Specifications

Version	Änderungen gegenüber der letzten Fassung:
01	ENGLISH VERSION
02	Update

1.1 Newborn screening

Target disease	Analyte	Material	Volume / Amount	
			Total amount: 5 spots	
Congenital Adrenal Hyperplasia (CAH)	17α-Hydroxyprogesterone	DBS	2-3 spots	**24 h**
Maple syrup urine diesease	(Iso)leucine	DBS	2-3 spots	**24 h**
Biotinidase deficiency	Biotinidase	DBS	2-3 spots	**24 h**
Carnitine-acylcarnitine translocase deficiency (CACT)	Free carnitine, several long chain acylcarnitines	DBS	2-3 Spots	**24 h**
Carnitine-palmitoyltransferase deficiency type 1 (CPT I)	Free carnitine, several long chain acylcarnitines	DBS	2-3 spots	**24 h**
Carnitine-palmitoyltransferase deficiency type 2 (CPT II)	Free carnitine, several long chain acylcarnitines	DBS	2-3 Spots	**24 h**
Classical galactosemia	Galactose-1-P-Uridyltransferase, second-tier: total galactose	DBS	2-3 spots	**24 h**
Glutaric aciduria type 1	Glutarylcarnitine	DBS	2-3 spots	**24 h**
Hypothyroidism	Thyroid-stimulating hormone (TSH)	DBS	2-3 Spots	**24 h**
Isovaleric acidemia	C5-carnitine, second-tier: pivaloyl-/isovaleryl-carnitine	DBS	2-3 spots	**24 h**
LCHAD- deficiency	several long chain acylcarnitines	DBS	2-3 spots	**24 h**
VLCAD- deficiency	several (very) long chain acylcarnitines	DBS	2-3 Spots	**24 h**
MCAD-deficiency	several medium chain acylcarnitines	DBS	2-3 Spots	**24 h**
Phenylketonuria & hyperphenylalaninemia	Phenylalanine	DBS	2-3 spots	**24 h**
Cystic fibrosis (CF)	Immunoreactive trypsinogen (IRT) Pancreatitis-associated protein (PAP) CF-genetics (31 mutations)	DBS DBS DBS	2-3 spots 2-3 spots 2-3 Spots	**24 h** **2-14 Days** **2-14 Days**
Severe combined immunodeficiency (SCID)	TREC	DBS	2-3 spots	**24 h**
Sickel cell disease (SCD)	Hemoglobin S, C, D, E, O	DBS	2-3 Spots	**24 h**
Spinal muscular atrophy (SMA)	SMN1	DBS	2-3 Spots	**24 h**

Tyrosinemia type I	Succinylacetone	DBS	2-3 spots	**24 h**

1.2 Selective metabolic diagnostics

Disease / Test	Analyte	Material	Volume / Amount		Processing Time	
					Routine	Express ***
Lysosomal enzymes			***Total amount: 3-5 spots***			
Pompe diesease	α-Glucosidase	E, DBS	2-3 spots		1-2 weeks	3 days
Gaucher disease	β-Glucosidase	E, DBS	2-3 spots	**	1-2 weeks	3 days †
Acid sphingomyelinase deficiency (Niemann-Pick A/B disease)	Acid sphingomyelinase	E, DBS	2-3 spots		1-2 weeks	3 days †
Fabry disease	α-Galactosidase	E, DBS	2-3 spots	**	1-2 weeks	3 days †
Mukopolysaccharidoses			***Total amount: 3-5 spots***			
MPS I	α-Iduronidase	E, DBS	2-3 spots	**	ca. 2 weeks	3 days †
MPS II	Iduronat-2-sulfatase	E, DBS	2-3 spots	**	ca. 2 weeks	3 days
Sanfilippo A-D (a,b,c)		E, DBS		**	ca. 2 weeks	3 days
a) MPS IIIA	Heparan-N-sulfatase	E		**	ca. 2 weeks	3 days
b) MPS IIIB	N-Acetylglucosaminidase	E, DBS	2-3 spots		ca. 2 weeks	3 days
c) MPS IIIC	Acetyl CoA: α-Glucosaminid-N-acetyltransfe-rase	E		**	ca. 2 weeks	
MPS IV A	N-Acetylgalactosamin-6-sulfatase	E, DBS	2-3 spots		ca. 2 Weeks	3 days
MPS VI	Arylsulfatase B	E, DBS	2-3 spots		ca. 2 weeks	3 days
MPS VII	β-Glucuronidase	E, DBS	2-3 spots	**	ca. 2 weeks	3 days
Mukolipidosis II/III	Mucolipidase II/III	E, DBS	2-3 spots	**	ca. 2 weeks	3 days
Multiplex (MPS II, IIIB, IVA, IVB, VI, VII)	Mucopolysaccharidoses	E, DBS	2-3 spots		ca. 2 weeks	3 days †
Neuronal ceroid lipofuscinoses			***Total amount: 3-5 spots***			
CLN1	PPT 1	E, DBS	2-3 spots	**	1-2 Weeks	3 days †
CLN2	TPP 1	E, DBS	2-3 spots	**	1-2 weeks	3 days †
Oligosaccharidoses			***Total amount: 3-5 spots***		ca. 2 weeks	
α-Mannosidosis	α-Mannosidase	E, DBS	2-3 spots	**	1-2 weeks	3 days †
β-Mannosidosis	β-Mannosidase	E, DBS	2-3 spots		1-2 weeks	3 days †
α-Fucosidosis	α-Fucosidase	E, DBS	2-3 spots		1-2 weeks	3 days †
Gangliosidoses			***Total amount: 3-5 spots***		ca. 2 weeks	

