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Center for Metabolic Diseases Heidelberg
Metabolic Laboratory
Im Neuenheimer Feld 669
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Phone +49 (0)6221-56 8276 (Mo-Thu: 08:00 - 16:30, Fr: 08:00 - 15:00)
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Outside the above mentioned hours:
Emergency phone/information desk +49 (0)6221-56 4002
Univ. Children's Hospital/Metabolic Laboratory
Im Neuenheimer Feld **430**
69120 Heidelberg - GERMANY

UNIVERSITY HOSPITAL **HEIDELBERG**

University Children's Hospital
Department of General Pediatrics
General Pediatrics, Neuropediatrics, Metabolism, Gastroenterology, Nephrology

Chairman of Pediatrics
Univ.-Prof. Dr. med. Prof. h. c. (RCH)
Georg F. Hoffmann

Request form Metabolic Laboratory

Patient data (block letters)

Name

First name

Date of birth . . ○ f ○ m ○ d

Patient ID-No.

Your lab No.

○ **Inpatient**

○ **Outpatient**

For patients from abroad, please enclose a copy of the cost coverage!

Billing address

ward

Clinical information/suspected diagnosis/diagnostic indications (essential for interpretation of test results!)

Infectious (CMV, HIV, MRSA etc.) ○ no ○ yes (details essential!), which:

Medicine/infusions ○ no ○ yes (details essential!), which:

Special diet ○ no ○ yes (details essential!), which: (medium-chain triglycerides: ○ no ○ yes)

Medical history

Birth____ weeks of gestation

Symptomatic from_____

Day/week/month/year of life

yes	no	
○	○	Consanguinity of parents
○	○	SGA
○	○	SIDS or unsolved illness in siblings: _____

General findings

○	○	Acute metabolic decompensation
○	○	Severe general illness
○	○	Coma or encephalitis
○	○	Cerebral hemorrhage
○	○	Psychomotor retardation
○	○	Progressive deterioration
○	○	Residual impairment
○	○	Failure to thrive
○	○	Recurrent vomiting

yes	no	
○	○	Hyperventilation
○	○	Dysmorphism/skeletal abnormalities _____
○	○	Short stature
○	○	Macrocephaly
○	○	Microcephaly
○	○	Abnormalities of skin/hair: _____
○	○	Abnormal odor

Organ dysfuntion

○	○	Hepatopathy
○	○	Cholestasis
○	○	Hepato-/splenomegaly
○	○	Nephropathy
○	○	Renal-tubular dysfunction
○	○	Cardiomyopathy
○	○	Ocular abnormalities: _____

yes	no	**Neurologic findings**
○	○	Seizures
○	○	Myoclonus
○	○	Muscular hypertonia
○	○	Muscular hypotonia
○	○	Ataxia, cerebellar dysfunction
○	○	Spasticity
○	○	Dystonia
○	○	Pyramidal signs
○	○	Extrapyramidal signs
○	○	Reflexes ↓/↑

Neuroradiological findings

○	○	Performed (MRI, CT, US mark as appropriate)
○	○	Normal
○	○	White matter abnormalities
○	○	Grey matter abnormalities
○	○	Malformation/lack of gyration
○	○	Supratent. atrophy
○	○	Infratent. atrophy

yes	no	**Laboratory findings**
○	○	Metabolic acidosis
○	○	Hyperlactatemia _____ mmol/l
○	○	Hypoglycemia _____ mg/dl (minimal/current)
○	○	Ketones urine/blood elevated
○	○	Anion gap increased
○	○	Hyperammonemia _____ µmol/l
○	○	CK elevated _____ U/l
○	○	ALAT/ASAT elevated
○	○	Coagulopathy
○	○	Anemia
○	○	Neutropenia
○	○	Pancytopenia

Miscellaneous:

_____

_____

_____

_____

Sender/recipient of report (stamp)

Name of referring physician (block letters), **phone-No.** (queries)

Date and signature

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Sampel data

Sample data:

Date (ddmmyy) | Time (hrmin)

