BLOODGENETICS
Diagnostics in Inherited Blood Diseases
Edificio Synlab
Verge de Guadalupe, 18, 08950 Esplugues de Llobregat, Spain
Tel. (+34) 636147238
info@bloodgenetics.com
www.bloodgenetics.com

PATIENT INFORMATION DOCUMENT (V.19-2022)

INFORMED CONSENT FOR GENETIC AND/OR BIOCHEMICAL DIAGNOSIS OF HEMATOLOGICAL HEREDITARY DISEASES AND DISEASES OF IRON METABOLISM

This document is intended to request **your written permission** to perform the genetic and / or biochemical diagnosis requested.
It is important that you read carefully this informed consent sheet, you understand its content and purpose and do all the questions you may have.

BLOODGENETICS (BG)

We carry out genetic studies and research biochemical measures relevant to the diagnosis and / or management of hereditary hematological diseases, related diseases and diseases of iron metabolism.

BG detects by genetic studies the presence or absence (yes / no) of pathogenic mutations in genes that cause the aforementioned diseases. A pathogenic mutation is any change, deletion, inversion or insertion of one or more nucleotides that alter any of the known functional regions of the studied gene product. The partial or total loss of the gene under study is also considered a pathogenic mutation.

Aim of the genetic studies

The purpose of this study is to better understand the causes of the deregulation of iron metabolism in the body and other erythropathologies diseases you may suffer in order to make an early diagnosis and avoid further complications.

Biological samples and associated information

To participate in the study you must donate a **biological sample** (small blood and/or urine sample and/or oral swab and/or aspirate/bone marrow biopsy). From this sample we isolate DNA for genotyping or sequence several genes whose alterations are suspected to be related to the disease. From the blood sample and / or oral swab and / or aspirate / bone marrow biopsy, the DNA will be isolated. All this to genotype or to sequence various genes whose alterations are suspected to be related to the disease it presents. From the urine sample, there will be biochemical measurement studies of compounds and other analyses of interest carried out. The collection of data and samples will be done by professionals of the Hospital.

The biological samples and associated information are kept under the conditions and guarantees of quality and safety required by current legislation and the codes of conduct approved by the Hospital's Clinical Research Ethics Committee (CEIC).
With these samples we will perform clinical, genetics and/or molecular biology studies and from them we may obtain information about your health and your relatives. All data will be protected (see section on data protection and confidentiality). This diagnosis is performed to complement clinical care that you are already routinely administered.

Discomfort and possible risks

Sampling will be performed by qualified professionals of the Hospital.
Sampling can cause a burning sensation at the point where the needle is inserted in the skin and cause a small bruise or a minor infection that goes away in a few days. More rarely dizziness can occur at the time of blood collection.
Aspirate/biopsy bone marrow sampling is a test whose welfare risks you will be explicitly informed.
Swab and urine tests are non-invasive procedures and risk free.

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Limitations of genetic testing

Due to technological or knowledge limitations is possible that the mutation responsible for the disease is not identified. This test does not provide information regarding other diseases not considered in this diagnosis test which may have a hereditary genetic component.

Benefits of genetic test

A positive genetic test result means that the mutation responsible for the disease has been identified and this can reduce the uncertainty about your own risk and your relatives. A negative result will be informative when knowing a specific mutation of a gene in your family, you are not a carrier of this mutation.

This test also offers benefits to the society since it contributes to a better understanding of the causes of these diseases, improve our ability to diagnose them and predict prognosis and eventually allow us to develop preventive procedures and/or better treatments.

Data protection and confidentiality

The purpose of the registry is to guarantee the quality, security and traceability of the data and biological samples stored as well as the associated procedures, complying at all times with the duty of secrecy and confidentiality, in accordance with current legislation on data protection for personal character.

The identification of the biological samples will undergo a coding process. Each sample is assigned an identification code separated from personal data. Only authorized personnel may relate their identity to the aforementioned codes.

If required for research projects in which the BG company participates, the samples and personal data will be kept in the **collection of biological samples** of human origin of BG for research purposes (responsible Dr. Cristian Tornador Antolín) registered with number C .0005855 in the Register of sample collections dependent on the Carlos III Health Institute.

