

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 : Man Woman

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

Relevant clinical and laboratory findings

Clinical laboratory geneticists:
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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):

Amino acid metabolism and transport disorders

- 2-Methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency (*HSD17B10/HADH2*)
- 3-hydroxyisobutyrate dehydrogenase deficiency (*HIBADH*)
- 3-hydroxyisobutyryl-CoA hydrolase deficiency (*HIBCH*)
- 3-Methylcrotonyl-CoA carboxylase deficiency (*MCC*) (all genes mentioned below)
 - MCCC1*
 - MCCC2*
- 3-methylglutacon aciduria type I (*AUH*)
- Argininosuccinate lyase (*ASL*)
- BOLA family member 3 deficiency (*BOLA3*)
- Branched chain aminotransaminase 2 deficiency (*BCAT2*)
- Carbamylphosphate synthase 1 deficiency (*CPS1*)
- Citrullinemia (all genes mentioned below)
 - type I (*ASS1*)
 - type II (*SLC25A13*)
- Cystathionine beta-synthase deficiency (*CBS*)
- Dihydrofolate dehydrogenase deficiency (*MSUD type III*) (*DLD*)
- Phenylketonuria/hyperphenylalaninemia, non-PKU mild (*PAH*)
- Glutamate dehydrogenase-1 deficiency (*GLUD1*)
- Glutaminase deficiency (*GLS*)
- Glutaredoxin 5 deficiency (*GLRX5*)
- Glycine N-methyltransferase deficiency (*GNMT*)
- Glycine transporter 1 deficiency (*SLC6A9*)
- Glycine transporter 2 deficiency (*SLC6A5*)
- Hartnup disease (*SLC6A19*)
- HMG-CoA lyase deficiency/
 - 3-Hydroxy-3-methylglutaryl-CoA lyase deficiency (*HMGCL*)
- Hyperlysinemia (*AASS*)
- Hyperphenylalaninemia, mild, non-BH4-deficient (*DNAJC12*)
- Isovaleryl-CoA dehydrogenase deficiency / Isovaleric acidemia (*IVD*)
- Lipoic Acid synthase deficiency (*LIAS*)
- Lysinuric protein intolerance (*LPI/ SLC7A7*)
- Maple syrup urine disease (all genes mentioned below)
 - type Ia (*BCKDHA*)
 - type Ib (*BCKDHB*)
 - type II (*DBT*)
- Methionine adenosyltransferase deficiency (*MAT1A*)
- Multiple mitochondrial dysfunctions syndrome 1 (*NFU1*)
- N-acetylglutamate synthase deficiency (*NAGS*)
- NKH (all genes mentioned below)
 - Aminomethyltransferase deficiency (*AMT*)
 - Glycine cleavage system H-protein deficiency (*GCSH*)
 - Glycine decarboxylase deficiency (*GLDC*)
- Ornithine transcarbamylase deficiency (*OTC*)
- Phosphoglycerate dehydrogenase deficiency (*PHGDH*)
- Phosphoserine aminotransferase deficiency (*PSAT1*)
- Phosphoserine phosphatase deficiency (*PSPH*)
- S-adenosylhomocysteine hydrolase (*SAHH*) deficiency (*AHCY*)
- Short-chain enoyl-CoA hydratase/Crotonase deficiency (*ECHS1*)
- Glutamate oxidase deficiency (*SUOX*)

Creatine metabolism and transport disorders

- Arginine:glycine amidinotransferase deficiency (*AGAT/ GATM*)
- Creatine transporter defect, X-linked (*SLC6A8*)
- Guanidinoacetate methyltransferase deficiency (*GAMT*)
- Monocarboxylate transporter 12 deficiency (*SLC16A12*)