- Spot urine from: _____ , _____ hour
- EDTA-plasma from: _____ , _____ hour
- Serum from: _____ , _____ hour
- Dried blood spot from: _____ , _____ hour
- CSF from: _____ , _____ hour

Date (ddmmyy)

- Collection urine from: _____ Collection duration: ____ hrs. Volume: ____ ml
- Body weight: ____ kg
- Height: ____ cm
- Last meal before: ____ hrs.
- EDTA-blood from*: _____
- Skin biopsy*/fibroblasts* from: _____ Fibroblasts + skin biopsy include cryopreservation + mycoplasma assay

Specific details for neurotransmitter diagnostics: *please provide exact sample data!*

CSF should be frozen immediately after spinal punture (-70°C). In case of blood contamination, centrifuge immediately, and freeze the supernatant!

- lumbar ○ _____ (specify type of puncture) blood-stained? ○ yes ○ no ○ xanthochrom centrifuged? ○ yes ○ no

Fractions of CSF **(20 droplets each, in infants min. 12 droplets)** ○ 1. (your lab) ○ 2. ○ 3. ○ 4. ○ 5. (2. - 5. metabolic laboratory)

Requests

Metabolic investigations (phone: +49 (0)6221-56 8276)

- **Basic investigations:** Organic acids (U)[1, 9], amino acids (P)[2, 7, 16], acylcarnitine profile (DBS)[11, 12], glycosaminoglycans (U)[1, 9], basic metabolic tests (U)[1, 9]

Metabolic tests:

- **Organic acids (U)**[1, 9] (total profile)

Exact quantification by stabile Isotopes

- Methylmalonic acid (MMA) (U)[1, 9]
- Methylmalonic acid (MMA) (P)[2, 7]
- Mevalonic acid (MVA) (U)[1, 9]
- 3-OH-Glutaric acid (3OHGA) + Glutaric acid (GA) (U)[1, 9]

- **Amino acids (P)**[2, 7, 16]
- Amino acids (L)[4, 8]
- Amino acids (U)[1, 9]
- **Acylcarnitine profile (DBS)**[11, 12]
- Acylcarnitine profile (P)[2, 7]
- Carnitine status (P)[2, 7]
- Free fatty acids/ketones (P)[2, 15]
- L-lactate (L)[4, 8]
- L-lactate/creatinine ratio (U)[1, 9]
- D-lactate (U)[1, 9]
- Homocysteine (P)[2, 7, 16]
- SAM + SAH (P)[2, 8]
- Orotic acid/orotidine (U)[1, 9]
- Homocitrulline (U)[1, 9]
- Total galactose (DBS)[11, 12]
- GALT activity (DBS)[11, 12] *

- Succinylacetone (DBS)[11, 12]
- Biotinidase activity (DBS)[11, 12] *
- Biotinidase activity (S)[2, 15, 18] *
- Purines/pyrimidines (U)[1, 9]
- Sulfocysteine (U)[1, 9]
- Polyols (U)[1, 9]
- Trimethylamine (U)[1, 14]
- Total bile acids (S)[2, 7, 18]
- Essential fatty acids (P)[2, 7, 16]
- Glutathione (EV)[3, 13, 14]
- Sterols (P)[2, 7]

- CDG diagnostics (S)[4, 7, 18] (in case of abnormalities in the initial analysis (MS-TOF), further analyses follow)

Creatine deficiency syndromes (guanidino compounds)

- **Urine**[1, 9] ○ Plasma[2, 7] ○ CSF[4, 8]

Therapy control for PKU or MSUD

- Phenylalanine, tyrosine (DBS)[11, 12]
- Phenylalanine, tyrosine (P)[4]
- Leucine, isoleucine, valine (P)[4]

Peroxisomal diagnostics and pyridoxine dependent epilepsy

- Very long chain fatty acids (VLCFA) (P)[2, 7]
- Phytanic acid (P)[2, 7]
- Plasmalogens (EV)[3, 13]
- Bile acid metabolites (U)[1, 9]
- Pipecolic acid + AASA/P6C (U)[1, 9]
- Pipecolic acid + AASA/P6C (P)[2, 7]
- Vitamine B_6-metabolites, incl. pyridoxal phosphate (L)[8]

