

INFORMED CONSENT FORM

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

S-202110258 Disorders of the Immune System [513 genes]

Primary Immunodeficiencies (PID)

S-202110259 Primary Immunodeficiencies (PID) [359 genes]

S-202110260 Primary Antibody Deficiencies (PAD) [51 genes]

S-202110261 Common Variable Immunodeficiency (CVID) [35 genes]

S-202008257 Agammaglobulinemia [10 genes]

S-201906890 Hyper-IgM Syndrome (HIGM) [8 genes]

S-202110262 Combined Immunodeficiencies (CID) [47 genes]

S-202008258 Bare Lymphocyte Syndrome (BLS) [13 genes]

S-202110433 Severe Combined Immunodeficiency (SCID) [20 genes]

S-202110434 Severe Combined Immunodeficiency [T- B+] SCID [11 genes]

S-202008260 Severe Combined Immunodeficiency [T- B-] SCID [8 genes]

S-202110263 Syndromes with Combined Immunodeficiency [81 genes]

S-202008261 Dyskeratosis Congenita (DKC) [16 genes]

S-202110264 Hyper-IgE Syndrome (HIES) [21 genes]

S-202008256 Ataxia-Telangiectasia (AT) [1 gen]

S-202110265 Defects in Intrinsic & Innate Immunity [138 genes]

S-202110266 Viral Infections, Predisposition [74 genes]

S-202110267 Mendelian Susceptibility to Mycobacterial Disease (MSMD) [38 genes]

S-202110268 Susceptibilidad mendeliana a enfermedades por micobacterias (SMEEM) [26 genes]

S-202110269 Fungal Infections, Predisposition [16 genes]

S-202110270 Invasive Bacterial Infections, Predisposition [8 genes]

S-201906649 Cystic Fibrosis (CF) [1 gen]

S-201907069 Phagocytes Defects, Congenital [44 genes]

S-202110271 Neutropenia, Syndromic [33 genes]

S-202110288 Chronic Granulomatous Disease (CGD) [8 genes]

S-202110272 Complement System Deficiencies [41 genes]

S-201805829 Atypical Haemolytic Uremic Syndrome (aHUS) [17 genes]

S-202008267 Disseminated Neisserial Infections [9 genes]

S-202008268 Systemic Lupus Erythematosus (SLE)-like Syndrome [8 genes]

S-202008269 Pyogenic Infections, Recurrent [6 genes]

S-202110273 Hereditary Angioedema (HAE) [5 genes]

Immune Dysregulation Diseases (IDD)

S-202110274 Immune Dysregulation Diseases (IDD) [274 genes]

S-202110275 Autoimmune Diseases (AD) [168 genes]

S-202110276 Systemic Lupus Erythematosus (SLE) [77 genes]

S-202110277 Autoimmune Nephropathy (AN) [50 genes]

S-202110278 Autoimmune Lymphoproliferative Syndrome (ALPS) [26 genes]

S-202110279 Autoimmune Enteropathy (AE) [23 genes]

S-202110280 Autoimmune Polyendocrinopathy (AP) [13 genes]

S-202110281 Autoinflammatory Diseases (AID) [157 genes]

S-202110282 Behçet's Disease (BD) [49 genes]

S-202008190 Inflammatory Bowel Disease (IBD) [28 genes]

S-202110283 Inflammatory Bowel Disease (IBD) [45 genes]

S-202110284 Autoinflammatory Diseases with Recurrent Fever [20 genes]

S-202110285 Síndrome CANDLE [7 genes]

S-201805369 Aicardi-Goutières Syndrome (AGS) [7 genes]

S-202110286 Hemophagocytic Lymphohistiocytosis (HLH) [46 genes]

S-202110287 HLH with Epstein Barr Virus Susceptibility [20 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).

> Immunology

Other genetic tests

- S-202109974 Secuenciación individualizada de genes (Sanger)
- S-202109975 Secuenciación masiva NextGenDx®
- S-202109976 Massive sequencing with CNVs

Whole exome

- S-202110014 Whole-exome – sequencing only (fastq)
 - S-202110013 Whole-exome – annotation of variants
 - S-202110336 Whole-exome – with report tool
 - S-202110015 Whole-exome – with clinical report
-
- S-202109977 Targeted exome:
 Gene/genes: _____
-
- S-202110133 Trio clinical exome

SNP array:

- S-201601485 Index case
- S-201702726 Family study or confirmation of CNVs

Array CGH:

- S-202008036 Prenatal array (37K)
- S-202109987 Postnatal array (60K)
- S-202109988 Postnatal array (180K)

- S-202109998 Variant segregation / Family studies
 Variant: _____

Kits (PCR real-time (qPCR):

- S-202009875 Imegen-HLA-B27
- S-202009876 Imegen-HLA-B57:01
- S-202009877 Imegen-Coeliac
- S-202009878 Imegen-IL28B

- S-202109983 MLPA and methylation-specific MLPA:
 Gene/genes: _____

Other services: _____

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

> Immunology

- 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

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