

Informed Consent Form - Immunogenetics

1 Patient

Informed consent is required for genetic testing. The patient (or parent or guardian in the case of minors under the age of 18 or adults lacking legal capacity) must sign the attached consent form. If the samples are anonymous, we will accept a statement from the physician responsible for the patient indicating that an appropriate informed consent has been obtained (section "Statement of the existence of informed consent").

Patient's full name

2 Genetic study requested

NGS panels:

<input type="checkbox"/> Disorders of the Immune System	458 genes		
<input type="checkbox"/> Primary Immunodeficiencies	301 genes		
<input type="checkbox"/> Primary Antibody Deficiencies	41 genes	<input type="checkbox"/> Agammaglobulinemia	10 genes
<input type="checkbox"/> Common Variable Immunodeficiency	25 genes	<input type="checkbox"/> Hyper-IgM Syndrome	8 genes
<input type="checkbox"/> Combined Immunodeficiencies	37 genes	<input type="checkbox"/> Bare Lymphocyte Syndrome	13 genes
<input type="checkbox"/> Severe Combined Immunodeficiency	19 genes		
<input type="checkbox"/> T(-)B(+) SCID	11 genes	<input type="checkbox"/> T(-)B(-) SCID	8 genes
<input type="checkbox"/> Syndromes with Combined Immunodeficiency	74 genes	<input type="checkbox"/> Hyper-IgE Syndrome	14 genes
<input type="checkbox"/> Dyskeratosis Congenita	16 genes	<input type="checkbox"/> Ataxia telangiectasia	1 gene
<input type="checkbox"/> Defects in Intrinsic & Innate Immunity	67 genes	<input type="checkbox"/> Fungal Infections, Predisposition	15 genes
<input type="checkbox"/> Viral Infections, Predisposition	21 genes	<input type="checkbox"/> Invasive Bacterial Infections, Predisposition	6 genes
<input type="checkbox"/> Mendelian Susceptibility to Mycobacterial Disease	17 genes	<input type="checkbox"/> Cystic fibrosis	1 gene
<input type="checkbox"/> Phagocyte Defects, Congenital	44 genes	<input type="checkbox"/> Neutropenia, Non-Syndromic	7 genes
<input type="checkbox"/> Neutropenia, Syndromic	21 genes	<input type="checkbox"/> Chronic Granulomatous Disease	6 genes
<input type="checkbox"/> Complement System Deficiencies	38 genes	<input type="checkbox"/> Systemic Lupus Erythematosus (SLE)-like Syndrome	8 genes
<input type="checkbox"/> Atypical Haemolytic Uremic Syndrome	13 genes	<input type="checkbox"/> Pyogenic Infections, Recurrent	6 genes
<input type="checkbox"/> Disseminated Neisserial Infections	9 genes	<input type="checkbox"/> Hereditary Angioedema	2 genes

The personal data provided in this form are subject to the current data protection regulations, specifically to Organic Law 3/2018, of December 5, on the Protection of Personal Data and Guarantee of Digital Rights ("LOPDGDD") and to Law 14/2007, of 3 July, on Biomedical Research. The data you provide will be included in files whose responsible is Health in Code. The purpose is the analysis and diagnosis of genetic diseases. Likewise, the data categories are the ones reflected in this form, along with the results obtained. Your personal data will be processed exclusively for the aforementioned purposes. This data processing is made legitimate by the express consent provided by accepting these terms. Your data will be retained for the whole duration of the relationship established with the entity and while the data fulfil their purposes for this service or until you decide to exercise your cancellation or suppression rights. Said data will not be transferred to third parties without a corresponding prior consent, or in cases other than those expressly defined in data protection legislation. You are hereby informed that you may exercise your rights to access, rectification, cancellation, and objection, as well as to restriction of data processing and to data portability by contacting Health in Code through written communication addressed to Edificio O Fortín, As Xubias, s/n., Campus de Oza, 15006 A Coruña, España, with the subject: "Data Protection", including a copy of your national ID card or passport. You also have the right to file your claim to the Spanish Data Protection Agency (Agencia Española de Protección de Datos).

- | | | | |
|---|-----------|---|----------|
| <input type="checkbox"/> Immune Dysregulation Diseases | 247 genes | | |
| <hr/> | | | |
| <input type="checkbox"/> Autoimmune Diseases | 156 genes | <input type="checkbox"/> Autoimmune Lymphoproliferative Syndrome | 21 genes |
| <input type="checkbox"/> Systemic Lupus Erythematosus | 69 genes | <input type="checkbox"/> Autoimmune Enteropathy | 18 genes |
| <input type="checkbox"/> Autoimmune Enteropathy | 52 genes | <input type="checkbox"/> Autoimmune Polyendocrinopathy | 13 genes |
| <hr/> | | | |
| <input type="checkbox"/> Autoinflammatory Diseases | 145 genes | <input type="checkbox"/> Autoinflammatory Diseases with Recurrent Fever | 12 genes |
| <input type="checkbox"/> Behçet's Disease | 27 genes | <input type="checkbox"/> Aicardi-Goutières Syndrome | 7 genes |
| <input type="checkbox"/> Inflammatory Bowel Disease | 26 genes | | |
| <hr/> | | | |
| <input type="checkbox"/> Hemophagocytic Lymphohistiocytosis | 29 genes | <input type="checkbox"/> HLH with Epstein Barr Virus Susceptibility | 13 genes |

