

Laboratory Clinical Genetics

Request form Enzyme Diagnostics

Postal address

Maastricht UMC+
Laboratory Clinical Genetics
PO Box 5800, 6202 AZ Maastricht
The Netherlands



Material delivery

Laboratory Clinical Genetics, Sample Reception
P. Debyelaan 25, 6229 HX Maastricht
North Building, 2nd floor, Route 14
The Netherlands

T: +31 43 3871345

E: cmo.klin.genetica@mumc.nl

W: klinischegenetica.mumc.nl

PATIENT DETAILS (REQUIRED TO FILL IN)

Initials and name _____
Date of birth _____ Gender M/ F Multiple birth _____
Address _____
Zip code and city _____
Country _____

PHYSICIAN DETAILS

Name _____
Hospital _____
Department _____
Address _____
Zip code and city _____
Country _____
Telephone _____
E-mail physician _____
E-mail secretariat _____
CC result to _____

Extern ref.no. _____

Informed consent

It must be indicated below whether the advice seeker DOES or DOES NOT grant permission for the use of their material/data for follow-up research within the same research question.

The material/data may be used for follow-up research if no cause is found during the current test.

A follow-up examination within the same research question can then be performed without a new request. For example, when a new gene/variant is discovered or a more accurate test becomes available. The requester will be informed. The laboratory will not submit a new bill. Any costs incurred via the treating physician fall under the deductible.

The material/data may **not** be used for follow-up research in line with the current research question.

Further use of the material/data has not (yet) been discussed.

► **URGENT REQUEST** via cmo.klin.genetica@mumc.nl.

Please check boxes of specimen of requested analyses

SPECIMEN TYPE	DATE OF SAMPLE	TIME	SIGNATURE
1 x 6 ml EDTA blood	_____	_____	_____
1 x 6 ml Li HEPARIN blood	_____	_____	_____
5 ml pleural fluid	_____	_____	_____
Fibroblasts	_____	_____	_____
Other _____	_____	_____	_____

See page 3 for the correct collection, storage and shipping conditions.

To be completed only by Clinical Genetics Sample Reception employee

Material received in accordance with acceptance/shipping protocol Yes No Date of receipt _____

	BLOOD	SER/PLA	BM	
EDTA	# _____	# _____	# _____	DNA
HEPARIN	# _____	# _____	# _____	UMBILICAL CORD
SERUM	# _____	# _____	# _____	FIBROBLASTS
URINE	# _____ fractions	# _____ UP	# _____ UV	MUSCLE
LIQUOR	# _____ fractions			STRECK (NIPT)
OTHERWISE:	_____			
Quality indicator	_____ Signature _____			

Laboratory Clinical Genetics

Laboratory tests

Result turnaround time 3 weeks, unless otherwise stated.

Sugar metabolism

Galactokinase (GALK)	2 x 6 ml EDTA blood
Galactose-1-phosphate uridyltransferase (GALT)	2 x 6 ml EDTA blood

Purine metabolism

Adenosine Deaminase (SCID_ADA) ¹	2 x 6 ml EDTA blood
Adenosine Deaminase (TB screening) ¹	5 ml pleural fluid
Purine Nucleoside Phosphorylase (PNP) ¹	2 x 6 ml EDTA blood
<i>PNP and ADA activities are always both determined</i>	
Adenylosuccinaat lyase (ADSL) ¹	2 x 6 ml EDTA blood
Hypoxanthine Guanine Phosphoribosyl Transferase (HGPRT, Lesch-Nyhan syndrome) ¹	2 x 6 ml EDTA blood
Adenine Phosphoribosyl Transferase (APRT) ¹	2 x 6 ml EDTA blood

Pyrimidine metabolism

Thymidine phosphorylase (TYMP) ¹	2 x 6 ml EDTA blood
Pyrimidine 5'-nucleotidase I & II ¹	2 x 6 ml EDTA blood
Fluxomics analysis ¹	Fibroblasts
Functional evaluation of pyrimidine metabolism (<i>Turnaround time 3 months</i>)	

Hemolytic anemias

Hemolytic anemias package: enzyme diagnostics and genetic screening for G6PD and PKLR. For glutathione reductase (only enzyme -/+ FAD). (<i>Turnaround time 2 weeks</i>)	3 x 6 ml EDTA blood
Pyrimidine 5'-nucleotidase I & II ¹	2 x 6 ml EDTA blood

Other

Biotinidase	1 x 6 ml EDTA blood or Li HEPARIN blood
-------------	---

¹ ISO 15189 accreditation does not apply to these analyses, for turnaround time contact the laboratory.

For enzyme diagnostics of enzymes within purine or pyrimidine metabolism not mentioned above, please contact the laboratory.

Laboratory Clinical Genetics

Collection/storage conditions

- Urgency requests: notify us via cmo.klin.genetica@mumc.nl.
- All samples must contain patient name, date of birth and type of material.
- Blood must be delivered to the Laboratory Clinical Genetics within 24 hours of collection.
- The Laboratory Clinical Genetics cannot guarantee to process material that arrives after 12:00 on Friday.
- Sampling date must be written at page 1 of the request form.
- Children at least 2 ml whole blood.
- Do not centrifuge or freeze blood tubes; ship at room temperature.
- Freeze pleural fluid after collection and ship on dry ice.
- Mention recent blood transfusion.

