NÁRODNÍ AKREDITAČNÍ ORGÁN

EA MLA Signatory
Český institut pro akreditaci, o.p.s.
Olšanská 54/3, 130 00 Praha 3

issues

according to section 16 of Act No. 22/1997 Coll., on technical requirements for products, as amended

CERTIFICATE OF ACCREDITATION

No. **730/2020**

Všeobecná fakultní nemocnice v Praze
with registered office U Nemocnice 499/2, 128 08 Praha 2, Company Registration No. 00064165

to the Medical laboratory No. **8097**
Department of Pediatrics and Inherited Metabolic Disorders of the First Medical Faculty of Charles University and the General University Hospital in Prague
Diagnostic Laboratories for Inherited Metabolic Disorders (DPM)

Scope of accreditation:

Laboratory examination and diagnostics of hereditary metabolic disorders in the field of clinical biochemistry and molecular genetics to the extent as specified in the appendix to this Certificate.

This Certificate of Accreditation is a proof of Accreditation issued on the basis of assessment of fulfillment of the accreditation criteria in accordance with

ČSN EN ISO 15189:2013

In its activities performed within the scope and for the period of validity of this Certificate, the Body is entitled to refer to this Certificate, provided that the accreditation is not suspended and the Body meets the specified accreditation requirements in accordance with the relevant regulations applicable to the activity of an accredited Conformity Assessment Body.

This Certificate of Accreditation replaces, to the full extent, Certificate No.: 483/2019 of 25. 9. 2019, or any administrative acts building upon it.

The Certificate of Accreditation is valid until: **4. 6. 2023**

Prague: 2. 12. 2020

Český institut pro akreditaci, o.p.s.
NÁRODNÍ AKREDITAČNÍ ORGÁN
– 6 –

Jiří Růžička
Director
Czech Accreditation Institute
Public Service Company

The Appendix is an integral part of
Certificate of Accreditation No. 730/2020 of 03/12/2020

Accredited entity according to ČSN EN ISO 15189:2013:

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Department of Pediatrics and Inherited Metabolic Disorders of the First Medical Faculty of Charles University and the General University Hospital in Prague
Diagnostic Laboratories for Inherited Metabolic Disorders (DPM)
Ke Karlovu 455/2, 128 08 Praha 2

Medical laboratory locations:

1. **Biochemical Laboratory of the DPM** Ke Karlovu 455/2, Praha 2
2. **DNA Diagnostic Laboratory of the DPM** Ke Karlovu 455/2, Praha 2
3. **Laboratory for Study of Mitochondrial Disorders** Ke Karlovu 455/2, Praha 2

1. **Biochemical Laboratory**

Examination:

Ordinal number	Examination procedure name	Examination procedure identification	Examined object
801 - Clinical Biochemistry			
1.	Determination of relative amount-of-substance concentration of lactate in urine by enzymatic photometric method [*U-Lactate*]	SOP-KDDL-DMP-B-10	Urine
2.	Determination of mucopolysaccharides in urine by screening method [*U-Mucopolysaccharides Screening*]	SOP-KDDL-DMP-B-14	Urine
3.	Determination of relative mass concentration of mucopolysaccharides in urine by photometric method [*U-Mucopolysaccharides*]	SOP-KDDL-DMP-B-15	Urine
4.	Determination of amount-of-substance concentration of creatinine in urine by Jaffé photometric method without deproteinization [*U-Creatinine*]	SOP-KDDL-DMP-B-18	Urine
5.	Determination of amount of substance concentration of uric acid in serum, plasma and urine by uricase/peroxidase enzymatic photometric method [*S,P,U-Uric Acid*]	SOP-KDDL-DMP-B-19	Serum, plasma, urine

