The Appendix is an integral part of
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Accredited entity according to ČSN EN ISO 15189:2013:

Fakultní nemocnice Hradec Králové
Laboratory of the Institute for Clinical Biochemistry and Diagnostics and the Department of Medical Genetics – germline genome
Sokolská 581, 500 05 Hradec Králové - Nový Hradec Králové

The Laboratory has a flexible scope of accreditation permitted as detailed in the Annex. Updated list of activities provided within the flexible scope of accreditation is available on the laboratory website http://ukbd.fnhk.cz/zamereni-laboratore.html and at the Quality Management Department.

Examinations:

Ordinal number	Examination procedure name	Examination procedure identification	Examined object
816 - Medical Genetics Laboratory			
1.	Analysis of congenital genetic CNV variants by ARRAY – CGH method [Array - CGH] [Array – CGH - prenatal]	4-36-0038 A	Incoagulable blood - peripheral, cultured amniocytes, CVS cells, tissue, bone marrow, native amniotic fluid
2.	Analysis of DNA by PCR method with electrophoretic detection of product [1)]	4-36-0060 F	Incoagulable blood - peripheral, cultured amniocytes, CVS cells, tissue, bone marrow
3.	Detection of methylation status of gene by methylation specific MLPA[2)]	4-36-0061 F	Incoagulable blood - peripheral, cultured amniocytes, CVS cells, tissue, bone marrow
4.	Detection of sequential variants in genes by real-time PCR method [3)]	4-36-0062 F	Incoagulable blood - peripheral, cultured amniocytes, CVS cells, buccal smear, tissue, bone marrow
5.	Analysis of fluorescently labelled DNA fragments by capillary electrophoresis method [4)]	4-36-0063 F	Incoagulable blood - peripheral, cultured amniocytes, CVS cells, tissue, bone marrow, amniotic fluid
6.	Determination of genomic alterations by MLPA method [5)]	4-36-0064 F	Incoagulable blood - peripheral, cultured amniocytes, CVS cells, tissue, bone marrow
7.	Detection of sequential variants in genes by Sanger sequencing [6)]	4-36-0065 F	Incoagulable blood - peripheral, cultured amniocytes, CVS cells, buccal smear, tissue, bone marrow
8.	Gene mutation analysis by massive parallel sequencing method [7)]	4-36-0066 F	Incoagulable blood - peripheral, Buccal smear, tissue, bone marrow, FFPE

Names in parentheses [ ] are the names of examinations shown in the reports.

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Annex:
Flexible scope of accreditation

Examination procedure ordinal numbers
2, 3, 4, 5, 6, 7, 8

The Laboratory is allowed to modify the examination procedures listed in the Annex within the specified scope of accreditation provided the measuring principle is observed.

The flexible approach to the scope of accreditation cannot be applied to the examinations not included in the Annex.

Explanatory notes on the scope of accreditation:

1)

Examination of polymorphism in apolipoprotein E gene, codons 112 and 158;
Procedure 4-36-0007 A; [ApoE]

Examination of genetic predisposition to coeliac disease;
Procedure 4-36-0052 A; [Genetic predisposition to coeliac disease]
Area: HLA-D2QA1*05-DQB*02, HLA-DQA1*03-DQB1*0302

Molecular genetic analysis of the trinucleotide expansion – expansion of CGG repeats in *FMR1* gene 5´ area;
Procedure 4-36-0018 A; [FRAXA-screening]

2)

Procedure 4-36-0044 A; [PWS-AS-UPD 14]
Locus: 15q11

3)

Examination of HFE gene C282Y, H63D and S65C mutations;
Procedure 4-36-0005 A; [HFE]

Examination of Thiopurine Methyltransferase (TPMT) gene mutations, alleles *2, *3A, *3B and *3C;
Procedure 4-36-0017 A; [Mutation TPMT]

Examination of blood coagulation Factor V gene Leiden mutation (G1691A);
Procedure 4-36-0028 A; [FV Leiden]

Examination of blood coagulation Factor II gene G20210A mutation (Prothrombin)
Procedure 4-36-0029 A; [G20210A FII]

Examination of C677T mutation in Methylenetetrahydrofolate Reductase (MTHFR) gene;
Procedure 4-36-0003 A; [C677T MTHFR]

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Examination of alpha-1 antitrypsin gene defect, mutations Z and S;
Procedure 4-36-0006 A; [Alpha1-AT]

Examination of variants in CYP2C9 and VKORC1 genes;
Procedure 4-36-0014 A; [Exam. polymorph. of CYP/VKOR gene]

HRM analysis of c.1138 in *FGFR3* gene;
Procedure 4-36-0055 A; [Achondroplasia_HRM]

4)

FRAXA – examination of Fragile X syndrome, *FMR1 gen;*
Procedure 4-36-0023A; [FRAXA FA]

