



Accr.Nr: 157
EN/ISO 15189

Laboratory Genetic Metabolic Diseases

Test request form DNA diagnostics



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To assure correct handling of your request, please fill in this form completely and send it in together with the sample(s) or return by fax.

Patient information

Family name :
First name :
Date of birth (mm/dd/yy) :
Sex : Male Female
Consanguinity :
Ethnic origin (optional) :

To be completed by Lab. GMD

Labnr.
Vorig Labnr.
Testcode
Declarabel
 S
 Research
Projectcode
Gereed
Autorisator

Family members or relatives analyzed previously?

No/unknown
 Yes: Family name, first name :
Date of birth (mm/dd/yy) :
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)

Relevant clinical and/or laboratory findings

Results should be sent to

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

Invoice should be sent to*

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

** Original E-112 forms (EU countries only) should be filled in 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*)
- Carnitine-acylcarnitine translocase deficiency (*SLC25A20/CACT*)
- Carnitine deficiency, primary (*SLC22A5/OCTN2*)
- Carnitine palmitoyltransferase 1 deficiency (*CPT1A*)
- Carnitine palmitoyltransferase 2 deficiency (*CPT2*)
- CHILD syndrome (*NSDHL*)
- Conradi-Hunermann-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*)
- 2-Methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency (*HADH2*)
- Hyper IgD syndrome/ Mevalonate kinase deficiency (*MVK*)
- Hyperoxaluria type I, primaire (*AGT*)
- Hypoxanthine-guanine phosphoribosyl transferase deficiency (*HPRT1*)
- Isovaleric acidemia (*IVD*)
- Lathosterolosis (*SC5DL*)
- 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
- Multiplex acyl-CoA dehydrogenase deficiency *ETFDH* *ETFA* *ETFB*
- Phosphoribosyl pyrophosphate synthetase 1 superactivity and deficiency (*PRPS1*)
- Purine nucleoside phosphorylase deficiency (*NP*)
- Refsum, disease of (*PHYH/PAHX*)
- Rhizomelic chondrodysplasia punctata (RCDP) type 1 (*PEX7*) type 2 (*DHAPAT*) type 3 (*AGPS*)
- Short-chain acyl-CoA dehydrogenase deficiency (*ACADS*)
- Sjögren-Larsson syndrome (*ALDH3A2/ALDH10*)
- Smith-Lemli-Opitz syndrome (*DHCR7*)
- SUCLA2 deficiency (*SUCLA2*)
- 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*
 - PEX12* *PEX13* *PEX14* *PEX16* *PEX19* *PEX26*
- Other request (only after contacting laboratory)