

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

► 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: 04-2023

To be filled in by the lab

Datum ontvangst:
Paraaf ontvangst:

Ingescande pagina's:

monstersticker(s)

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

► 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

► 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.

► Clinical information

Birth weight

Current length

Age of diagnosis

Clinical relevant information

Current weight

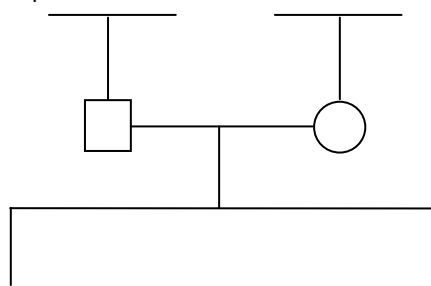
Head circumference

Consanguinity

► Expected inheritance

► 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 ¹) 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!**
- 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: |
| | Rapid trio/de novo analysis of proband and both parents: <15 business days | - 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) |
| 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 | Gene/array diagnostics: 2 x 3-6ml EDTA blood Chromosome diagnostics (karyotyping): 2 x 5ml Heparin blood in Natrium- or Lithium-Heparin tubes (neonates 1-2ml) |
| Farmacogenetics | 1-8 weeks* | 2 x 3-6ml EDTA blood |
| Chromosome diagnostics | 2-5 weeks | Karyotyping: 2 x 5ml Heparin blood in Natrium- or Lithium-Heparin tubes (neonates 1-2ml) QF-PCR: neonates: 1-2ml EDTA blood |
| FISH | 2-5 weeks | 2 x 5ml Heparin blood in natrium- of lithium-heparin tubes |
| Urgent request | Please, contact: gen@radboudumc.nl 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