



**Metabolic Unit / Enzyme Diagnostics
Department of Clinical Chemistry
VU University Medical Center**

invoer	
brief	
autorisatie	
type	PED/PEV/ G
Projectcd.	
declaratie	BL / N
DNA nummer	

To be filled in by our laboratory:

Lab nummer:.....
Onderzoek:
Datum ontv:.....
Mat code:

Please fill in the following fields (see also page 2):

Patient information:	Material:	Sampling date:
Family name:	<input type="checkbox"/> Fibroblasts
First name: <input type="checkbox"/> Male <input type="checkbox"/> Female	<input type="checkbox"/> Lymphoblasts
Date of birth: (dd/mm/yyyy):	<input type="checkbox"/> Plasma (heparine), volume: ml
	<input type="checkbox"/> Blood (ACD/EDTA), volume: ml
Family members previously tested?	<input type="checkbox"/> Other:.....
<input type="checkbox"/> No <input type="checkbox"/> Yes: Our lab nr(s):		
Name:	Your reference nr:	

Clinical and biochemical details: (please fill in detailed information)

.....
.....
.....
.....

Requested enzyme assay

	Material		Material
<input type="checkbox"/> 4-Aminobutyrate aminotransferase (GABA-T)	LYB*, LYC	<input type="checkbox"/> L-2-hydroxyglutaric dehydrogenase (L2HGDH)	F, LYB, LYC
<input type="checkbox"/> Arginine;glycine amidinotransferase (AGAT)	LYB, LYC	<input type="checkbox"/> Methylene tetrahydrofolate reductase (MTHFR)	F, LYC
<input type="checkbox"/> Biotinidase (BIO)	P	<input type="checkbox"/> Ribose-5-phosphaat isomerase (RPI)	LYB*, F
<input type="checkbox"/> Creatine transporter (CTR)	F	<input type="checkbox"/> Ribulose-5-phosphaat epimerase (RPE)	**
<input type="checkbox"/> Cystathionine β-synthase (CBS)	F	<input type="checkbox"/> Succinic semialdehyde dehydrogenase (SSADH)	LYB*, LYC
<input type="checkbox"/> Dihydrofolate reductase (DHFR)	LYB	<input type="checkbox"/> Transaldolase (TALDO)	F, LYB
<input type="checkbox"/> Guanidinoacetate methyltransferase (GAMT)	F, LYB	<input type="checkbox"/> Transketolase (TK)	**
<input type="checkbox"/> D-2-hydroxyglutaric dehydrogenase (D2HGDH)	F, LYB	<input type="checkbox"/> Other:.....	

F = fibroblasts (skin biopsy); LYB = lymphoblasts; LYC = send (full) blood for lymphocyte isolation; P = plasma

* Preferred material, ** only after consultation

Clinical Biochemical Geneticists
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Information requesting physician:

Requesting physician:

Department:.....

Hospital/institute:

Address:

City and Zip-code:

Phone/ Fax :

E-mail address:

Copy of results to:

Please indicate if the patient or carekeeper does **NOT** give consent for storage of materials for future diagnostics or research.

Invoice to (if different from above) Alternatively an E-112 form can be attached, only European countries.

Name:.....

Department and hospital/institute:

Address:

City and Zip code:.....

Country:.....

In the case of payment by credit card:

Name:.....

No:.....

Type:.....

Exp date:.....

Conditions for sampling and shipment:

- Sampling: Plasma: 1 ml heparin plasma
- Lymphocytes: 15 ml ACD or EDTA (full) blood, for neonates minimal 6 ml. Provide also blood from a control subject.
- Fibroblasts and lymphoblasts: sent in complete culture media.
- Shipment: Plasma: Sent frozen on dry ice with courier service.
- Fibroblasts, lymphoblasts and blood for lymphocytes sent at room temperature with courier service (**never** freeze or cool).
- Sent samples from Monday to Wednesday. Samples should arrive at the laboratory within 24-48 hours after sampling.

Please contact Prof. Dr. C. Jakobs prior to sending blood or cells:

c.jakobs@vumc.nl
Phone: +31 (0)20 4442416

Shipment address:

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