

The fields marked with (*) are required to carry out the genetic test.

→ REQUESTOR *

FIRST NAME SURNAME (S)

HOSPITAL / CLINIC / CENTER

TEL. NO. CONTACT EMAIL

Please check this box if the person requesting the study and the recipient of the report are the same person. Otherwise, please fill in the following details of the recipient.

EMAIL ADDRESS FOR THE REPORT

FIRST NAME SURNAME (S)

→ PATIENT DETAILS (INDEX CASE)

It is mandatory to include details about the patient's clinical indication and family history. For family testing, use separate request and informed consent forms for each person.

FULL NAME / REFERENCE *

SEX ASSIGNED AT BIRTH * DATE OF BIRTH * MRN *

AFFECTED: YES NO CONSANGUINITY: YES NO

TYPE OF SAMPLE *:

DNA BLOOD SALIVA BUCCAL SWAB TISSUE AMNIOTIC FLUID CHORIONIC VILLUS SAMPLING

If a DNA sample is submitted, please specify if its origin is different from blood *:

DATE OF SAMPLE COLLECTION (if the period is >24 h and the sample is not refrigerated):

BLOOD TRANSFUSION LESS THAN 60 DAYS AGO: YES NO BONE MARROW TRANSPLANT: YES NO

The patient or legal representative has agreed to being informed about any results relative to **secondary findings** (ACMG)¹** and/or **incidental findings**.

** Reporting these secondary findings entails the activation of a service with an added cost. Please contact us for more information.

¹Miller et al. Genet Med. 2022 24(7):1407-1414

→ CLINICAL INDICATION* AND FAMILY HISTORY (IF APPLICABLE)

Please attach the study indication and all clinical information considered relevant. Partial or missing information may compromise the correct interpretation of the results in the clinical context.

Clinical information attached to this form.

→ **TYPE OF TEST ***

INDIVIDUAL TESTING	FAMILY SCREENING	PRENATAL STUDY: Maternal contamination has been ruled out
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→ **TEST REQUESTED * (SELECT THE TYPE OF ANALYSIS AMONG THE OPTIONS BELOW)**

01 GENOME SEQUENCING

<p>CLINICAL GENOME TESTING Sequencing + Report with clinical/diagnostic guidance</p> <p>Individual Duo Trio</p> <p>Describe clinical indication, phenotype, HPOs, etc. in the corresponding section[†].</p> <p>Do you require access to analysis software? *: Yes No</p>	<p>WHOLE GENOME SEQUENCING (WGS) WGS wet lab + Raw Data</p> <p>100Gb 30X 200Gb 60X</p> <p>Deliverable: Sequencing files only Access to analysis software</p>
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02 NGS GENE PANEL

View our list of [available panels](#) by clinical specialty.

S- If you know the reference no. of the desired panel, please insert it here.

Pathology / Phenotype *:

Do you require access to analysis software? *: Yes No

03 PHENOTYPE-BASED EXOME ANALYSIS

View our [specific designs](#) by clinical specialty.

S- If you know the reference no. of the desired study, please insert it here.

Disease, phenotype or condition *: OMIM

Do you require access to analysis software? *: Yes No

04 EXOME SEQUENCING

<p>WHOLE EXOME</p> <p>Individual Duo Trio</p> <p>Describe clinical indication, phenotype, HPOs, etc. in the corresponding section[†].</p> <p>INCLUDING REPORT REPORT NOT INCLUDED</p> <p>Access to analysis software Deliverable: Sequencing files only Access to analysis software</p>	<p>EXPRESS CLINICAL EXOME </p> <p>Individual Duo Trio</p> <p>Describe clinical indication, phenotype, HPOs, etc. in the corresponding section[†].</p> <p>Do you require access to analysis software? *: Yes No</p>
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05 ANALYSIS OF SPECIFIC GENETIC VARIANT

A copy of the original report describing the mutation is required, in addition to a sample from a family member as a positive control.