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Ordinal number	Examination procedure name	Examination procedure identification	Examined object
6.	Determination of amount of substance concentration of creatinine in serum and plasma by enzymatic photometric method [*S,P-Creatinine*]	SOP-KDDL-DMP-B-20	Serum, plasma
7.	Determination of amount of substance concentration of lactate in deproteinized blood and CSF by enzymatic photometric method [*B, L – Lactate*]	SOP-KDDL-DMP-B-23	Deproteinized blood, CSF
8.	Determination of amount of substance concentration of pyruvate in deproteinized blood by enzymatic photometric method [*B-Pyruvate*]	SOP-KDDL-DMP-B-24	Deproteinized blood
9.	Determination of amount of substance concentration of 3-hydroxybutyrate in deproteinized blood by enzymatic photometric method [*B-3-hydroxybutyrate*]	SOP-KDDL-DMP-B-25	Deproteinized blood
10.	Determination of amount of substance concentration of total homocysteine in plasma, serum by enzymatic photometric method [*P, S – Total homocysteine enzymat.*]	SOP-KDDL-DMP-B-28	Plasma, serum
11.	Profiling examination of amino acids in serum, plasma and CSF on amino acid analyzer by ion exchange chromatography method with ninhydrin detection [1] [*S, P, L – Amino acids*]	SOP-KDDL-DMP-B-30	Serum, plasma, cerebrospinal fluid
12.	Profiling examination of amino acids in urine on amino acid analyzer by ion exchange chromatography method with ninhydrin detection [2] [*U– Amino acids*]	SOP-KDDL-DMP-B-31	Urine

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Ordinal number	Examination procedure name	Examination procedure identification	Examined object
13.	Determination of amino acids and acylcarnitines in dried blood spot using tandem mass spectrometry for the purpose of newborn screening [3)] [*KP-AMK, AC Newborn Screening*]	SOP-KDDL-DMP-B-33	Dried blood spot
14.	Determination of amino acids and acylcarnitines in dried blood spot using tandem mass spectrometry for the purpose of selective screening [4)] [*KP-AMK, Acylcarnitines MS/MS, Pregnancy Screening MS/MS, Dietary Compensation MS/MS; B-Phenylalanine, B-Tyrosine*]	SOP-KDDL-DMP-B-34	Dried blood spot
15.	Determination of relative amount of substance concentration of orotic acid in urine by capillary electrophoresis [*U-Orotic Acid CE*]	SOP-KDDL-DMP-B-57	Urine
16.	Determination of relative amount of substance concentration of galactitol in urine by gas chromatograph with flame ionization detection [*U-Galactitol*]	SOP-KDDL-DMP-B -21	Urine
17.	Profiling examination of purines and pyrimidines by HPLC method with UV detection [*U -P/P*]	SOP-KDDL-DMP-B-32	Urine
18.	Determination of catalytic biotinidase activity in dried blood spot using fluorimetric detection [*KP-BTD Newborn Screening, KP- Biotinidase qualitative*]	SOP-KDDL-DMP-B-60	Dried blood spot

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Ordinal number	Examination procedure name	Examination procedure identification	Examined object
19.	Determination of catalytic activity of α-galactosidase by fluorimetric method [*α-galactosidase in leukocytes, plasma, serum*; *Patient*, α- *galactosidase – control enzyme; Patient*]	SOP-KDDL-DMP-E-02	Leukocytes, plasma, serum
20.	Determination of catalytic activity of β-galactosidase by fluorimetric method [*β-galactosidase in leukocytes, plasma, serum*; *Patient*]	SOP-KDDL-DMP-E-03	Leukocytes, plasma, serum
21.	Determination of catalytic activity of chitotriosidase by fluorimetric method [*chitotriosidase in serum, plasma*; *Patient*]	SOP-KDDL-DMP-E-07	Plasma, serum
22.	Determination of catalytic activity of acid α-1,4-glucosidase by fluorimetric method [*acid α-1,4-glucosidase in leukocytes*; *Patient*]	SOP-KDDL-DMP-E-31	Leukocytes
23.	Determination of catalytic activity of acid α-1,4-glucosidase in dried blood spot by fluorimetric method [*acid α-1,4-glucosidase on dried blood spot*; *Patient*]	SOP-KDDL-DMP-E-33	Dried blood spot
24.	Determination of catalytic activity of α-galactosidase in dried blood spot by fluorimetric method [*α-galactosidase in dried blood spot*; *Patient*]	SOP-KDDL-DMP-E-34	Dried blood spot

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

Explanatory notes:

[1] Taurin, Phosphoethanolamine, Aspartic acid, Hydroxyproline, Threonine, Serine, Asparagine, Glutamic acid, Glutamine, Glu+Gln, α-aminoadipic acid, Proline, Glycine, Alanine, Citrulline, α-aminobutyric acid, Valine, Cystien, Methionine, Allo-isoleucine, Cystathionine, Isoleucine, Leucine, Tyrosine, Phenylalanine, Free homocystine, β-alanine, β-aminoisobutyric, γ-aminobutyric, δ- aminolevulinic acid, Free

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hydroxylysine, Ethanolamine, Ornithine, Lysine, Histidine, Homocarnosine (in CSF only), 1-Methylhistidine, 3-Methylhistidine, Arginine.

