Print Form

REQUEST FORM CONSTITUTIONAL GENETIC ANALYSIS

version 2/20251001

Label sample

CENTRE FOR MEDICAL GENETICS
UZ Brussel
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Universitair Ziekenhuis Brussel

https://www.uzbrussel.be/web/centrum-voor-medische-genetica/

BELAC 141-MED accredited according to quality standard ISO15189

Identification patient *

* Mandatory data

Name: Sticker
First name:
Date of birth: Gender (M/F):
Residential address: identification patient

Invoice address:

Email address:
Phone:
National registry N°:
Ethnic origin:

Identification referring physician *

Name: Stamp
First name:
Referring service:
Address: referring physician

Email address:
Phone:
RIZIV/INAMI number:
Signature *:

Request date *:
Your reference:
Copy result to:
Address:

☐ Genetisch rapport in Nederlands ☐ Genetics report in English

Sample data *

Sample type
☐ Blood
☐ Dried blood spots
☐ Biopsy Specify:
☐ DNA from Specify:
☐ Tissue culture ☐ Fibroblast culture or skin biopsy
☐ Other Specify:
☐ Other Specify:

☐ Stock sample Reason:
Collection date:

Indication *

☐ Diagnostic analysis
☐ Check familial DNA variant/chromosomal aberration (1)/(2) required
☐ Presymptomatic analysis 2nd independent sample required
☐ Research Specify:
☐ Other Specify:
☐ Urgent Reason:

Urgent = minimal turn-around-time
Final decision urgency is determined by lab

Clinical information * add in capitals please

☐ Symptomatic ☐ Asymptomatic

Extra data in attachment

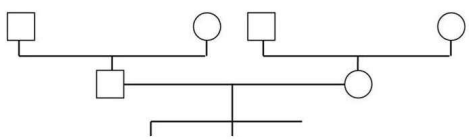
☐ Informed consent
☐ Pedigree
☐ Clinical report/checklist
☐ Genetic report (index patient)

Family data

☐ Family member followed elsewhere Specify:
Name family member:
First name family member:
Date of birth family member: Gender (M/F):
Relation patient to family member:
Clinical findings family member (1) :

Genetic defect in family member (2) : addition genetic report required

Consanguinity
☐ Between partner and patient
☐ Between parents of patient
☐ Other Specify:



☐ man
◯ woman
◇ fetus
☐◯◇ normal
◧◑◈ carrier
■●◆ affected
╱ deceased

Additional information other family members:

Chromosomal analysis

Sampling	Chromosomal analysis	Specification	TAT
H	☐ Conventional karyotyping	:	4-10 weeks
E	☐ Molecular karyotyping	:	2-12 weeks
E	☐ QF-PCR (chr X, Y, 13, 18, 21)	:	2- 6 weeks
H	☐ FISH	:	2-12 weeks

Biochemical analysis

Sampling	Lysosomal storage disease	Enzyme	Specification	TAT
S E	☐ Chitotriosidase activity	chitotriosidase		2-3 months
H	☐ Fabry (only analysed for male)	α-galactosidase		2-3 months
H	☐ Gaucher	β-glucosidase		2-3 months
H	☐ MPS1-Hurler-Scheie	α-L-iduronidase		2-3 months
F ☏/@	☐ Pompe	α-glucosidase		2-3 months

REQUEST FORM CONSTITUTIONAL GENETIC ANALYSIS

Identification patient *

Name & first name patient: _____

Date of birth: _____ Gender (M/F): _____

Legend

- E EDTA blood min. 4ml
- F only fibroblasts
- H Na-Hep blood min. 7ml
- S tube without additive 5ml (serum)
- * mandatory data
- IC informed consent required
- KV clinical report required
- T trio (index+parents) required
- ☏ /@ only after phone/mail consult

Print Form

Molecular (DNA) analysis

(Detailed information gene panels see http://www.brightcore.be/gene-panels)

