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Test request form
Rare genetic diseases

INTERNATIONAL DIVISION
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Client no.

Find all the information on single gene tests, gene panels and Whole Exome Sequencing offered by Eurofins Biomnis in the field of rare genetic diseases on :
wwww.eurofins-biomnis.com > Services section > Genetic test guide.

CUSTOMISE YOUR GENETIC TEST ORDER IN 2 STEPS

1 Type of test

2 "Plus" option (Del/Dup CNV)

☐ Single gene *(specify the gene name):*		☐
☐ Gene panel *(specify the disease)*		☐
☐ Customised gene panel *(specify the gene list)*		☐
☐ Exome : please use the specific request form «Exome sequencing» (Ref. B34-INTGB)		☐
☐ Other *(specify)*:		☐

REFERRING PHYSICIAN

Last name: ..... First name: .....
Address: .....
Postcode: ⌊_⌊_⌊_⌊_⌊_⌋ City: ..... Counrty: .....
Email:.....
Tel.: ⌊_⌊_⌋⌊_⌊_⌋⌊_⌊_⌋⌊_⌊_⌋⌊_⌊_⌋ Fax: ⌊_⌊_⌋⌊_⌊_⌋⌊_⌊_⌋⌊_⌊_⌋⌊_⌊_⌋

Physician's stamp

PATIENT (INDEX CASE)

Last name: ..... First name: .....
Birth name: .....
Date of birth: ⌊_⌊_⌋⌊_⌊_⌋⌊_⌊_⌊_⌊_⌋ Gender: ☐ F ☐ M
Address: .....
Postcode: ⌊_⌊_⌊_⌊_⌊_⌋ City: ..... Counrty: .....

SAMPLE

Type: ☐ EDTA Whole blood ☐ DNA sample **Date of sampling:** ⌊_⌊_⌋⌊_⌊_⌋⌊_⌊_⌋

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OTHER CLINICAL SYMPTOM(S)

Imaging and cerebral malformation

- ☐ Agenesis of the corpus callosum
- ☐ Cerebral atrophy
- ☐ Cortical dysplasia
- ☐ Heterotopia
- ☐ Hydrocephalus
- ☐ Leukodystrophy
- ☐ Other:.....

Other craniofacial and sensory impairments

- ☐ Physical facial features
  (please specify: .....)
- ☐ Vision impairment
  - ☐ severe refractive error
  - ☐ optic atrophy
  - ☐ pigmentary retinopathy
- ☐ Other ophthalmological impairments:
  - ☐ cataracts (age: .....)
  - ☐ coloboma
  - ☐ glaucoma (age: .....)
- ☐ Hearing impairment:
  age started: .....
  type: .....
- ☐ Malformation of the ear
- ☐ Unilateral or bilateral cleft soft palate / lip and palate
- ☐ Dental abnormalities
- ☐ Other:.....

Skin and skin appendage symptoms

- ☐ Pigmentation abnormalities
  (please specify: .....)
- ☐ Nail abnormalities
  (please specify: .....)
- ☐ Ichthyosis
- ☐ Vascular/capillary abnormalities
  (please specify: .....)

Skeletal symptoms

- ☐ Upper limb abnormalities
- ☐ Lower limb abnormalities
- ☐ Vertebral abnormalities
- ☐ Scoliosis
- ☐ Polydactylism
- ☐ Syndactylia
- ☐ Other:.....

Metabolism

- ☐ Abnormal CPK levels
- ☐ High alanine levels
- ☐ Low carnitine levels
- ☐ Ketosis
- ☐ Lactose acidosis (CSF)
- ☐ Organic aciduria
- ☐ High pyruvate levels
- ☐ Other:.....

Digestive system

- ☐ Imperforate anus/anal stenosis/ displacement of the anus
- ☐ Constipation
- ☐ Diarrhoea
- ☐ Oesophageal atresia
- ☐ Liver failure
- ☐ Hepatomegaly
- ☐ Hirschsprung's disease
- ☐ Umbilical hernia
- ☐ Pyloric stenosis
- ☐ Vomiting
- ☐ Other:.....

