List of activities within the flexible scope of accreditation

Accredited Body: Všeobecná fakultní nemocnice v Praze

CAB Name: Department of Pediatrics and Inherited Metabolic Disorders (KPDPM) of the General University Hospital in Prague and 1st Medical Faculty of Charles University, Diagnostic Laboratories for Inherited Metabolic Disorders (DPM)

CAB Number: 8097

Certificate of Accreditation No. 532/2024

Field of Accreditation: Medical Laboratory - ČSN EN ISO 15189 ed. 3:2023

Updated on: 17.12.2024

1. **DPM Biochemistry Laboratory** Ke Karlovu 455/2, Praha 2

Examinations:

Ordinal Number	Analyte/ parameter/diagnostics	Principle of examination	Identification of method procedure/ equipment	Examined material	Degrees of freedom[1]
801 – Clinical Biochemistry					
1.	Amino acids and acylcarnitines	Tandem Mass Spectrometry	SOP-KPDPM-DMP-B-33, 27/03/2023; SOP-KPDPM-DMP-B-34, 27/03/2023; Sciex API 4500 Triple Quad; Sciex API 3200 Triple Quad; Sciex API 3200 QTrap	Dry blood spot	A, B, C
2.	Biotinidase	Fluorimetry	SOP-KPDPM-DMP-B-60, 01/03/2023; BioTek Synergy 2	Dry blood spot	A, B
3.	Amino acid profile	Ion-exchange chromatography	SOP-KPDPM-DMP-B-30, 01/03/2023; SOP-KPDPM-DMP-B-31, 01/03/2023; AAA 400	Serum, plasma, cerebrospinal fluid, urine	A, B, C
4.	Orotic acid	Capillary electrophoresis	SOP-KPDPM-DMP-B-57, 01/03/2023; Agilent Technologies G7100A;	Urine	A, B

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Ordinal Number	Analyte/ parameter/diagnostics	Principle of examination	Identification of method procedure/ equipment	Examined material	Degrees of freedom[1]
5.	Galactitol	Gas chromatography	SOP-KPDPM-DMP-B-21, 01/03/2023; Thermo Scientific TRACE1610	Urine	A, B
6.	Profile of purines and pyrimidines	Liquid chromatography	SOP-KPDPM-DMP-B-32, 01/03/2023; Waters Alliance 2695	Urine	A, B, C
7.	Mucopolysaccharides	Spectrophotometry	SOP-KPDPM-DMP-B-15, 01/03/2023; Thermo Evolution 60S	Urine	A, B
8.	Lactate	Spectrophotometry	SOP-KPDPM-DMP-B-10, 01/03/2023; SOP-KPDPM-DMP-B-23, 01/03/2023; Erba XL-200	Urine, blood deproteinate, cerebrospinal fluid deproteinate	A, B
9.	Vacant				
10.	Homocysteine	Spectrophotometry	SOP-KPDPM-DMP-B-28, 01/03/2023; Erba XL-200	Plasma, serum	A, B
11.	Creatinine	Spectrophotometry	SOP-KPDPM-DMP-B-18, 01/03/2023; SOP-KPDPM-DMP-B-20, 01/03/2023; Erba XL-200	Serum, plasma, urine	A, B
12.	Uric acid	Spectrophotometry	SOP-KPDPM-DMP-B-19, 01/03/2023; Erba XL-200	Serum, plasma, urine	A, B

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Ordinal Number	Analyte/ parameter/diagnostics	Principle of examination	Identification of method procedure/ equipment	Examined material	Degrees of freedom[1]
13.	Enzymes	Fluorimetry	SOP-KPDPM-DMP-E-02, 15/03/2023; SOP-KPDPM-DMP-E-03, 15/03/2023; SOP-KPDPM-DMP-E-31, 15/03/2023; SOP-KPDPM-DMP-E-33, 15/03/2023; SOP-KPDPM-DMP-E-34, 15/03/2023; Perkin Elmer LS 55; BioTek Synergy 2	Leukocytes, serum, plasma, dry blood spot	A, B, C, D
816 – Medical Genetics Laboratory					
1.	Newborn screening for SCID and SMA	Real-time PCR	SOP-KPDPM-DMP-NS-01, 01/01/2024; Janus G3 Workstation; QuantStudio Dx	Dry blood spot	A, B