Neurotransmitter diagnostics (NT diagnostics)

- **NT basic diagnostics:** biogenic amines (L)[8], 5-MTHF (L)[8], pterins (biopterin + neopterin) (L)[8] and amino acids (L[8] + P[2, 8, 16])

- Biogenic amines (L)[8]
- 5-MTHF (L)[8]
- Pterins (L)[8]
- Pterins and DHPR activity (DBS)[11, 12] *
- Sepiapterin (L)[8]
- DHPR activity (DBS)[11, 12] *
- Pterins (U)[1, 8, 10]
- Serotonin (EV)[3, 8]
- AADC activity (P)[3, 8] *
- 3-*o*-methyldopa (3-OMD) (DBS)[11, 12]

For lysosomal diagnostics (please see page 3)

*** Requires declaration of consent according to the Gene Diagnostics Act §8, para. 1;** not necessary for the follow-up of individuals with known diagnosis.

Informations on individual requests

U	Urine	P	EDTA plasma	S	Serum[18]	DBS	Dried blood spots (3-5 circles)	F	Fibroblasts
24U	24-hour urine	EV	EDTA whole blood	L	CSF			H	Skin biopsy

1. 10 ml
2. 1 ml
3. 2 ml
4. 0.5 ml
5. 5 - 10 ml infants 2.5-5 ml
6. 2 x 10 ml
7. If possible, store at -20°C until shipment; send the frozen sample wrapped in a frozen cold pack or similar.
8. Put directly on dry ice; shipment on dry ice
9. Preservation with 4-6 droplets diclhloromethane, shipment at room temperature
10. Protect from light immediately
11. Please allow to dry at room temperature for 2 hrs.
12. Shipment at room temperature
13. Shipment within 24 hrs. at room temperature
14. Request for detailed instructions
15. Shipment on dry ice
16. 4 hrs. after last meal
17. Please ask the laboratory for a list of analysis days!
18. After centrifugation transfer clear supernatant into a labelled fresh tube; shipment as specified in the analysis

Optimal shipping method for EDTA plasma, serum and urine is shipping on dry ice.

UK HD

LYSOSOMAL DIAGNOSTICS

LYSOSOMAL SCREENING DIAGNOSTICS IN URINE

Oligosaccharide thin layer chromatography (U)	Suspicion of oligosaccharidosis (e.g. α-mannosidosis), as well as GM1-gangliosidosis, sialidosis, galactosialidosis
Total glycosaminoglycans (GAGs) (U) (if abnormal, then MPS electrophoresis)	Suspicion of mucopolysaccharidosis (MPS)
MPS-electrophoresis (+ GAGs) (U)	
Neuraminic acid (free + bound) (U)	Suspicion of free sialic acid storage disease (FSASD), sialidosis, galactosialidosis, sialuria, GM1-gangliosidosis
Sulfatides (U)	Suspicion of metachromatic leukodystrophy (MLD), saposin B defect, multiple sulfatase deficiency

LYSOSOMAL SCREENING DIAGNOSTICS IN BLOOD [a, b]

Neurodegenerative screening (EV+DBS)[c]	[chitotriosidase, Gaucher, MLD, Krabbe, GM1, GM2, MPS IIIB, MPS VII, α- and β-mannosidosis, fucosidosis, aspartylglucosaminuria, Schindler, mucolipidosis II/III, NCL1, NCL2][d]
Liver screening (EV+DBS)[e]	[chitotriosidase, Gaucher, ASMD (Niemann-Pick A/B), Niemann-Pick C, GM1, GM2, MPS IIIB, MPS VII, α- and β-mannosidosis, fucosidosis, mucolipidosis II/III, LAL-D (Wolman)]
Hydrops screening (EV)	[chitotriosidase, Gaucher, ASMD (Niemann-Pick A/B), GM1, GM2, MPS I, MPS II, MPS IIIB, MPS IVA, MPS VI, MPS VII, α- and β-mannosidosis, fucosidosis, mucolipidosis II/III, LAL-D (Wolman), multiple sulfatase deficiency][f, g]