Growth and endocrine disorder

- ☐ Type I diabetes
- ☐ Type II diabetes
- ☐ Hyperparathyroidism
- ☐ Hyperthyroidism
- ☐ Hypoparathyroidism
- ☐ Hypothyroidism
- ☐ Obesity
- ☐ Age: ..... years
- ☐ Birth weight (g, SD or %):
  .....
- ☐ Birth length (cm, SD or %):
  .....
- ☐ Head circumference (cm, SD or %):
  .....
- ☐ Other:.....

Genito-urinary symptoms

- ☐ Abnormal kidney morphology
- ☐ Kidney cysts
  (please specify: .....)
- ☐ Hydronephrosis
- ☐ Kidney agenesia/hypoplasia
- ☐ Tubulopathy
  (please specify: .....)
- ☐ Disorders of sex development
- ☐ Other:.....

Cardiovascular impairment

- ☐ Malformation
  (please specify: .....)
- ☐ Muscle impairment (hypertrophic cardiomyopathy, dilated cardiomyopathy)
- ☐ Arrhythmia
- ☐ Conduction disorders
- ☐ Hypertension
- ☐ Other:.....

GENETIC TESTS ALREADY PERFORMED

☐ YES ☐ NO

Please specify the type of analysis / genes and results obtained

.....

.....

.....

.....

.....

.....

Note: Some types of genetic abnormalities are not detectable such as repeated regions and methylation abnormalities. Mosaics are not sought after. Regions with strong homologies are eliminated at the time of alignment (multiple match) and variants potentially present in these regions are not detectable.

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CLINICAL SYMPTOM(S)

Family tree

Geographical origin*: .....
*(*The frequency and distribution of genetic mutations differs according to the ethnic/geographical origins of the patient)*
Consanguinity: ☐ YES *(please indicate on the family tree)* ☐ NO

CLINICAL SYMPTOM(S)

Please provide a detailed clinical report for the patient and his/her medical history.
Detailed clinical information is essential for accurately interpreting results.

.....
.....
.....
.....
.....
.....

Current weight: ..... kg Current height: ..... cm Head circumference: ..... cm

OTHER CLINICAL SYMPTOM(S)

Perinatal case history

- ☐ Abnormalities during pregnancy
- ☐ Analyses conducted during pregnancy
- ☐ Foetal-placental hydrops
- ☐ Amniotic fluid abnormality
  - ☐ Oligohydramnios
  - ☐ Polyhydramnios
- ☐ Malformations (system, to be described in later sections): .....
- ☐ Gestational age
- ☐ Birth weight (g, SD or %): .....
- ☐ Birth length (cm, SD or %): .....
- ☐ Head circumference (cm, SD or %): .....
- ☐ Other:.....

Neurological and psychiatric symptoms

- ☐ Ataxia
- ☐ Spasticity
- ☐ Other pyramidal symptoms
- ☐ Chorea
- ☐ Dystonia
- ☐ Other extrapyramidal symptoms
- ☐ Muscle weakness
- ☐ Exercise intolerance
- ☐ Migraine
- ☐ Lethargy
- ☐ Oculomotor dysfunction
- ☐ Epilepsy
- ☐ Other:.....

Neurodevelopmental and behavioural symptoms

- ☐ Hypotonia
- ☐ Motor delay (age started walking: .....)
- ☐ Delayed speech
- ☐ Global development delay / intellectual impairment
  - ☐ mild
  - ☐ moderate
  - ☐ serious
  - ☐ severe
- ☐ Psychomotor regression
- ☐ Autism spectrum disorders or autistic traits
- ☐ Stereotypies
- ☐ Attention deficit disorder with/ without hyperactivity
- ☐ Other behavioural disorders and associated psychiatric disorders: *(please specify: .....)*

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Declaration of genetic consultation and informed consent

INTERNATIONAL DIVISION
Tel: 04 72 80 23 85 • Fax: 04 72 80 73 56 • Email: international@biomnis.eurofinseu.com

CONSENT FOR THE GENETIC CHARACTERISTICS TEST ON AN INDIVIDUAL AND THE PRESERVATION OF SAMPLES.

Patient information Last name: .....First name: .....
Date of birth: |__|__| |__|__| |__|__|__|__|

Legal representative(s) information
(as appropriate) Last name: .....First name: .....
(as appropriate) Last name: .....First name: .....