Specification of the scope of accreditation:

Field Nr. / Ordinal Number	Detailed information on activities within the scope of accreditation
801/1	SOP-KPDPM-DMP-B-33: Ala, Phe, Tyr, Val, Xle, Cit, Arg, Met, Orn, 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, Arg/Orn, Arg/Phe, Cit/Phe, Orn/Cit, Met/Phe C5DC/C8, C5/C0, C8/C2, C14:1/C2, C14:1/C16, (C16+C18)/C0, (C16+C18:1)/C2; SOP-KPDPM-DMP-B-34: Ala, Phe, Tyr, Val, Xle, Cit, Arg, 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, Arg/Orn, C3/C2, C4/C3, C5DC/C8, C5/C0, C8/C2, C14:1/C2, (C16+C18)/C0, (C16+C18:1)/C2.
801/3	SOP-KPDPM-DMP-B-30: 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, Homocarnosine (in CSF only), 1-Methylhistidine, 3-Methylhistidine, Arginine; SOP-KPDPM-DMP-B-31: 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,

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	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.
801/6	Hypoxanthine, Xanthine, Uracil, 2,8–Dihydroxyadenine, Deoxyadenosine, Deoxyinosine, Deoxyguanosine, Thymine, Uridine, Adenosine, Succinyladenosine, SAICAr
801/13	α-galactosidase, β-galactosidase, acid α-1,4-glucosidase

List of activities within the flexible scope of accreditation

2. **DPM Molecular Genetics Laboratory** Ke Karlovu 455/2, Praha 2

Examinations:

Ordinal Number	Analyte/ parameter/diagnostics	Principle of examination	Identification of method procedure/ equipment	Examined material	Degrees of freedom[1]
816 – Medical Genetics Laboratory					
1.	Examination of germline genome variants	Massive parallel sequencing	SOP-KPDPM-DMP-S-01, 26/8/2024; PP-KPDPM-DMP-S-02, 07/01/2022; PP-KPDPM-DMP-S-03, 26/8/2024; PP-KPDPM-DMP-S-06, 21/12/2023; Miseq	Biological material containing genomic DNA	A, B, C
2.	Examination of germline genome variants	Sanger sequencing	SOP-KPDPM-DMP-G-62, 02/01/2023; Annex SOP-KPDPM-DMP-G-062, 20/01/2023; PP-KPDPM-DMP-G-01, 02/01/2023; PP-KPDPM-DMP-G-03, 02/01/2023; PP-KPDPM-DMP-G-09, 02/01/2023; PP-KPDPM-DMP-G-10, 06/10/2022; 3500xL	Biological material containing genomic DNA	A, B, C

Specification of the scope of accreditation:

Field Nr. / Ordinal Number	Detailed information on activities within the scope of accreditation
816/1	**Cardiology panel:** *ABCC9, ABCG5, ABCG8, ACTA1, ACTA2, ACTC1, ACTN2, AKAP9, ALMS1, ANK2, ANKRD1, APOA4, APOA5, APOB, APOC2, APOE, BAG3, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2 , CALM3, CALR3, CASQ2, CASZ1, CAV3, CBL, CETP, COL3A1, COL4A5, COL5A1, COL5A2, CREB3L3, CRELD1, CRYAB, CSRP3, CTF1, CTNNA3, DES, DMD, DNAJC19, DOLK, DPP6, DSC2, DSG2, DSP, DTNA, EFEMP2, ELN, EMD, EYA4, FBN1, FBN2, FHL1, FHL2, FKRP, FKTN, FLNC, FXN, GAA, GATA4, GATA5, GATAD1, GJA5, GLA, GPD1, GPD1L, GPIHBP1, HCN4, HFE, JAG1, JPH2, JUP, KCNA5, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, KLF10, LAMA2, LAMA4, LAMP2, LDB3, LDLR, LDLRAP1, LMF1, LMNA, LOX, LPL, LTBP2, MAP2K1, MAP2K2, MIB1, MT-TL1, MURC, MYBPC3, MYH11, MYH6, MYH7, MYL2, MYL3, MYLK, MYLK2, MYOM1, MYOZ2, MYPN, NEBL, NEXN, NKX2-5, NODAL, NOS1AP, NOTCH1,*