Voluntary participation and withdrawal of consent

Your participation in this study is completely voluntary and it is possible to withdraw from it at any time, without giving any reason. In the case of withdrawal, this does not entail any change in the doctor-patient relationship or prejudice in the diagnosis, treatment and / or monitoring.

Your transfer of biological samples is free. You will not get now or in the future any economic benefit.
If you would like to withdraw your consent in the future or ask for the anonymization of the samples you must request it in writing by contacting Dr. Mayka Sanchez de BG at the following address: BLOODGENETICS. Edificio Synlab, C/Verge de Guadalupe, 18, 08950 Esplugues de Llobregat, Barcelona, Spain. The effects of this withdrawal or anonymization could not be extended to the diagnoses that had already been carried out.

Information about the genetic and/or research biochemical results

The results of the requested diagnostic tests will be sent to the doctor who requested them. He will inform you of the results except in case of cancellation or anonymization.
Regarding the results of the genetic diagnosis, relevant information could be obtained not only for you but also for your family. In this case, it will be up to you to decide whether or not you wish to inform your relatives. If you do not wish to inform them, please note that the law states that if the obtained information is necessary to avoid serious harm to the health of your biological relatives, a welfare expert committee will study the case and decide whether to inform the affected parties or their legal representatives.

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Incidental findings

During the genetic study, unexpected genetic variants which have not been requested and not related to the main medical reason for which this test is prescribed may be found. These incidental findings are limited to only those genes that BG offers and are also listed on the ACMG recommendation list. These findings may have potential health implications but are not related to the disease symptoms for which the test was requested. These changes could be relevant to initiate a monitoring in order to obtain an early diagnosis in the corresponding hospital unit. If you do NOT wish to be informed of these incidental findings, you must mark it on this informed consent.

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Name and surname of the donor.....
ID number or passport ..... Age.....

Reporting person (medical doctor, nurse) and Center/Hospital's name
..... ID number or passport .....

If you have understood the information given to you, you have resolved any questions you might have and you decide to perform this diagnostic study requested by your medical doctor in the terms explained above, please **read and sign** this form below.

The undersigned authorizes BG to include biological material in the collections record of biological samples of human origin for the diagnosis of hereditary hematological diseases, related diseases and diseases of iron metabolism, which are subject to the approval of the competent Ethics Committee on Biological Research. This authorization is granted after having been verbally informed and having read the enclosed information.

Please remember that your participation in this study is voluntary; you can withdraw from this study at any time, without explanation and with no impact on your health care.

I freely give my approval for this requested genetic and/or biochemical study and authorize donated biological material and associated clinical information to be used for diagnosis in the terms included in the Patient Information Document.

○ PLEASE CHECK THIS BOX IF YOU DO **NOT** WISH TO BE INFORMED OF POSSIBLE INCIDENTAL FINDINGS

PATIENT	REPORTING PERSON (medical doctor, nurse)
Signature	Signature
LEGAL REPRESENTATIVE IN CASE THAT THE PATIENT IS MINOR	
Please, check as appropriate ☐ Father ☐ Mother ☐ Guardian or legal representative	
Signature	

....., (city).....20..... (date)

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DIAGNOSIS INFORMED CONSENT REVOCATION

If you have understood the information given to you, you have resolved any questions you may have and decide to **revoke your participation in this diagnosis** requested by your doctor, please read and sign this form

Donor:

I, ... with ID Number/Passport..... nullify the informed consent.
Date: ..... (day).....(month) .....(year) and I do not wish to continue the voluntary donation to this diagnostic test, which I end today.

☐ I REQUEST THE ELIMINATION OF THE SAMPLE ONLY
☐ I REQUEST THE ELIMINATION OF PERSONAL DATA ONLY. The sample will be irreversible anonymised and may be used in research projects after having requested a new informed consent for research.
☐ I REQUEST THE ELIMINATION OF MY DATA AND SAMPLE

Signature:

....., (city)..... .....20..... (date)

Guardian or legal representative of the donor:

I, ... with ID Number/Passport ..... nullify the informed consent.
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PATIENT	REPORTING PERSON (medical doctor, nurse)
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☐ Father ☐ Mother ☐ Guardian or legal representative

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