Cholesterol/isoprenoid biosynthesis defects

- CHILD syndrome (*NSDHL*)
- Conradi-Hünemann-Happle syndrome/CDPX2 (*EBP*)
- Desmosterolosis (*DHCR24*)
- Greenberg skeletal dysplasia (*LBR*)
- Hyper IgD syndrome/ Mevalon aciduria/
 - Mevalonate kinase deficiency (*MVK*)
- Lathosterolosis (*SC5DL*)
- Smith-Lemli-Opitz Syndrome (*DHCR7*)

GABA metabolism disorder

- GABA-transaminase deficiency (*GABA-T/ ABAT*)
- Succinate semialdehyde dehydrogenase deficiency (*SSADH/ ALDH5A1*)

Hypophosphatemic rickets

- Dentin matrix acidic phosphoprotein 1 deficiency (*DMP1*)
- Ectonucleotide pyrophosphatase/phosphodiesterase 1 deficiency (*ENPP1*) Hypophosphatemic rickets, X-linked dominant (*PHEX*)
- Sodium phosphate cotransporter, member 3 deficiency (*SLC34A3*)

Ketolysis defects

- Monocarboxylate transporter member 1 (*MCT1*) deficiency (*SLC16A1*)
- β -ketothiolase deficiency (*ACAT1*)
- Succinyl CoA:3-oxoacid CoA transferase deficiency (*OXCT1/SCOT*)

Carbohydrate metabolism and transport disorders

- Fructokinase/ ketohexokinase deficiency (*KHK*)
- Fructose intolerance, hereditary (*ALDOB*)
- Fructose-1,6-biphosphatase-1 deficiency (*FBP1*)
- Galactokinase deficiency (*GALK1*)
- Galactosemia (*GALT*)
- Glucose transporter deficiency (*GLUT1/ SLC2A1*)
- Glucose-6-phosphate dehydrogenase deficiency (*G6PD*)
- Glycogen storage disease type IV (*GBE1*)
- Glycogen storage disease type VII (Tauri) (*PFKM*)
- Glycogen storage disease type 0, GSD-0 (*GYS2*)
- Phosphoglycerate mutase deficiency, muscle (*PGAM2*)
- Ribose 5-phosphate isomerase deficiency (*RPIA*)
- Sedoheptulokinase deficiency (*SHPK*)
- Transaldolase deficiency (*TALDO1*)
- Transketolase deficiency (*TKT*)
- Triosephosphate isomerase deficiency (*TPI1*)

Clinical DNA testing is available for the following disorders (gene):

Lysosomal storage disorders

- Arylsulfatase A deficiency (*ARSA*)
- Galactosialidosis (*CTSA*)
- Glycogen storage disease type II/ Pompe disease (*GAA*)
- GM1-gangliosidosis (*GLB1*)
- Krabbe disease (*GALC*)
- Lysosomal acid lipase deficiency (*LIPA*)
- Mannosidase, beta A, lysosomal deficiency (*MANBA*)
- Mannosidosis, alpha-, type I and II (*MAN2B1*)
- Mucopolysaccharidose type 4/ mucopolipin-1 deficiency (*MCOLN1*)
- Mucopolysaccharidose type 3/ Sanfilippo type A (*SGSH*)
- Neuraminidase deficiency / Sialidosis (*NEU1*)
- Sulfatase modifying factor 1 deficiency / Multiple sulfatase deficiency (*SUMF1*)
- Tay-Sachs disease (*HEXA*)

Mitochondrial fatty acid oxidation defects

- Carnitine deficiency, primary (*SLC22A5/OCTN2*)
- Carnitine palmitoyltransferase 1 deficiency (*CPT1A*)
- Carnitine palmitoyltransferase 2 deficiency (*CPT2*)
- Carnitine-acylcarnitine translocase deficiency (*SLC25A20/CACT*)
- Medium-chain acyl-CoA dehydrogenase deficiency (*ACADM*)
- Mitochondrial trifunctional protein deficiency (all genes mentioned below)
 - HADHA* *HADHB* *LCHAD* (c.1528G>C mutation)
- Multiple acyl-CoA dehydrogenase deficiency (all genes mentioned below)
 - ETFA* *ETFB* *ETFDH*
- Short-chain acyl-CoA dehydrogenase deficiency (*ACADS*)
- Very long-chain acyl-CoA dehydrogenase deficiency (*ACADVL*)

