

## Laboratory Genetic Metabolic Diseases

### 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 : .....

Reference number : .....

#### Requested test(s) (see page 3, 4, 5 and [www.labgmd.nl](http://www.labgmd.nl))

Enzyme and/or disorder: .....

Prenatal analysis

#### Material\*

For prenatal testing please contact the laboratory before sending samples

#### sampling:

<input type="checkbox"/> Blood (EDTA)*	date .....	time .....	<input type="checkbox"/> Chorion villi sample	date .....	time .....
<input type="checkbox"/> Erythrocytes	date .....	time .....	<input type="checkbox"/> Chorion villi fibroblasts	date .....	time .....
<input type="checkbox"/> Plasma	date .....	time .....	<input type="checkbox"/> Amniocytes	date .....	time .....
<input type="checkbox"/> Blood spot	date .....	time .....	<input type="checkbox"/> Tissue ; specify	:	date .....
<input type="checkbox"/> Skin biopsy	date .....	time .....			time .....
<input type="checkbox"/> Skin fibroblasts	date .....	time .....	<input type="checkbox"/> Other ; specify	:	date .....
					time .....

\* Blood (whole blood at room temperature) must arrive within 48 hours after collection. For detailed specification of transport conditions see [www.labgmd.nl](http://www.labgmd.nl)

#### Relevant clinical and laboratory findings

#### Clinical laboratory geneticists:

Dr. S. Ferdinandusse      Dr. A.B.P. van Kuilenburg  
Dr. M.S. Ebberink         Dr. A. Pop, trainee

Amsterdam UMC, location AMC  
Lab GMD (F0-132)  
Meibergdreef 9  
1105 AZ Amsterdam  
The Netherlands

[www.labgmd.nl](http://www.labgmd.nl)  
[gmz\\_enzym@amc.nl](mailto:gmz_enzym@amc.nl)  
Tel: +31(0)20-566 5393



---

**Results should be sent to**

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

\* 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 : .....  
Financial reference 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

<input type="checkbox"/> 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)
<input type="checkbox"/> Very long-chain fatty acids	F	ZSD, XALD, ACOX1, DBP
<input type="checkbox"/> C26:0 lysoPC	F	ZSD, XALD, ACOX1, DBP
<input type="checkbox"/> Dihydroxyacetonephosphate-acyltransferase (DHAPAT)	F	ZSD, RCDP
<input type="checkbox"/> Immunofluorescence catalase	F	ZSD, ACOX1, DBP
<input type="checkbox"/> Immunofluorescence ALDP (adrenoleukodystrophy protein)	F	XALD
<input type="checkbox"/> Acyl-CoA oxidase 1 (ACOX1)	F	ACOX1 deficiency
<input type="checkbox"/> D-Bifunctional protein (DBP/MFP2)	F,BL	DBP deficiency
<input type="checkbox"/> Peroxisomal thiolase branched-chain (Sterol Carrier Protein X)	F	SCPx deficiency
<input type="checkbox"/> Phytanic acid $\alpha$ -oxidation	F	Refsum disease
<input type="checkbox"/> Peroxisomal $\beta$ -oxidation	F	ZSD, XALD, AMACR, SCPx, ACOX1, DBP
<input type="checkbox"/> Immunoblot peroxisomal proteins	F	ZSD, RCDP
<input type="checkbox"/> Plasmalogens	F	RCDP, ZSD

### Mitochondrial fatty acid oxidation

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

### Amino acid metabolism

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

BL = EDTA blood

Pla = EDTA plasma

Ery = Erythrocytes

Bsp = Blood spot

F = Fibroblasts

L = Liver

\* = This test is not covered by the accreditation of the laboratory

**For prenatal testing please contact the laboratory before sending samples**

Tests Enzyme diagnostics continued

*Purine and Pyrimidine metabolism*

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

*Carbohydrate degradation*

<input type="checkbox"/> Galactose-1-phosphate uridylyltransferase (GALT)	BL,F	Galactosemia type 1, classic galactosemia
<input type="checkbox"/> Galactokinase (GALK)	BL	Galactosemia type 2
<input type="checkbox"/> UDP galactose-4-epimerase (GALE)	BL,F	Galactosemia type 3
<input type="checkbox"/> Glucose 6-phosphate dehydrogenase (G6PD)	BL	G6PD deficiency
<input type="checkbox"/> Pyruvate kinase (PK)	BL	Pyruvate kinase deficiency

*Ketolysis defects*

<input type="checkbox"/> Succinyl-CoA : 3-oxoacid transferase (SCOT)	F,BL	SCOT deficiency
<input type="checkbox"/> β-ketothiolase (2-methyl-acetoacetyl-CoA specific)	F,BL	β-ketothiolase deficiency

*Cholesterol/isoprenoid biosynthesis*

<input type="checkbox"/> <b>Screening cholesterol biosynthesis defects (Sterol profiling) *</b>	F	Smith-Lemli-Opitz syndrome (SLO), Desmosterolosis, Conradi-Hunermann syndrome, Lathosterolosis and Greenberg dysplasia
<input type="checkbox"/> Mevalonate kinase (MVK)	F,BL	Hyper IgD syndrome/mevalonic aciduria

*Neurotransmitter metabolism*

<input type="checkbox"/> Aromatic amino acid decarboxylase (AADC) *	Pla	Aromatic amino acid decarboxylase deficiency
---	-----	--

*Hyperoxaluria*

<input type="checkbox"/> Glyoxylate reductase (GR) *	BL,L	Hyperoxaluria type 2
--	------	----------------------