GM1-Gangliosidosis	β-Galactosidase	E, DBS	2-3 spots	**	1-2 weeks	3 days †
GM2-Gangliosidoses	Total hexosaminidase	E, DBS	2-3 spots	**	1-2 weeks	3 days †
	Hexosaminidase A	E, DBS	2-3 spots	**	1-2 Weeks	3 3 days †
Leukodystrophies			***Total amount: 3-5 spots***		ca. 2 weeks	
Metachromatic leukodystrophy	Arylsulfatase A	E		**	1-2 weeks	3 days †
Krabbe disease	β-Galaktocerebrosidase	E, DBS	2-3 Spots		1-2 weeks	3 days †
Other						
Lysosomal acid lipase deficiency (Wolman disease / CESD)	Lysosomal acid lipase	E, DBS	2-3 Spots		1-2 weeks	3 days †
Additional enzymes						
Biotinidase deficiency	Biotinidase	E, DBS P	2-3 Spots 1-2 ml		2-4 Weeks	48-72 h
Galactosemia	Galactose-1-P-uridyltransferase	DBS	2-3 Spots		24 h	24 h
Fatty acid oxidation disorders / organic acids						
Several diseases	Acylcarnitines	E, DBS	1 Spot		24-48 h	24 h
Tyrosinemia type I	Succinyl acetone	E, DBS	1 Spot		24-48 h	24 h
Several diseases	Org. acids (a,b,c)	U	5-10 ml		1-2 Weeks	24-48 h
Multistix	Multistix	U	5-10 ml		1-2 Weeks	24-48 h
Amino acids						
Several diseases	Amino acids	E, DBS P CSF	2-3 Spots 1 ml, centrifuged 1 ml	*	24-72 h	24 h
Several diseases	Amino acids	U	5-10 ml		2-4 days	24 h
Phenylketonuria	Phenylalanine	E, DBS P	2-3 Spots 1 ml, centrifuged		24 h 24-72 h	24 h
Fatty acids analyses						
Follow-up of known patients e.g. under dietary treatment	Essential fatty acids	P	1-2 ml		2-3 weeks	48-72 h
Peroxisomal disorders	Very long chain fatty acids	P	1-2 ml		2-3 weeks	48-72 h
Peroxisomal disorders	Phytanic acid	P	1-2 ml		2-3 weeks	48-72 h
Special diagnostics in urine						
Lysosomal storage diseases (MPS / multiple sulfatase deficiency)	GAGs	U	1-2 ml		2-3 weeks	48-72 h
Additional diagnostics						
Sweat test *(Kinder-UKE only) for cystic fibrosis (CF)*	Chloride	SW	50 – 100 µl		24 h	24 h
Several liver diseases	Total bile acids	S	1 ml		48-72 h	24 h

Additional diagnostics in blood						
Galactokinase deficiency (and other galactosemias)	Total galactose	E, DBS	2-3 spots		24 h	24 h
Congenital disorders of glycosylation	CDG-diagnostics, Transferrin-IEF	S	1-2 ml		2-3 weeks	48 h

* preferably cooled, only ship supernatant

** please send blood samples by express mail at the beginning of the week

EDTA-blood is required, among other, to isolate leukocytes. Please send the sample material at the beginning of the week:

- Children 3-5 ml
- Adults 5-10 ml

*** Express samples can be processed in metabolic diagnostics. Samples must be received by 11:00 a.m.

**** dripped on dried blood spot card

† An express analysis is unsually clinically unnecessary. Therefore prior agreement is required.

Processing times are not corrected for weekends and holidays.

(E- EDTA-blood, DBS- dried blood sample card, F- fibroblasts, S- serum, P- EDTA plasma, U- urine, CSF- cerebrospinal fluid, SW – sweat)

Sample vessels/materials to be used:

E – Blood collection tube (with EDTA as anticoagulant)

P - Blood collection tube (with anticoagulants like EDTA or heparin)

S – Serum tube (without anticoagulants)

DBS – Dried blood spot card

U – Tubes / screw cap container