 2347delG, Q890X, R553X, G551D, S549R, S549N, M1101K, G542X, 3905insT, Y1092X, S1251N, 444delA, 1811+1.6kbA>G, 1717-1G>A, R117H, R117C, N1303K, Y122X, 394delTT, G85E, R1066C, 1898+1G>A, W846X, 2184delA, D1152H, CFTRdele2_3(21kb), P67L, 2143delT, E60X, 3659delC, 3272-26A>G, 621+1G>T, A455E, R1162X, R1158X.<br>AZF microdeletion on Y chromosome; Examination of UGT1A1*28 polymorphism;<br>Expansion of CGG repeat in *FMR1* gene |
| 816/4 | Examination of chromosomes 13, 15, 16, 18, 21, 22, X and Y |
| 816/6 | *ATM1, ATM2, BRCA1, CATSPER2, CFTR, COL1A1, COL1A2, DHCR7, CHEK2, MLH1, MSH2, MSH6, MUTYH, OTOA, PMS2, PTEN, SMN1, SMN2, STK11, STRC, SHOX* |
| 816/7 | Examination of collagen: *COL1A1, COL1A2,*<br>AR panel genes: *AR, INSL3, INSL3R, SRY, SOX9, DHH, NR5A1, MAP3K1, ZFPM2, NR2F2,*<br>Genes and polymorphisms in the Small Panel genes (which includes, among others, the above):<br>*NR0B1, cbx2, AKR1C2, AKR1C4, BMP1, CCDC134, CREB3L1, CRTAP, FKBP10, IFITM5, KDELR2, MBTPS2, MESD, P3H1, PPIB, SERPINF1, SERPINH1, SP7, SPARC, TENT5A, TMEM38B, WNT1, DHCR7, FGFR3, RPE65, ACE, ACTN3, ADORA2A, ADRB2, ADRB2, AGT, AMPD1, BCO1, COL1A1, COL5A1,CREM, CRP, CYP1A2, CYP2R1, FTO, FUT2, GALNT13, GC, GDF5, GSTT1, HFE, HIF1A, IL6, IL6R, LINC01721, MMP3, MTHFD1, MTHFR, NOS3, NRF2, PCBP3, PEMT, PPARA, PPARG, PPARGC1A, SLC30A8, SOD2, TCF7L2, TF, TMPRSS6, TNF, TNFAIP8, VDR, VEGFA, WWP2, HLA-B; DQA1, NOD2, DRB1, HNF4A, RNF186, SERPINA1, DQB1, CFTR, HFE, PNPLA3, PTPN22, INS, TMEM18, CDKAL1, IGF2BP2, MC4R, SLC30A8, PPARG, KCNJ11, HHEX, TCF7L2, FTO, VDR, CASC8, BRCA1, PARP1, APOE, ABCA7, LRP1, HDAC9, PITX2, ZFHX3, MIA3, PHACTR1, CDKN2B-AS1, MRAS, LPA, HNF1A, CELSR2, CXCL12, LPL, SMARCA4, F5, ABO, F2, ARMS2, CFH, WNT16, LRP5*<br>GENkomp genes: *ABCA3, ABCA4, ABCC6, ABCC8, ABCD1, ACADM, ACADS, ACADVL, ACAT1, ADGRV1, AFF2, AGA, AGL, AGXT, AHI1, AIRE, ALDOB, ALPL, ANO10, AR, ARG1, ARSA, ARX, ASL, ASPA, ASS1, ATM, ATP7B, BBS1, BBS2, BCKDHA, BCKDHB, BCS1L, BLM, BTD, CAPN3, CBS, CC2D2A, CCDC88C, CDH23, CEP290, CFTR, CLCN1, CLN3, CLRN1, CNGB3, COL4A3, COL4A5, COL7A1, CPT1A, CPT2, CTNS, CYP11A1, CYP21A2, CYP27A1, CYP27B1, DBT, DHCR7, DHDDS, DLD, DMD, DYNC2H1, ELP1, ERCC2, EVC2, F11, F2, F5, F8, F9, FAH, FANCA, FANCC, FKRP, FKTN, FMO3, FMR1, FSHR, FXN, G6PC1, GAA, GALC, GALT, GBA1, GBE1, GCDH, GJB2, GLA, GLB1, GNPTAB, GRIP1, HADHA, HBA1, HBA2, HBB, HEXA, HFE, HLA-B, HPS1, HPS3, CHRNE, IDUA, IL2RG, IVD, L1CAM, LAMB3, LRP2, MCCC1, MCCC2, MCOLN1, MCPH1, MEFV, MID1, MLC1, MMACHC, MMUT, MTHFR, MTM1, MVK, MYO7A, NAGA, NAGLU, NBN, NEB, NPC1, NPC2, NPHS1, NPHS2, NR0B1, OCA2, OTC, PAH, PCDH15, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX2, PEX6, PEX7, PKHD1, PLP1, PMM2, POLG, PPT1, PRF1, PROP1, RARS2, RNASEH2B, RPGR, RS1, SBDS, SCO2, SERPINA1, SGSH, SLC19A3, SLC25A20, SLC26A2, SLC26A4, SLC37A4, SLC6A8, SMN1, SMPD1, SRY, STS, TF, TGM1, TMEM216, TNXB, TPP1, TSHR, TYR, USH1C, USH2A, XPC*<br>Hereditary tumor predisposition: *AIP; ALK; APC; APEX1; ATM; ATMIN; ATR; ATRIP; AURKA; AXIN1; BABAM1; BAP1; BARD1; BLM; BMPR1A; BRAP; BRCA1; BRCA2; BRCC3; BRE; BRIP1; BUB1B; C11orf30; C19orf40; casp8; CCND1; CDC73; CDH1; CDK4; CDKN1B; CDKN1C; CDKN2A; CEBPA; CEP57; CLSPN; CSNK1D; CSNK1E; CWF19L2; CYLD; DCLRE1C; DDB2; DHFR; DICER1; DIS3L2; DMBT1; DMC1; DNAJC21; DPYD; EGFR; EPCAM; EPHX1; ERCC1; ERCC2; ERCC3; ERCC4;* |

