Amsterdam UMC
University Medical Centers

Laboratory Genetic Metabolic Diseases

Print Form

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	:	.....

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): .....

☐ 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
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The Netherlands

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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	:
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.

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)*
- ☐ β-ketothiolase deficiency (*ACAT1*)
- ☐ β-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 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!