List of activities within the flexible scope of accreditation

	NPPA, OBSCN, PCSK9, PDLIM3, PKP2, PLN, PRDM16, PRKAG2, PRKG1, PTPN11, RAF1, RANGRF, RBM20, RIT1, ROBO4, RYR2, SALL4, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SDHA, SGCD, SGCG, SHOC2, SLC2A10, SLMAP, SMAD3, SMAD4, SNTA1, SOS1, SYNE1, SYNE2, TAB2, TAZ, TBX20, TBX3, TBX5, TCAP, TECRL, TGFB2, TGFB3, TGFBR1, TGFBR2, TMEM43, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRDN, TRIM63, TRPM4, TTN, TTR, TXNRD2, VCL, ZIC3 **<u>Metabolic panel:</u>** **Glycogen metabolism disorders (29 genes)*:*** *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 aciduria (19 genes):** *ARG1, ASL, ASS1, CAD, CPS1, DHODH, FTCD, NAGS, OTC, SHMT1, SHMT2, SLC25A13, SLC25A15, SLC25A2, SLC46A1, SLC7A7, TYMP, TYMS, UMPS;* **Peroxisomal disorders (34 genes):** *ABCD1, ABCD3, ACBD5, 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 (53 genes):** *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, PDXK, PDXP, PNPO, SLC19A1, SLC46A1, SQOR, SUCLA2, SUOX, TCN1, TCN2, THAP11, TST, ZNF143;* **Leucinosis (4 genes):** *BCKDHA, BCKDHB, DBT, DLD;* **Rhabdomyolysis and fatty acid metabolism disorders (47 genes):** *ACADM, ACADVL, AGL, ALDOA, AMPD1, ANO5, ATP2A1, CACNA1S, CASQ1, CAV3, CHKB, CPT1A, CPT2, CTDP1, DGUOK, DYSF, ENO3, ETFA, ETFB, ETFDH, FKRP, FLAD1, HADHA, HADHB, ISCU, LAMP2, LDHA, LPIN1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PYGM, RYR1, SCN4A, SIL1, SLC16A1, SLC22A5, SLC25A20, SLC25A32, SLC52A1, SLC52A2, SLC52A3, TANGO2, TSEN54;* **Neurotransmitter metabolism disorders (26 genes):** *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 (13 genes):** *ATP13A2, CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, DNAJC5, GRN, KCTD7, MFSD8, PPT1, TPP1;* **Hyperuricemia (5 genes and selected SNP):** *ABCG2, HPRT1, PRPS1, REN, UMOD;* examined SNP: *GCKR – rs1260326, ALDH2 – rs671, ALDH16A1 – rs150414818, ADH1B – rs1229984;* **Other disorders:** *CADASIL (NOTCH3),* cystinuria *(SLC3A1, SLC7A9),* isovaleric aciduria *(IVD),* propionic aciduria *(PCCA, PCCB);* **<u>Skeletal disorder genes panel</u>**: *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, CYP24A1, CYP27B1, DDR2, DHCR7, DHCR24, DHODH, DLL3, DLL4, DLX3, DLX5, DMP1, DOCK6, DVL1, DYM, DYNC2H1, EBP, EFNB1,*

