Amsterdam UMC
University Medical Centers

Laboratory Genetic Metabolic Diseases

Print Form

Test request form Enzyme diagnostics

Please fill out this form completely (grey fields are mandatory) and send it in together with the sample(s).

Patient information

Family name	:	.....
First name	:	.....
Date of birth	:	Day ..... Month ..... Year .....
Sex	:	Male/Female
Address	:	.....
ZIP code	:	.....
Country	:	.....

Requested test(s) (see page 3, 4, 5 and www.labgmd.nl)

Enzyme and/or disorder: .....

☐ Prenatal analysis

Material*

For prenatal testing please contact the laboratory before sending samples

	sampling:			sampling:	
☐ Blood (EDTA)*	date	time	☐ Chorion villi sample	date	time .....
☐ Erythrocytes	date	time	☐ Chorion villi fibroblasts	date	time .....
☐ Plasma	date	time	☐ Amniocytes	date	time .....
☐ Blood spot	date	time	☐ Tissue ; specify :		
☐ Skin biopsy	date	time		date	time .....
☐ Skin fibroblasts	date	time	☐ Other ; specify :		
				date	time .....

* Blood must arrive within 48 hours after collection. For detailed specification of transport conditions see www.labgmd.nl

Relevant clinical and laboratory findings

Clinical laboratory geneticists:
Dr. S. Ferdinandusse *Dr. A.B.P. van Kuilenburg*
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Lab GMD (F0-132)
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Fax: +31(0)20-696 2596

TESTING RvA M 154

Results should be sent to

Name	:
Department	:
Hospital/institute	:
Address	:
City and Zip-code	:
Country	:
Phone	:
Fax	:
E-mail*	:

* *For privacy reasons results will be faxed. Results can only be sent by email if a secure email option is provided. Please provide email address for correspondence.*

Copy results should be sent to

Name	:
Department	:
Hospital/institute	:
Address	:
City and Zip-code	:
Country	:
E-mail	:

Invoice should be sent to*

Name	:
In case of institution	
Department	:
Hospital/institute	:
Address	:
City and Zip-code	:
Country	:
E-mail of financial contact	:
VAT number	:

* *Be sure to include all information needed by the financial department of your institution.*

* *For EU countries only:*
VAT number of your institution must be provided.
Original S2 forms (formerly E 112) should be filled out completely and can be sent in together with the sample(s) or separately.

Form completed by

Name	:
Function/Department	:
Date	:
Signature	:

Please note that without the above requested information the requested test(s) cannot be performed.

Tests Enzyme diagnostics

Peroxisomal metabolism

☐ **Screening peroxisomal defects**	F	Zellweger spectrum defects (ZSD), Acyl-CoA oxidase 1 deficiency (ACOX1), D-Bifunctional protein deficiency (DBP), X-linked adrenoleukodystrophy (XALD), Rhizomelic Chondrodysplasia Punctata (RCDP)
☐ Very long-chain fatty acids	F	ZSD, XALD, ACOX1, DBP
☐ C26:0 lysoPC	F	ZSD, XALD, ACOX1, DBP
☐ Dihydroxyacetonephosphate-acyltransferase (DHAPAT)	F	ZSD, RCDP
☐ Immunofluorescence catalase	F	ZSD, ACOX1, DBP
☐ Immunofluorescence ALDP (adrenoleukodystrophy protein)	F	XALD
☐ Acyl-CoA oxidase 1 (ACOX1)	F	ACOX1 deficiency
☐ D-Bifunctional protein (DBP/MFP2)	F,BL	DBP deficiency
☐ Peroxisomal thiolase branched-chain (Sterol Carrier Protein X)	F	SCPx deficiency
☐ Phytanic acid α -oxidation	F	Refsum disease
☐ Peroxisomal β-oxidation	F	ZSD, XALD, AMACR, SCPx, ACOX1, DBP
☐ Immunoblot peroxisomal proteins	F	ZSD, RCDP
☐ Plasmalogens	F	RCDP, ZSD

Mitochondrial fatty acid oxidation

☐ **Screening mitochondrial β-oxidation (acylcarnitine profiling)**	F	Deficiency of VLCAD, LCHAD/MTP, CPT2, CACT, MCAD, SCAD, Multiple acyl-CoA dehydrogenase deficiency (MADD/Glutaric aciduria type 2)
☐ Oleate β-oxidation (flux assay)	F	Deficiency of VLCAD, LCHAD/MTP, CPT2, CACT, MADD
☐ Plasmamembrane carnitinetransporter (OCTN2)	F	Systemic/primary carnitine deficiency
☐ Carnitine palmitoyltransferase 1 (CPT1)	F	CPT1 deficiency
☐ Carnitine palmitoyltransferase 2 (CPT2)	F,BL	CPT 2 deficiency
☐ Mitochondrial carnitine/acylcarnitine transporter (CACT)	F	CACT deficiency
☐ Very long-chain acyl-CoA dehydrogenase (VLCAD)	F,BL	VLCAD deficiency
☐ Mitochondrial trifunctional protein (MTP)	F,BL	LCHAD/MTP deficiency
Long-chain 3-hydroxy-acyl-CoA dehydrogenase (LCHAD)		
Long-chain 3-ketothiolase		
☐ Medium-chain acyl-CoA dehydrogenase (MCAD)	F,BL	MCAD deficiency
☐ Short-chain acyl-CoA dehydrogenase (SCAD)	F,BL	SCAD deficiency
☐ Short-chain enoyl-CoA hydratase/Crotonase (ECHS1)	F,BL	ECHS1/Crotonase deficiency
☐ Short-chain 3-hydroxy-acyl-CoA dehydrogenase (SCHAD)	F,BL	SCHAD deficiency

