

# **Requestform Genomediagnostics Nijmegen**

#### Postal address

Radboudumc Genome Diagnostics, route 815 Head laboratory: Dr. H. Yntema Postbus 9101 6500 HB Nijmegen The Netherlands

Opening hours laboratory: 08:30 - 16:30

Tel: +31(0)24 3613799 Fax: +31(0)24 3616658 @: gen@radboudumc.nl

• https://order.radboudumc.nl/en/genetics

## ► 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\* E-mail Address\* Vat number

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

Hospital / Institute\*

Address Email\* Specialism Department\* Country

Fax

Copy results

### Test(s) requested

Telephone

## 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: 12-2022

## To be filled in by the lab

Datum ontvangst:

Paraaf ontvangst:

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

Ingescande pagina's:

monstersticker(s)							

#### 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 diagostic request.

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

#### Clinical information

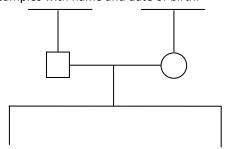
Birth weight Current weight
Current length Head circumference
Age of diagnosis Consanguinity

Clinical relevant information

## Expected inheritance

#### Pedigree chart

Please mark the individual of this request with an arrow ( $\rightarrow$ ). Designate affected family members as  $\blacksquare$  / $\bullet$  and indicate previous sent family samples with name and date of birth.



#### Collaboration MUMC+

The laboratory Genomediagnostics 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 Genomediagnostics 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.

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## **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/afdelingen/genetica/about-us/genomediagnostics

Service	Turnaround time	Required material		
Exome sequencing diagnostics (WES)	Exome gene panel analysis: 2-3 months  Exome gene panel analysis followed by exome wide analysis (in one report): 2-3 months	Preferably: - 2 x 3-6ml EDTA blood - 20 ml amniotic fluid (only Rapid) or >30 ml (Rapid + QF-PCR growing cells)  If there is no blood and/or amniotic fluid available: - Desired amount DNA: >1u - Absolute minimum (e.g prenatal material) is 450 ng (30 ul o 15 ng/ul). Minimum concentration is 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	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.nlor Tel: +310243613799, TAT depends on technique	Please, see material under requested service		
DNA 4-12 weeks		Cooled (none frozen) urine, we prefer 50-100ml		

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

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