List of activities within the flexible scope of accreditation

	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, GNS, GPC6, HDAC8, HESX1, HOXA13, HOXD13, HRAS, HSPG2, ~~IDS~~, IFITM5, IFT43, IFT80, IFT122, IFT140, IFT172, IGF1, IGF1R, IGF2, 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, NAGLU, 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, SMAD6, 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, TRPC3, TRPS1, TRPV4, TTC21B, TWIST1, TYROBP, VDR, VIPAS39, WDR19, WDR34, WDR35, WDR60, WISP3, WNT1, WNT5A, WNT7A, XRCC2, XRCC4, XYLT1 **<u>Familial hypercholesterolaemia</u>** (11 genes and selected SNP): *ABCG5, ABCG8, APOB, APOE, LDLR, LDLRAP1, LIPA, LIPC, LIPG, PCSK9, STAP1;* *ABCG8 – rs4299376, APOB – rs1367117, APOE – rs429358, APOE – rs7412, CELSR2 – rs629301, HFE – rs1800562, LDLR – rs6511720, MYLIP – rs3757354, NYNRIN – rs8017377, PCSK9 – rs2479409, SLC22A1 – rs1564348, ST3GAL4 – rs11220462;* *CELSR2 – rs646776, LPA – rs3798220, LPA – rs10455872, SLCO1B1 – rs4149056.* **<u>Inherited diseases of the posterior segment of the eye</u>** (314 genes): *ABCA4, ABCC6, ABCD1, ABHD12, ACO2, ADAM9, ADAMTS18, ADGRV1, ADIPOR1, AGBL5, AHI1, AIPL1, ALMS1, AMACR, ARHGEF18, ARL13B, ARL2BP, ARL3, ARL6, ARMC9, ARR3, ARSG, ATF6, ATOH7, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C1QTNF5, CFAP410, PCARE, CPLANE1, CFAP418, CA4, CABP4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CDH23, CDH3, CDHR1, CEP104, CEP120, CEP164, CEP19, CEP250, CEP290, CEP41, CEP78, CEP83, CERKL, CHM, CIB2, CISD2, CLN3, CLN5, CLN6, CLN8, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL11A2, COL18A1, COL2A1, COL9A1, COL9A2, COL9A3, COQ2, CPE, CRB1, CRX, CSPP1, CTC1, CTNNA1, CTNNB1, CTSD, CWC27, CYP4V2, WHRN, DHDDS, DHX38, DNAJC5, DRAM2, DTHD1, DYNC2H1, EFEMP1, ELOVL4, EMC1, ESPN, EXOSC2, EYS, FAM161A, FDXR, FLVCR1, FRMD7, FZD4, GNAT1, GNAT2, GNB3, GNPTG, GPR143, GPR179, GRK1, GRM6, GUCA1A, GUCY2D, HARS, HGSNAT, HK1, HMX1, IDH3A, IDH3B, IFT140, IFT172, IFT27, IFT81, IMPDH1, IMPG1, IMPG2, INPP5E, INVS, IQCB1, CRPPA, JAG1, KCNJ13, KCNV2, KATNIP, KIAA0586, KIAA0753, KIAA1549, KIF11, KIF7, KIZ, KLHL7, LAMA1, LCA5, LRAT, LRIT3, LRP2, LRP5, LZTFL1, MAK, MERTK, MFN2, MFRP, MFSD8, MKKS, MKS1, MMACHC, MTTP, MVK, MYO7A, NAGLU, NDP, NEK2, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NR2F1, NRL, NYX, OAT, OCA2, OFD1, OPA1, OPA3, OPN1SW, OTX2, P3H2, PANK2, PAX2, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6D, PDE6G, PDE6H, PDSS1, PDSS2, PDZD7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, PISD, PITPNM3, PLA2G5, PLK4, PNPLA6, POC1B, POMGNT1, PPT1, PRCD, PRDM13, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, RAB28, RAX2, RBP3, RBP4, RCBTB1, RD3, RDH11, RDH12, RDH5, REEP6, RGR, RGS9, RGS9BP, RHO, RIMS1,*