Neurotransmitter metabolism disorders

- Aromatic amino acid decarboxylase deficiency (*DDC*)
- GTP cyclohydrolase I deficiency (*GCH1*)
- Tyrosine hydroxylase deficiency (*TH*)

Purine and pyrimidine metabolism disorders

- Adenine phosphoribosyl transferase deficiency (*APRT*)
- Adenosine deaminase 2 deficiency (*ADA2/CECR1*)
- Adenosine deaminase deficiency (*ADA*)
- Adenosine kinase deficiency (*ADK*)
- Adenylosuccinate lyase deficiency (*ADSL*)
- Dihydropyrimidinase deficiency (*DPYS*)
- Dihydropyrimidine dehydrogenase deficiency (*DPYD*)
- Hypoxanthine-guanine phosphoribosyltransferase deficiency (*HPRT1*)
- Inosine triphosphatase deficiency (*ITPA*)
- Phosphoribosyl pyrophosphate synthetase superactivity and deficiency (*PRPS1*)
- Purine nucleoside phosphorylase deficiency (*PNP*)
- β -ureidopropionase deficiency (*UPB1*)
- Thymidine phosphorylase deficiency (*TYMP*)

Organic acidurias

- 2-methylbutyryl glycinuria /
- 2-methylbutyryl-CoA dehydrogenase deficiency (*ACADSB*)
- 3-methylglutaconaciduria type 3 / Optic atrophy (*OPA3*)
- 2-methylglutaconyl-CoA hydratase deficiency / 3-methylglutaconaciduria type I (*AUH*)
- Acyl-CoA synthetase family, member 3 deficiency (*ACSF3*)
- Canavan disease (*ASPA*)
- D-2- and L-2-hydroxyglutaric aciduria/ mitochondrial citrate transporter (*SLC25A1*)
- D-2-hydroxyglutaric acidurie met metaphyseal chondromatosis (*IDH1*)
- D-2-hydroxyglutaric aciduria (all genes mentioned below)
 - type I (*D2HGDH*) type II (*IDH2*)
- Ethylmalonic aciduria (Ethylmalonic encephalopathy) (*ETHE1*)
- Glutaric aciduria type 1 (*GCDH*)
- L-2-hydroxyglutaric aciduria (*L2HGDH*)
- Malonyl-CoA-decarboxylase deficiency (*MLYCD*)
- Methylmalonate semialdehyde dehydrogenase deficiency (*ALDH6A1*)
- Methylmalonyl CoA mutase deficiency (*MUT*)
- Methylmalonyl-CoA epimerase deficiency (*MCEE*)
- Mitochondrial DNA depletion syndrome 2 (*SUCLA2*)
- Propionic acidemia / Propionyl CoA carboxylase deficiency (all genes mentioned below)
 - alpha subunit (*PCCA*) beta subunit (*PCCB*)
- Succinate-CoA ligase (*SUCLG1*)

Peroxisomal disorders

- Acyl-CoA oxidase deficiency (*ACOX1*)
- α -methylacyl-CoA racemase deficiency (*AMACR*)
- Complementation analysis (PEX gene unknown; skin fibroblasts required)
- D-Bifunctional protein deficiency (*HSD17B4/DBP/MFE2*)
- Hyperoxaluria (all genes mentioned below)
 - type 1 (*AGXT*) type 2 (*GRHPR*) type 3 (*HOGA1*)
- Refsum disease (all genes mentioned below)
 - PEX7* *PHYH/PAHX*
- Rhizomelic chondrodysplasia punctata (*RCDP*)
 - type 1 (*PEX7*) type 2 (*GNPAT*) type 3 (*AGPS*)
- X-linked adrenoleukodystrophy (*ABCD1/X-ALD*)
- Zellweger spectrum (Zellweger syndrome, *NALD, IRD*)
 - PEX1* *PEX2* *PEX3*
 - PEX5* *PEX6* *PEX10*
 - PEX11B* *PEX12* *PEX13*
 - PEX14* *PEX16* *PEX19*
 - PEX26*

