

## Requestform Clinical Genetics Maastricht

## ▶ Postal address

Maastricht Universitair Medisch Centrum  
 Laboratorium Klinische Genetica  
 Postbus 5800  
 6202 AZ Maastricht  
 The Netherlands  
 Opening hours laboratory: 08:30 - 16:30  
 Head laboratory: Dr. A. van den Wijngaard  
 Tel: +31(0)43-3871345  
 Fax: +31(0)43-3877901  
 @: [cmo.klin.genetica@mumc.nl](mailto:cmo.klin.genetica@mumc.nl)

## ▶ Patient information (\* =required field)

Initials\*  
 Last name\*  
 Date of birth\*  
 Gender\*  
 Ethnical background  
 Address  
 Postal code  
 City  
 Reference / MDN  
 Is patient deceased?  
 Date of death

## ▶ Invoice address (\* =required field)

Name\*  
 Address\*

E-mail  
 Vat number

## ▶ Referring physician (\* =required field)

Name\*  
 Hospital / Institute\*  
 Address  
 Email\*  
 Specialism  
 Department\*  
 Country  
 Telephone

Fax  
 Copy results

## ▶ Test(s) requested

## ▶ Material

Material already available in our lab?  
 Date of sample collection  
 Which kind of material will be included?

DNA isolated from:

## ▶ Reason of the request

Reason of referral  
 Gene involved and mutation  
 Has material of family member(s) been sent previously?  
 Is this an urgent request?  
 Reason of urgency

Name(s)

Date of birth

Date of request:

Version: 09-2024

**To be filled in by the lab**

Datum ontvangst:  
 Paraaf ontvangst:

Ingescande pagina's:

monstersticker(s)

Materiaal:	EDTA	HEP	DNA	Vlok	Vruchtw.	Overig:
Aantal:						

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► **Family members**

Relation to current patient

Other familial information

Will you include parental samples?

Name father

Name mother

Parental clinical details

Which kind of parental material?

Date of birth father

Date of birth mother

Reason including parental samples

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► **Informed consent**

**I have informed my patient that the knowledge of genetic conditions is likely to improve in the future. He/She understands that, using this knowledge, the diagnostic laboratory is able to maintain an active search to identify the genetic cause of the disease for which the test was performed.**

The patient *ALLOWS* the material and/or data to be used for analysis aimed at the identification of the genetic cause of disease. In case a diagnosis is obtained, I (the medical specialist) will be informed by the laboratory, and I will communicate the outcome with the patient. The patient understands that possible additional costs can follow when medical care is needed. The laboratory do not send an additional invoice.

The patient *DOES NOT ALLOW* the material and/or data to be used for additional analysis in line with the original diagnostic request.

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

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► **Clinical information**

Birth weight

Current length

Age of diagnosis

Clinical relevant information

Current weight

Head circumference

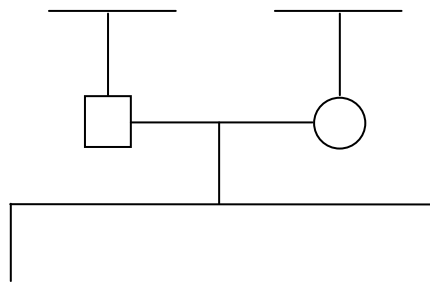
Consanguinity

► **Expected inheritance**

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► **Pedigree chart**

Please mark the individual of this request with an arrow (->). Designate affected family members as ■ / ● and indicate previous sent family samples with name and date of birth.



► **Collaboration MUMC+**

The laboratory Genomediagnosics Nijmegen closely collaborates with the Clinical Genomics Laboratory of the MUMC+ in Maastricht. A limited part of the test offer (marked <sup>1</sup>) will be carried out by the partner laboratory in Maastricht. In that case, the diagnostic report will be issued directly by the Maastricht laboratory, but the payment will be issued by Genomediagnosics Nijmegen. Direct analyses of variants in family members detected by whole exome sequencing (WES) will be carried out by the laboratory that reported the variant originally in the index.

## Material withdrawal form

### ► Patient information/sticker

Initials

Date of birth

Last name

Gender

### ► Material and shipping conditions

- Put name, gender and date of birth on each blood- or DNA tube. Improperly labelled samples will be refused. **Excess tubes will not be stored!**
- Ship samples at room temperature. **Do not freeze blood samples!**
- Do not submit material if patient has undergone bone marrow or stem cell transplant. Please contact lab.
- Upon cancellation the requested test will be charged completely.
- Additional information for requesting diagnostic testing at the Division of Genome Diagnostics can be found at <https://www.radboudumc.nl/en/afdeling/genetica/about-us/genomediagnosics>

Service	Turnaround time	Required material
Exome sequencing diagnostics (WES)	Exome gene panel analysis: 2-3 months	Preferably: - 2 x 3-6ml EDTA blood - 20 ml amniotic fluid (only Rapid) or >30 ml (Rapid + QF-PCR + growing cells)
	Exome gene panel analysis followed by exome wide analysis (in one report): 2-3 months	If there is no blood and/or amniotic fluid available: - Desired amount DNA: 5ug - Absolute minimum: 3ug (minimum concentration is 25 ng/ul, volume 30 ul). Prenatal material minimum is 450 ng (30 ul of 15 ng/ul)
	Rapid trio/de novo analysis of proband and both parents: <15 business days	
Interpretation of exome data	2 months	n/a
Array diagnostics (genome wide)	5 weeks	2 x 3-6ml EDTA blood
Multiple gene diagnostics (Gene panels)	3-8 weeks*	2 x 3-6ml EDTA blood
Single gene diagnostics	4-8 weeks*	2 x 3-6ml EDTA blood
Mutation diagnostics (carrier testing)	4 weeks	<b>Gene/array diagnostics:</b> 2 x 3-6ml EDTA blood  <b>Chromosome diagnostics (karyotyping):</b> 2 x 5ml Heparin blood in Natrium- or Lithium-Heparin tubes (neonates 1-2ml)
Pharmacogenetics	1-8 weeks*	2 x 3-6ml EDTA blood
Chromosome diagnostics	2-5 weeks	<b>Karyotyping:</b> 2 x 5ml Heparin blood in Natrium- or Lithium-Heparin tubes (neonates 1-2ml)  <b>QF-PCR:</b> neonates: 1-2ml EDTA blood
FISH	2-5 weeks	2 x 5ml Heparin blood in natrium- of lithium-heparin tubes
Urgent request	Please, contact: <a href="mailto:gen@radboudumc.nl">gen@radboudumc.nl</a> or Tel: +310243613799, TAT depends on technique	Please, see material under requested service
mtDNA	4-12 weeks	Cooled (none frozen) urine, we prefer 50-100ml

\* In our application system you will find the exact turnaround times for each individual test