Among the ethical standards required of medical professionals in Colombia by Law 23 of 1981 is the duty to inform adequately and promptly inform all patients of the risks that may arise from the treatment that will be performed, requesting your consent in advance (article 15 and 16). Therefore, with this written document we seek inform you about the procedure or test to be performed, for which we request that you fill out the spaces in blank your own handwriting.

MOLECULAR BIOLOGY TECHNIQUES

1. **Analysis of a mutation, a gene or several genes:** A molecular analysis of a gene or genes seeks to detect specific changes, called mutations among the analyzed genes. These genes are associated with a specific syndromes or diseases. These tests have a general sensitivity of 96% DEPENDING ON THE TECHNIQUE USED and the condition studied.
2. **Exome analysis:** Exome sequencing consists of analyzing the coding region of approximately 22,000 genes known to date. Not all of them have been related to known pathologies or phenotypes. The test is designed for the detection of point mutations, deletions or small insertions in the exome, not large deletions or duplications. The latter must be confirmed using other techniques. The genome of each person has millions of genetic variants. However, most of these variants are not related to a disease, or they have no defined relevance so far or they may be associated with a disease, but without relation to the medical question of the case. These findings are not the focus of the examination and will not be actively pursued. However, secondary findings of high relevance to the patient's health can be identified in rare cases, even when our analysis is guided by the clinical picture and medical indication. In these cases, the report will contain the information found. Variants of uncertain significance may be included in the result at the discretion of the Genetix medical team.
3. **Microarray Analysis:** The purpose of the microarray is to determine changes in copy number in the genome (CNV`s) by high resolution deletions and duplications, also allows the analysis of loss of heterozygosity and single nucleotide polymorphisms. Variants of uncertain significance may be included in the result at the discretion of the Genetix medical team.
4. **MLPA:** The purpose of an MLPA test is to determine deletions or duplications ate the exon gene level by multiplex ligation probe amplification techniques. Internal controls are used for interpretation.

MOLECULAR TESTING

Objectives of molecular testing: answer a clinical question and respond to the request of the treating physician. The information contained in the report will necessarily be combined with the clinical history, physical examination and/or other diagnostic tests in the definition of diagnosis and medical conduct.

Risks of molecular testing: It is important to know that there are factors that can alter the results such as DNA quality, contamination and amplification failures. Genomic analysis techniques are new and subject to adjustments periodically to increase the accuracy of the test. In addition to that, the clinical significance of the variants is constantly updated accompanying the advancement of scientific and medical knowledge. Annually they are

hundreds of new findings have been published on the relationship between genes, health and disease. In some cases interpretation may change with such scientific findings and therefore the results should not be treated as immutable. Current genomic analysis techniques cover between 95 and 99% of the sequences of interest.

An analysis may not identify the cause of the disease being investigated. At your doctor's request, a new test may be be performed in Genetix. Genetix may charge, at its discretion, for this reanalysis or other analysis not related to the initial question.

Another possibly unwanted information is the rare identification of non-paternity. This type of results will not be published since it is not the objective of the test, except when it is directly relevant to the medical request.

Complications:

1. In prenatal diagnosis, approximately 1% of the cases the sample obtained is contaminated with cells of maternal origin, which will give rise to a false result. This is especially common when the sample is bloody. For this reason, we perform maternal cell contamination tests in order to ensure that the sample to be studied is fetal DNA.

2. On some occasions the extracted DNA does not pass concentration and purity criteria, in these cases a new sample will be requested to ensure the result.

3. When the quality controls of the molecular tests are not optimal, reprocessing will be carried out.

4. One of the limitations that may arise is the little clinical information available about the patient, in these cases the analysis of variants is difficult because lack of propper relevant information.

Alternatives: Some molecular tests use another molecular test as an alternative, for example MLPA with Array, NGS sequencing with sanger sequencing. These will be suggested in cases they are required.

Benefits: Making the decision to undergo this analysis can directly benefit you and your family. The results of this genetic test could help you and your doctor make better and informed decisions about your health care, such as screening tests, risk-reducing surgeries, medications and preventive strategies.

Identification of a certain mutation(s) in a gene in a family allows others relatives determine whether or not they share the same cancer risk, or risk of having an recessive disorder.

During genetic counseling you will be able to consult about how the specific disease is inherited and find out about the likelihood that your children and blood relatives may have inherited the same mutation (or mutations) in the gene or genes analyzed. The patient's closest relatives share genetic information and this analysis will be able to reveal it.

TEST RESULTS: Test results will be sent to the healthcare provider when referred

by a specific entity, if it is an individual, it will use your username and password to download the result from the web, take the result to your treating doctor for appropriate management.

Genetix will not disclose personal information or test results to third parties unless required by local, state, federal or national law.

USE OF INFORMATION AND REMAINING SAMPLES: In accordance with best laboratory practices and standards, leftover sample (unless prohibited by law), as well as genetic information and other information learned from your testing, may be used by Genetix or others on its behalf for quality control, operations, laboratory tests, research studies, laboratory test development and laboratory improvement. All these uses will be in accordance with applicable laws. We may use your leftover sample and health information, including Genetic information. Such uses may result in the development of products, commercial services or scientific publications.

You will not receive notice of specific uses and you will not receive any compensation for these uses. All these uses will be in accordance with applicable law.

You can revoke this consent whenever you wish, to do so you only have to request the revocation from the laboratory, which

consists of signing the LAG-FOR-30 REVOCATION OF INFORMED CONSENT form.

Consent:

I, _____, identified with citizenship card number _____ of

_____, as patient and/or I, _____, identified with identity card

_____ citizenship of _____, as guardian of the patient noted above, I DECLARE that I have read and understood what the test that I am going to perform consists of.

I authorize YES____ NO____ GENETIX SAS to carry out the MOLECULAR GENETICS test.

If you have marked NO, it is understood that you have disagreed with taking the test. To do so, it is important that you keep in mind the consequence of not performing it and is not being able to determine genetic alterations and not being able to provide appropriate medical management.

In these cases, consult your treating doctor.