2) Taurin, Phosphoethanolamine, Aspartic acid, Hydroxyproline, Threonine, Serine, Asparagine, Glutamic acid, Glutamine, Glu+Gln, α-aminoadipic acid, Proline, Glycine, Alanine, Citrulline, α-aminobutyric acid, Valine, Cystien, Methionine, Allo-isoleucine, Cystathionine, Isoleucine, Leucine, Tyrosine, Phenylalanine, Free homocystine, β-alanine, β-aminoisobutyric, γ-aminobutyric, δ- aminolevulinic acid, Free hydroxylysine, Ethanolamine, Ornithine, Lysine, Histidine, 1-Methylhistidine, Arginine, Cystin-lithogenity.

3) Ala, Phe, Tyr, Val, Xle, C2, C5, C5DC, C6, C8, C10, C10:1, C12, C14, C14:1, C14:2, C14OH, C16, C16OH, C16:1, C18, C18:1, C18:1OH, C18OH, C0 and ratios: Phe/Tyr, Xle/Ala, C5DC/C8, C5/C0, C8/C2, C14:1/C2, (C16+C18)/C0, (C16+C18:1)/C2

4) Ala, Phe, Tyr, Val, Xle, Cit, C2, C3, C3DC, C4, C4DC, C5, C5:1, C5DC, C5OH, C6, C8, C10, C10:1, C12, C14, C14:1, C14:2, C14OH, C16, C16:OH, C16:1, C18, C18:1, C18:1OH, C18OH, C0 and ratios: Phe/Tyr, Xle/Ala, C3/C2, C4/C3, C5DC/C8, C5/C0, C8/C2, C14:1/C2, (C16+C18)/C0, (C16+C18:1)/C2.

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2. **DNA Diagnostic Laboratory of the DPM**

The Laboratory has a flexible scope of accreditation permitted as detailed in the Annex. Updated list of activities provided within the required flexible scope of accreditation is available from the Laboratory Quality Manager and in the Appendix 1 to the Metabolic Handbook available at https://www.vfn.cz/pacienti/kliniky-ustavy/klinika-detskeho-a-dorostoveho-lekarstvi/laborator/

Examination:

Ordinal number	Examination procedure name	Examination procedure identification	Examined object
816 - Medical Genetics Laboratory			
1.	Analysis of rare disease genes by Sanger sequencing [5)]	SOP-KDDL-DMP-G-62	Peripheral blood, isolated DNA
2.	Microscopic evaluation (detection) of cell growth in tissue culture	SOP-KDDL-DMP-T-01	Skin biopsy
3.	Analysis of human genome variants by massive parallel sequencing [6)] (MPS)	SOP-KDDL-DMP-G-61	Peripheral blood, isolated DNA

Annex:
Flexible scope of accreditation

Examination procedure ordinal numbers
1, 3

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:

[5)] genes: *NOTCH3, ASPA, ACADM, BTD, HADHA, OTC, GLA, GBA, GALC, CLN2 (TPP1), CLN3, CLN7 (MFSD8), NPC1, NPC2, IDS, GCDH, ABCD1*

[6)] **Cardiomyopathies**: *ABCC9, ABCG5, ABCG8, ACTA1, ACTA2, ACTC1, ACTN2, AKAP9, ALMS1, ANK2, ANKRD1, APOA4, APOA5, APOB, APOC2, APOE, BAG3, BRAF, CACNA1C, CACNA2D1, CACNB2, CALM1, CALR3, CASQ2, CAV3, CBL, CBS, CETP, COL3A1, COL5A1, COL5A2, COX15, CREB3L3, CRELD1, CRYAB, CSRP3, CTF1, DES, DMD, DNAJC19, DOLK, DPP6, DSC2, DSG2, DSP, DTNA, EFEMP2, ELN, EMD, EYA4, FBN1, FBN2, FHL1, FHL2, FKRP, FKTN, FXN, GAA, GATAD1, GCKR, GJA5, GLA, GPD1L, GPIHBP1, HADHA, HCN4, HFE, HRAS, HSPB8, ILK, JAG1, JPH2, JUP, KCNA5, KCND3, KCNE1, KCNE2, KCNE3,*

