The Appendix is an integral part of
Certificate of Accreditation No. 245/2021 of 20/04/2021

Accredited entity according to ČSN EN ISO 15189:2013:

Fakultní nemocnice v Motole
IBMG Laboratories
V Úvalu 84/1, 150 06 Praha 5

Examinations:

Ordinal number	Examination procedure name	Examination procedure identification	Examined object
816 - Medical Genetics Laboratory			
1.	Molecular genetic examination of the 50 most common mutations and T(n) variants of the *CFTR* gene by ARMS using a diagnostic assay	IISOP_UBLG_06/2013	Peripheral blood, amniotic fluid and chorionic villi
2.	Molecular genetic examination of thrombophilia mutations by ARMS using a diagnostic assay	IISOP_UBLG_09/2013	Peripheral blood
3.	Molecular genetic examination of aneuploidy and sex determination by QF-PCR using a diagnostic assay	IISOP_UBLG_12/2013	Peripheral blood, umbilical cord blood, amniotic fluid and chorionic villi
4.	Molecular genetic examination of germline mutations in various rare diseases by the Sanger DNA sequencing method	IISOP_UBLG_19/2013	Peripheral blood, amniotic fluid and chorionic villi
5.	Molecular genetic examination of the fragile X syndrome by fragment analysis method using a diagnostic assay	IISOP_UBLG_01/2015	Peripheral blood, amniotic fluid and chorionic villi
6.	Molecular genetic examination of germline mutations of selected genes responsible for rare genetic diseases by Sanger sequencing method	IISOP_UBLG_02/2015	Peripheral blood, blood spots, amniotic fluid and chorionic villi
7.	Examination of DNA copy number changes by MLPA using a diagnostic assay	IISOP_UBLG_01/2017	Peripheral blood, blood spots, and amniotic fluid
8.	Molecular genetic examination of germline mutations of genes associated with syndromes with hereditary predisposition to cancer by massive parallel sequencing using a diagnostic assay	IISOP_UBLG_02/2017	Peripheral blood

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Ordinal number	Examination procedure name	Examination procedure identification	Examined object
9.	Examination of the human genome by oligonucleotide array CGH assay	IISOP_UBLG_03/2015	Peripheral blood, umbilical cord blood, amniotic fluid, chorionic villi and tissue of aborted fetuses
10.	Karyotyping by conventional chromosome staining methods	IISOP_UBLG_16/2013a	Peripheral blood, fetal blood, umbilical cord blood, amniotic fluid, chorionic villi, tissue of aborted fetuses and skin biopsy cells
11.	Examination of chromosomal breaks by conventional chromosome staining methods	IISOP_UBLG_16/2013b	Peripheral blood
12.	Analysis of chromosomal aberrations by fluorescent in-situ hybridization (FISH)	IISOP_UBLG_17/2013	Peripheral blood, fetal blood, umbilical cord blood, amniotic fluid, chorionic villi, tissue of aborted fetuses, skin biopsy cells and buccal swab
IVF Examinations Laboratory			
1.	Complex sperm analysis by their macroscopic assessment and microscopic evaluation	IISOP_UBLG_30/2013	Ejaculate

Explanatory notes:

ARMS - Amplification Refractory Mutation System

QF-PCR - Quantitative Fluorescence-Polymerase Chain Reaction

MLPA - Multiplex Ligation-dependent Probe Amplification

IISOP_UBLG_06/2013
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(T>G), S549N, M1101K, G542X, 3905insT, Y1092X(C>A), S1251N, 444delA, 1811+1.6kbA>G, 1717-1G>A, R117H, R117C,
N1303K, Y122X, 394delTT, G85E, R1066C, 1898+1G>A, W846X, 2184delA, D1152H, CFTRdele2,3, P67L, 2143delT, E60X, 3659delC, 3272-26A>G, 621+1G>T, A455E, R1162X and R1158X, IVS8-5T, IVS8-7T, IVS8-9T

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IISOP_UBLG_09/2013
Factor V Leiden mutation, mutation 20210G> A in the 3´untranslated region of the coagulation factor II gene and variants C677T and 1298A> C in the methylenetetrahydrofolate reductase (MTHFR) gene.

IISOP_UBLG_12/2013
13q12.12 (D13S742), 13q21.32-q21.33 (D13S634), 13q31.1 (D13S628), 13q13.3 (D13S305), 13q22.1 (D13S800), 13q12.2 (D13S252), 13q21.1 (D13S1492), 18q12.3 (D18S978), 18q12.3 (D18S535), 18q22.1 (D18S386), 18q22.1 (D18S386), 18q11.2 (D18S1002), 18p11.31 (D18S976), 18p11.32 (GATA178F11), 18q22.1 (D18S1364), 21q21.3 (D21S1435), 21q21.1 (D21S11), 21q22.3 (D21S1411), 21q22.13 (D21S1444), 21q21.3 (D21S1442), 21q22.2 (D21S2055), Xq21.31/Yp11.31 (DXYS267), Xp22.33/Yp11.31 (DXYS218), Xq26.2 (DXS1187), Xq13.1 (DXS981), Xq26.2-26.3 (XHPRT), Xq27.1-q27.2 (DXS2390), Xp22.2 (AMELX), Yp11.2 (AMELY), Yp11.31/Xp22.1 (ZFY, ZFX), Yp11.31 (SRY), 7q34/Xq13, 3p24.2/Xq21.1

IISOP_UBLG_19/2013
exon 10 of *FGFR3* gene in achondroplasia, exons 13 and 15 of *FGFR3* gene in hypochondroplasia, exons 7, 10, 15 and 19 of *FGFR3* gene in thanatophoric dysplasia, exon 7 of *FGFR3* gene in Muenke craniosynostosis, exon 8 of *FGFR2* gene in Apert syndrome, exons 8 and 10 of *FGFR2* in Crouzon syndrome, Pfeiffer syndrome and nonsyndromic craniosynostosis, entire *VHL* gene in von Hippel-Lindau disease, entire *PANK2* gene in pantotenatkinase neurodegeneration (PKAN), entire *GCH1* gene in dopa-responsive dystonia, entire *AR* gene in androgene insensitivity, entire *SRY* gene in disturbances of sexual development and entire *GJB2* gene in non-syndromic deafness.

IISOP_UBLG_02/2015

CFTR, BRCA1, BRCA2

IISOP_UBLG_01/2017

CFTR, BRCA1, BRCA2

IISOP_UBLG_02/2017

APC, BRCA1, BRCA2, PALB2, TP53, ATM, CDH1, CHEK2, NBN, RAD51C, RAD51D, BARD1, BRIP1, MLH1, MRE11A, MSH2, MSH6, MUTYH, PMS2, PMS2CL, PTEN, RAD50, STK11, EPCAM, FAM175A, PIK3CA and XRCC2