Complementary services:

- Familial study** (please identify the index case if tested at Health in Code) Details of the index case:
- Gene/variant Gene/variant
- Single-gene sequencing**
- Gene/variant: Gene/variant:
- Study extension**
- Specify the name of the new panel:
- Genetic variants report without sequencing**

Other services

- Exome**
- Sequencing + FASTQ Sequencing + FASTQ + variant annotation Sequencing + FASTQ + variant annotation + interpretation
- MLPA**
- Gene:

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3 Patient's authorization

I declare that I have been informed of, that I understand, and that I am in agreement with the type of genetic study indicated above and in which I am voluntarily participating.

I understand that I may be affected by or be a carrier of a hereditary genetic disorder, the diagnosis of which may be confirmed by a laboratory study analyzing DNA obtained from my biological samples. I hereby give my consent to have my sample sent to Health in Code S.L., a company with a level of data protection in accordance with European legislation, to carry out the indicated genetic study, as well as to the center or centers designated by it, complying with ethical considerations and current legal regulations:

Yes No

I understand that:

- Genetic disorders may be inherited by family members and that the results of my test may have implications for my own family.
- In the case of a genetic study of a mutation, the determination of the mutation is diagnostic, while non-determination does not exclude the pathology. A negative test does not exclude the possibility of having the disease (some diseases have multiple causes and it is not possible to test for all of them).
- Occasionally, there may be unusual alterations in the DNA structure of certain individuals that may yield results that are difficult to interpret, making the diagnosis difficult and even making it impossible to obtain conclusive results.
- Although the methods used to perform this diagnostic testing are extremely sensitive and specific, there is always a small chance of failure of the technique or of an interpretation error. For this reason, repeating the test or performing additional ones may be necessary in some cases, which may or may not require obtaining new samples, particularly in those cases where quality of the biological sample is suboptimal.
- Given the complexity of genetic studies based on DNA and the important implications of the results of a genetic study, I will be informed of said results by a physician or genetic expert, always with the highest confidentiality level from both medical and laboratory personnel.
- I may change my mind at any time and withdraw the authorization for the genetic study given by me in this document, thereby revoking my decision to continue with the analysis.
- The only people who will have access to the test results will be members of the Health in Code, S.L. team and health service professionals involved in patient care.
- It is possible to obtain unexpected information during the sample analysis process, and I hereby declare that I want to be informed about it:

Yes No

- It is possible that information concerning the relatives of the sample donor will be obtained. We recommend that the latter (or his/her legal representative) should be the person who shares said information. In any event, the approval of each family member will be required.

Current legislation requires [Health in Code, S.L.](#) to keep clinical documentation under conditions that ensure its proper maintenance and security for purposes of due patient care for at least five years after the assistance process has ended. I am aware and accept that a DNA aliquot will be kept in the laboratory for subsequent studies and/or confirmation tests:

Yes No

In addition, I consent to the biological sample being used by the entity Health in Code, S.L. for research purposes approved by the relevant ethics committee after the termination of the study, always maintaining the patient's anonymity.

Yes No

In which case, you are informed of:

- The purpose of the research related to the pathology whose diagnosis is intended and to other related lines of research.
- The expected benefits of the research, which will consist of a greater understanding of the pathologies studied, their development, and related population studies.
- The possibility that you will be contacted later for the purpose of collecting new data or obtaining new samples.
- The right to revoke this consent at any time and without any justification whatsoever and to decide to have the sample destroyed or anonymized.
- The obligation of Health in Code, S.L. to destroy or anonymize the sample once the research has finished and after the statutory storage period, unless authorization for longer storage has been given.
- Your right to know the genetic data obtained from the analysis of your biological samples.
- The confidentiality of the information obtained, with solely members of the Health in Code, S.L. research team having access to personal data.
- The possibility that information concerning the relatives of the sample donor may be obtained. We recommend that the latter (or his/her legal representative) be the person who shares said information. In any event, the approval of each family member will be required.

If applicable, I hereby authorize the extraction of biological samples and the genetic study of dependent minor/s in my care to be used under the terms and conditions previously described for the genetic test for the aforementioned disease.

Name of the patient or legal representative*

**If the patient is a minor or lacks legal capacity*

National Identification Number of the patient or legal representative

Signature of patient or legal representative

Date

4 Statement of the existence of informed consent

- I hereby declare that the patient identified on this request is aware of the information on said request and has signed the Informed Consent form to permit this genetic study to be carried out and that this has been included in his/her clinical record.

Physician's signature

Date