

# Test request form Constitutional molecular genetics

**INTERNATIONAL DIVISION**

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Customer number:

Date :        
EDTA whole blood sample

Laboratory's stamp  
or bar code sticker

**PRESCRIBING CLINICIAN**

First name(s): ..... Surname : .....  
Address: .....  
Post code:     City : ..... Country: .....  
Tel.:       Fax:

**PATIENT**

First name(s): ..... Surname : .....  
Date of birth\* :       Gender:  F  M  
Address: .....  
Post code:     City: .....  
Country: ..... Tel.:        
Geographical origin\*\* :  Europe/North Africa  Sub-Saharan Africa and the Caribbean  
 Asia  Other (e.g. mixed-race): .....

\* If the patient is a minor, consent must be given by the holders of parental authority.  
\*\* This information is essential only for the tests marked [1] below.

**CLINICAL SIGNS**

**SAMPLE TYPE - This section must be completed**

EDTA whole blood  Other - *please specify* : .....

**MOLECULAR GENETICS TEST REQUEST FORM**

**INDICATIONS: This information must be provided (Biomnis group code)**

- ABCB1 (identification of variants rs2032583 et rs2235015) (ABCB1)
- Alpha 1-antitrypsin (S and Z variants) (A1BM)
- Angelman syndrome (Postnatal: SNRPS / Prenatal : SNRPL)
- Uniparental disomy (DUPRE)
- Factor II (mutation g.20210G>A) (F2M)
- Factor V Leiden (mutation p.Arg506Glu) (F5L)
- Familial Mediterranean Fever (FMF) or periodic disease (by New Generation Sequencing method)<sup>[1]</sup> (FMF) : *please attach the completed clinical information form which is available on [www.eurofins-biomnis.com](http://www.eurofins-biomnis.com)*
- IL-28B genotyping (IL28B)
- RHD genotyping (BMGR1)
- Gilbert syndrome (polymorphism UGT1A1\*28) (GILB)
- Haemochromatosis: mutation p.Cys282Tyr (HMC)
- Haemochromatosis: mutation p.His63Asp (H63D)
- Haemochromatosis: mutation p.Ser65Cys (S65C)
- HLA class I (loci A, B)<sup>[1]</sup> (HLA1) :  A\*29  B\*27  B\*51  B\*57
- HLA classe II (loci DQ, DR)<sup>[1]</sup> (HLA2) :  DR4  DQ2  DQ8  DQB1\*0602
- HLA-B\*27<sup>[1]</sup> (B27)
- Lactose intolerance (LCT)
- Y chromosome microdeletions (loci AZFa, AZFb and AZFc) (DELY)
- MTHFR (Methylene Tetrahydrofolate Reductase - thermolabile variant, mutation c.677C>T) (MTHFR)
- Cystic fibrosis (CFTR, screening of most frequent mutations)<sup>[1]</sup> (MUCO) : *specific request form required, please see [www.eurofins-biomnis.com](http://www.eurofins-biomnis.com)*
- Cystic fibrosis, whole gene - next generation sequencing<sup>[1]</sup> (MUCON) : *specific request form required, please see [www.eurofins-biomnis.com](http://www.eurofins-biomnis.com)*
- Prader-Willi syndrome (Postnatal: SNRPS / Prenatal: SNRPL)
- Fragile X syndrome (Postnatal: XFRA / Prenatal: XFRAP)
- Other - *please specify* : .....

[1] The geographical origin of the patient must be specified for these tests-of "Patient" section

**CONSENT PRIOR TO THE CARRYING OUT OF GENETIC EXAMINATIONS OF AN INDIVIDUAL**

(Pursuant to articles R. 1131-4 and R. 1131-5 of the French Public Health Code).

I, the undersigned .....  
born on       hereby  
declare that I had a consultation with Dr:

.....  
where information on the genetic tests to be performed for the reasons listed below was provided:

- To confirm or otherwise the diagnosis of a genetic disease in relation to my symptoms,
- To confirm or otherwise the pre-symptomatic diagnosis of a genetic disease,
- To identify the healthy carrier status of an individual (heterozygote screen or chromosomal rearrangement),
- To assess my genetic susceptibility of being afflicted with a genetic disease or undertaking a medical treatment.

As such, I consent to:

- sample(s) being collected from me.
- sample(s) being collected from my child (for minors) or an adult under guardianship.
- sample(s) being collected from my foetus.

I have been informed that the results of these genetic tests will be communicated to me by the aforementioned Doctor during an individual consultation. If the exam reveals any results other than those specified on the original request, the aforementioned Doctor will determine the appropriate steps to be taken during the individual consultation.

Should any of the sample remain unused following examination:

- I consent to this sample being used, if needs be, for scientific research purposes. In this case, all personal medical data will be protected by it being made totally anonymous. Consequently, I am conscious that the scientific studies performed will not provide me with any advantage or prejudice.

Signed in (city) .....  
on

Patient's signature, signature of the holders of the parental authority of the child or the guardian of the adult under guardianship:

**DECLARATION OF MEDICAL CONSULTATION**

(French Decree n° 2008-321 dated 4 April 2008 - French Decree dated 27 May 2013).

I, the undersigned .....  
R.1131-5 of the French Public Health Code, hereby certify that the patient mentioned above was received for a consultation today where information on the characteristics of the disease to be screened, the methods used to detect it and details on the possibilities of prevention and treatment were provided.

Signed in (city) .....  
on

Physician's signature: