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 Tampere
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*

Name	: FIMLAB Laboratories
In case of institution	
Department	: For electronic billing: OVT 003723925196, Basware Oyj Operator: BAWCFI22
Hospital/institute	:
Address	: P.O. Box 1008
City and Zip-code	: FIN-00071 Ostolaskut
Country	: FINLAND
E-mail of financial contact	: genetiikka@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!