Print Form

REQUEST FORM CONSTITUTIONAL GENETIC ANALYSIS

version1/20210901

Label sample

CENTRE FOR MEDICAL GENETICS
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tel. +32 (0)2 477 64 79
https://www.uzbrussel.be/web/centrum-voor-medische-genetica/

Universitair Ziekenhuis Brussel

BELAC 141-MED **BELAC 141-MED accredited according to quality standard ISO15189:2012**

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

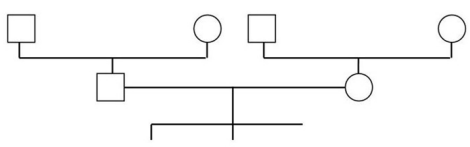
☐ Informed consent
☐ Pedigree
☐ Clinical report/checklist
☐ Genetic report (index patient)

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.

INFORMED CONSENT FOR GENETIC ANALYSIS

Universitair Ziekenhuis Brussel

CENTRE FOR MEDICAL GENETICS - UZ Brussel
Laarbeeklaan 101 - 1090 Brussel
email: cmg.laboratory@uzbrussel.be - tel. +32 (0)2 477 64 79

Identification patient *

*** Mandatory data**

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

Email address:
Phone:
National registry N°:

GENETIC TEST

1. Diagnostic genetic test

I, the undersigned, agree to perform a diagnostic genetic test on a blood sample, or other samples, of the above person for the following condition:

.....

The diagnostic genetic test that will be performed is:

☐ a limited analysis or 'targeted' analysis of gene(s)
☐ a broad analysis or 'non-targeted' genome-wide analysis**

2. Scientific research

After a diagnostic genetic test is done for a condition, some material usually remains. This material can be preserved as it can be useful for further diagnostic testing with broader genetic analyses** at a later date and/or for scientific research (see the explanation below).
With regard to the preservation of remaining samples, the genetic data, and medical data for later scientific research:

☐ I agree
☐ I do not agree

**Broad genetic analysis:
Broad genetic analyses can lead to an incidental and/or secondary discovery of genetic results unrelated to the condition for which the test was performed. I realize that such results can have implications for myself and my family. I would like to be informed about genetic results that present an increased risk for diseases for which:

☐ appropriate follow-up, prevention, or treatment is available (such as a risk for cancer, heart disease)
☐ no prevention or treatments exist (such as for dementia; NB only adult mentally competent persons may choose this option)

With regard to the storage and exchange of data/ samples as part of the diagnostic process and scientific research, I understand that:

- the exchange of medical and genetic data between experts is important to improve knowledge of genetic diseases.
- this exchange can be done in the context of diagnostic testing and/or scientific projects approved by the relevant Ethics Committee.
- the exchange of data may lead to improved diagnosis for myself or others, improved healthcare, improved prevention, improved therapeutic means; and may be published in scientific journals, or presented at scientific meetings.
- my samples, genetic data, and relevant medical data are labelled with a code (see explanation on the next page).
- my encoded genetic samples can be used as control material for the general improvement or development of tests.
- genetic and relevant medical data can be re-analyzed in the context of diagnostic tests that are available at a later stage and/or within approved research projects, without me being informed in advance.
- the knowledge and possibilities for analysis and interpretation of genetic research will increase in the future and re-analysis can reveal a (new) diagnosis. There is currently no systematic re-analysis of data.
- if my health insurance does not reimburse the costs for the original genetic test, these will be invoiced to me in full.
- I reserve the right to change my consent at any time, for one or more of the various points described. The withdrawal of consent will not adversely affect my general medical treatment (unrelated to the genetic test for which this consent was given). I understand that my withdrawal cannot be applied to the results and data collected before my request for withdrawal.
- my participation is voluntary and will not be linked to financial benefits.