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KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, KLF10, KRAS, LAMA2, LAMA4, LAMP2, LDB3, LDLR, LDLRAP1, LMF1, LMNA, LPL, LTBP2, MAP2K1, MAP2K2, MIB1, MURC, MYBPC3, MYH11, MYH6, MYH7, MYL2, MYL3, MYLK, MYLK2, MYO6, MYOZ2, MYPN, NEBL, NEXN, NKX2-5, NODAL, NOTCH1, NPPA, NRAS, PCSK9, PDLIM3, PKP2, PLN, PRDM16, PRKAG2, PRKAR1A, PTPN11, RAF1, RANGRF, RBM20, RYR1, RYR2, SALL4, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCO2, SDHA, SEPN1, SGCB, SGCD, SGCG, SHOC2, SLC25A4, SLC2A10, SMAD3, SMAD4, SNTA1, SOS1, TAZ, TBX20, TBX3, TBX5, TCAP, TGFB2, TGFB3, TGFBR1, TGFBR2, TMEM43, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TRDN, TRIM63, TRPM4, TTN, TTR, TXNRD2, VCL, ZBTB17, ZHX3, ZIC3

Metabolic disorders:
Glycogen storage disorders: *AGL, ALDOA, ALDOB, ALDOC, ENO3, FBP1, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, KHK, PC, PFKM, PGAM2, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAB1, PRKAB2, PRKAG2, PYGL, PYGM, RBCK1, SLC2A2, SLC37A4*
Urea cycle disorders, orotic acidurias: *ARG1, ASL, ASS1, CAD, CPS1, DHODH, FTCD, NAGS, OTC, SHMT1, SHMT2, SLC25A13, SLC25A15, SLC25A2, SLC46A1, SLC7A7, TYMP, TYMS, UMPS*
Peroxisomal disorders: ABCD1, ABCD3, ACOX1, AGPS, AGXT, AMACR, BAAT, CAT, DNM1L, FAR1, GDAP1, GNPAT, HSD17B4, MFF, PEX1, PEX2, PEX26, PEX3, PEX5, PEX5L, PEX6, PEX7, PEX10, PEX11A, PEX11B, PEX11G, PEX12, PEX13, PEX14, PEX16, PEX19, PHYH, SCP2
Hyperhomocysteinemia: *ABCD4, ADK, AHCY, ALDH7A1, AMN, CBS, CD320, CDO1, CTH, CUBN, DHFR, ETHE1, FOLH1, FOLR1, FOLR2, FOLR3, FTCD, FUT2, GIF, GNMT, GPHN, HCFC1, LMBRD1, LRP2, MAT1A, MAT2A, MAT2B, MCEE, MMAA, MMAB, MMACHC, MMADHC, MOCS1, MOCS2, MTHFD1, MTHFR, MTHFS, MTR, MTRR, MUT, PCCA, PCCB, PDXK, PDXP, PNPO, SLC19A1, SLC25A32, SLC46A1, SQOR, SUCLA2, SUOX, TCN1, TCN2, THAP11, TST, ZNF143*
Leucinoses: *BCKDHA, BCKDHB, DBT, DLD*
Rhabdomyolyses and disorders of the metabolism of fatty acids: *ACADM, ACADVL, AGL, ALDOA, AMPD1, ANO5, ATP2A1, CACNA1S, CASQ1, CAV3, CHKB, CPT1A, ACADM, ACADVL, AGL, ALDOA, AMPD1, ANO5, ATP2A1, CACNA1S, CASQ1, CAV3, CHKB, CPT1A, CPT2, CTDP1, CYP2C8, DGUOK, DYSF, ENO3, ETFA, ETFB, ETFDH, FDX1L, FKRP, FLAD1, HADHA, HADHB, HRAS, ISCU, LAMP2, LDHA, LPIN1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, POLG, PYGM, QARS, RYR1, SCN4A, SIL1, SLC16A1, SLC25A20, SLC25A32, SLC52A1, SLC52A2, SLC52A3, TANGO2, TSEN54, TSFM*
Disorders of the metabolism of neurotransmitters: *ABAT, ALDH5A1, ALDH7A1, AMT, DBH, DDC, DHFR, DNAJC12, FOLR1, GCH1, GCSH, GLDC, GLUL, MAOA, PCBD1, PHGDH, PNPO, PSAT1, PSPH, PTS, QDPR, SLC18A2, SLC46A1, SLC6A3, SPR, TH*
Neuronal ceroid lipofuscinosis: *ATP13A2, CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, DNAJC5, GRN, KCTD7, MFSD8, PPT1, TPP1*
Other disorders: *CADASIL (NOTCH3), cystinuria (SLC3A1, SLC7A9), isovaleric aciduria (IVD)*