Sampling	Blood disorders	Gene	Specification	TAT
E	☐ Hemochromatosis	HFE p.His63Asp & p.Cys282Tyr	Serum ferritine*: _____ Transferrine sat*: _____	2-4 weeks
E	☐ Hemoglobinopathy	☐ HbS ☐ HbC ☐ α-thal ☐ β-thal	α-thal and β-thal: prior Hb-electrophoresis required	1-3 months

Sampling	Blood coagulation problems	Gene (variant)	Specification	TAT
E	☐ Antithrombine deficiency	☐ gene SERPINC1 ☐ targeted	: _____	1-3 months
E	☐ Complement factor H	☐ gene CFH ☐ targeted	: _____	1-3 months
E	☐ Factor II/Prothrombine	F2 c.*97G>A		2-4 weeks
E	☐ Factor V/APC-cofactor	F5 c.1601G>A p.Arg506Gln	APC resistence*: _____	2-4 weeks
E IC	☐ Protein C deficiency	☐ gene PROC ☐ targeted	: _____	1-3 months

Sampling	Cardiac disorders	Gene panel/targeted	Specification	TAT
E IC KV	☐ Cardiomyopathie	☐ gene panel ☐ targeted	: _____	2-6 months
E IC KV	☐ Primary cardiac arrhythmia	☐ gene panel ☐ targeted	: _____	2-6 months

Sampling	Endocrinological disorders	Gene/gene panel/targeted	Specification	TAT
E	☐ Androgen receptor	☐ gene AR ☐ targeted	: _____	1-3 months
E	☐ Calcium-sensing receptor	☐ gene CASR ☐ targeted	: _____	1-3 months
E	☐ Combined pituitary hormone deficiency	☐ gene PROP1 ☐ gene POU1F1 ☐ targeted	_____	1-6 months
E IC	☐ Hypogonadotropic hypogonadism	☐ gene panel ☐ targeted	: _____	2-6 months
E IC	☐ Obesitas, monogenic, early onset	☐ gene panel ☐ targeted	: _____	2-6 months
E IC	☐ Thyroid dysgenesis	☐ gene panel ☐ targeted	: _____	2-6 months
E	☐ Thyroid hormone resistance	☐ gene THRB ☐ targeted	: _____	1-3 months

Sampling	Familial cancer	Gene/gene panel/targeted	Specification	TAT
E IC	☐ Breast and/or ovarium carcinoma	☐ gene panel ☐ targeted	: _____	1-3 months
E IC	☐ Colon carcinoma (Lynch/polyposis)	☐ gene panel ☐ targeted	: _____	1-3 months

Sampling	Metabolic disorders	Gene/gene panel/targeted	Specification	TAT
E	☐ Aldolase B/fructose intolerance	☐ gene ALDOB ☐ targeted	: _____	2-4 weeks
E IC	☐ Congenital defects of glycosylation	☐ gene panel ☐ targeted	: _____	2-6 months
E IC	☐ Glycogen storage disease	☐ gene panel ☐ targeted	: _____	2-6 months
E IC	☐ Lysosomal storage disease	☐ gene panel ☐ targeted	: _____	2-6 months
E IC	☐ Neurotransmitter aandoening	☐ gene panel ☐ targeted	: _____	2-6 months
E IC	☐ Organic aciduria	☐ gene panel ☐ targeted	: _____	2-6 months
E IC	☐ Peroxisomal disorder	☐ gene panel ☐ targeted	: _____	2-6 months
E IC	☐ Metabolic disorder (other)	☐ gene panel ☐ targeted	: _____	2-6 months