Mutation (HGVS nomenclature) Gene NM

Is interpretation of the variant required? *: Yes No *If the index case has been tested at Health in Code, please indicate it below.*

Index case[Ⓢ]

Ⓢ You may enter the "HIC sample number", "Medical Record Number" or any other information that may help us identify the index case, such as the patient's full name.

06 CYTOGENETIC TESTING

180K array CGH 60K array CGH Karyotype [View sample requirements.](#)

07 OTHER TYPES OF GENETIC TESTS / For information on gene availability and advice on cost-efficient technology, please contact customercare@healthincode.com

WHOLE-GENE SEQUENCING	MLPA	EXPANSIONS	OTHER TESTS
Description: Gene <input type="text"/> Region <input type="text"/> Mutation (HGVS nomenclature) <input type="text"/>			

[†]The analysis and interpretation is carried out in line with the clinical indication provided.

STATEMENT BY THE REQUESTING PHYSICIAN *

(1) I hereby certify that the information provided in this form is correct to the best of my knowledge and that I have requested the indicated genetic test based on my professional judgment and the patient's clinical and family history. (2) I have explained the limitations of this test and I have answered any questions using my clinical judgment. If an informed consent form signed by the patient or legal guardian is not provided, (3) I hereby certify that a copy of said consent form is secured with the patient's medical records at my medical center. I understand that Health in Code, S.L. may require additional clinical and familial information for the correct interpretation of the obtained results within the indicated clinical context, and I agree to provide said information if necessary.

DATE SIGNATURE / print and sign manually or attach a signature image. (or) DIGITAL SIGNATURE

Patient information sheet

Purpose and implications of genetic testing.

Genetic tests are a type of tests that examine DNA to identify genetic variations that can affect health, physical features, and predisposition to certain diseases. These tests can aid in the diagnosis, treatment, and prevention of genetic diseases, as well as in family planning and health-related decision-making.

Genetic testing is performed by extracting DNA from biological samples, such as blood, saliva, or prenatal samples (e.g. amniotic fluid), among others.

Genetic test results can have implications for you, your offspring, and other members of your family, as they could reveal details about the risk of developing or transmitting a genetic disease, the possible response to a certain treatment, or previously unknown kinship details (e.g. non-biological paternity). It is important to bear in mind that the severity of a genetic condition or the age at which symptoms could occur cannot be accurately predicted. In the case of prenatal testing, the results could influence decisions about pregnancy, such as terminating it or making preparations for the birth of a baby with certain genetic conditions.

You have the right not to be informed of the results of this genetic test. If you choose not to, we may inform your family members (or legal guardians) with your consent and/or if there are medically ethical grounds, when this information is necessary to avoid serious harm to their health, as determined by the treating physician. Communication shall include only the necessary information.

Pharmacogenetic testing: these tests are for informative and investigational purposes and are being developed to provide information about how certain drugs could act in your body and support the treatment of specific conditions and pathologies.

Possible genetic test results.

- ↳ **Positive result:** One or more genetic variants have been identified that could explain the reason for requesting the test.
- ↳ **Negative result:** No genetic variants related to the condition have been identified. This does not mean that there is no predisposition to any diseases, as some genetic disorders have multiple causes and may not be detected by the test performed. A negative result could also occur as a result of scientific, technical, and/or knowledge-based limitations.
- ↳ **Non-conclusive result:** One or more genetic variants have been identified whose clinical significance is currently uncertain. These variants are known as VUS ("variants of uncertain significance") and cannot be used as the basis for medical decisions. In certain cases, additional testing may be recommended for yourself or for other members of your family. The report will only include variants of unknown significance if they are considered clinically relevant by the clinical team based on the currently available scientific evidence.
- ↳ **Non-informative result:** Exceptionally, due to possible sample quality/quantity issues or to sample contamination, the test may not produce any results. If this occurs, a new sample may be requested.

The identified genetic variants are classified into five different categories of pathogenicity, as per the standards developed by the ACMG (American College of Medical Genetics and Genomics).

Reanalysis.

Scientific advances may change the significance of these variants; therefore, **it is recommended to reanalyze the results after some time.** DNA sequencing data are not routinely reanalyzed at our laboratory, but it is possible to do so for certain genetic tests (e.g., clinical exome or genome testing) upon request.