Requests (specific suspicion of lysosomal storage disease, if necessary in addition to urine investigation)

Disease	**Enzyme(s)** [a] (incl. one reference enzyme per material; preferred material in **bold** letters)	**Metabolites (P)** (also for monitoring the follow-up of a known diagnosis)
α-mannosidosis	α-mannosidase (**EV**, DBS, H/F)	
ASMD (Niemann-Pick A/B)	Acid sphingomyelinase (**EV**, H/F)	lysoSM + lysoSM509
Aspartylglucosaminuria	Aspartylglucosaminidase (**EV**, H/F)	
β-mannosidosis	β-mannosidase (**EV**, DBS, H/F)	
Cystinosis		Cystine in **leukocytes (PFA)**
Fabry	α-D-galactosidase A (**EV**, DBS, H/F)	lysoGL-3 (lysoGb3)
Fucosidosis	α-L-fucosidase (**EV**, DBS, H/F)	
Galactosialidosis	β-galactosidase + neuraminidase (**H/F**)	
Gaucher	Glucocerebrosidase (**EV**, H/F)	lysoGL-1 (lysoGb1)
GM1-gangliosidosis	β-galactosidase (**EV**, DBS, H/F)	lysoGM1
GM2-gangliosidosis - type Sandhoff	β-hexosaminidase total (**EV**, DBS, H/F) (N-acetyl-β-D-glucosaminidase total)	lysoGM2
GM2-gangliosidosis - type Tay-Sachs	β-hexosaminidase A (**EV**, DBS, H/F) (N-acetyl-β-D-glucosaminidase A)	lysoGM2
GM2-gangliosidosis - AB-variant (GM2-activator-defect)		lysoGM2
Krabbe	Galactocerebrosidase (**EV**, H/F)	Psychosine
LAL-D (Wolman, CESD)	Acid lipase (EV, **DBS,** H/F)	
Metachromatic leukodystrophy (MLD)	Arylsulfatase A (**EV**, H/F)	Sulfatides (U, see above)
Mucolipidosis II (I-Cell-disease)	Several lysosomal enzymes (EV, **P**, DBS, H/F)	
Mucolipidosis III	Several lysosomal enzymes (EV, **P**, DBS, H/F)	
MPS I (Hurler/Scheie)	α-iduronidase (**EV**, DBS, H/F)	
MPS II (Hunter)	Iduronate-sulfatase (**EV**, DBS, H/F)	
MPS IIIA (Sanfilippo A)	Heparan-N-sulfatase (**EV**, H/F)	
MPS IIIB (Sanfilippo B)	α-N-acetyl-glucosaminidase (**EV**, DBS, H/F)	
MPS IIIC (Sanfilippo C)	Acetyl-CoA: α-glucosamine-acetyltransferase (**EV**, H/F)	
MPS IIID (Sanfilippo D)	α-N-acetyl-glucosamine-6-sulfatase (**EV**, H/F)	
MPS IVA (Morquio A)	Galactose-6-sulfatase (**EV**, H/F)	
MPS IVB (Morquio B)	β-galactosidase (**EV**, DBS, H/F)	
MPS VI (Maroteaux-Lamy)	Arylsulfatase B (**EV**, DBS, H/F)	
MPS VII (Sly)	β-glucuronidase (**EV**, DBS, H/F)	
Multiple sulfatase deficiency	Several sulfatases (**EV**, DBS, H/F)	Sulfatides (U, see above)
Neuronal ceroid lipofuscinosis infantile (NCL1)	Palmitoyl-protein-thioesterase 1 (PPT1) (EV, **DBS**, H/F)	
Neuronal ceroid lipofuscinosis late infantile (NCL2)	Tripeptidylpeptidase 1 (TPP1) (EV, **DBS**, H/F)	
Niemann-Pick (NP) C		lysoSM509 + lysoSM
Pompe (GSD II)	α-glucosidase (acid maltase) (EV, **DBS**, H/F)	
Schindler	N-acetyl-α-D-galactosaminidase (**EV**, H/F)	
Sialidosis	Neuraminidase (**H/F**)	Neuraminic acid (U, see above)
Sialic acid storage disease (FSASD)		Neuraminic acid (U, see above)
Sphingolipidoses, screening of (e.g. Gaucher, ASMD)	Chitotriosidase (EV, DBS, **P**)	lysoSphingolipids[h]