CZECANCA:
AIP; ALK; APC; APEX1; ATM; ATMIN; ATR; ATRIP; AURKA; AXIN1; BABAM1; BAP1; BARD1; BLM; BMPR1A; BRAP; BRCA1; BRCA2; BRCC3; BRE; BRIP1; BUB1B; C11orf30; C19orf40; casp8; CCND1; CDC73; CDH1; CDK4; CDKN1B; CDKN1C; CDKN2A; CEBPA; CEP57; CLSPN; CSNK1D; CSNK1E; CWF19L2; CYLD; DCLRE1C; DDB2; DHFR; DICER1; DIS3L2; DMBT1; DMC1; DNAJC21; DPYD; EGFR; EPCAM; EPHX1; ERCC1; ERCC2; ERCC3; ERCC4; ERCC5; ERCC6; ESR1; ESR2; EXO1; EXT1; EXT2; EYA2; EZH2; FAM175A; FAM175B; FAN1; FANCA; FANCB; FANCC; FANCD2; FANCE; FANCF; FANCG; FANCI; FANCL; FANCM; FBXW7; FH; FLCN; GADD45A; GATA2; GPC3; GRB7; HELQ; HNF1A; HOXB13; HRAS; HUS1; CHEK1; CHEK2; KAT5; KCNJ5; KIT; LIG1; LIG3; LIG4; LMO1; LRIG1; MAX; MCPH1; MDC1; MDM2; MDM4; MEN1; MET; MGMT; MLH1; MLH3; MMP8; MPL; MRE11A; MSH2; MSH3; MSH5; MSH6; MSR1; MUS81; MUTYH; NAT1; NBN; NCAM1; NELFB; NF1; NF2; NFKBIZ; NHEJ1; NSD1; OGG1; PALB2; PARP1; PCNA; PHB; PHOX2B; PIK3CG; PLA2G2A; PMS1; PMS2; POLB; POLD1; POLE; PPM1D; PREX2; PRF1; PRKAR1A; PRKDC; PTEN; PTCH1; PTTG2; RAD1; RAD17; RAD18;

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RAD23B; RAD50; RAD51; RAD51AP1; RAD51B; RAD51C; RAD51D; RAD52; RAD54B; RAD54L; RAD9A; RB1; RBBP8; RECQL; RECQL4; RECQL5; RET; RFC1; RFC2; RFC4; RHBDF2; RNF146; RNF168; RNF8; RPA1; RUNX1; SBDS; SDHA; SDHAF2; SDHB; SDHC; SDHD; SETBP1; SETX; SHPRH; SLX4; SMAD4; SMARCA4; SMARCB1; SMARCE1; STK11; SUFU; TCL1A; TELO2; TERF2; TERT; TLR2; TLR4; TMEM127; TOPBP1; TP53; TP53BP1; TSC1; TSC2; TSHR; UBE2A; UBE2B; UBE2I; UBE2V2; UBE4B; UIMC1; VHL; WRN; WT1; XPA; XPC; XRCC1; XRCC2; XRCC3; XRCC4; XRCC5; XRCC6; ZNF350; ZNF365