Occasionally, Health in Code may reanalyze the issued results in the future, and the patient or the requesting party may be contacted in case of new findings related to the clinical indication for the test.

Incidental/non-requested findings.

Tests that analyze the exome or genome could detect genetic variants not related to the main reason for the study, but with clinical implications. These findings, known as incidental or non-requested, will only be reported if you have explicitly consented to be informed about them. Some of these findings may indicate a risk of developing an unpreventable or untreatable genetic disorder.

Health in Code will not report any incidental finding related to adult-onset neurodegenerative disorders for which no treatment is available. Likewise, any carrier status unrelated to the reason for the study will not be reported unless explicitly consented to in the study request form.

Secondary findings.

With exome and genome testing, the ACMG guidelines recommend additional analysis of certain pathogenic (or possibly pathogenic) genetic variants associated with diseases for which prevention or treatment is available a priori. These findings are known as secondary findings and will only be reported if you have explicitly stated that you would like to be made aware of them in this consent form.

Limitations and potential risks.

- ↳ **Blood draw:** If genetic testing is performed on a blood sample, you could experience small bleeding, temporary discomfort, bruising at the puncture site, dizziness, or loss of consciousness.
- ↳ **Prenatal testing:** If amniotic fluid or chorionic villus are sampled, the procedure (amniocentesis) entails a small risk of bleeding, infection or, rarely, miscarriage.
- ↳ **Psychological impact:** Results from genetic testing can cause stress or anxiety to you or your family.
- ↳ **Limitations of the technique:** There are various types of genetic alterations, and no technique is capable of detecting all of them. Every technique has its own specific limitations, which will be duly indicated in the results report.
- ↳ **Errors:** Under exceptional circumstances, the genetic test may return inaccurate results due to errors during sample collection, labeling, and processing or during data analysis and interpretation.
- ↳ **Scientific advances:** The analysis and clinical interpretation of the genetic test are based on the currently available knowledge and technology. As scientific knowledge evolves, the analysis and interpretation of the test may change or require supplementation.
- ↳ Genetic variants may not explain the described medical condition and may not result in any change in the current therapeutic and/or pharmacological treatment or approach.

Limitations of whole-exome and clinical genome testing.

These tests provide a vast amount of information about genetic variants. The report will only include those genetic changes that could be associated with the patient's clinical findings, along with secondary and/or incidental findings, should you consent to be informed about them, as the available data on the clinical significance of most non-targeted variants detected are currently insufficient.

Genetic counseling.

The test results report is not a substitute for a medical diagnosis or genetic counseling. The results must be explained by a physician or healthcare professional.

Health in Code is not responsible for any misuse of the test data by the patient, the requesting party, or any third party. Health in Code remains at the disposal of the patients and their physicians for any questions related to the genetic test.

Personal data protection and use.

Your personal data will be used to manage your relationship with Health in Code and to provide clinical laboratory services in accordance with applicable laws. Only authorized personnel will have access to your health-related data, which are considered particularly sensitive, and will be able to share them with other organizations when necessary for the delivery of the requested services or for the compliance with our legal obligations. The transfer of your personal data outside the European Economic Area is not foreseen.

Your data will be kept during our relationship and as long as required by the different laws and regulations, namely Act 41/2002, of 14 November, Regulating Patient Autonomy and Health Documentation and Information-Related Rights and Obligations. Once the applicable legal deadlines have been met, we shall proceed to eliminate them in a secure manner.

At any time you may contact us to know the information we have about you, rectify it if incorrect, request its transfer to another organization, and eliminate it once our relationship has ended, when legally possible. To exercise any of these rights, you should submit a written request to our email address, along with a photocopy of your ID for your correct identification: info@healthincode.com

You can also contact our Data Protection Officer at: fgomez@audidat.com

Should you have any reason to believe that your rights have not been adequately protected by our organization, you can lodge a complaint with the Spanish Data Protection Agency (www.agpd.es).

For more information about the use of your data, please refer to our website (healthincode.com).

