NIMGenetics
New Integrated Medical Genetics

COD.

To be filled by the laboratory

INFORMED CONSENT
POSTNATAL Genetic Study

Conventional Karyotype	☐	MLPA/MS-MLPA	☐	One-off mutation	☐
FISH	☐	TromboNIM®	☐	NIMSeq - NGS panel	☐
KaryoNIM® - CGH array	☐	Triplet expansion	☐	ExoNIM® - Exonic sequencing	☐
ReproNIM®	☐	Single gene sequencing	☐	Mitochondrial DNA sequencing	☐

Identification Code:	**Name and Surname:**
Reason for Consultation:	**Date of Birth:**
Full address with post code and city:	**ID:**
Patient E-mail:	**Telephone Number:**
Name of Medical Practitioner:	**E-mail:**
Clinic /Hospital /Laboratory:	**Telephone Number:**

1. I have received information on the indication, purpose, characteristics, scope and limitations of the Genetic Diagnostic Test, and I have had the opportunity to read the information provided about the test through the information sheet attached to this document, and my questions have been answered satisfactorily.
2. I declare that the personal and medical information I have provided is true and reliable.
3. I understand that I may be asked for a new sample if the diagnosis complexity makes other genetic tests necessary or if the sample obtained is not optimal in quality or quantity. I have also understood that there is a possibility that a blood sample may be required from the biological parents or other relatives in order to assist in a better interpretation of the tests performed on the sample.
4. NIMGenetics assumes that, in the studies aimed to identifying the origin of the genetic alterations being studied, the medical practitioner has confirmed that the samples from the parents submitted correspond to the biological parents.

   If not, specify: _____.
5. I understand and agree that the clinical team may contact me for additional clinical data.
6. I understand that the results of this test do not replace the medical diagnosis made within a medical visit, nor the genetic counselling given by your physician, recommending that these results be shared in a medical visit, where, in addition, the genetic counselling described in the attached information sheet should be carried out. NIMGenetics, S.L. shall not be liable for any use made by you or your physician of the results obtained, nor for any harmful consequences that may derive from the use of such information.
7. I understand that the normality study does not guarantee the complete diagnosis of your condition with regard to all possible genetic alterations, due to the limitations described in this document, as well as in the report of results.
8. I understand that by performing this test, genetic information may be obtained that is not related to the suspected diagnosis for which this test has been requested. These findings, which would be included in the report of results, may require further testing.

   ☐ I check this box to indicate that I **DO NOT WANT** this information to be communicated to me.
9. I understand that the information obtained may also have implications for other family members, as well as the desirability of transmitting such information to them myself in this case.

For all these reasons, I declare the above and give my consent to carry out the Genetic Diagnosis Test.

Signature of Patient/Legal Guardian: **Date:** **Signature of Medical Practitioner:**

_____ _____ _____

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ADDITIONALLY,

☐ I consent to the use of my clinical data and the obtained results under a pseudonym (i.e. without name or surname) for research purposes, scientific publications, quality studies, and databases in the healthcare field, in which strict confidentiality shall be preserved regarding my identity.

☐ I consent to the assignment of my clinical data and results under a pseudonym (i.e. without your name or surname) to third parties for scientific research.

☐ I authorise NIMGenetics, or its affiliates, to send me information related to its products and services.

Signature of Patient/Legal Guardian: **Date:**

Genetic diagnosis test: purpose and risks

This test is intended to detect a change in DNA that can determine the following situations:

1. The genetic alteration is responsible for a syndrome or disorder that is the reason for the study;
2. The genetic alteration predisposes the individual to the development of a syndrome or disorder that could affect them;
3. The genetic alteration confers a carrier status, and consequently can be transmitted hereditarily, even if the carrier does not suffer from the disorder or syndrome under study (autosomal recessive or recessive X-linked diseases).