Bone dysplasias:
ACAN, ACP5, ACTB, ACTG1, ACVR1, ADAMTS10, ADAMTS17, ADAMTSL2, AGPS, AIFM1, AKT1, ALPL, ALX3, ALX4, AMER1, ANKH, ANKRD11, ANO5, ARHGAP31, ARSB, ARSE, ATP6V0A2, ATR, B3GALT6, B3GAT3, B4GALT7, BCS1L, BGN, BHLHA9, BMP1, BMP2, BMPER, BMPR1B, BRAF, BRCA2, BRIP1, CA2, CANT1, CASR, CBL, CCDC8, CDC6, CDC45, CDKN1C, CDT1, CENPJ, CEP63, CEP152, CHST3, CHST14, CHSY1, CKAP2L, CLCN5, CLCN7, COL1A1, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, COL10A1, COL11A1, COL11A2, COMP, CREB3L1, CREBBP, CRTAP, CSPP1, CTSK, CUL7, CYP27B1, DDR2, DHCR7, DHCR24, DHODH, DLL3, DLL4, DLX3, DLX5, DMP1, DOCK6, DVL1, DYM, DYNC2H1, EBP, EFNB1, EFTUD2, EIF2AK3, ENAM, ENPP1, EOGT, EP300, ERCC4, ESCO2, EVC, EVC2, EXT1, EXT2, EXTL3, EZH2, FAM20A, FAM20C, FAM58A, FAM83H, FAM111A, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FBN1, FBN2, FGD1, FGF10, FGF23, FGFR1, FGFR2, FGFR3, FKBP10, FLNA, FLNB, GALNT3, GDF5, GH1, GHR, GHRHR, GJA1, GLI2, GLI3, GNAS, GNPAT, GPC6, HDAC8, HESX1, HOXA13, HOXD13, HRAS, HSPG2, IDS, IFITM5, IFT43, IFT80, IFT122, IFT140, IFT172, IGF1, IGF1R, IGFALS, IHH, IMPAD1, INPPL1, INSR, IRS1, KAT6B, KIF7, KIF22, KMT2A, KRAS, LARP7, LBR, LEMD3, LHX3, LHX4, LIFR, LMNA, LMX1B, LONP1, LRP4, LRP5, LTBP2, LTBP3, LZTR1, MAFB, MAP2K1, MAP2K2, MATN3, MBTPS2, MESP2, MGP, MMP2, MMP9, MMP13, MSX2, MYCN, NANS, NEK1, NF1, NFIX, NIPBL, NKX3-2, NOG, NOTCH1, NOTCH2, NPR2, NRAS, NSD1, NSDHL, OBSL1, ORC1, ORC4, ORC6, OSTM1, OTX2, P3H1, PALB2, PAPSS2, PCNT, PCYT1A, PDE4D, PEX7, PEX14, PEX19, PGM3, PHEX, PIK3CA, PITX2, PLOD2, PLS3, POC1A, POLR1C, POLR1D, POR, POU1F1, PPIB, PRKAR1A, PROP1, PTDSS1, PTH1R, PTHLH, PTPN11, PYCR1, RAB33B, RAD21, RAD51C, RAF1, RASA2, RBBP8, RBM8A, RBPJ, RECQL4, RIT1, RMRP, RNU4ATAC, ROR2, RRAS, RTTN, RUNX2, SALL1, SALL4, SBDS, SEC24D, SERPINF1, SERPINH1, SETBP1, SF3B4, SH3BP2, SH3PXD2B, SHOC2, SHOX, SKI, SLC26A2, SLC29A3, SLC34A3, SLC35D1, SLC39A13, SLCO2A1, SLX4, SMAD3, SMAD4, SMARCAL1, SMC1A, SMC3, SNX10, SOS1, SOST, SOX2, SOX3, SOX9, SP7, SPARC, SRCAP, STAMBP, STAT5B, TBX3, TBX4, TBX5, TBX6, TBX15, TBX19, TCF12, TCIRG1, TCOF1, TCTN3, TGFB1, TGFB2, TGFB3, TGFBR1, TGFBR2, TMEM38B, TNFRSF11A, TNFRSF11B, TNFSF11, TP63, TRAPPC2, TRIM37, TRIP11, TRPS1, TRPV4, TTC21B, TWIST1, TYROBP, VDR, VIPAS39, WDR19, WDR34, WDR35, WDR60, WISP3, WNT1, WNT5A, WNT7A, XRCC2, XRCC4, XYLT1

