

## 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: Family name, first name : .....

No/unknown 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  
1105 AZ Amsterdam  
The Netherlands

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



---

**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 : .....  
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.*

**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*)
  - $\alpha$ -Methylacyl-CoA racemase deficiency (*AMACR*)
  - Argininosuccinate lyase deficiency (*ASL*)
  - Aromatic amino acid decarboxylase deficiency (*DDC*)
  - B-ketothiolase deficiency (*ACAT1*)
  - B-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-Hünemann-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*
    - ETFPA*
    - 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 deficiency (*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](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!**