Clinical DNA testing is available for the following disorders (gene):

Vitamin, co-factor or metal disorder

- 5-Methyltetrahydrofolate-homocysteine methyltransferase deficiency, type cblG (*MTR*)
- 5-Methyltetrahydrofolate-homocysteine methyltransferase reductase deficiency, type cblE (*MTRR*)
- Brown-Vialetto-Van Laere syndrome (all genes mentioned below)
 - type 1 (*SLC52A3*)
 - type 2 (*SLC52A2*)
 - type 3 (*SLC52A1*)
- Ceruloplasmine deficiency/ ferroxidase (*CP*)
- Cytosolic iron-sulfur assembly component (*CIAO1*)
- Dihydrofolate reductase deficiency (*DHFR*)
- Folate malabsorption, hereditary (*SLC46A1*)
- Folate transport deficiency (*FOLR1*)
- Gastric intrinsic factor deficiency (*GIF*)
- Holocarboxylase synthetase deficiency (*HLC5*)
- Hypercarotenemia and vitamin A deficiency (*BCO1*)
- Hyperphenylalaninemia, BH4-deficiency, A (*PTS*)
- Hyperphenylalaninemia, BH4-deficiency, C / Dihydropteridine reductase deficiency (*QDPR*)
- Imerlund-Grasbeck syndrome 2 / Megaloblastic anemia-1, Norwegian type (*AMN*)
- Methylenetetrahydrofolate dehydrogenase 1 deficiency (*MTHFD1*)
- Methylenetetrahydrofolate reductase deficiency (*MTHFR*)
- Methylmalonic acidemia and homocysteinemia, cblX type (X-linked) (*HCFC1*)
- Methylmalonic aciduria and homocystinuria (all genes mentioned below)
 - cblC type (*MMACHC*)
 - cblD type (*MMADHC*)
 - cblF type (*LMBRD1*)
 - cblJ type (*ABCD4*)
- Methylmalonic aciduria, vitamin B12 responsive (all genes mentioned below)
 - cblA type (*MMAA*)
 - cblB type (*MMAB*)
 - transcobalamin receptor defect (*TCBIR / CD320*)
- Molybdenum cofactor deficiency (all genes mentioned below)
 - type A (*MOCS1*)
 - type B (*MOCS2*)
 - type C (*GPHN*)
- Pyridoxal phosphate binding protein (*PLPBP*)
- Pyridoxamine 5'-phosphate oxidase deficiency (*PNPO*)
- Pyridoxine dependent epilepsy (*ALDH7A1*)
- Thiamine transporter deficiency (*SLC19A3*)
- Zinc transporter deficiency/ Acrodermatitis enteropathica (*SLC39A4*)

Other defects

- Alexander disease (*GFAP*)
- Alkaline phosphatase, liver deficiency (*ALPL*)
- B-Cell receptor associated protein 31 deficiency (*BCAP31*)
- Carbonic anhydrase VA deficiency (*CA5A*)
- Chitotriosidase deficiency c.1049_1072dup (*CHIT1*)
- Collectrin/ Transmembrane protein 27 (*TMEM27*)
- Congenital disorder of glycosylation, type II h, CDG IIh (*COG8*)
- Galactosyltransferase 1 / Congenital disorder of glycosylation, type II d (*B4GALT1*)
- Hypomyelination/ phosphatidylinositol (PI) 4-kinase deficiency (*PI4KA*)
- LIPIN 3 (*LPIN3*)
- Majeed syndrome (*LPIN2*)
- Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities (NEDHSB) /Spermatogenesis-associated protein 5 deficiency (*SPATA5*)
- Nucleotide-binding protein-like protein deficiency (*NUBPL*)
- Rhabdomyolysis, acute recurrent (*LPIN1*)
- Sjögren-Larsson syndrome (*ALDH3A2/ALDH10*)
- Sodium-dependent citrate transporter, member 5 deficiency (*SLC13A5*)
- Spermine synthase deficiency (Snyder-Robinson) (*SMS*)

