Amsterdam UMC
University Medical Centers

Laboratory Genetic Metabolic Diseases

Test request form DNA-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	:	.....

Family members or relatives analyzed previously?

☐ Yes, namely:
☐ No/unknown

Family name, first name : .....
Date of birth (dd/mm/yyyy) : .....
Relation : .....
Findings : .....

Requested test (see page 3 and www.labgmd.nl)

Disease and/or gene (s): DPYD sequencing and copy number variation (MLPA)

■ Whole gene analysis ☐ Carrier analysis ☐ Prenatal analysis

Material

For prenatal testing please contact the laboratory before sending samples

■ Blood (EDTA preferred)	sample date	(>2 ml; ship at room temperature; receipt <4 days)
☐ DNA	sample date	(ship at room temperature)
☐ Skin fibroblasts	sample date	(in T25 flask filled with medium; ship at room temperature)
☐ Chorion villi sample	sample date	(in medium; ship at room temperature)
☐ Chorion villi fibroblasts	sample date	(in T25 flask filled with medium; ship at room temperature)
☐ Amniocytes	sample date	(in T25 flask filled with medium; ship at room temperature)
☐ Other, i.e.....	sample date	(frozen; ship on dry ice)

Relevant clinical and laboratory findings

Clinical laboratory geneticists:
Prof.dr. H.R. Waterham Dr. A.B.P. van Kuilenburg
Dr. M.S. Ebberink

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

www.labgmd.nl
gmz_dna@amc.nl
Tel: +31(0)20-566 5393
Fax: +31(0)20-696 2596

TESTING
RvA M 154

Results should be sent to

Name	: FIMLAB Laboratories
Department	: Laboratory of Clinical Genetics
Hospital/institute	:
Address	: P.O. Box 66
City and Zip-code	: FIN-33013 FIMLAB
Country	: FINLAND
Phone	:
Fax	: +358-9-4257 8283
E-mail*	: genetiikka@fimlab.fi

* *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*

Please do not include any confidential information such as personal identity codes or health information on your invoice. If any such information is necessary for invoicing, it should be sent via encrypted e-mail to: ostolaskut@fimlab.fi

Name	**FIMLAB Laboratories**
In case of institution Department Hospital/institute	• E-invoice address: 003723925196 / operating ID 003708599126 / Open Text Oy • Invoices in .PDF format: Send to **scanning.europe@liaison.com** including the following information: Fimlab Laboratoriot Oy, 16812778, PL 940 00019 SSC
Address	
City and Zip-code	
Country	
E-mail of financial contact	**ostolaskut@fimlab.fi**
VAT number	**FI23925196**
Financial reference number	**Always refer to Fimlab sample number Nxxxxxx**

* *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.

Clinical DNA testing is available for the following disorders (*gene*):