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Diagnostic Laboratories for Inherited Metabolic Disorders (DPM)
Ke Karlovu 455/2, 128 08 Praha 2

3. Laboratory for Study of Mitochondrial Disorders

The Laboratory has a flexible scope of accreditation permitted as detailed in the Annex. Updated list of activities provided within the required flexible scope of accreditation is available from the Laboratory Quality Manager and in the Laboratory Manual available at https://www.vfn.cz/pacienti/kliniky-ustavy/klinika-detskeho-a-dorostoveho-lekarstvi/laborator/

Examination:

Ordinal number	Examination procedure name	Examination procedure identification	Examined object
816 - Medical Genetics Laboratory			
1.	Detection of mtDNA mutations associated with LHON syndrome by RFLP	SOP-KDDL-DMP-M-4	Biological material containing genomic DNA [9)]
2.	Detection of mutations in genomic DNA by Sanger sequencing [7)]	SOP-KDDL-DMP-M-5	Biological material containing genomic DNA [9)]
3.	Detection of MECP2 gene mutations by HRM and Sanger sequencing	SOP-KDDL-DMP-M-9	Biological material containing genomic DNA [9)]
4.	Gene analysis by massively parallel sequencing [8)]	SOP-KDDL-DMP-M-20	Biological material containing genomic DNA [9)]
801 - Clinical Biochemistry			
5.	Screening of congenital disorders of glycosylation by transferrin sialylated isoforms profile analysis using electrophoretic separating techniques	SOP-KDDL-DMP-M-10	Clotted blood, serum

Annex:
Flexible scope of accreditation

Examination procedure ordinal numbers
2, 4

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

Accredited entity according to ČSN EN ISO 15189:2013:

Všeobecná fakultní nemocnice v Praze

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Explanatory notes:

[7] genes: SURF1, SCO2, *OPA1, EXT1, EXT2*

[8] **mitochondrial DNA:**
MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY
Autoinflammatory syndromes (periodic fevers):
ADAM17, AP3B1, C1QA, C1QB, C1QC, C1R, C2, C3, C4A, C5, C6, C7, C8A, C8B, C9, CARD14, CARD8, CASP10, CASP8, CECR1 (ADA2), CFH, CFHR5, CFI, CFP, COL3A1, COL5A1, COL5A2, CTLA4, DNASE2, DOCK8, ELANE, ELN, FAS, FASLG, FOXP3, G6PC3, HAX1, IKBKG, IL10, IL10RA, IL10RB, IL1RN, IL21, IL22, IL36RN, LPIN2, LRBA, LYST, MALT1, MASP2, MBL2, MEFV, MVK, NCF2, NLRC4, NLRP12, NLRP3, NLRP6, NLRP7, NOD2, NRAS, OTULIN (FAM105B), PLCG2, PLOD1, PRF1, PRG4, PSMA3, PSMB4, PSMB8, PSMB9, PSTPIP1, RAB27A, SEC16A, SERPING1, SH2D1A, SLC29A3, STX11, STXBP2, TMEM173, TNFAIP3, TNFRSF11A, TNFRSF1A, TRAP1, TRNT1, TTC7A, UNC13D, WAS, WDR1, XIAP

