Department of
Newborn Screening and Metabolic Diagnostics

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Medical management

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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 tests

☐ Acylcarnitines (E,DBS,P)
☐ Succinyl acetone (E, DBS)
☐ Organic acids (U)
☐ Berry-Spot test (U)
☐ Sakagushi-reaction (U)
☐ Multistix (U)

Amino acids

☐ Amino acids (P)
☐ Amino acids (U)
☐ Amino acids (L)
☐ Amino acids (E,DBS)
☐ Phenylalanine (E,DBS)

Fatty acids analyses

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

Special diagnostics in urine

☐ Glykosaminoglycans (GAGs)
☐ Sulfatides (qual.) (24h-U)

Additional special diagnostics

☐ Sweat test *(Kinder-UKE only)* (SW)
☐ Total bile acids (S)
☐ Lipoprotein X (S)
☐ TIMP (S)

Additional diagnostics in blood / cerebrospinal fluid

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