This genetic test is carried out from the extraction of DNA from various fluids such as saliva or blood, or from tissues such as skin, buccal scraping or fetal remains, among others. It is your doctor's responsibility to inform you of the risks associated with the procedures necessary to obtain the sample. The consequences of obtaining peripheral blood by venipuncture are mild and infrequent, and include the possibility of developing a hematoma at the puncture site, fainting, or dizziness.

The proposed study shall be performed on the sample applying the technology requested by your physician, as it is considered the best diagnostic strategy currently available for the clinical case under study. However, new clinical or scientific evidence may emerge indicating the need for further testing.

As medical knowledge advances and new discoveries are made, the interpretation of results may change. It is possible that, in the future, a new interpretation of their results could lead to new information about the medical condition under study.

In some cases, it may be necessary to perform complementary tests on the submitted sample, or to request a blood sample from family members in order to complete the study.

A new sample shall be required if the sample obtained is not optimal in quality or quantity, or if the diagnosis complexity requires other genetic tests to be performed.

Location where the analysis will be carried out and use of the biological sample at the end of the analysis

This test shall be performed by technical personnel at NIMGenetics in the laboratory owned by this company located in Madrid or, depending on the type of test to be performed, in a collaborating laboratory with whom a partnership agreement has been signed in accordance with the terms and legal requirements stipulated by current legislation.

If there is sufficient sample quantity left after the test, an aliquot of the excess sample and/or extracted DNA shall be stored in a coded form at NIMGenetics' headquarters, for a maximum period of 5 years, to be used in cases where a repeat study is necessary for diagnostic confirmation.

Only authorised NIMGenetics personnel shall have access to the connection between your biological sample, your DNA and information obtained from its processing, and the code assigned in each case.

Test Results

Before the test is carried out, you should consider the implication of the possible results. There are five possible outcomes:

- **Positive result:** One or more alterations considered to be the cause of the syndrome or genetic disorder that led to the study are detected. In some cases, this result does not imply that the syndrome or disorder associated with that genetic alteration is present, but rather that there is a higher risk of suffering from it than the general population. This finding would confirm or clarify the diagnosis.
- **Carrier status result:** A variant is detected in a healthy individual that can or has been transmitted to their offspring. In diseases associated with a recessive inheritance pattern, the disease or condition associated with alterations in the affected gene will only develop if an individual who is a carrier of a change has a child with someone who is also a carrier. In X chromosome-linked recessive diseases, the syndrome or disorder will develop only when the carrier mother transmits the DNA changes to a son.
- **Inconclusive result:** One or more alterations of uncertain meaning are detected. In this case it may be necessary to request additional tests or to study other family members in order to try to confirm whether the findings are related to the pathology or genetic alteration that led to the study being carried out.
- **Negative result:** No mutations are detected that could explain the pathology or genetic alteration. A negative result does not imply the absence of a genetically-caused pathology, since it depends on the scope of the study requested according to the diagnostic suspicion and the used technique's own limitations.
- **Non-informative result:** Exceptionally, a contamination of the sample, its bad quality or low quantity can determine no results being obtained.

It should be noted that the test results could have implications for your family members.

The report of results shall be sent to the requesting medical practitioner to be explained in consultation. The report's average availability period varies depending on the type of study requested.

Limitations of the Test

All genetic tests, regardless of the technique used for the specific case, have limitations that can affect the efficiency and reliability of the results obtained.

The following situations make it impossible to obtain a reliable test result:

- Genetic alterations that affect a limited number of the individual's cells (germline mosaicism).
- Genetic alterations in regions that are not analysed or whose analysis has not been included in the study requested by the medical practitioner (scope of the study).
- Genetic abnormalities whose size is below the technique's resolution limits.
- Genetic abnormalities that by their nature or because they are located in complex or repetitive regions are not identified, due to the limitations of the technology used.
- With the exception of karyotype and FISH, no other technology of those used in the diagnostic routine is capable of detecting balanced chromosomal rearrangements.
- In this diagnostic routine context, technologies capable of detecting polyploidies are karyotype, FISH, QF-PCR and microarrays based on single nucleotide variations.
- Changes in the individual's genetic sequencing (polymorphisms) that, because of their low frequency, are not considered in the test design.
- Individuals whose genetic origin does not correspond to the analysed parents (egg donation, semen donation, non-paternity, etc.)
- Type of biological samples: Unsuitable, scarce and/or poor quality samples can affect a result's efficiency and reliability.
- The time that elapses from when a biological sample is collected to when it is processed can affect the integrity of the DNA.