(U: Urine[1, 9], EV: EDTA blood[5, 13], P: EDTA plasma[2, 7], DBS: Dried blood spot[11, 12], PFA: PFA blood[3, 13, 17], H/F: Skin biopsy/fibroblasts[a, 13, 14])

a: Requires informed consent according to the Gene Diagnostics Act §8, para. 1; not necessary in case of follow-up of known diagnosis.
b: If there are abnormalities in the plasma/serum and/or dried blood spot further examination in leukocytes and/or dried blood spot is carried out if possible.
c: Enzyme analysis only
d: In addition, MPS electrophoresis and neuraminic acid in urine recommended
e: Enzyme analysis and lysoSM509 + lysoSM
f: In addition, neuraminic acid in urine recommended
g: In addition, oligosaccharide thin layer chromatography and neuraminic acid in urine recommended
h: Contains all lysosomal metabolites in plasma indicated in the column “Metabolites”.

Analysis	Indication	Material (preferred material in bold letters)
Acylcarnitine profile	**Basic investigation** Suspicion of fatty acid oxidation disorders, organoacidemia (e.g . methylmalonic aciduria, propionic aciduria, isovaleric aciduria and glutaric aciduria type I)	**DBS** EDTA plasma
Amino acids in plasma	**Basic investigation** Suspicion of aminoaciduria, hyperammonemia, disorders of energy metabolism	**EDTA-Plasma,** 4 hrs. after last meal
Amino acids in CSF	Suspicion of neurometabolic disease, especially epileptic encephalopathies	CSF and EDTA plasma
Amino acids in urine	Renal tubular dysfunction, suspicion of cystinuria, lysinuric protein intolerance	Urine
Biotinidase activity	Metabolic acidosis, neurological abnormalities, muscular hypotonia, developmental retardation, eczema; K_m-variants, hepatic glycogenoses	DBS Serum
Carnitine status	Primary or secondary carnitine deficiency	**EDTA plasma**/serum additional urine in case of special questions
CDG diagnostics	N-/O-proteinglycosylation defects: multi systemic disease, hepatopathy, psychomotor retardation	Serum
Creatine deficiency syndromes	Suspicion of creatine deficiency syndromes: guanidinoacetate methyltransferase (GAMT) deficiency, arginine:glycine amidinotransferase (AGAT) deficiency, creatine transport defect	**24-hour urine/urine** EDTA plasma (CSF)
Essential fatty acids	Follow-up on a low-fat diet	EDTA plasma, preprandial
Free fatty acids/ ketones	Assessment of endogenous lipolysis in hypoglycemia or as part of a fasting test	EDTA plasma
GALT activity (Gal-1-P-uridyltransferase)	Neonatal hepatopathy, suspicion of classical galactosemia	DBS
Galactose total	Disorders of galactose metabolism, follow-up of galactosemia	DBS
Homocitrulline	Suspicion of HHH-syndrome	Urine
Homocysteine	Suspicion of homocystinuria or hyperhomocysteinemia (thrombembolism, early vascular disorder); suspicion of remethylation defects (progressive neuropathy, unclear developmental retardation, megaloblastic anemia, microcephaly); suspicion of vitamine B_{12} deficiency	EDTA plasma, preprandial
D-lactate	Suspicion of bacterial overgrowth in short bowel syndrome	Urine
L-lactate in CSF	Suspicion of energy metabolism disorder	CSF
L-lactate/creatinine in urine	Suspicion of energy metabolism disorder, hepatic glycogenoses	Urine
Lysosomal diagnostics	See page 3	
Neurotransmitter: **Basic investigation (biogenic amines, 5-MTHF, pterins, amino acids)**	Dystonia, extrapyramidal movement disorders, severe encephalopathies, (axial) muscular hypotonia, okulogyric crises	Always CSF + EDTA plasma *see sample data!*