Sampling	Mitochondrial disorders	Gene/gene panel/targeted	Specification	TAT
E	☐ Aminoglycoside induced deafness	MT-RNR1	: _____	2-6 months
E	☐ Leigh or NARP syndrome		: _____	2-6 months
E	☐ LHON syndrome	MT-ND1 m.3460, MT-ND4 m.11778, MT-ND6 m.14484		2-4 weeks
E	☐ MERRF/MELAS (incl.MIDD)	MT-TK tRNALys, MT-TL1 tRNALeu		2-6 months
E IC KV	☐ Mitochondrial disorder, nuclear	☐ gene panel ☐ targeted	: _____	2-6 months
E	☐ MNGIE		: _____	2-6 months
E IC	☐ mtDNA deletions		: _____	2-6 months
E IC	☐ mtDNA depletie syndrome (MDDS)	☐ gene panel ☐ targeted	: _____	2-6 months
E IC KV	☐ mtDNA resequencing	complete mtDNA	: _____	2-6 months
E	☐ Polymerase G	☐ gene POLG ☐ targeted	: _____	2-6 months

Sampling	Neurological/neurodegenerative disorder	Gene/gene panel/targeted	Specification	TAT
E IC KV	☐ Epilepsy (incl. EIEE) T	☐ gene panel ☐ targeted	: _____	2-6 months
E	☐ GTP cyclohydrolase I deficiency (Segawa AD)	☐ gene GCH1 ☐ targeted	: _____	1-3 months
E	☐ PLA2G6-ass neurodegenerative disorder	☐ gene PLA2G6 ☐ targeted	: _____	1-3 months
E	☐ Spinocerebellar ataxia+DRPLA	SCA 1, 2, 3, 6, 7, 8, 17 + ATN1		3-6 months
E	☐ Tyrosine hydroxylase (Segawa AR)	☐ gen TH ☐ targeted	: _____	1-3 months
E	☐ Huntington disease	HTT CAG-repeat		2-3 months
E	☐ Kennedy disease	AR CAG-repeat		2-3 months

Sampling	(Neuro)development- and growth disorders	Gene/gene panel/targeted	Specification	TAT
E	☐ Achondroplasia	FGFR3 c.1123G>T, p.Gly375Cys & c.1138G>A, p.Gly380Arg & c.1138G>C, p.Gly380Arg		2-4 weeks
E IC KV	☐ Congenital malformation(s)/MCA T	☐ gene panel ☐ targeted	: _____	2-6 months
E IC KV	☐ Cortical malformations	☐ gene panel ☐ targeted	: _____	2-6 months
E	☐ Fragiele-X syndrome	FMR1 CGG-repeat		2-4 weeks
E KV	☐ Hydrocephaly, X-linked	☐ gene L1CAM ☐ targeted	: _____	1-3 months
E IC KV	☐ Neurological development disoders T	☐ gene panel ☐ targeted	: _____	2-6 months
E IC KV	☐ Skelet dysplasia T	☐ gene panel ☐ targeted	: _____	2-6 months

Sampling	(Neuro)muscular disorders	Gene/gene panel/targeted	Specification	TAT
E	☐ AMP deaminase	☐ gene AMPD1 ☐ targeted	: _____	2-4 weeks
E	☐ Becker-Thomsen myotonia	☐ gene CLCN1 ☐ targeted	: _____	1-3 months
E	☐ Congenital (para)myotonia	☐ gene SCN4A ☐ targeted	: _____	1-3 months
E	☐ Myotonic dystrophy/Steinert disease	DMPK CTG-repeat		2-3 months
E IC	☐ Neuromuscular disorder	☐ gene panel ☐ targeted	: _____	2-6 months
E	☐ Spinal muscular atrophy	SMN1 del ex7		2-4 weken

Sampling	Diverse	Gene/gene panel/targeted	Specification	TAT
E	☐ Mucoviscidosis	CFTR frequent variants		2-4 weeks
E	☐ Incontinentia pigmenti	☐ gene IKBKG ☐ targeted	: _____	1-3 months
E	☐ X-inactivation		: _____	1-3 months
E ☏/@	☐ Other		: _____	1-6 months

We strive to complete the analyses within the set turnaround times (TAT). In exceptional situations, we may deviate from the standard turnaround time.

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D01-E
INFORMED CONSENT FOR GENETIC ANALYSIS AND RESEARCH

IDENTIFICATIE PATIËNT (* verplicht)

[Sticker ID patiënt]

I. PERMISSION FOR:

I consent to a diagnostic genetic test being performed on a blood sample, or other samples for the following condition:

.....