Acyl-CoA oxidase deficiency (*ACOX1*)
Adenine phosphoribosyl transferase deficiency (*APRT*)
Adenosine deaminase deficiency (*ADA*)
Adenosine deaminase 2 deficiency (*ADA2/CECR1*)
__ α-Methylacyl-CoA racemase deficiency *(AMACR)*
Argininosuccinate lyase deficiency (*ASL*)
Aromatic amino acid decarboxylase deficiency *(DDC)*
ß-ketothiolase deficiency (*ACAT1*)
ß-ureidopropionase deficiency (*UPB1*)
Brown-Vialetto-Van Laere syndrome
type 1 (SLC52A3) *type 2 (SLC52A2)* *type 3 (SLC52A1)*
Carnitine-acylcarnitine translocase deficiency *(SLC25A20/CACT)*
Carnitine deficiency, primary *(SLC22A5/OCTN2)*
Carnitine palmitoyltransferase 1 deficiency (*CPT1A*)
Carnitine palmitoyltransferase 2 deficiency (*CPT2*)
CHILD syndrome *(NSDHL)*
Chitotriosidase deficiency c.1049_1072dup (*CHIT1*)
Conradi-Hünermann-Happle syndrome/CDPX2 (*EBP*)
Cytosolic iron-sulfur assembly component 1 (*CIAO1*)
D-Bifunctional protein deficiency (*HSD17B4/DBP/MFE2*)
Desmosterolosis (*DHCR24*)
Dihydropyrimidinase deficiency (*DPYS*)
Dihydropyrimidine dehydrogenase deficiency (*DPYD*)
Ethylmalonic aciduria (Ethylmalonic encephalopathy) (*ETHE1*)
__ Fructose intolerance, inherited *(ALDOB)*
Galactokinase deficiency (*GALK1*)
Galactosemia (*GALT*)
Glutaminase deficiency (*GLS*)
__ Glutaryl-CoA dehydrogenase deficiency /
Glutaric aciduria type I (*GCDH*)
Glycogen storage disease type 0, GSD-0 *(GYS2)*
Greenberg skeletal dysplasia (*LBR*)
GTP cyclohydrolase I deficiency *(GCH1)*
HMG-CoA lyase deficiency (*HMGCL*)
Holocarboxylase synthetase deficiency (*HLCS*)
3-Hydroxyisobutyrate dehydrogenase deficiency (*HIBADH*)
3-Hydroxyisobutyryl-CoA hydrolase deficiency (*HIBCH*)
Hyper IgD syndrome/ Mevalonate kinase deficiency (*MVK*)
Hyperlysinemia (*AASS*)
Hyperoxaluria
type 1 (AGXT) *type 2 (GRHPR)* *type 3 (HOGA1)*
Hypoxanthine-guanine phosphoribosyl transferase
deficiency (*HPRT1*)
Isovaleryl-CoA dehydrogenase deficiency /
Isovaleric acidemia *(IVD)*
Lathosterolosis *(SC5DL)*
L2-hydroxyglutaric aciduria (*L2HGDH*)
Medium-chain acyl-CoA dehydrogenase deficiency *(ACADM)*
2-Methyl-3-hydroxybutyryl-CoA dehydrogenase
deficiency (*HSD17B10/HADH2*)
3-Methylcrotonyl-CoA carboxylase deficiency
MCCC1 *MCCC2*
3-Methylglutaconyl-CoA hydratase deficiency /
3-Methylglutaconic aciduria type I (*AUH*)
Methylmalonyl-CoA epimerase deficiency *(MCEE)*
Mevalonic aciduria/Mevalonate kinase deficiency (*MVK*)
Mitochondrial DNA depletion syndrome 2 *(SUCLA2)*
Mitochondrial trifunctional protein deficiency
HADHA *HADHB* *LCHAD* c.1528G>C mutation
Monocarboxylate transporter member 1 (MCT1) deficiency
(*SLC16A1*)
Multiple acyl-CoA dehydrogenase deficiency
ETFDH *ETFA* *ETFB*
Neuraminidase deficiency / Sialidosis (*NEU1*)
Phosphoglycerate mutase deficiency, muscle (*PGAM2*)
Phosphoribosyl pyrophosphate synthetase 1 superactivity
and deficiency (*PRPS1*)
Purine nucleoside phosphorylase deficiency (*PNP*)
Refsum disease (*PHYH/PAHX*)
Rhabdomyolysis, acute recurrent (*LPIN1*)
__ Rhizomelic chondrodysplasia punctata (RCDP)
type 1 *(PEX7)* type 2 *(GNPAT)* type 3 *(AGPS)*
Short-chain acyl-CoA dehydrogenase deficiency (*ACADS*)
Short-chain enoyl-CoA hydratase/Crotonase deficiency (*ECHS1*)
Sjögren-Larsson syndrome (*ALDH3A2/ALDH10*)
Smith-Lemli-Opitz syndrome (*DHCR7*)
__ Succinate-CoA ligase (*SUCLG1*)
__ Succinyl CoA:3-oxoacid CoA transferase deficiëntie (*OXCT1/SCOT*)
Thymidine phosphorylase deficiency (*TYMP*)
Thymidylate synthase deficiency (*TYMS*)
Tyrosine hydroxylase deficiency *(TH)*
Very long-chain acyl-CoA dehydrogenase deficiency (*ACADVL*)
X-linked adrenoleukodystrophy *(ABCD1/X-ALD)*
Zellweger spectrum disorders (Zellweger syndrome, NALD, IRD)
PEX1 *PEX2* *PEX3*
PEX5 *PEX6* *PEX10*
PEX11B *PEX12* *PEX13*
PEX14 *PEX16* *PEX19*
PEX26
Complementation analysis
(*PEX* gene unknown; skin fibroblasts required)
Peroxisomal Gene Array - Ion Torrent * (*PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, ACOX1, ACOX2, HSD17B4, SCP2, ABCD1, ABCD2, ABCD3, ACBD5, AMACR, PHYH, GNPAT, AGPS*)

Other request (only after contacting laboratory) : .....

* This test is temporarily outsourced to the Metabolic Unit of the Amsterdam UMC, location VUmc
** This method is not covered by the accreditation of the laboratory

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!