The Appendix is an integral part of
Certificate of Accreditation No. 154/2015 of 02/03/2015

Accredited entity according to ČSN EN ISO 15189: 2013

Institut reprodukční medicíny a genetiky s.r.o.
Medical Genetics Laboratory
Bělehradská 14, 360 01 Karlovy Vary

Examination:

Ordinal number	Examination procedure name	Examination procedure identification	Examined object
816 - Medical Genetics Laboratory			
1.	Cytogenetic examination of caryotype from biological material	SOP 01	fetal and peripheral blood, amniotic fluid
2.	Examination of chromosome aneuploidies, microdeletions and structural changes by FISH method	SOP 02	fetal and peripheral blood, blastomere, amniotic fluid
3.	A - Molecular genetic examinations of 29 mutations in CFTR gene by Elucigenen 29v2 method B - Additional examination of slavonic mutation of CFTRdele2,3 (21kb) using allele specific PCR reaction and analysis in agarose gel	SOP 03	peripheral blood, buccal smear
4.	Molecular genetic examination of thrombophilic mutations by REAL-time PCR	SOP 04	peripheral blood, buccal smear
5.	Molecular genetic examination of chromosome Y microdeletions using allele specific PCR followed by agarose gel electrophoresis	SOP 05	peripheral blood, buccal smear
6.	Molecular genetic examination of 34 mutations and 1 polymorphism in CFTR gene by StripAssay method	SOP 06	peripheral blood, buccal smear
7.	Molecular genetic examination of chromosome Y microdeletions using YChromStrip kit	SOP 09	peripheral blood, buccal smear

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List of mutations examined:

CFTR

F508del, 1717-1G>A, G542X, W1282X, N1303K, 3849+10kbC>T, 394delTT, 621+1G>T, G551D, R117H, R1162X, R334W, A455E, 2183AA>G, 3659delC, 1078delT, dI507, R347P, R553X, 3120+1G>A, 2789+5G>A, 1898+1G>A, 711+1G>T, G85E, 2184delA, R560T, CFTR dele 2,3(21kb), 2143delT, 2184insA, 3905insT, Y122X, R347H, 3272-26A>G, Y1092X, IVS8 5T/7T/9T

MDY

AZFa- loci sY84, sY86
AZFb- loci sY127, sY134
AZFc- loci sY254, sY255
ZFY/ZFX gene - check amplification

TM

A1298C mutation in MTHFR gene
C677T mutation in MTHFR gene
Factor V G1691A Leiden mutation
20210A mutation in Prothrombin gene (Factor II)
4G/5G mutation in PAI-1 gene (Plasminogen Activator Inhibitor-1)