Amino acid metabolism

☐ Glutaryl-CoA dehydrogenase (GCDH)	F,BL	Glutaric aciduria type 1
☐ Short-chain enoyl-CoA hydratase/Crotonase (ECHS1)	F,BL	ECHS1/Crotonase deficiency
☐ 3-Hydroxy-isobutyryl-CoA hydrolase (HIBCH)	F	HIBCH deficiency
☐ 3-Hydroxy-isobutyric acid dehydrogenase (HIBADH)	F	HIBADH deficiency, 3-Hydroxy-isobutyric aciduria
☐ Methylmalonate semialdehyde dehydrogenase (MMSDH)	F,BL	MMSDH deficiency, 3-Hydroxy-isobutyric aciduria
☐ Propionyl-CoA carboxylase (PCC)	F,BL	Propionic acidemia
☐ Short branched-chain acyl-CoA dehydrogenase (SBCAD)	F,BL	SBCAD deficiency
☐ 2-Methyl-3-hydroxybutyryl-CoA dehydrogenase (MHBD)/ Short-branched-chain hydroxyacyl-CoA dehydrogenase (SBCHAD)	F,BL	MHBD/SBCHAD deficiency
☐ Isovaleryl-CoA dehydrogenase (IVD)	F,BL	Isovaleric acidemia
☐ 3-Methyl-crotonyl-CoA carboxylase (MCC)	F	MCC deficiency
☐ 3-Methyl-glutaconyl-CoA hydratase (MGH)	F,BL	3-Methylglutaconic aciduria type 1
☐ 3-Hydroxy-3-methylglutaryl-CoA lyase (HMGCoA lyase)	F,BL	HMGCoA lyase deficiency

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See next page for Lysosomal Storage Disorders

Tests Enzyme diagnostics continued

Purine and Pyrimidine metabolism

☐	Dihydropyrimidine dehydrogenase (DPD)	F,BL,L	DPD deficiency
☐	Dihydropyrimidinase (DHP)	L	DHP deficiency
☐	ß-Ureidopropionase (ß -UP)	L	ß-UP deficiency
☐	Thiopurine methyltransferase (TPMT)	BL,Ery	TPMT deficiency
☐	UMP synthase	BL,Ery	UMP synthase deficiency, orotic aciduria
☐	Phosphoribosyl pyrophosphatesynthetase (PRPPs)	BL,Ery	PRPPs deficiency and PRPPs superactivity
☐	Thymidine phosphorylase (TP)	BL,Bsp	Mitochondrial neurogastrointestinal encephalopathy (MNGIE)
☐	Adenosine deaminase (ADA)	BL,Bsp	Severe combined immunodeficiency (SCID)
☐	Adenosine deaminase 2 (ADA2)	BL,Pla	Adenosine deaminase 2 deficiency
☐	Purine nucleoside phosphorylase (PNP)	BL,Ery	Severe combined immunodeficiency (SCID)
☐	Hypoxanthine-guanine phosphoribosyltransferase (HGPRT)	BL,Ery	Lesch-Nyhan syndrome

Carbohydrate degradation

☐	Galactose-1-phosphate uridyltransferase (GALT)	BL	Galactosemia type 1, classic galactosemia
☐	Galactokinase (GALK)	BL	Galactosemia type 2
☐	UDP galactose-4-epimerase (GALE)	BL	Galactosemia type 3
☐	Glucose 6-phosphate dehydrogenase (G6PD)	BL	G6PD deficiency
☐	Pyruvate kinase (PK)	BL	Pyruvate kinase deficiency

Ketolysis defects

☐	Succinyl-CoA : 3-oxoacid transferase (SCOT)	F,BL	SCOT deficiency
☐	ß -ketothiolase (2-methyl-acetoacetyl-CoA specific)	F,BL	ß-ketothiolase deficiency

Cholesterol/isoprenoid biosynthesis

☐	**Screening cholesterol biosynthesis defects**	F	Smith-Lemli-Opitz syndrome (SLO), Desmosterolosis,
	(Sterol profiling)		Conradi-Hunermann syndrome, Lathosterolosis and Greenberg dysplasia
☐	Mevalonate kinase (MVK)	F,BL	Hyper IgD syndrome/mevalonic aciduria

