The Appendix is an integral part of
Certificate of Accreditation No. 598/2018 of 15/11/2018

Akreditovaný subjekt podle ČSN EN ISO 15189:2013

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

Examination:

Ordinal number	Exact examination procedure name	Examination procedure identification	Examined object
816 - Medical Genetics Laboratory			
1.	Molecular genetic examination of the F508del CFTR gene mutation by fragment analysis	IISOP_UBLG_01/2013	Peripheral blood, amniotic fluid and chorionic villi
2.	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
3.	Reserved		
4.	Molecular genetic examination of thrombophilia mutations by ARMS using a diagnostic assay	IISOP_UBLG_09/2013	Peripheral blood
5.	Molecular genetic examination of aneuploidy and sex determination by QF-PCR using a diagnostic assay	IISOP_UBLG_12/2013	Peripheral blood, amniotic fluid and chorionic villi
6.	Molecular genetic examination of Y chromosome microdeletions by fragment analysis using a diagnostic assay	IISOP_UBLG_15/2013	Peripheral blood
7.	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
8.	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
9.	Analysis of the human genome by oligonucleotide array CGH assay	IISOP_UBLG_03/2015	Peripheral blood, amniotic fluid, chorionic villi and tissue of aborted fetuses

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Ordinal number	Exact examination procedure name	Examination procedure identification	Examined object
10.	Karyotyping from cultured peripheral and fetal blood cells, amniotic fluid cells, chorionic villi, samples from aborted fetuses or other tissues by conventional chromosome colouring methods	IISOP_UBLG_16/2013	Peripheral blood, fetal blood, amniotic fluid, chorionic villi, tissue of aborted fetuses and skin biopsy cells
11.	Analysis of chromosomal aberrations by fluorescent in-situ hybridization (FISH)	IISOP_UBLG_17/2013	Peripheral blood, fetal blood, amniotic fluid, chorionic villi, tissue of aborted fetuses, skin biopsy cells and buccal swab
12.	Molecular genetic examination of germline mutations of selected genes responsible for rare genetic diseases by Sanger sequencing method	IISOP_UBLG_02/2015	Peripheral blood, amniotic fluid and chorionic villi
13.	Examination of DNA copy number changes by MLPA using a diagnostic assay	IISOP_UBLG_01/2017	Peripheral blood and amniotic fluid
14.	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
Laboratory examination for IVF			
1.	Complex sperm analysis by their macroscopic assessment and microscopic evaluation	IISOP_UBLG_30/2013	Ejaculate

Primary sampling:

	Exact primary sampling procedure name	Primary sampling procedure identification	Primary sample
	Procedure for taking primary samples	IISOP_UBLG_28/2013	Blood

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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 a R1158X, IVS8-5T, IVS8-7T, IVS8-9T (according to legacy nomenclature; www.cftr2.org)

IISOP_UBLG_09/2013

Factor V Leiden mutation, Prothrombin (Factor II) 20210G>A mutation and MTHFR C677T polymorphism

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_15/2013

AZFa: sY86, sY625, sY84, M259

AZFb: sY127, sY131, sY134

AZFc: sY254, sY255, sY15.

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 pantothenate kinase 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

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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 a XRCC2