(tick one choice)

- ○ LIMITED genetic analysis or TARGETED analysis of gene(s)
- ○ WIDE genetic analysis or NON-TARGETED genome wide analysis

A wide genetic analysis could lead to incidental and/or secondary findings of genetic results unrelated to the condition for which the analysis was originally performed (such as risk for cancer, heart disease, or other conditions for which an appropriate prevention, follow-up or treatment is available).
I realise that such a diagnosis may have consequences for my family as well as for myself.

- ○ I **do not** wish to be informed of such incidental and/or secondary genetic results.

II. RIGHT TO CHANGE PERMISSION

I have the right to withdraw my consent at any *time**. The withdrawal of my consent will be recorded in writing and added to my patient record. The healthcare provider will inform me of any possible consequences of withdrawing my consent on my further medical treatment.

**The change, however, does not apply to data and/or results already collected before my request to withdraw my consent.*

III. STORAGE AND EXCHANGE OF DATA/SAMPLES for DIAGNOSTICS and RESEARCH

I understand that:

- my medical data and genetic samples are coded (security method) so that they can be used for diagnostic, and/or scientific research approved by the Ethics Committee.
- the exchange of coded medical and genetic data between experts is important to improve (general) knowledge of genetic disorders and/or development of tests. This could lead to:
  - a better diagnosis for myself and/or others,
  - better healthcare and therapeutic options,
  - better prevention.

  In addition, the results may be published in scientific journals or presented at scientific meetings.
- my data can be analysed again and more widely (re-analysis) in the context of improved diagnostics and approved research projects, without having to inform me in advance. Please note that no systematic re-analysis of data currently takes place.

IV. COSTS of diagnostic genetic analysis

If my health insurance does not cover the cost of genetic testing, I will be held liable.

V. VOLUNTARY participation

My participation is entirely voluntary and will in no way provide any financial benefits to me.

NOTES

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To be completed by the PATIENT or REPRESENTATIVE:

- I confirm that I have been properly informed regarding the objectives and nature of the analysis related to my condition.
- I received the necessary info from the healthcare provider and/or I received, read and understood the accompanying information leaflet.
- I have had time and opportunity to ask questions, and I am satisfied with the answers and additional explanations.

I give this consent informed, knowingly and freely.

Date:

Signature:

*If representative:

Relationship to patient:

Name and first name:

To be completed by the professional:

I hereby confirm that I have informed the signed patient/ representative and answered questions in the best possible way regarding the possible results, limitations and options for the above genetic testing

Date:

Signature: [Stamp health care provider]

Notes on storage and use of medical (personal) data and bodily material

After genetic testing as part of your diagnostics, some material is often left over (residual material). This material can be stored for possible future use. The possible uses are explained below:

- **Diagnostic testing - new diagnostic test:** Your stored material can be reused later to investigate a genetic cause of a disorder of yourself or your family. Your data will be encrypted to protect your privacy. If an important discovery is made for you and your health, you *may* be informed through your treating physician. This finding will then have to be confirmed for you with an independent test.
- **Scientific research:** The material can also be used for scientific research, for example to develop new knowledge about genetic diseases. This may eventually become important for the treatment of other patients. Results are generally not reported back.
- **General genetic research:** The material may contribute to broader scientific research that helps others, for example in developing new treatments or testing new diagnostic devices in the laboratory. Your material can serve as control material in general research, without directly applying to you as an individual. This can help compare genetic data from patients and healthy individuals.
- **New genetic questions (from you or your relatives):** If new questions arise about the heredity of conditions, the material can be used again for genetic testing.

In conclusion

We hope this explanation will help you make an informed decision about the storage and use of your body material. For more information, please contact the Medical Genetics Centre at UZ Brussel at https://www.uzbrussel.be/en/web/genetics

For more information on privacy, please visit: https://www.uzbrussel.be/web/international