I, the undersigned, declare that I have been informed by:

☐ Dr .....

☐ Genetic Counsellor ..... under the responsibility of Dr .....
and on their behalf

about the genetic characteristics test which will conducted on a sample/samples taken from:

☐ Myself
☐ My child or an adult under my guardianship

For: *(mandatory statement of the name of the pathology or name of the test conducted according to an aetiological, predictive or healthy carrier diagnosis)*

.....

ACKNOWLEDGEMENT OF THE FOLLOWING INFORMATION:

I declare that I have received the information needed to understand this test and its purpose.
I consent to this test being performed.

The results of the test will be provided to me and explained based on the current state of knowledge by the doctor/genetic counsellor who prescribed it as part of an individual consultation. The doctor/genetic counsellor will explain the necessary treatment methods where appropriate.

I understand that if a genetic abnormality that could be responsible for a predisposition or a serious affliction is identified, I must allow this information to be passed on to the rest of my/their family. I have been warned that remaining silent could pose a risk to them and their descendants, where preventive measures, including genetic counselling or treatment, could be proposed. I can either share this genetic information with members of my/their family myself, or permit the prescribing physician to do so.

I authorise, in compliance with medical confidentiality: The transmission of information from my/their medical file to the doctors involved with this test.

I acknowledge that my/the personal data relevant for making a diagnosis and the results report for my/their test will be kept, in paper form or in a digital database, by the prescribing physician and the medical biology laboratory authorised to conduct this test, in accordance with the regulations in force.

I have been informed that, in accordance with the current laws, my/their sample will be destroyed once the legal retention period has expired or, unless requested otherwise by myself in writing sent to the Eurofins Biomnis administrative office, used and transferred, anonymously and according to medical confidentiality, for scientific or quality control purposes.

In addition, cross out any of the following paragraphs that you disagree with:

* I wish to be informed of the results of the test conducted.
* Genetic information not directly linked to my/their pathology but which may have an impact on my/their care and/or treatment or that of my/their relatives may be disclosed. I wish for this information to be disclosed to me:
  ☐ YES ☐ NO ☐ Not applicable
* I agree for the transmission and use of my/their results for the genetic analysis of other members of my family who may wish for a consultation.
* I agree for a sample of a biological material from me/them to be kept and used at a later date to continue the investigation as part of this diagnostic approach, according to developments in medical knowledge.

Signed in..... ,
On |__|__| |__|__| |__|__|

Signature of the patient or legal representative(s) for a minor or adult under guardianship

REFERRING PHYSICIAN DECLARATION OF CONSULTATION**

I certify that I have informed the patient named above or their legal representative of the characteristics of the disease being tested for, the means for identifying it, the reliability of the analyses, options for prevention and treatment and how the disease in question can be transmitted genetically, along with its potential consequences for other members of the family.
I certify that I have received the consent of the patient named above or their legal representative according to the conditions laid down in the regulations in force.

Signed in ..... on .....

Signature and stamp

***REMINDER OF THE REGULATIONS**

***The referring physician** must keep:*

- *The written consent*
- *Duplicates of the prescription and declaration*
- *The reports of medical biology analyses with discussion and which have been signed (Art. R1131-5).*

***The authorised laboratory** conducting the tests must:*

- *Ensure that there is a prescription, referring physician declaration and written consent from the patient*
- *Send, to the referring physician, who alone is authorised to communicate the results to the individual concerned, the medical biology analysis report with discussion and which is signed by an approved practitioner*
- *Send, where appropriate, to the laboratory that transmitted the sample and was involved in the analysis, the medical biology analysis report with discussion and which is signed by an approved practitioner*

Law no. 2011-814 of 7 July 2011 *on bioethics*

Order of 27 May 2013 *defining the rules of good practice applicable to the genetic characteristics test on an individual for medical purposes*

Decree no. 2013-527 of 20 June 2013 *on the conditions for informing biological relative in relation to genetic characteristics tests for medical purposes*

Decree no. 2008-321 of 4 April 2008 *on genetic characteristics tests on an individual or their identification via genetic fingerprinting for medical purposes.*

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