Neurotransmitter metabolism

☐	Aromatic amino acid decarboxylase (AADC) *	Pla	Aromatic amino acid decarboxylase deficiency

Hyperoxaluria

☐	Glyoxylate reductase (GR) *	BL,L	Hyperoxaluria type 2

Miscellaneous enzymes

☐	Fatty aldehyde dehydrogenase (FALDH, SLS)	F,BL	Sjögren Larsson syndrome
☐	Steroidsulfatase (arylsulfatase C, ARYC)	BL,F	X-linked ichthyosis
☐	Biotinidase	BL,Pla	Biotinidase deficiency

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Tests Lysosomal Storage Disorders

Mucopolysaccharidoses

☐ α-L-iduronidase	BL,F	MPS Type IH/IS (Hurler/Scheie)
☐ Iduronate sulfatase	BL,F	MPS Type II (Hunter)
☐ Sulfamidase	BL,F	MPS Type III A (Sanfilippo A)
☐ N-acetyl-α -D-glucosaminidase	BL,F	MPS Type III B (Sanfilippo B)
☐ AcetylCoA-glucosamine acetyltransferase	BL,F	MPS Type III C (Sanfilippo C)
☐ N-acetylglucosamine-6-sulfatase	BL,F	MPS Type III D (Sanfilippo D)
☐ N-acetylgalactosamine-6-sulfatase	BL,F	MPS Type IV A (Morquio A)
☐ ß-D-galactosidase	BL,F	MPS Type IV B (Morquio B)
☐ Arylsulfatase B	BL,F	MPS Type VI (Maroteaux-Lamy)
☐ ß-D-glucuronidase	BL,F	MPS Type VII (Sly)

Mucolipidoses

☐ N-acetyl-α-D-neuraminidase	F	Mucolipidosis Type I
☐ N-acetyl-ß-D-glucosaminidase	BL,F	Mucolipidosis Type II and III (I-cell disease, pseudo-Hurler polydystrophy)

Oligosaccharidoses

☐ α-L-fucosidase	BL,F	Fucosidosis
☐ α-D-mannosidase	BL,F	α-Mannosidosis
☐ ß-D-mannosidase	BL,F	ß-Mannosidosis
☐ N-acetyl-α -D-galactosaminidase	BL,F	Schindler / Kanzaki
☐ Aspartylglucosaminidase	BL,F	Aspartylglucosaminuria
☐ Protective protein / Cathepsine A	BL,F	Galactosialidosis

Sphingolipidoses

☐ Arylsulfatase A	BL,F	Metachromatic leukodystrophy
☐ Arylsulfatase A+B	BL,F	Mucosulfatidosis / Multiple sulfatase deficiency
☐ α-D-galactosidase	BL,F	Fabry
☐ ß-D-galactosidase	BL,F	GM-1 gangliosidosis
☐ N-acetyl-ß-D-glucosaminidase A	BL,F	Tay-Sachs / GM-2 gangliosidosis B variant
☐ N-acetyl-ß-D-glucosaminidase A+B	BL,F	Sandhoff / GM-2 gangliosidose 0 variant
☐ Sphingomyelinase	BL,F	Niemann-Pick Type A/B
☐ Filipinestaining	F	Niemann-Pick Type C
☐ ß-D-glucosidase	BL,F	Gaucher
☐ Chitotriosidase	BL,Pla	Gaucher and several other LSDs
☐ Galactocerebrosidase	BL,F	Krabbe
☐ Acid Lipase	BL,F,Bsp	Wolman/ Cholesteryl ester storage disease (CESD)

Glycogenoses

☐ α-D-glucosidase	BL,F,Bsp	Glycogenose Type II (Pompe)

Neuronal Ceroid Lipofuscinoses

☐ Palmitoyl-protein thioesterase	BL,F	NCL type I (Infantile NCL)
☐ Tripeptidyl peptidase I	BL,F	NCL type II (Late infantile NCL)

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INSTRUCTIONS

- Please use the appropriate request form:
  (Metabolite-, Enzyme- or DNA- diagnostics)
  See www.labgmd.nl (Protocols & Forms).
- Be sure to fill out the test request form completely **in English** (grey fields are mandatory).
- Please include copies of relevant correspondence concerning the request.
- Please include all information needed by the financial department of your institution
- In case of urgent requests (e.g. prenatal testing) please contact a staff member of the laboratory BEFORE sending the sample.
- Samples should arrive Monday through Thursday from 8:30 AM to 4:00 PM and Friday or the day prior to a national holiday before 12:00 AM. Our website www.labgmd.nl lists national holidays on which our laboratory is closed.
- For test-specific information about material/shipment please visit our website www.labgmd.nl

Please use the address label on the next page for shipment

Use this as address label

Laboratory Genetic Metabolic Diseases (F0-132)

Amsterdam UMC, location AMC

Meibergdreef 9

1105 AZ Amsterdam

The Netherlands

DIAGNOSTISCH MATERIAAL

SPOED!