List of activities within the flexible scope of accreditation

Accredited Body: Cytogenetická laboratoř Brno, s.r.o.
CAB Name: Cytogenetic Laboratory Brno
CAB Number: 8067
Certificate of Accreditation No. 319/2023
Field of Accreditation: Medical Laboratory - ČSN EN ISO 15189:2013
Updated: 03.06.2024

Examinations:

Ordinal Number	Analyte/ parameter/diagnostics	Principle of examination	Identification of method procedure/ equipment	Examined material	Degrees of freedom[1]
816 – Medical Genetics Laboratory					
1.	Examination of constitutional karyotype	Conventional cytogenetic analysis	SOP-C1, Issue 4	Amniotic fluid, peripheral blood, umbilical blood, chorionic villi, tissue of aborted fetus, cultivated tissue culture	A
2.	Examination of germline genome variants	PCR-fragment analysis	SOP-M1, Issue 2; I-15, 18/01/2023; I-19, 15/06/2022; I-28, 15/05/2023	DNA, amniotic fluid, chorionic villi, tissue of aborted fetus, umbilical blood, peripheral blood, buccal swab	A,B,C,D
3.	Examination of germline genome variants	PCR-fragment analysis	SOP-M2, Issue 2; I-19, 15/06/2022	DNA, amniotic fluid, chorionic villi, tissue of aborted fetus, umbilical blood, peripheral blood, buccal swab	A,B,C,D
4.	Examination of germline genome variants	NGS-MPS	SOP-M3, Issue 5; I-19, 15.06.2022; I-28, 15.05.2023 I-36, 01.02.2024 I-40, 09.01.2024	DNA, amniotic fluid, chorionic villi, tissue of aborted fetus, umbilical blood, peripheral blood, buccal swab	A,B,C,D

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Ordinal Number	Analyte/ parameter/diagnostics	Principle of examination	Identification of method procedure/ equipment	Examined material	Degrees of freedom[1]
5.	Examination of germline genome variants	HRM	SOP-M4, Issue 3; I-14, 06/01/2016; I-19, 15/06/2022; I-28, 15/05/2023	DNA, peripheral blood, buccal smear	A,B,C,D
6.	Examination of germline genome variants	MLPA	SOP-M6, Issue 2; I-19, 15/06/2022	DNA, amniotic fluid, chorionic villi, tissue of aborted fetus, umbilical blood, peripheral blood, buccal swab	A,B,C,D
7.	Examination of unbalanced chromosomal aberrations	Comparative genomic hybridization on a biochip	SOP-M7, Issue 4; I-19, 15/06/2022; I-23, 09/11/2016; I-28, 15/05/2023; I-31, 25/04/2023	DNA, amniotic fluid, chorionic villi, tissue of aborted fetus, umbilical blood, peripheral blood, buccal swab	A,B,C,D
8.	Non-invasive prenatal test (NIPT) of genomic variants	NGS-MPS	SOP-M8, Issue 4; I-17, 27/01/2023	Free fetal DNA, maternal peripheral blood, maternal peripheral blood plasma	A,B,C,D
9.	Examination of germline genome variants	dTP-PCR	SOP-M17, Issue 4; I-19, 15/06/2022; I-23, 09/11/2016; I-28, 15/05/2023	DNA, amniotic fluid, chorionic villi, tissue of aborted fetus, umbilical blood, peripheral blood, buccal swab	A,B,C,D
10.	Examination of germline genome variants	RealTime PCR	SOP-M20, Issue 2; I-19, 15/06/2022; I-28, 15/05/2023	DNA, peripheral blood, buccal swab	A,B,C,D

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Specification of the scope of accreditation:

Field Nr. / Ordinal Number	Detailed information on activities within the scope of accreditation
816/2	QF-PCR method using diagnostic kits: • Devyser Resolution 21 v2: for the diagnosis of chromosome 21 aneuploidy • Devyser Complete v2: for the detection of aneuploidies of chromosomes 13, 18, 21, X and Y • Devyser Extend M1 v2: for the detection of aneuploidies of chromosomes 15, 16 and 22
816/3	Testing for the most common mutations in the *CFTR* gene by ARMS method: • Devyser CFTR Core / Devyser CFTR Italia v2 (67M): Traditional nomenclature: *F508del, CFTRdele2,3(21kb), G551D, N1303K, G542X, 1898+1 G>A, 2143delT, R347P, W1282X, 1717-1G>A, R1162X, 3849+10kb C>T, 2184insA, G85E, 621+1G>T, R334W, R553X, 2183AA>G, 2789+5G>A I336K, CFTR-dele2, Q552X, D1152H, R1158X, S549R(A>C), 3272-26A>G, CFTRdele22,23, D110H, 3659delC, Q39X, R117C, 711+1G>T, R347H, I507del, E585X, 2184delA, R1066C, R117H, 1078delT, 1677delTA, R650T, 3120+1G>A, Y1092X(C>A), T388I, L1077P, L1065P, M1V, P5L, Dele17a_18, 852del22, CFTRdele1, D110E, R1066H, G1244E, c.1584+18672A>G, 1259insA, 711+5G>A, R352Q, G178R, D579G, 1898+3A>G, 4382delA, 4016insT, Dele14b_17b, Dele2ins182, Dele22_24, G1349D* HGVS nomenclature: *c.1521_1523delCTT, c.54-5940_273+10250del21080, c.1652G>A. c.3909C>G, c.1624G>T, c.1766+1G>A c.2012delT, c.1040G>C, c.3846G>A, c.1585-1G>A, c.3484C>T, c.3718-2477C>T, c.2052_2053insA, c.254G>A, c.489+1G>T, c.1000C>T, c.1657C>T, c.2051_2052delAAinsG, c.2657+5G>A, c.1007T>A, c.54-1161_164+1603del2875, c.1654C>T, c.3454G>C, c.3472C>T, c.1645A>C, c.3140-26A>G, c.3964-78_4242+577del, c.328G>C, c.3528delC, c.115C>T, c.349C>T, c.579+1G>T, c.1040G>A, c.1519_1521delATC, c.1753G>T, c.2052delA, c.3196C>T, c.350G>A, c.948delT, c.1545_1546delTA, c.1679G>C, c.2988+1G>A, c.3276C>A, c.1013C>T, c.3230T>C, c.3194T>C, c.1A>G, c.14C>T, c.2988+1173_3468+2111del, c.720_741delAGGGAGAATGATGATGAAGTAC, c.4+53+69delins299, c.330C>A, c.3197G>A, c.3731G>A, c.1585-9412A>G(c.1584+18672A>G), c.1127_1128insA, c.579+5G>A, c.1055G>A, c.532G>A, c.1736A>G, c.1753G>T, c.4251delA, c.3884_3885insT, c.2620-674_3367+198del, c.54-5811_164+2186del8108ins182, c.3964-3890_3143delinsTAACT, c.4046G>A* • Devyser CFTR 68: Traditional nomenclature: *711+ 1G>T, 2043delG, 1677delTA, W1282X, R1283M, K710X, 3849+10kbC>T, 2789+5G>A, M1101K, G85E, 3905insT, 1525-1G>A, 2184delA, 3659delC, N1303K, 2184insA, 1812-1G->A, CFTRdele2,3, 2143delT, Y569D, R1162X, A561E, S1251N, P67L, R1158X, 1609delCA, Q493X, E60X, 1898+1G>A, 1898+5G>T, I507del, F508del, V520F, 394delTT, D1152H, V232D, L218X, 621+2T>C, 1717-1G>A, L206W, E92X, 3120+1G>A, G542X, S549N, G551D, 712-1G>T, R553X, 3272-26A>G, R560T, 2183AA>G, R117H, R117C, 1811+1.6kbA>G, 2869insG, Y122X, Q890X, R1066C, R347H, R347P, 1161delC, 1154ins TC, E92K, I336K, R334W, Y1092X (C>A), 621+1G>T, 1078delT, A455E* HGVS nomenclature: *c.579+1G>T, c.1911delG, c.1545_1546delTA, c.3846G>A, c.3848G>T, c.2128A>T, c.3718-2477C>T, c.1519_1523ATCTT, c.2657+5G>A, c.3302T>A, c.254G>A, c.3773_3774insT, c.1393-1G>A, c.2052delA, c.3528delC, c.3909C>G, c.2052_2053insA, c.1680-1G>A, c.54-5940_273+10250del21kbc.2012delT, c.1705T>G, c.3484C>T, c.1682C>A, c.3752G>A, c.200C>T, c.3472C>T, c.1477_1478delCA, c.1477C>T, c.178G>T, c.1766+1G>A, c.1766+5G>T, c.1519_1521delATC, c.1521_1523delCTT, c.1558G>T, c.262_263delTT, c.3454G>C, c.695T>A, c.653T>A, c.489+2T>C, c.1585-1G>A, c.617T>G, c.274G>T, c.2988+1G>A, c.1624G>T, c.1646G>A, c.1652G>A, c.580-1G>T, c.1657C>T, c.3140-26A>G, c.1679G>C, c.2051_2052delAAinsG, c.350G>A, c.349C>T, c.1680-886A>G, c.2737_2738insG, c.366T>A,*

