Department of
Newborn Screening and Metabolic Diagnostics

Prof. Dr. med. G. Gramer
Medical Head
Newborn Screening and Metabolic Diagnostics

Center of Obstetrics, Pediatrics and Adolescent Medicine

University Hospital Hamburg-Eppendorf
Newborn Screening and Metabolic Diagnostics
Building N22, EG
Martinistr. 52
20246 Hamburg

Tel.: +49 (0) 40 7410-53737
Tel.: +49 (0) 40 7410-56717
Fax: +49 (0) 40 7410-57318
stw-labor@uke.de
www.uke.de

Request form metabolic diagnostics (external sender)

Sample number (is filled out by the laboratory)

Patient data (or label):

Name: _____

First name: _____

Date of birth: ___ / ___ / _____

Sample date: ___ / ___ / _____

Ext. Laboratory no.: _____

Payer / health insurance: _____

Private health insurance: yes ☐ no ☐

Address (patients with private health insurance):

Sender (stamp if applicable, please specify hospital ward)

Date

_____

Phone-number for queries

_____

Name of referring physician

_____

(Name, signature)

Billing address (if different from the sender)

Clinical information / diagnostic indications: *(essential for interpretation of the test results!)*

_____

_____

_____

Medication: no ☐ yes ☐ _____

Infusion: no ☐ yes ☐ _____

Special diet: no ☐ yes ☐ _____

Request: *(E- EDTA-blood, DBS- dried blood spot card, F- fibroblasts[1], S- serum, P- EDTA plasma, U- urine, CSF- cerebrospinal fluid, SW – sweat)* [2]

[1] *In case of fibroblast culture, please consult +49(0)40 7410-53737*
[2] *Information about sample volumes, sample vessels to be used and forms to be submitted can be found online in the specifications*

☐ Basic metabolic investigations (organic acids analysis and quick tests from urine, acylcarnitines from DBS, amino acids from plasma)

☐ Specific suspected diagnosis: _____

☐ Follow-up of patient (diagnosis): _____

Lysosomal enzymes

☐ Pompe disease / α-Glucosidase (E, DBS)
☐ Gaucher disease / β-Glucosidase (E, DBS)
☐ Acid sphingomyelinase deficiency (Niemann-Pick A/B-disease) / acid sphingomyelinase (E,DBS)
☐ Fabry disease / α-Galactosidase (E, DBS)

Mukopolysaccharidoses

☐ Multiplex (MPS II, IIIB, IVA, IVB, VI, VII)
- MPS II / Iduronat-2-sulfatase
- MPS IIIB / N-acetylglucosaminidase
- MPS IV A (N-acetylgalactosamine-6-sulfatase)
- MPS VI / Arylsulfatase B
- MPS VII / β-Glucuronidase
- Mukolipidose II/III

☐ MPS I / α-Iduronidase (E, DBS)
☐ MPS IIIA/ Heparan-N-sulfatase (E)
☐ MPS IIIC/ Acetyl CoA: α-Glucosaminid-N-acetyltransferase (E)

Neuronal ceroid lipofuscinoses

☐ CLN1 / PPT 1 (E, DBS)
☐ CLN2 / TPP 1 (E, DBS)

Glycoproteinosis

☐ α-Mannosidosis / α-Mannosidase (E, DBS)
☐ β-Mannosidosis / β-Mannosidase (E, DBS)
☐ α-Fucosidosis / α-Fucosidase (E, DBS)

Gangliosidoses

☐ GM1-Gangliosidosis / β-Galactosidase (E, DBS)
☐ GM2-Gangliosidosis / total-hexosaminidases (E, DBS)
☐ Hexosaminidase A (E, DBS)

Leukodystrophies

☐ Metachromatic leukodystrophy / Arylsulfatase A (E)
☐ Krabbe disease / β-Galactocerebrosidase(E, DBS)

others

☐ Lysosomal acid lipase deficiency (Wolman disease, CESD) / acid lipase (E, T)

Additional enzymes

☐ Biotinidase (E, P, DBS)
☐ Galactose-1-P-Uridyltransferase (E, DBS)

Fatty acid oxidation disorder / organic acids / quick test

☐ Acylcarnitines (E, DBS, P)
☐ Organic acids (U)
☐ Multistix (U)

Amino acids

☐ Amino acids (P)
☐ Amino acids (U)
☐ Amino acids (CSF)
☐ Diagnosis of Argininosuccinic aciduria: Argininosuccinate (DBS)
☐ Diagnosis / monitoring tyrosinemia type I: succinylacetone (DBS)
☐ Therapy monitoring MSUD: leucine, isoleucine, valine (E, P)

Fatty acids analyses

☐ Essential fatty acids (P)
☐ Very long-chain fatty acids (P)
☐ Phytanic acid (P)

Special diagnostics in urine

☐ Glykosaminoglycans (GAGs)

Additional special diagnostics

☐ Sweat test *(Kinder-UKE only)* (SW)

Additional diagnostics in blood

☐ Total galactose (E, DBS)
☐ CDG-diagnostics, T-IEF (S)