List of activities within the flexible scope of accreditation

	RLBP1, ROM1, RP1, RP1L1, RP2, RPE65, RPGR, RPGRIP1, RPGRIP1L, RS1, RTN4IP1, SAG, SAMD11, SCAPER, SCLT1, SDCCAG8, SEMA4A, SGSH, SLC24A1, SLC25A46, SLC45A2, SLC7A14, SNRNP200, SPATA7, SPP2, SRD5A3, TCTN1, TCTN2, TCTN3, TEAD1, TIMM8A, TIMP3, TMEM107, TMEM126A, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TOPORS, TPP1, TRAF3IP1, TREX1, TRIM32, TRPM1, TSPAN12, TTC21B, TTC8, TTLL5, TTPA, TUB, TUBB4B, TUBGCP4, TUBGCP6, TULP1, TYR, TYRP1, USH1C, USH1G, USH2A, VCAN, VPS13B, WDPCP, WDR19, WFS1, YME1L1, ZNF408, ZNF423, ZNF513
816/2	*NOTCH3, ASPA, ACADM, BTD, HADHA, OTC, GLA, GBA, GALC, CLN2 (TPP1), CLN3, CLN7 (MFSD8), NPC1, NPC2, IDS, GCDH, ABCD1, BEST1, RS1, OVOL2, TTR.*

List of activities within the flexible scope of accreditation

3. **Laboratory for the study of mitochondrial disorders** Ke Karlovu 455/2, Praha 2

Examinations:

Ordinal Number	Analyte/ parameter/diagnostics	Principle of examination	Identification of method procedure/ equipment	Examined material	Degrees of freedom[1]
801 – Clinical Biochemistry					
1.	Determination of the profile of sialic forms of transferrin	Isoelectric focusing	SOP-KPDPM-DMP-M-10, 20/03/2023; Isoelectric focusing Model 111 Mini IEF Cell	Coagulable blood, serum	A, B, D
816 – Medical Genetics Laboratory					
1.	Examination of germline genome variants	Massive parallel sequencing	SOP-KPDPM-DMP-M-20, 9/12/2024; PP-KPDPM-DMP-M-MZ99, 1/8/2024; PP-KPDPM-DMP-M-MZ100, 9/12/2024; PP-KPDPM-DMP-M-MZ101, 1/8/2024; N-KPDPM-DMP-M-48, 1/8/2024; MiSeq	Biological material containing genomic DNA	A, B, C

List of activities within the flexible scope of accreditation

Ordinal Number	Analyte/ parameter/diagnostics	Principle of examination	Identification of method procedure/ equipment	Examined material	Degrees of freedom[1]
2.	Examination of germline genome variants	Sanger sequencing	SOP-KPDPM-DMP-M-5, 9/12/2024. PP-KPDPM-DMP-M-MM5, 1.3.2024 PP-KPDPM-DMP-M-MM8, 01/03/2024. PP-KPDPM-DMP-M-MM17, 01/03/2024; PP-KPDPM-DMP-M-MZ31, 01/03/2024; PP-KPDPM-DMP-M-MZ32, 01/03/2024; PP-KPDPM-DMP-M-MZ50, 01/03/2024; PP-KPDPM-DMP-M-MZ51, 01/03/2024; PP-KPDPM-DMP-M-MZ66, 01/03/2024; PP-KPDPM-DMP-M-MZ103, 9.12.2024 3500xL	Biological material containing genomic DNA	A, B, C
3.	Investigation of mtDNA mutations associated with LHON syndrome	RFLP	SOP-KPDPM-DMP-M-4, 01/03/2023; N-KPDPM-DMP-M-22, 01/03/2021; N-KPDPM-DMP-M-23, 20/03/2023; N-KPDPM-DMP-M-25, 01/03/2023; N-KPDPM-DMP-M-38, 01/03/2023; Thermocyclers: C1000, S1000; TProfessional	Biological material containing genomic DNA	A, B, C

List of activities within the flexible scope of accreditation

Ordinal Number	Analyte/ parameter/diagnostics	Principle of examination	Identification of method procedure/ equipment	Examined material	Degrees of freedom[1]
4.	Examination of germline genome variants	HRM	SOP-KPDPM-DMP-M-18, 9/12/2024; Annex SOP-KPDPM-DMP-M-18, 9/12/2024; N-KPDPM-DMP-M-23, 20/03/2023; N-KPDPM-DMP-M-25, 01/03/2023; N-KPDPM-DMP-M-36, 09/12/2024; Light Scanner	Biological material containing genomic DNA	A, B, C