Examination of *CFTR* gene mutations;
Procedure 4-36-0004 A; [CFTR]
An expanded range of mutations in the CFTR gene: *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 and IVS9: 5T variant (including TG9-13 identification)/7T/9T.*

Examination of post transplant chimerism using STR loci and amelogenin gene;
List of genetic loci used:
CSF1PO, D2S1338, D18S51, D5S818, D7S820, D3S1358, D19S433, FGA, D8S1179, D13S317, TPOX, D21S11, D16S539, vWA, TH01, Amelogenin
Procedure 4-36-0012 A; [Chimerism]

Molecular genetic analysis - CTG trinucleotides in *3´ UTR DMPK* gene;
Procedure 4-36-0031 A; [Myotonic dystrophy-basic]

Molecular genetic analysis - CTG trinucleotides in *3´ UTR DMPK* gene;
Procedure 4-36-0031 A; [Myotonic dystrophy-full]

13, 18, 21, X and Y chromosome aneuploidies;
Procedure 4-36-0076 A; [QF-PCR-fragment analysis on ABI3130/3500]

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5)

MLPA-fragment analysis;
genes *NF1, NF2, APC, ATM, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, CHEK2, FH, GREM, EPCAM, MLH1, MSH2, MSH6, MUTYH, NBN, PALB2, PMS2, POLD1, POLE, PTCH1, PTEN, RAD50, RAD51C, RAD51D, SMAD4, STK11, SUFU, TP53, SHOX, RUNX2, CYBA, CYBB, NCF2, NCF4, FOXF1, FGD1,* syndromes - *CMT1A1,* microdeletion syndromes 1
Procedure 4-36-0049 A; [MLPA GM]

MLPA BRCA1;
Procedure 4-36-0033 A; [MLPA BRCA]

Spinal muscular atrophy;
Procedure 4-36-0059 A; [SMA I]

6)

Investigation of gene mutations by Sanger sequence analysis;
genes *NF1, NF2, APC, ATM, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, CHEK2, FH, GREM, EPCAM, MLH1, MSH2, MSH6, MUTYH, NBN, PALB2, PMS2, POLD1, POLE, PTCH1, PTEN, RAD50, RAD51C, RAD51D, SMAD4, STK11, SUFU, TP53, PMP22, DHCR7, GJB2, PHOX2B, RUNX2, SHOX, SOX2, KAT6B, FGFR3, HCCS, SPRED1 and TTN*
Procedure 4-36-0048 A; [Seq. analysis of gene exon Y] , Y = number of amplicons

Predictive diagnosis of BRCA1/2 genes, BRCA1/2 gene exon amplicons;
Procedure 4-36-0025 A; [BRCA ½ prediction]

Examination of connexin 26 gene (GJB2) mutations;
Procedure 4-36-0039 A; [Mutations-GJB2]

7)

Examination of mutations in NF1 and NF2 genes by MPS method;
Procedure 4-36-0053 A; [Neurofibromatosis -NGS]

MPS-ENRICHMENT SURESELECT;
genes *ATM, APC, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, MUTYH, NBN, PALB2, PMS2, PTEN, RAD50, RAD51C, RAD51D, SMAD4, STK11, TP53+BAP1, BLM, CDK4, CDKN2A, FH, GREM, POLD1, POLE, PTCH1 and SUFU*
Procedure 4-36-0056 A; [SSEL_34G]

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Examination of minimal residual disease in IGVH rearrangements using MPS;
Procedure 4-36-0057 A; [NGS SM]

MPS-digitalMLPA;
genes: *APC, ATM, BAP1, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, CHEK2, EPCAM, GREM1, MLH1, MSH2, MSH6, MUTYH, NBN, PALB2, PMS2, PTEN, RAD51C, RAD51D, SCG5, SMAD4, STK11 and TP53*
Procedure 4-36-0078 A; [MPS-digitalMLPA]

MPS-ENRICHMENT SURESELECT $^{XT\ HS}$SOMATIC MUTATIONS;
genes: *ABL1, ANKRD26, ASXL1, ATM, BCL2, BCOR, BIRC3, BRAF, BTK, CALR, CARD11, CBL, CEBPA, CSF3R, DDX41, DNMT3A, ETNK1, ETV6, EZH2, FBXW7, FLT3, GATA1, GATA2, IDH1, IDH2, IKZF3, IRF4, JAK2, KIT, KRAS, MAP2K1, MCL1, MED12, MPL, MYD88, NFKBIE, NOTCH1, NPM1, NRAS, PHF6, PLCG2, POT1, PTPN11, RPS15, RUNX1, SAMHD1, SETBP1, SF3B1, SRSF2, STAG2, TET2, TP53, TRAF2, TRAF3, U2AF1, WT1, XPO1, ZRSR2*
Procedure 4-36-0080 A; [NGS panel1]