*Miscellaneous enzymes*

<input type="checkbox"/> Fatty aldehyde dehydrogenase (FALDH, SLS)	F,BL	Sjögren Larsson syndrome
<input type="checkbox"/> Steroidsulfatase (arylsulfatase C, ARYC)	BL,F	X-linked ichthyosis
<input type="checkbox"/> Biotinidase	BL,Pla	Biotinidase deficiency

- BL = EDTA blood
- Pla = EDTA plasma
- Ery = Erythrocytes
- Bsp = Blood spot
- F = Fibroblasts
- L = Liver

\* = This test is not covered by the accreditation of the laboratory

**For prenatal testing please contact the laboratory before sending samples**

## Tests Lysosomal Storage Disorders

---

### Mucopolysaccharidoses

<input type="checkbox"/> $\alpha$ -L-iduronidase	BL,F	MPS Type IH/IS (Hurler/Scheie)
<input type="checkbox"/> Iduronate sulfatase	BL,F	MPS Type II (Hunter)
<input type="checkbox"/> Sulfamidase	BL,F	MPS Type III A (Sanfilippo A)
<input type="checkbox"/> N-acetyl- $\alpha$ -D-glucosaminidase	BL,F	MPS Type III B (Sanfilippo B)
<input type="checkbox"/> AcetylCoA-glucosamine acetyltransferase	BL,F	MPS Type III C (Sanfilippo C)
<input type="checkbox"/> N-acetylglucosamine-6-sulfatase	BL,F	MPS Type III D (Sanfilippo D)
<input type="checkbox"/> N-acetylgalactosamine-6-sulfatase	BL,F	MPS Type IV A (Morquio A)
<input type="checkbox"/> $\beta$ -D-galactosidase	BL,F	MPS Type IV B (Morquio B)
<input type="checkbox"/> Arylsulfatase B	BL,F	MPS Type VI (Maroteaux-Lamy)
<input type="checkbox"/> $\beta$ -D-glucuronidase	BL,F	MPS Type VII (Sly)

---

### Mucopolipidoses

<input type="checkbox"/> N-acetyl- $\alpha$ -D-neuraminidase	F	Mucopolipidosis Type I
<input type="checkbox"/> N-acetyl- $\beta$ -D-glucosaminidase	BL,F	Mucopolipidosis Type II and III (I-cell disease, pseudo-Hurler polydystrophy)

---

### Oligosaccharidoses

<input type="checkbox"/> $\alpha$ -L-fucosidase	BL,F	Fucosidosis
<input type="checkbox"/> $\alpha$ -D-mannosidase	BL,F	$\alpha$ -Mannosidosis
<input type="checkbox"/> $\beta$ -D-mannosidase	BL,F	$\beta$ -Mannosidosis
<input type="checkbox"/> N-acetyl- $\alpha$ -D-galactosaminidase	BL,F	Schindler / Kanzaki
<input type="checkbox"/> Aspartylglucosaminidase	BL,F	Aspartylglucosaminuria
<input type="checkbox"/> Protective protein / Cathepsin A	BL,F	Galactosialidosis

---

### Sphingolipidoses

<input type="checkbox"/> Arylsulfatase A	BL,F	Metachromatic leukodystrophy
<input type="checkbox"/> Arylsulfatase A+B	BL,F	Mucopolysaccharidosis / Multiple sulfatase deficiency
<input type="checkbox"/> $\alpha$ -D-galactosidase	BL,F	Fabry
<input type="checkbox"/> $\beta$ -D-galactosidase	BL,F	GM-1 gangliosidosis
<input type="checkbox"/> N-acetyl- $\beta$ -D-glucosaminidase A	BL,F	Tay-Sachs / GM-2 gangliosidosis B variant
<input type="checkbox"/> N-acetyl- $\beta$ -D-glucosaminidase A+B	BL,F	Sandhoff / GM-2 gangliosidosis O variant
<input type="checkbox"/> Sphingomyelinase	BL,F	Niemann-Pick Type A/B
<input type="checkbox"/> Filipinestaining	F	Niemann-Pick Type C
<input type="checkbox"/> $\beta$ -D-glucosidase	BL,F	Gaucher
<input type="checkbox"/> Chitotriosidase	BL,Pla	Gaucher and several other LSDs
<input type="checkbox"/> Galactocerebrosidase	BL,F	Krabbe
<input type="checkbox"/> Acid Lipase	BL,F,Bsp	Wolman / Cholesteryl ester storage disease (CESD)

---

### Glycogenoses

<input type="checkbox"/> $\alpha$ -D-glucosidase	BL,F,Bsp	Glycogenose Type II (Pompe)
--	----------	-----------------------------

---

### Neuronal Ceroid Lipofuscinoses

<input type="checkbox"/> Palmitoyl-protein thioesterase	BL,F	NCL type I (Infantile NCL)
<input type="checkbox"/> Tripeptidyl peptidase I	BL,F	NCL type II (Late infantile NCL)

---

BL = EDTA blood  
Pla = EDTA plasma  
Ery = Erythrocytes  
Bsp = Blood spot  
F = Fibroblasts  
L = Liver

\* = This test is not covered by the accreditation of the laboratory

**For prenatal testing please contact the laboratory before sending samples**

# INSTRUCTIONS

- Please use the appropriate request form: (Metabolite-, Enzyme- or DNA- diagnostics) See [www.labgmd.nl](http://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](http://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](http://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



**BIOLOGICAL SUBSTANCE  
CATEGORY B**

DIAGNOSTISCH MATERIAAL

**SPOED!**