No genetic study technique is capable of identifying all the possible genetic alterations associated with a certain pathology. Therefore, each technology has its own specific instructions and limitations that will be reflected in the report of results.

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Incidental Findings

It is important to understand that sometimes we can identify genetic alterations that are not related to the diagnostic suspicion behind this study, and that could have relevant implications for the health of the individual under study and/or their family members. This incidental finding, if you consent, shall be included in the report of results. These findings may require additional invasive or imaging tests. You must decide whether or not you wish to receive this additional information by completing the appropriate section at the beginning of the document.

Genetic Counselling

The medical practitioner who requests/advises this test undertakes to provide information about the purpose of the analysis and provide genetic counselling once the analysis results have been obtained and evaluated. NIMGenetics is available to this professional to clarify any questions that may arise.

Data Protection and Confidentiality

Personal data and the genetic test results can only be accessed by duly authorised NIMGenetics technical and health personnel. This information will be confidential and shall be processed in accordance with the European Data Protection Regulation (Regulation (EU) 2016/679). The personal data that you provide us with, as well as those obtained from the analyses carried out, will be included and registered within the information processing systems of NIMGenetics for the diagnostic purpose described throughout this document. If you are not satisfied with the aforementioned processing, we will be unable to carry out the requested analysis. Likewise, we inform you that your personal information will not be disclosed to any person outside NIMGenetics, except for those who you authorise as requested in the following section “Further use of data”, or as required by law. However, your personal information will not be used for any purpose other than that reported here, or that is incompatible with it.

In compliance with Law 14/2007, of 3 July, on Biomedical Research, this test result shall be stored for a period of five (5) years after the analysis has been completed and, after such period has expired, it shall be pseudonymised if you have not exercised your right to cancel this genetic information. Test results shall only be kept as identified beyond the five (5) years mentioned above, in cases where it is necessary to safeguard your health, or that of your relatives.

The results of sample sequencing may be transferred to recipients outside the European Economic Area. NIMGenetics guarantees that this transfer shall be carried out in accordance with and under the warranties provided for in Regulation (EU) 2016/679, and that it shall not include any identification data.

You may exercise your rights of access, rectification, erasure, limitation of any specific processing, portability and opposition to the processing described by contacting the following address: NIMGenetics GENÓMICA Y MEDICINA S.L.: Parque Científico de Madrid C/ Faraday, 7 Campus Cantoblanco, 28049 Madrid. In the event that you require additional information on the processing of your personal data, you may check the additional information on our website: **https://www.nimgenetics.com/en/privacy-policy/**

Further use of the data

The results may contribute to increasing the analytical capacity of the test and the current state of knowledge, providing benefits for new studies. Therefore, NIMGenetics requests your consent to use your clinical data and results under a pseudonym, (i.e. without your name or surname) for research purposes, scientific publications, quality studies, and databases in the healthcare field, maintaining strict confidentiality regarding your identity, which will not be revealed under any circumstances.

Furthermore, NIMGenetics requests your consent for the assignment of your clinical data and results under a pseudonym (i.e. without your name or surname) to third parties for scientific research.

NIMGenetics contact details

Feel free to contact NIMGenetics by calling +34 91 037 83 54 to clarify any doubts that may arise in connection with the content of this informed consent.

In order to perform the requested genetic test, the informed consent document must be signed and dated.