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| | *ERCC5; ERCC6; ESR1; ESR2; EXO1; EXT1; EXT2; EYA2; EZH2; FAM175A; FAM175B; FAN1; FANCA; FANCB; FANCC; FANCD2; FANCE; FANCF; FANCG; FANCI; FANCL; FANCM; FBXW7; FH; FLCN; GADD45A; GATA2; GPC3; GRB7; HELQ; HNF1A; HOXB13; HRAS; HUS1; CHEK1; CHEK2; KAT5; KCNJ5; KIT; LIG1; LIG3; LIG4; LMO1; LRIG1; MAX; MCPH1; MDC1; MDM2; MDM4; MEN1; MET; MGMT; MLH1; MLH3; MMP8; MPL; MRE11A; MSH2; MSH3; MSH5; MSH6; MSR1; MUS81; MUTYH; NAT1; NBN; NCAM1; NELFB; NF1; NF2; NFKBIZ; NHEJ1; NSD1; OGG1; PALB2; PARP1; PCNA; PHB; PHOX2B;PIK3CA; PIK3CG; PLA2G2A; PMS1; PMS2; POLB; POLD1; POLE; PPM1D; PREX2; PRF1; PRKAR1A; PRKDC; PRSS1, PTEN; PTCH1; PTTG2; RAD1; RAD17; RAD18; RAD23B; RAD50; RAD51; RAD51AP1; RAD51B; RAD51C; RAD51D; RAD52; RAD54B; RAD54L; RAD9A; RB1; RBBP8; RECQL; RECQL4; RECQL5; RET; RFC1; RFC2; RFC4; RHBDF2; RINTI, RNF146; RNF168; RNF8; RPA1; RUNX1; SBDS; SDHA; SDHAF2; SDHB; SDHC; SDHD; SETBP1; SETX; SHPRH; SLX4; SMAD4; SMARCA4; SMARCB1; SMARCE1; STK11; SUFU; TCL1A; TELO2; TERF2; TERT; TLR2; TLR4; TMEM127; TOPBP1; TP53; TP53BP1; TSC1; TSC2; TSHR; UBE2A; UBE2B; UBE2I; UBE2V2; UBE4B; UIMC1; VHL; WRN; WT1; XPA; XPC; XRCC1; XRCC2; XRCC3; XRCC4; XRCC5; XRCC6; ZNF350; ZNF365* |
| 816/8 | Thrombophilic mutations - genes: *F2, F5, MTHFR, PAI-1*<br>Ankylosing spondylitis (Bechterew's disease): HLA-B*27<br>A1AT deficiency – *SERPINA* gene<br>Hemochromatosis –*HFE* gene |
| 816/9 | *APC, ATM, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDKN2A, COL1A1, COL1A2, DHCR7, EPCAM, FANCC, FANCM, FGFR3, GJB2, HOXB13, CHEK2, KIT, MLH1, MLH3, MRE11A, MSH2, MSH6, MUTYH, NBN, PALB2, PIK3CA, PMS2, POLD1, POLE, PRSS1, PTEN, RAD51C, RAD51D, RECQL, RECQL4, RINT1, SLX4, SMAD4, SMARCA4, STK11, TP53, VLH, XRCC2*<br>+ genes listed in NGS examination (816/7) (GENkomp, hereditary tumor predispositions, small panel, AR panel) |
| 816/10 | Coeliac sprue: DQA1*02,*05;DQB1*02,*03:02 |

## Explanatory notes:

**1** Established degrees of freedom according to MPA 00-09-..:

A – Flexibility concerning the documented examination/ sample collection procedure

B – Flexibility concerning the technique

C – Flexibility concerning the analytes / parameters

D – Flexibility concerning the examined material

If no degree of freedom is specified, the laboratory cannot apply a flexible approach to the scope of accreditation for this examination.

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|---|---|
| aCGH | Oligonucleotide array comparative genomic hybridization |
| FISH | Fluorescence In Situ Hybridization |
| MLPA | Multiplex Ligation-Dependent Probe Amplification |
| NGS-MPS | Massively Parallel Sequencing |
| QF PCR | Quantitative Fluorescence PCR |

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| PCR | Polymerase Chain Reaction |
| Real-Time PCR | Real-Time PCR |