5-MTHF	Suspicion of disorder of folate cycle, progressive encephalopathy/neuropathy	CSF see sample data!
Serotonin	Suspicion of VMAT2 deficiency or serotonin deficiency	EDTA blood
3-OMD	Suspicion of AADC deficiency	DBS
AADC activity (aromatic L-amino acid decarboxylase)	Confirmation of suspected AADC deficiency with typical CSF results only!	EDTA plasma *request instruction sheet*
Pterins + **DHPR activity**	Unclear hyperphenylalaninemia suspicion of BH4 defects	CSF, urine, DBS DBS
Organic acids	**Basic investigation** Suspicion of organoaciduria or other disorders of intermediar metabolism (hypoglycemia, unclear coma), neurometabolic disorders, tyrosinemia type I	**Urine**
Orotic acid, orotidine	Suspicion of urea cycle disorder (especially OTC deficiency), hereditary orotic aciduria	Urine
Peroxisomal diagnostics	Multi systemic disease with muscular hypotonia, encephalopathy, epilepsy, hepatopathy, skeletal abnormalities, dysmorphism	
Very long chain fatty acids (VLCFA)	Suspicion of X-ALD/AMN; peroxisomal biogenesis defect (Zellweger spectrum)	EDTA plasma
Phytanic acids	Suspicion of Refsum disease	EDTA plasma
Plasmalogens	Suspicion of rhizomelic chondrodysplasia punctata; peroxisomal biogenesis defect (Zellweger spectrum)	EDTA blood
Bile acid metabolites	Suspicion of bile acid synthesis disorder, peroxisomal biogenesis defect (Zellweger spectrum)	Urine
Pipecolic acid, aminoadipinic acid semialdehyde (AASA), Piperideine-6-carboxylate (P6C)	Suspicion of antiquitin 1 deficiency (pyridoxin dependent epilepsy) (P, U) Suspicion of molybdenum cofactor deficiency, sulfite oxidase deficiency (U)	EDTA plasma, urine Urine
Polyoles	Disorders of pentose phosphate pathway and polyols (e.g. transaldolase deficiency)	Urine
Purines/pyrimidines	Suspicion of Lesch-Nyhan syndrome, molybdenum cofactor deficiency; multi systemic disorder with neurologic, renal and/or immunologic manifestation (developmental retardation, epilepsy, movement disorder, myopathy, urinary stones, immune deficiencies)	Urine, if possible 24-hour urine
SAM and SAH S-adenosyl-methionine and S-adenosyl-homocysteine	Suspicion of methylation disorders (MAT I/III deficiency, GNMT deficiency, SAHH deficiency, ADK deficiency)	**EDTA plasma**, CSF
Sterols	Suspicion of Smith-Lemli-Opitz syndrome (SLOS), cerebrotendinous xanthomatosis (CTX), desmosterolosis, lathosterolosis, Conradi-Hünermann syndrome (CDPX2), sitosterolemia	EDTA plasma
Succinyl acetone	Suspicion of tyrosinemia type I, unclear hepatopathy, hypertyrosinemia	DBS Urine (organic acids)
Sulfocysteine	Suspicion of molybdenum cofactor deficiency, sulfite oxidase deficiency	Urine
Trimethylamine	Suspicion of trimethylaminuria	Urine, *request instruction sheet!*

UNIVERSITY
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Angelika-Lautenschläger-Klinik
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Informed consent for enzymatic/genetic analyses (Please send with the request form!)

(Mandatory for enzyme activity determinations, skin biopsies/fibroblast cultures and genetic analyses!)

Patient ☐ **female** ☐ **male** ☐ **diverse**	**Sender:** Hospital/ward/ambulance/physician, incl. phone/Fax
Name: First name: DOB: Address:	

Requested analyses (Enter below or refer to request form dated .....)

