Laboratory Genetic Metabolic Diseases

Test request form DNA diagnostics

Accr.Nr: 157
EN/ISO 15189

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Lab. Genetic Metabolic Diseases (F0-132)
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Print Form

Dr. H.R. Waterham – clinical molecular geneticist
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Dr. A.B.P. van Kuilenburg – clinical biochemical geneticist
Dr. M.S. Ebberink – clinical biochemical geneticist, trainee

To assure correct handling of your request, 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

Family members or relatives analyzed previously?

☐ No/unknown
☐ Yes: Family name, first name : .....
Date of birth (dd/mm/yyyy) : .....
Relationship : .....
Findings : .....

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

Disease and/or gene (s) : .....

☐ Whole gene analysis ☐ Carrier analysis ☐ Prenatal analysis

Material

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

For detailed specification of transport conditions see www.labgmd.nl

Relevant clinical and laboratory findings

Results should be sent to

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

** Results will be sent per email when fax is unavailable, unless disagreed.* ☐ No

Copy results should be sent to

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

Invoice should be sent to*

Name	: .....
In case of institution	
Department	: .....
Hospital/institute	: .....
Address	: .....
City and Zip-code	: .....
Country	: .....

** For EU countries only: 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*)
- ☐ Alpha-methylacyl-CoA racemase deficiency *(AMACR)*
- ☐ Argininosuccinate lyase deficiency (*ASL*)
- ☐ Aromatic amino acid decarboxylase deficiency *(DDC)*
- ☐ Beta-ureidopropionase deficiency (*UPB1*)
- ☐ Brown-Vialetto-Van Laere syndrome (*C20orf54*)
- ☐ 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*)
- ☐ D-Bifunctional protein deficiency (*DBP/MFE2*)
- ☐ Desmosterolosis (*DHCR24*)
- ☐ Dihydropyrimidinase deficiency (*DPYS*)
- ☐ Dihydropyrimidine dehydrogenase deficiency (*DPYD*)
- ☐ Ethylmalonic aciduria (Ethylmalonic encephalopathy) (*ETHE1*)
- ☐ Fructose intolerance, inherited *(ALDOB)*
- ☐ Galactosemia (*GALT*)
- ☐ Galactokinase deficiency (*GALK1*)
- ☐ Glutaryl-CoA dehydrogenase deficiency / Glutaric aciduria type I (*GCDH*)
- ☐ Glycogen storage disease type 0, GSD-0 *(GYS2)*
- ☐ GTP cyclohydrolase I deficiency *(GCH1)*
- ☐ Greenberg skeletal dysplasia (*LBR*)
- ☐ Holocarboxylase synthetase deficiency (*HCLS*)
- ☐ 3-hydroxyisobutyrate dehydrogenase deficiency (*HIBADH*)
- ☐ 3-hydroxyisobutyryl-CoA hydrolase deficiency (*HIBCH*)
- ☐ Hyper IgD syndrome/ Mevalonate kinase deficiency (*MVK*)
- ☐ Hyperlysinemia (*AASS*)
- ☐ Hyperoxaluria type I (*AGXT*)
- ☐ Hypoxanthine-guanine phosphoribosyl transferase deficiency (*HPRT1*)
- ☐ Isovaleryl-CoA dehydrogenase deficiency / Isovaleric acidemia *(IVD)*
- ☐ Lathosterolosis *(SC5DL)*
- ☐ 2-Methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency (*HADH2*)
- ☐ 3-Methylcrotonyl-CoA carboxylase deficiency
  - ☐ *MCCC1* ☐ *MCCC2*
- ☐ 3-methylglutaconyl-CoA hydratase deficiency /
  - ☐ 3-methylglutaconic aciduria type I (*AUH*)
- ☐ Medium-chain acyl-CoA dehydrogenase deficiency *(ACADM)*
- ☐ Methylmalonyl-CoA epimerase deficiency *(MCEE)*
- ☐ Mevalonic aciduria/Mevalonate kinase deficiency (*MVK*)
- ☐ Mitochondrial trifunctional protein deficiency
  - ☐ *HADHA* ☐ *HADHB* ☐ LCHAD c.1528G>C mutation
- ☐ Multiple acyl-CoA dehydrogenase deficiency
  - ☐ *ETFDH* ☐ *ETFA* ☐ *ETFB*
- ☐ Phosphoglycerate mutase deficiency, muscle (*PGAM2*)
- ☐ Phosphoribosyl pyrophosphate synthetase 1 superactivity and deficiency (*PRPS1*)
- ☐ Purine nucleoside phosphorylase deficiency (*NP*)
- ☐ 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*)
- ☐ Sjögren-Larsson syndrome (*ALDH3A2/ALDH10*)
- ☐ Smith-Lemli-Opitz syndrome (*DHCR7*)
- ☐ SUCLA2 deficiency *(SUCLA2)*
- ☐ SUCLG1 deficiency (*SUCLG1*)
- ☐ Thymidine phosphorylase deficiency (*TYMP*)
- ☐ 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)
  - ☐ Complementation analysis (*PEX* gene unknown; skin fibroblasts required)
    - ☐ *PEX1* ☐ *PEX2* ☐ *PEX3*
    - ☐ *PEX5* ☐ *PEX6* ☐ *PEX10*
    - ☐ *PEX11β* ☐ *PEX12* ☐ *PEX13*
    - ☐ *PEX14* ☐ *PEX16* ☐ *PEX19*
    - ☐ *PEX26*

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

Use this as address label

Laboratory Genetic Metabolic Diseases (F0-132)

Academic Medical Center

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The Netherlands

For Couriers only:
Meibergdreef 9
1105 AZ Amsterdam
The Netherlands

DIAGNOSTISCH MATERIAAL

GRAAG MET SPOED NAAR DE POSTKAMER BRENGEN!

INSTRUCTIONS

- Please use the test request form that applies:
- 1) metabolite/tumor-, 2) enzyme-, 3) DNA- diagnostics.
  See www.labgmd.nl/forms
- To assure correct handling of your request, please fill out the test request form completely **in English** and send it together with the sample(s). Grey fields are mandatory.
- Please include copies of all relevant correspondence with our laboratory concerning the request.
- In case of urgent requests (e.g. prenatal testing) please contact a staff member of the laboratory BEFORE sending the sample.
- Our laboratory is open on working days Monday to Friday from 8.30 AM to 5.00 PM. Our website www.labgmd.nl lists national holidays on which our laboratory is closed.
- Please make sure that sample(s) arrive on Friday before 12 AM. Otherwise we cannot guarantee that we can process the samples appropriately.
- For test-specific information about material/shipment please visit our website www.labgmd.nl