Mitochondrial disease panel, leukodystrophy:
Mitochondrial diseases: *AARS2, ACAD9, ACO2, ADCK3, AGK, AIFM1, APOPT1, ATAD3A, ANT1, ATP5A1, ATP5D, ATP5E, ATP5F1D, ATPAF2, BCS1L, BOLA3, C10orf2, C12orf62, C12orf65, C19orf70, C20orf7, C2orf64, C8orf38, CEP89, CLPB, CLPP, COA3, COA5, COA6, COA7, COA7, COASY, COQ2, COQ4, COQ5, COQ6, COQ9, COX10, COX15, COX4I1, COX4I2, COX6B1, COX7B, CTBP1, CYC1, DARS2, DGUOK, DIAPH1, DLAT, DLD, DNA2, DNAJC12, DNAJC19, DNM1L, E4F1, EARS2, ECHS1, ELAC2, ETHE1, FAM36A, FARS2, FASTKD2, FBXL4, FDX1L, FDXR, FLAD1, FOXG1, FOXRED1, GARS, GFM1, GFM2, GTPBP3, HARS2, HTRA2, CHCHD10, IARS2, ISCA1, ISCA2, ISCU, KARS, LARS, LARS2, LIPT2, LONP1, LRPPRC, LYRM4, LYRM7, MARS2, MDH2, ME2, MFF, MGME1 (C20orf72), MICU1, MIEF2, MPV17, MRM2, MRPL3, MRPL44, MRPS16, MRPS2, MRPS22, MRPS34, MRPS7, MSTO1, MTFMT, MTO1, MTPAP, NAXE, NBAS, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA4, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF6, NDUFAF7, NDUFB11, NDUFB3, NDUFB8, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFU1, CSNU3, NUBPL, OPA1, OPA3, OXA1L, PC, PCK2, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PET100, PITRM1, PMPCB, PNPT1, POLG, POLG2, PPA2, PUS1, RARS2, RMND1, RMRP, RNASEH1, RRM2B, SARS2, SCO1, SCO2, SDHA, SDHAF1, SDHAF2, SDHB, SDHC, SDHD, SERAC1, SFXN4, SLC19A3, SLC25A10, SLC25A19, SLC25A24, SLC25A26, SLC25A3, SLC25A4, SLC25A4, SLC25A46, SLC39A8, SPG20, SSBP1, SUCLA2, SUCLG1, SURF1, TACO1, TARS2, TAZ, TIMM50, TIMMDC1, TK2, TMEM126A, TMEM126B, TMEM70, TPK, TRAK1, TRIT1, TRMT5, TRMU, TRNT1, TSFM, TTC19, TUFM, TXN2, TYMP, UNG, UQCC2, UQCRB, UQCRC2, UQCRFS1, UQCRQ, USMG5, VARS2, WARS2, XPNPEP3, YARS2*
Leukodystrofie: *AARS, AARS2, ABCD1, ADAR1, AGPS, AIFM1, AIMP1, ALDH3A2, AMT, APOPT1, APP, ARSA, ASPA, ATAD3A, ATAD3B, ATN1, ATRN, AUH, BCAP31, BCKDHA, BCKDHB, BOLA3, BPIFA2, CBS, CLCN2, CLPP, CNTNAP1, COL4A1, COL4A2, COX6B1, CSF1R, CST3, CTC1, CTSA, CYP27A1, D2HGDH, DARS, DARS2, DBT, DHAPAT, EARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EPRS, ERCC2, ERCC3, ERCC6, ERCC8, FAM126A, FBXL4, FOLR1, FUCA1, GALC, GAN, GBE1, GCDH, GCSH, GFAP, GJA1, GJB1, GJC2, GLA, GLB1, GLDC, GLRX5, GM2A, GSN, GTF2H5, HEXA, HEXB, HMBS, HMGCL, HSPD1, HTRA1, IBA57, IDH1, IDH2, IKBKAP (ELP1), ISCA2, ITM2B, KARS, L2HGDH, LAMA2, LAMB1, LIAS, LMBRD1, LMNB1, LYRM7, MLC1, MMADHC, MMACHC, MOG, MTFMT, MTHFR, MTR, MTRR, NDUFA2, NDUFS1, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NFU1, NKX6-2, NOTCH3, NUBPL, PCCA, PCCB, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHGDH, PHYH, PLP1, POLR1C, POLR3A, POLR3B, POLR3D, PSAP, PSAT1, RARS, RARS2, RMND1, RNASEH2A,*

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RNASEH2B, RNASEH2C, RNASET2, RPIA, SAMHD1, SDHA, SDHAF1, SDHB, SLC16A2, SLC17A5, SLC19A3, SLC1A4, SLC25A1, SLC25A12, SNORD118, SOX10, SPTAN1, SUMF1, SURF1, TMEM106B, TREM2, TREX1, TTR, TUBB4A, TYMP, TYROBP, UFM1, VPS11

[9] blood, muscle, fibroblasts, hair, buccal swab, urine sediment, autoptic tissues, chorionic villi, isolated DNA

Abbreviations:
mtDNA – mitochondrial deoxyribonukleic acid
LHON – Leber´s hereditary optic neuropathy
RFLP – restriction fragment length polymorphism
HRM – high resolution melting