☐ I agree with the above
☐ I do not agree with the above

To be completed by patient or representative*

I confirm that I am well informed about the objectives and nature of the analyses related to my condition. I received the necessary information from the healthcare provider and/or I read the corresponding information leaflet. I have had the time and opportunity to ask questions and I am satisfied with the answers and supplemented explanations.

Date:
Signature*:

If representative
Relation to patient
Name:
First name:

To be completed by healthcare provider *

I hereby confirm that I have informed the undersigned patient and answered questions in the best possible way with regard to the possible results, limitations and options for the test(s) mentionned above.

Date:
Signature*:

Stamp
healthcare provider

Name:
First name:

INFORMED CONSENT FOR GENETIC ANALYSIS (continued)

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EXPLANATION ON STORAGE AND USE OF SAMPLES

After a diagnostic genetic test is done, a part of the material remains for which there is no immediate purpose. This material could be destroyed, but often it is useful not to do so. In some cases, it can be used in a meaningful way. The following four possibilities are considered:

1) a different diagnostic test within the scope of your original question;

2) scientific research within the scope of your original question;

3) genetic research of a general nature, with which you mainly help other people;

4) you and/or your descendants have a new question or condition that requires genetic testing.

Explanation
It is possible that a different diagnostic test is possible at a later date, for a condition that affects you or your family (1). Moreover, scientific research could be carried out in order to search for more understanding on this condition (2). The material that was previously obtained from you can be used for these tests. This material, and any encrypted medical data, will then be used further and examined at a national or international level. For research into rare diseases, such an approach can speed up the identification of an explanation for the condition. When performed, your data will always be encrypted in order to fully protect the privacy of you and your genetic and medical information.
Body material is often valuable for developing new scientific knowledge, or for testing diagnostic devices in the laboratory (3). This scientific knowledge is usually not directly applicable in practice at the beginning, but can become important for patients at a later date. A great deal of knowledge that is now used daily by doctors in patient care has arisen from such scientific research, of which the practical significance was initially not entirely clear.

Examples of further use
1) and 2) After diagnostic genetic testing the remaining material is stored after use but identified via a code. This means that your personal data will be replaced by a random number. The list that indicates which number (code) belongs to which patient is stored by an administrator in a safe place. The people who use the material only see the random number (code) associated with the material. The code can be traced back to your personal data if a researcher - sometimes years later -finds a genetic change in a coded sample, which explains your original question or may be of interest to your state of health. An example is an inherited predisposition to cancer or to heart disease, for which prevention, treatment or surveillance options are available. The likelihood of finding such a genetic change is usually small. There is also a chance that we may find a genetic change that could affect your treatment, such as an adjustment in your medication. The researcher who makes such a discovery passes on the code number to the administrator who can link the code to the name of the patient and to the name of the practitioner/doctor with whom that patient has been in contact. Subsequently, an assessment is made on whether the genetic change is indeed important for you and your health. This assessment is done in consultation with an independent committee of doctors and other experts, which helps to decide whether the genetic change should be linked back to you. If so, you will be contacted by your treating physician to inform you of the genetic change. This finding will then have to be confirmed with a new independent test.
3) Your material can also be used for scientific research that only provides general knowledge and can not be individually applied. An example of this is when your material is used as a control sample for a test, which has nothing to do with the condition for which you had genetic testing in the first place. Samples and encrypted data from groups of patients are then compared with those of other groups of patients or healthy individuals. The results of such scientific research are usually not reported back to you. In the case that there would be feedback, it could be many years later.
4) After your original question has been answered, you and/or your descendants may have a new question concerning genetics. In that case, your sample can be used for a new genetic test.

In conclusion
We hope to have given you sufficient information to make an informed decision about the storage and use of your samples and of your medical and genetic data. For more information, you may wish to contact the Medical Genetics Centre of UZ Brussel.https://www.uzbrussel.be/web/genetics
More information on privacy may be found at: https://www.uzbrussel.be/web/neem-zelf-uw-zorg-in-handen-/patiëntenrechten

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