Clinical DNA testing is available for the following disorders (gene):

Aminoacyl tRNA synthetases (Mitochondrial)

- Alanyl-tRNA synthetase 2 (*AARS2*)
- Arginyl-tRNA synthetase 2 (*RARS2*)
- Asparaginyl-tRNA synthetase 2 (*NARS2*)
- Aspartyl-tRNA synthetase 2 deficiency /
Leukoencephalopathy with brain stem and spinal cord
involvement and lactate elevation "LBSL" (*DARS2*)
- Cysteinyl-tRNA synthetase 2 (*CARS2*)
- Glutamyl-t-RNA synthetase 2 deficiency /
Leukoencephalopathy with thalamus and brainstem
involvement and high lactate "LTBL" (*EARS2*)
- Histidyl-tRNA synthetase 2 (*HARS2*)
- Isoleucyl-tRNA synthetase 2 (*IARS2*)
- Leucyl-tRNA synthetase 2 (*LARS2*)
- Methionyl-tRNA synthetase 2 (*MARS2*)
- Phenylalanyl-tRNA synthetase 2 (*FARS2*)
- Prolyl-tRNA synthetase 2 (*PARS2*)
- Seryl-tRNA synthetase 2 (*SARS2*)
- Threonyl-tRNA synthetase 2 (*TARS2*)
- Tryptophanyl-tRNA synthetase 2 (*WARS2*)
- Tyrosyl-tRNA synthetase 2 (*YARS2*) V
- Valyl-tRNA synthetase 2 (*VAR2*)

Aminoacyl tRNA synthetases (Cytoplasmic)

- Alanyl-tRNA synthetase (*AARS*)
- Arginyl-tRNA synthetase (*RARS*)
- Asparaginyl-tRNA synthetase (*NARS*)
- Aspartyl-tRNA synthetase deficiency /
Hypomyelination with brainstem and spinal cord
involvement and leg spasticity "HBSL" (*DARS*)
- Glutamyl-Prolyl-tRNA synthetase (*EPRS*)
- Cysteinyl-tRNA synthetase (*CARS*)
- Glutaminyl-tRNA synthetase (*QARS*)
- Glycyl-tRNA synthetase (*GARS*)
- Histidyl-tRNA synthetase (*HARS*)
- Isoleucyl-tRNA synthetase (*IARS*)
- Leucyl-tRNA synthetase (*LARS*)
- Lysyl-tRNA synthetase (*KARS*)
- Phenylalanyl-tRNA synthetase (all genes mentioned below)
 - alpha subunit (*FARSA*)
 - beta subunit (*FARSB*)
- Seryl-tRNA synthetase (*SARS*)
- Threonyl-tRNA synthetase (*TARS*)
- Tryptophanyl-tRNA synthetase (*WARS*)
- Tyrosyl-tRNA synthetase (*YARS*)
- Valyl-tRNA synthetase (*VAR2*)

Aminoacyl tRNA synthetases (Related)


- Aminoacyl-tRNA synthetase complex-interactive multifunctional protein 1 (*AIMP1*)
- Aminoacyl-tRNA synthetase complex-interactive multifunctional protein 2 (*AIMP2*)
- D-tyrosyl-tRNA deacylase (*DTD1*)
- Eukaryotic translation elongation factor epsilon (*EEF1E1*)
- Threonyl-tRNA synthetase-like 2 (*TARS2*)

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 following address label for shipment



<p>Laboratory Genetic Metabolic Diseases (F0-132) Amsterdam UMC, location AMC Meibergdreef 9 1105 AZ Amsterdam The Netherlands</p>	
<p><u>DIAGNOSTISCH MATERIAAL</u> SPOED!</p>	