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	c.2668C>T, c.3196C>T, c.1040G>A, c.1040G>C, c.1029delC, c.1022_1023insTC, c.274G>A, c.1007T>A, c.1000C>T, c.3276C>A, c.489+1G>T, c.948delT, c.1364C>A
816/4	• NGS Cardio - list of genes (https://www.cytogenetika.cz/sluzby/dedicna-onemocneni-srdce) *A2ML1, ABCC9, ACTA1, ACTA2, ACTC1, ACTN2, ACVR2B, AKAP9, ANK2, ANKRD1, BAG3, BGN, BRAF, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CALR3, CASQ2, CAV3, CBL, CCDC11, CFC1, CFHR5, CHD7, CITED2, COL1A1, COL1A2, COL3A1, COL4A3, COL4A4, COL4A5, COL5A1, COL5A2, CRELD1, CRYAB, CSRP3, CTF1, CTNNA3, DES, DCHS1, DMD, DOLK , DPP6, DSC2, DSE, DSG2, DSP, DTNA, ELN, EMD, EVC, EVC2, FBN1, FBN2, FHL1, FHL2, FHOD3, FLNA, FKBP14, FKTN, FLNC, FOG2, FOXE3, FOXH1, GAA, GATA4, GATA5, GATA6, GDF1, GJA1, GJA5, GLA, GATAD1, GPD1L, HAND1, HAND2, HCN4, HEY2, HRAS, JAG1, JUP, JPH2, KAT6b, KCNA5, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, KRAS, LAMA2, LAMA4, LAMP2, LDB3, LEFTY2, LMNA, LOX, LZTR1, MAP2K2, MAT2A, MED12, MED13L, MEK1, MFAP5, MIB1, MIB2, MOG1, MRAZ, MYBPC3, MYH6, MYH7, MYH7b, MYH9, MYH11, MYL2, MYL3, MYLK, MYLK2, MYLK3, MYOCD, MYOM1, MYOZ2, MYPN, NEBL, NEXN, NF1, NF2, NKX2-5, NKX2-6, NODAL, NOTCH1, NOTCH2, NOS1AP, NPPA, NRAS, NR2F2, NUP155, OBSCN , PDLIM3, PKP2, PLOD1, PLN, PPP1CB, PPP1R13L, PPCS, PRKAG2, PRKG1, PRDM16, PSEN2, PTPN11, RAF1, RASA1, RASA2, RBM8A, RBM10, RBM20, RIT1, RRAS, RYR1, RYR2, SALL4, SEMA3E, SLC2A10, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCO2, SDHA, SGCD, SHOC2, SHROOM3, SLMAP, SMAD2, SMAD3, SMAD4, SMAD6, SMARCB1, SNTA1, SOS1, SOS2, SPRED1, TAB2, TANGO2, TAZ, TBX1, TBX3, TBX5, TBX20, TCAP, TFAP2B, TECRL, TGFBR1, TGFBR2, TGFBR3, TGFB2, TGFB3, TLL1, TMEM43, TMPO, TNNI3, TNNI3K, TNNC1, TNNT2, TPM1, TRDN, TRIM54, TRIM63, TRPM4, TTN, TTR, TXNRD2, VCL, ZFPM2, ZIC3)* • NGS Rasopathy, a list of genes (https://www.cytogenetika.cz/sluzby/ngs-panel-rasopatie) *PTPN11, SOS1, RAF1, RIT1, BRAF, KRAS, NRAS, MAP2K1, RRAS, CBL, SHOC2, A2ML1, LZTR1, RASA2, SOS2, MAP2K2, HRAS, SPRED1, NF1, NF2, PPP1CB, KAT6b, MRAS, RASA1, SMARCB1* • Heredity - clinical exome (*G:\1_Molekularni laborator\1_VYSETRENI\HEREDITY PANEL\Info\KAPA_Heredity KAPA\HyperCap Heredity_gene_list)* A list of 3332 genes can be provided upon request by the referring physician.
816/5	Examination of 3 thrombophilic mutations (HRM): - mutations in the gene for Factor V (1691 G>A) - mutations in the gene for Factor II (20210G>A) - mutations in the gene for MTHFR (677 C>T)
816/6	Selected diagnoses: • Spinal muscular atrophy – copy numbers of exons 7 and 8 of genes: *SMN1 and SMN2* • Syndrom DiGeorge – gene copy numbers: *PPIL2, SLC25A18, DGCR8, HIRA, SNRPD3, TBX1, MICAL3, CLTCL1, CLDN5, ZNF74, GP1BB, GNAZ, SMARCB1, USP18, TXNRD2, TBX1, RSPH14, KLHL22, TOP3B, HIC2, MED15, IL17RA, RAB36, BID, SNAP29, SMARCB1, LZTR1, RSPH14, CDC45* in 22q11 region • NF1 – copy numbers of exons 1 to 58 of the *NF1* gene
816/7	Format 8x60K, AMADID (18,851 probes in ISCA regions + 40,208 backbone probes) practical median probe spacing is approx. 100 kb (with higher resolution in ISCA regions)
816/8	• Chromosomes 16, 18, 21, X, Y Trisomy test • Trisomy XY test • Trisomy test +
816/9	Expansion of CGG repeat in *FMR1* gene

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816/10	Examination of 3 thrombophilic mutations (real-time PCR) - mutations in the gene for Factor V (1691 G>A) - mutations in the gene for Factor II (20210G>A) - mutations in the gene for MTHFR (677 C>T)

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.

MLPA	Multiplex Ligation-Dependent Probe Amplification
NGS-MPS	Massive parallel sequencing (Next Generation Sequencing – NGS)
HRM	High Resolution Melting Curve Analysis
QF PCR	Quantitative Fluorescence Polymerase Chain Reaction
ARMS	Allele-specific amplification
Real-Time PCR	Real-Time Polymerase Chain Reaction
dTP-dCR	Direct Triplet-Primed PCR method