With your signature below you confirm the following statements:

- You have been informed by your doctor about the significance and consequences of the above-mentioned investigation.
- You were given sufficient time to think about it before agreeing to the investigation.
- You consent to the necessary sampling and biochemical/genetic analysis, which are carried out to clarify the (suspected) diagnosis

I agree that

yes	no	
☐	☐	the test results obtained after the reports have been made may be stored for more than the maximal necessary period of 10 years without making a claim. *(Storage of results after reporting according to GenDG §12)*
☐	☐	the sample material is stored for the purpose of a possibly required or desired check of the result or further examinations required for diagnosis. *(Use and discard of genetic samples according to GenDG §13)*
☐	☐	the sample material may be used pseudonymised for laboratory analysis quality control measures or scientific purposes. *(Use and discard of genetic samples according to GenDG §13)*
☐	☐	written notification of the investigation results is also sent to the following treating physicians: _____ (enter name(s)). *(Notification of the results of genetic investigations and analyses according to GenDG §11).*

We would like to point out that you can withdraw this consent at any time. In this case, the investigation will be discontinued and only the service provided up to that point will be billed.

City and date	**Name** (block letters)	**Signature of patient or of the legal representative**

City and date	**Name** (block letters)	**Signature of the informing physician according to GenDG §8 para. 1**

Patient information sheet for our informed consent form for enzymatic and genetic laboratory tests

Genetic examinations are subject to the regulations of the Gene Diagnostics Act (GenDG, effective: 01.02.2010). In order to carry out the corresponding investigations, the laboratory commissioned must have a patient consent form. In our declaration of consent, we also ask you to decide on the storage of the sample material and the data obtained in the requested investigation. The explanations below serve as a decision-making aid.
The consent for storage can be withdrawn at any time without giving reasons and without personal disadvantages. In the event of withdrawal, the examination material and/or the examination result will be destroyed immediately.
All information provided on consent forms, request forms or accompanying slips as well as all examination results are subject to medical confidentiality. They will only be passed on with the written consent of the patient/legal representative.

Storage of sample material

According to the terms of the Gene Diagnostics Act, surplus examination material may only be stored with the explicit consent of the patient/legal representative after the examination has been completed. However, surplus sample material could serve to verify our results and is also needed in diagnostics for necessary quality controls. Furthermore, surplus sample material is an important source for research and development work in the field of medical-genetic/biochemical diagnostics. For these purposes, the sample material is pseudonymised.

Storage of examination results

Many genetic diseases have consequences for other family members and children they may have in the future.
The results of a genetic examination of a family member can be significant for the examinations of further members or descendants of this family. This data is lost if it is destroyed. However, storage of the examination results beyond the legally prescribed period of 10 years is only possible with the consent of the patient/legal representative.

Notes on enzymatic/genetic tests

General notes

The results of enzymatic/genetic laboratory tests can have far-reaching consequences for life and family planning. However, no technical procedure is completely free of sources of error. We would therefore like to explain to you in the following which errors are possible in the collection and interpretation of biochemical/genetic laboratory results.

A major source of errors in medical laboratory diagnostics are sample mix-ups. Every effort is made to avoid these. Two important peculiarities come into play here:

- Often, members of the family are examined in addition to individuals. A valid test interpretation is then dependent on the correct indication of the family relationships.
- When obtaining cell material for prenatal examination, a mixing of fetal and maternal tissue cannot be completely ruled out, so that the subsequent analysis can lead to misleading results as a consequence.

Data processing/protection

Responsible for data processing:
Center for Metabolic
Diseases Heidelberg
Metabolic Laboratory
Im Neuenheimer Feld 669
69120 Heidelberg - GERMANY
Phone: +49 (0)6221-56 8276
Fax: +49 (0)6221-56 5565

Responsible Data Protection Officer:
Data Protection Officer
University Hospital Heidelberg
Im Neuenheimer Feld 672
69120 Heidelberg - GERMANY
E-Mail: Datenschutz@med.uni-heidelberg.de

Responsible regulatory institution:
The State Commissioner for Data Protection and
Freedom of Information of Baden-Württemberg
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Königstraße 10a, 70173 Stuttgart - GERMANY
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E-Mail: poststelle@lfdi.bwl.de
Internet: http://www.baden-wuerttemberg.datenschutz.de

You can find this form on our homepage on the internet under request form and informed consent.