Specification of the scope of accreditation:

Field Nr. / Ordinal Number	Detailed information on activities within the scope of accreditation
816/1	**Mitochondrial DNA genes**: *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 diseases (periodic fevers):** *ACP5, ADA2 (CECR1), ADAM17, ADAR, ALPI, ALPK1, ANGPT1, AP1S3, AP3B1, AP3D1, ARF1, ARHGAP42, ARPC1B, ATAD3A, BACH2, BLOC1S6, C1QA, C1QB, C1QC, C1R, C1S, C2, C2orf69, C3, C4A, C4B, C5, C6, C7, C8A, C8B, C8G, C9, CARD14, CARD8, CASP10, CASP8, CDC42, CEBPE, CFB, CFD, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CFP, COL7A1, COPA, CSF2, CTLA4, DHX38, DNASE1L3, DNASE2, DOCK11, DOCK8, DPP9, ELANE, ELF4, F12, FAS, FASLG, FCN3, FMNL2, FOXP3, G6PC3, HAVCR2, HAX1, HCK, HPS1, HPS4, HPS6, HS3ST6, HSPA1L, HTR1A, IFIH1, IFIT1, IKBKG, IL10, IL10RA, IL10RB, IL18BP, IL1R1, IL1RN, IL21, IL36RN, IL37, ITGAM, JAK1, LACC1, LPIN2, LRBA, LSM11, LYN, LYST, MALT1, MASP2, MBL2, MEFV, MVK, MYOF, NCF2, NCKAP1, NCKAP1L, NCSTN, NFAT5, NFKB1, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, NPC1, NRAS, OAS1, OTULIN, PIK3CG, PLCG2, PLG, PMVK, POLA1, POMP, PRF1, PRG4, PSEN1, PSENEN, PSMA3, PSMB10, PSMB4, PSMB8, PSMB9, PSMG2, PSTPIP1, PTPN2, RAB27A, RBCK1, RC3H1, RELA, RHOG, RIPK1, RNASEH2A, RNASEH2B, RNASEH2C, RNF213, RNF31, RNU7-1, SAMHD1, SERPING1, SH2D1A, SH3BP2, SHARPIN, SLC29A3, SLC7A7, STAT2, STAT4, STX11, STXBP2, STXBP3, SYK, TAP1, TAP2, TAPBP, TBK1, TGFB1, TLR7, TLR8, STING1 (TMEM173), TNFAIP3, TNFRSF1A, TREX1, TRIM22, TRNT1, TTC7A, UBA1, UNC13D, USP18, WAS, WDR1, XIAP, ZNFX1* **Mitochondrial disorder, leukodystrophy panel:** **Mitochondrial disorders***: 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,*

List of activities within the flexible scope of accreditation

	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; **Leukodystrophy**: *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, 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;*
816/2	*ATP7B, DNAJC30, EXT1, EXT2, MECP2, OPA1, SCO2, SURF1, UROD*
816/3	*NC_012920.1:m.3460G>A; NC_012920.1:m.11778G>A; NC_012920.1:m.14484T>C*
816/4	*ATP7B (p.His1069Gln), HFE (p.His63Asp, p.Ser65Cys, p.Cys282Tyr), MECP2; mtDNA (MT-ND1: m.3460G>A, MT-ND4: m.11778G>A, MT-ND6: m.14484T>C)*

Explanatory notes:

[1] Established degrees of freedom according to MPA 00-09-..:

A – Flexibility concerning the documented examination/ sample collection procedure

B – Flexibility concerning the technique

C – Flexibility concerning the analytes / parameters

D – Flexibility concerning the examined material

If no degree of freedom is specified, the laboratory cannot apply a flexible approach to the scope of accreditation for this examination.

SCID – Severe Combined Immunodeficiency

List of activities within the flexible scope of accreditation

SMA – Spinal Muscular Atrophy
mtDNA – Mitochondrial Deoxyribonucleic Acid
LHON – Leber Hereditary Optic Neuropathy
RFLP – Restriction Fragment Length Polymorphism
HRM – High Resolution Melting