The genetic data obtained may be used for research purposes to expand scientific knowledge by their inclusion in scientific publications or in genomic databases, unless you explicitly object to such use. These research activities provide new evidence for reclassifying variants, enabling a more accurate interpretation of the results and enhancing diagnosis, prevention, and treatment of genetic diseases.

If you consent to the use of your anonymized data, please bear in mind that the risk of identification, although low, cannot be entirely excluded due to the unique nature of genetic information.

You understand that you will not receive any financial benefit from any research carried out or any products developed.

Health in Code, S.L. is not responsible for any harmful consequences that may arise from the use of the test data by yourself, the requesting party, or any third parties.

Informed consent form

I hereby declare that:

- ☑ I have been informed about the purpose of the test, the procedure to be carried out, the limitations of the test, and the potential results and implications.
- ☑ I have carefully read the informed consent form, I have understood the information I have been given, and all my questions have been answered.
- ☑ All the personal and medical data provided are true, and I understand that the team at Health in Code, S.L. may need additional clinical data, for which my physician may contact me if necessary.
- ☑ I understand that a new sample may be requested if the quality or quantity of the previous sample is insufficient or if the complexity of the diagnosis requires additional genetic tests; likewise, I understand that samples may also be required from my family members to better interpret the results and complete the study.
- ☑ I understand that there are various types of genetic alterations, and all techniques have certain limitations; therefore, the detection of all possible alterations cannot be guaranteed. The findings included in the results report may require the performance of additional tests.
- ☑ I understand that the test results report is not a substitute for medical diagnosis or genetic counseling and that the results should be explained and interpreted by a healthcare professional.
- ☑ I understand that the results of this genetic test may have implications for me, my offspring, and other members of my family. In that case, I agree to transmit the information directly to my family members, or, if I do not wish to be notified of the results, the physician can directly communicate them to those family members that could be affected.
- ☑ I consent to the use of my personal data, or those of the patient I legally represent, with the purposes of performing the requested test.
- ☑ I consent to the use of my clinical data and test results, or those of the patient I legally represent, in anonymized form, for research purposes, scientific publications, inclusion in genomic databases, and quality controls, unless I explicitly object to such uses below.

I do not authorize the use of my data for research purposes or for reclassifying variants and updating and enhancing diagnostic processes.

- ☑ I understand that the test may obtain genetic information that is unrelated to the reason for the test and that has potential implications for my health or that of the patient I legally represent.

I hereby consent to being informed about secondary findings.

I hereby consent to being informed about incidental findings in case they indicate a risk of developing a genetic disorder that can be prevented or treated.

I hereby consent to being informed about incidental findings in case they indicate a risk of developing a genetic disorder that CANNOT be prevented or treated.

By signing this document, I authorize Health in Code to perform the genetic test and agree to the above statements.

In the case of a minor or a person without legal capacity, I authorize the performance of the above indicated genetic test as the person's Legal guardian. I confirm that the signatory is the sole legal guardian or that the other parent does not oppose this test being performed on our child.

NAME _____ SURNAME(S) _____ ID _____
 TELEPHONE _____ EMAIL _____

Place: _____, Date: ___/___/___
(day/month/year format)

Signed

GENETIC TEST REQUESTED

Please specify the requested genetic test (mandatory)

Reference (optional)

IDENTIFICATION OF THE REQUESTING PHYSICIAN

NAME _____ SURNAME(S) _____

EMAIL _____

HOSPITAL / CLINIC / CENTER _____

Your informed consent may be revoked at any time by notifying Health in Code, S.L. in writing.



