

## INFORMATION FOR GENETIC CHARACTERS INVESTIGATION

Genetic and pharmacogenetic predisposition

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AUTHORIZED LABORATORY FOR PERFORMING GENETIC CHARACTERS INVESTIGATION IN HUMAN

PATIENT	LEGAL ASPECTS: mandatory documents →
Name: .....	<input type="checkbox"/> Certificate stating that the prescribing physician collected the informed consent from the patient <u>OR</u> , if not available, the patient's consent <input type="checkbox"/> Name, address and phone number of the prescribing physician to whom results must be sent.
First name: .....	
Date of birth : .....	

### THROMBOPHILIA

#### Context of request

- Exploration of an index case (the patient proved to have a venous thrombosis history or a placental vascular pathology)  
 Exploration of a related patient (family investigation)

**Required Test:**    **FV Leiden (R506Q)**    **FII Prothrombin mutation 20210 G>A**    **Thermolabile *MTHFR* variant (677 C>T)**

### HEMOCHROMATOSIS

#### Context of request

- Patient with clinical, biological, radiological or histological signs suggesting a haemochromatosis  
 Familial investigation (1<sup>er</sup> degree related with homozygote C282Y mutation)

**Required test:**    ***HFE1* C282Y** (p.Cys282Tyr)    ***HFE1* H63D** (p.His63Asp)    ***HFE1* S65C** (p.Ser65Cys)  
 **Ferroportin\***    **Transferrin receptor 2\***    **Other\*:** .....   \* transmitted test

### HLA

#### Context of request (specify the pathology) :

- Auto-immune disease: .....  
 HIV+ patient: see PHARMACOGENETIC  
 Other : .....

#### Antigen or allele of interest:

HLA class I :    HLA A29    HLA B27    HLA B51(5)    other: .....  
 HLA class II :    DR1    DR4    DR3    DR2    DR5    DR7    DQ2    DQ8    DQB1\*0602    other.....

**Required test:**    **HLA Class I**    **HLA Class II**    **HLA-B\*27** (genotype)

### PHARMACOGENETIC

#### Context of request

- Pre-therapy check-up    Toxicity etiology    Therapeutic failure etiology  
 Other indication (specify): .....

**Molecule implicated:**    Abacavir    Peginterferon    Ribavirine    Irinotecan    Other (specify) : .....

**Required test:**    **HLA-B\*57:01** (genotype)    ***UGT1A1*** (genotype)    ***IL28B/IFNL4*** (genotype)    ***ITPA*** (genotype)  
 **m.1555A>G mitochondrial mutation**    Other (specify): .....

### METABOLIC AND NEUROLOGIC DISORDERS

#### Context of request

- Neurodegenerative disease    Dyslipoproteinemia    Clinical suspicion: digestive disorders    Other: .....

**Test required :**    ***APOE*** (genotype) )    Primary lactase deficiency (***LCT***)