Dispatch

- *By post*
Maastricht UMC+
Laboratory Clinical Genetics
P.O. Box 5800
6202 AZ Maastricht
The Netherlands
- *By courier (Monday to Thursday 08:30 - 16:00, Friday 08:30 - 12:00)*
Maastricht UMC+
Laboratory Clinical Genetics, Sample Reception
P. Debyelaan 25, 6229 HX Maastricht
North Building, 2nd floor, Route 14
The Netherlands

Questions?

Call +31 43 3871345 (option 1) if you have questions about purchase, storage and shipping conditions. Avoid delays and always use the latest version of the application form, which you can find on:

klinischegenetica.mumc.nl/aanvraagformulieren

Laboratory Clinical Genetics

General terms and conditions

1. Use of patient data and materials

The requestor's name must be entered on the first page of the request form. The requestor is responsible for fulfilling the requirement to inform the patient and/or the patient's parent(s) or legal representative of the ways in which patient data and materials are used. If the requestor's name is missing, the request may be refused.

I, the requestor, have informed this patient, and/or this patient's parent(s) or legal representative of the following:

- Where necessary, the personal data required for this test are stored in the MUMC+ systems.
- The MUMC+ Clinical Genetics Laboratory works closely with Radboudumc's partner laboratory. The test may be carried out by the partner laboratory and, in that case, the personal data will where necessary also be stored in Radboudumc's systems.
- In specific cases, the test is referred to a laboratory other than the ones mentioned above and the personal data are processed there.
- The security and privacy of the personal data and the material are guaranteed during this process.
- In order to develop new techniques and improve existing ones, Clinical Genetics uses anonymised patient material, e.g. for quality controls and validation. If the patient objects to the anonymous use of body material, he/she can make this known by ticking the appropriate box on page 1.

2. Requests

- 2.1 In order to avoid errors and delays, requests should be submitted in a clear and unambiguous manner. When completed, this request form contains all the desired data.
- 2.2 Requests may be refused if they contain insufficient information to achieve a result that meets the applicable quality criteria.
The minimum requirements are:
 - Patient identification, including gender, date of birth, address/contact details and unique identification.
 - Name or other unique identification of clinician, care provider or other person legally authorised to request tests or use medical information.
- 2.3 Relevant medical information on the patient must be provided to enable the test to be performed and the result to be interpreted. In case of doubt, a laboratory specialist in clinical genetics can be contacted.
- 2.4 Clinical Genetics must be given the opportunity to discuss the requested test with the requestor/clinician.
- 2.5 A laboratory specialist in clinical genetics is available during office hours to give advice on how to request tests.
- 2.6 By accepting a request, the Clinical Genetics department undertakes to perform the requested work with care and diligence in accordance with the quality criteria applicable to the department.
- 2.7 Before submitting patient material, the requestor is asked to check whether the patient concerned is insured for clinical genetic care. If, after a procedure has been performed, the patient proves not to be insured, the account will be sent to the patient.

Laboratory Clinical Genetics

3. Samples

- 3.1 The requestor will submit the samples to be tested to Clinical Genetics under the correct conditions, accompanied by details of material type and identification (name, gender and date of birth) and a request form completed in full.
- 3.2 The materials are collected from each patient as indicated on the request form. Other materials or quantities will only be considered after consultation by telephone.
- 3.3 It is possible to register a cito/urgent request by telephone. See page 1 of the request form.
- 3.4 Samples must be kept and transported in accordance with the conditions stated.
- 3.5 Clinical Genetics may refuse the sample submitted if:
 - the requirements set out in 1, 3.1, 3.2 and 3.4 are not satisfied;
 - the sample does not meet the quality criteria set.
- 3.6 Unless otherwise agreed at the time of submission of the request, Clinical Genetics will keep the (tested) samples or the remainder thereof after testing, in accordance with its own regulations, for an indefinite period.

4. Execution

- 4.1 Clinical Genetics determines the manner in which the work is performed and the method and equipment used to perform it.
- 4.2 All work will be performed in accordance with the applicable norms, standards and rules. Upon request, Clinical Genetics will provide the requestor with the relevant information.
- 4.3 If a request extends to include work in an area of which it has no knowledge or experience, Clinical Genetics will contact the requestor to arrange the outsourcing of this work.
- 4.4 All actions and storage prior to taking receipt of a sample fall outside the responsibility of Clinical Genetics.

5. Results

- 5.1 Results in the form of test reports, advice, information or otherwise will be presented by Clinical Genetics in writing to the person requesting the test.
- 5.2 The deadlines for results are as stated on the request form. In the event of an urgent request, alternative deadlines may be arranged by mutual agreement.
- 5.3 A laboratory specialist in clinical genetics is available during office hours to give advice on how to interpret test results.

6. Confidentiality

- 6.1 Personal information is protected in accordance with the General Data Protection Regulation (GDPR) and ISO-27001. Confidentiality of data is guaranteed and laid down in the hospital regulations of Maastricht UMC+.