Capture NGS sequencing panels *

Disorders of the immune system (global panel) / Ref.: S-202110258	+info	549 genes
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→ PRIMARY IMMUNODEFICIENCIES (PID)

Primary immunodeficiencies (PID) / Ref.: S-202110259	+info	381 genes
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/ Adaptive immunity defects

Primary antibody deficiencies (PAD) / Ref.: S-202110260	+info	51 genes
Common variable immunodeficiency (CVID) / Ref.: S-202110261	+info	35 genes
Agammaglobulinemia / Ref.: S-202008257	+info	10 genes
Hyper-IgM (HIGM) syndrome / Ref.: S-201906890	+info	8 genes
Combined immunodeficiency (CID) / Ref.: S-202110262	+info	47 genes
Bare lymphocyte syndrome (BLS) / Ref.: S-202008258	+info	13 genes
Severe combined immunodeficiency (SCID) / Ref.: S-202110433	+info	20 genes
T-B+ severe combined immunodeficiency (SCID) / Ref.: S-202110434	+info	12 genes
T-B- severe combined immunodeficiency (SCID) / Ref.: S-202008260	+info	8 genes
Combined immunodeficiency syndromes / Ref.: S-202110263	+info	81 genes
Hyper-IgE syndrome (HIES) / Ref.: S-202110264	+info	21 genes
Ataxia telangiectasia / Ref.: S-202008256	+info	1 gene

/ Innate immunity defects

Defects in intrinsic and innate immunity / Ref.: S-202110265	+info	138 genes
Natural Killer cell deficiency / Ref.: S-202110266	+info	74 genes
Susceptibility to viral infections / Ref.: S-202110267	+info	38 genes
Mendelian susceptibility to mycobacterial diseases (MSMD) / Ref.: S-202110268	+info	26 genes
Susceptibility to fungal infections / Ref.: S-202110269	+info	16 genes
Susceptibility to invasive bacterial infections / Ref.: S-202110270	+info	8 genes
Congenital phagocytic defects / Ref.: S-201907069	+info	44 genes
Neutropenia / Ref.: S-202110271	+info	33 genes
Chronic granulomatous disease (CGD) / Ref.: S-202110288	+info	8 genes
Complement system deficiencies / Ref.: S-202110272	+info	41 genes
Disseminated gonococcal infection / Ref.: S-202008267	+info	9 genes
Systemic lupus erythematosus syndrome (SLE-like) / Ref.: S-202008268	+info	8 genes
Recurrent pyogenic infections / Ref.: S-202008269	+info	6 genes
Hereditary angioedema (HAE) / Ref.: S-202110273	+info	5 genes

→ IMMUNE DYSREGULATION DISORDERS (IDD)

Immune dysregulation disorders (IDD) / Ref.: S-202110274	+info	273 genes
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/ Autoimmune diseases

Autoimmune diseases (extended panel) / Ref.: S-202110275	+info	168 genes
Autoimmune diseases (basic panel) / Ref.: S-202110277	+info	50 genes
Genetic lupus-like syndrome / Ref.: S-202110276	+info	77 genes
Autoimmune lymphoproliferative syndrome (ALPS) / Ref.: S-202110278	+info	26 genes
Autoimmune enteropathy (AIE) / Ref.: S-202110279	+info	23 genes
Autoimmune polyendocrinopathy / Ref.: S-202110280	+info	13 genes



/ Autoinflammatory diseases

Autoinflammatory diseases (extended panel) / Ref.: S-202110281	+info	158 genes
Autoinflammatory diseases (basic panel) / Ref.: S-202110282	+info	50 genes
Inflammatory bowel disease (IBD) / Ref.: S-202110283	+info	45 genes
Behcet's disease / Ref.: S-202008190	+info	28 genes
Autoinflammatory diseases with recurrent fever / Ref.: S-202110284	+info	20 genes
CANDLE syndrome / Ref.: S-202110285	+info	7 genes
Aicardi-Goutières syndrome (<i>Exome</i>) / Ref.: S-201805369	+info	8 genes

/ Hemophagocytic Lymphohistiocytosis (HLH)

Hemophagocytic lymphohistiocytosis (HLH) / Ref.: S-202110286	+info	45 genes
Hemophagocytic lymphohistiocytosis (HLH) with Epstein-Barr virus (EBV) susceptibility / Ref.: S-202110287	+info	20 genes

/ Hypersensitivity diseases

Hereditary alpha-tryptasemia (HαT) (<i>PCR</i>) / Ref.: S-202414299	+info	2 genes
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(*) Except for those where a different technique is specified.