Maastricht UMC+

Laboratory Clinical Genetics

Request form Biochemical Diagnostics

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

TESTEN RvA M 150

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 _____

PATIENT STICKER

PHYSICIAN DETAILS

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

PUT YOUR STAMP HERE

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 **Li HEPARIN** blood	_____	_____	_____
☐ Urine	by _____ to _____	_____	_____
☐ Liquor / CSF	_____	_____	_____
☐ Other	_____	_____	_____

See page 4 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 _____

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Quantitative analyses *

Result turnaround time 2-3 weeks

☐	Amino acids	6 ml **Li HEPARIN** blood / 5 ml urine / CSF / dried blood spot
☐	Organic acids (incl. acylglycines)	5 ml urine
☐	Purines and pyrimidines	5 ml urine / 6 ml **Li HEPARIN** blood
☐	Very long chain fatty acids	6 ml **Li HEPARIN** blood
☐	Sugar alcohols (incl. galactitol)	5 ml urine
☐	Sugars	5 ml urine
☐	Targeted Urine Metabolomics	5 ml urine
☐	LysoGb3 (Biochemical screening for Fabry)	6 ml **Li HEPARIN** blood
☐	Kidney stone screening	5 ml urine

* If multiple indications are checked for the same material type during the single analyses, the material only needs to be collected once.

Indications and questions

Result turnaround time 3-6 weeks

☐	**Biochemical screening for IEM**[1]	1 x 6 ml **Li HEPARIN** blood and 15 ml urine

Fill in clinical specification below.

1 Our partner metabolic laboratory at Radboudumc in Nijmegen (Metabolic laboratory, Genome diagnostics) handles the technical execution of several analyses.

Specification of clinical data / medication / nutrition

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Clinical indications

I general physical characteristics

100-103 P 10 50 90 length
110-113 P 10 50 90 weight to height ratio
120-123 P 10 50 90 head circumference
130 ☐ abnormal appearance/dysmorphia*
131 ☐ hepatomegaly
132 ☐ splenomegaly
133 ☐ pre-/dysmature
134 ☐ edema
135 ☐ jaundice
136 ☐ abnormal blood pressure
137 ☐ hair abnormalities
138 ☐ skin abnormalities
139 ☐ eye abnormalities
140 ☐ deafness
141 ☐ body odor
142 ☐ (near)-SIDS/ALTE
143 ☐ tachypnea
144 ☐ hyperventilation
145 ☐ xanthomatose
146 ☐ hydrops
147 ☐ ascites
148 ☐ hypertension
149 ☐ hypotension
150 ☐ speech disorder
151 ☐ gait disturbance
152 ☐ extreme fatigue
154 ☐ exercise tolerance

II neurological/muscle abnormalities

200 ☐ intellectual disability
201 ☐ motor development delay
202 ☐ abnormal EEG/CT/MRI scan*
203 ☐ spasticity
204 ☐ hypertonia
205 ☐ convulsions/seizures/epilepsy*
206 ☐ abnormal eye movements
207 ☐ hypotension
209 ☐ ataxia/athetosis*
212 ☐ myoclonus
213 ☐ autism/solitary behavior*
214 ☐ cardiological problems*
215 ☐ dystonia
216 ☐ unexplained leukoencephalopathy
217 ☐ myopathy
218 ☐ muscular dystrophy
219 ☐ muscle weakness
220 ☐ inappropriate crying
221 ☐ psychosis
222 ☐ impulsive aggressive behavior
223 ☐ coma/decreased consciousness
224 ☐ lethargy
225 ☐ pyramidal syndrome
226 ☐ extrapyramidal syndrome
227 ☐ cerebellar syndrome
228 ☐ peripheral neuropathy
229 ☐ stroke

III gastrointestinal abnormalities

300 ☐ vomiting
301 ☐ diarrhea
302 ☐ oral aversion*
303 ☐ colic
304 ☐ delayed bowel transit
305 ☐ nutritional status*
306 ☐ growth disorder

IV nephrological abnormalities

400 ☐ nephrolithiasis
401 ☐ polyuria
402 ☐ abnormal urinary odor/color*
404 ☐ renal insufficiency
405 ☐ anuria

V radiological abnormalities

500 ☐ bone age delay
501 ☐ skeletal abnormalities
502 ☐ osteoporosis
503 ☐ rickets

VI immuno-/hematological abnormalities

600 ☐ recurrent infections
601 ☐ vaccination triggered symptoms
602 ☐ immunodeficiency
603 ☐ vascular abnormalities
604 ☐ hemolysis
605 ☐ anemia*
606 ☐ neutropenia*
607 ☐ lymphopenia*
608 ☐ thromboembolic
609 ☐ bleeding tendency
610 ☐ abnormal morphological*
611 ☐ premature atherosclerosis
612 ☐ vacuolated lymphocytes
613 ☐ granular leukocyte
614 ☐ thrombocytopenia
615 ☐ sepsis

VII laboratory abnormalities

700 ☐ cloudy serum
701 ☐ hyperlipidemia
702 ☐ hormones*
703 ☐ electrolytes*
704 ☐ liver enzymes*
705 ☐ muscle enzymes*
706 ☐ trace elements/vitamins*
707 ☐ hypoglycemia
708 ☐ hypouricemia/hypouricosuria*
709 ☐ hyperuricemia/hyperuricosuria*
710 ☐ hyperammonemia
711 ☐ acidosis/ketosis*
712 ☐ hypoimmunoglobulinemia
713 ☐ abnormal urea/creatinine*
714 ☐ reducing substance in urine
715 ☐ proteinuria

VIII genetics

800 ☐ consanguinity
801 ☐ metabolic disease in family*
802 ☐ SIDS of sibling
803 ☐ pharmacogenetic defect

IX ophthalmological abnormalities

930 ☐ retinitis pigmentosa
931 ☐ cataract
932 ☐ corneal opacity
933 ☐ nystagmus
934 ☐ strabismus
935 ☐ lens luxation

X food/medication

900 ☐ oral standard diet
901 ☐ parenteral nutrition*
910 ☐ 1 g/kg protein intake
911 ☐ 2 g/kg protein intake
912 ☐ 3 g/kg protein intake
913 ☐ 4 g/kg protein intake
914 ☐ 5 g/kg protein intake
920 ☐ medication

* specification required

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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.
- Sampling date must be written at page 1 of the request form.
- For children we require at least 1 ml whole blood and/or 2 ml urine.
- When several dedicated assays are requested for the same sample type, only one volume is needed.
- Shipment must be sent on dry-ice.

Instructions for applicants outside the MUMC+			
Material type	**Action**	**Storage conditions**	**Shipping condition**
Li HEPARIN blood	Separate plasma	-20ºC	dry ice
EDTA blood	Separate plasma	-20ºC	dry ice
Urine sample/collection	Freeze	-20ºC	dry ice
Blood spot	-	room temperature	room temperature
Liquor	Freeze	-20ºC	dry ice
Other	Please contact the lab by phone (+31 43 3871345, option 1) before sending the material.		

Dispatch

Applicants outside the MUMC+ follow the instructions in the table above.

- *By post*
  Maastricht UMC+
  Laboratory Clinical Genetics
  P.O. Box 5800
  6202 AZ Maastricht
  The Netherlands
- *By courier (Monday to Friday 08:30 - 17:00)*
  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 at:
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.

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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.

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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+.