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.

GENETIC SCREENING

Screening for genetic conditions seeks to identify carriers of genetic diseases. Careful genetic counseling is recommended by a geneticist since there is family and personal history as well as demographic characteristics such as race, sex or place of origin that may suggest the existence of a transmissible genetic disease. Likewise, the limitations of genetic tests must be known, and reassurance provided about the low risk that assisted reproduction techniques represent.

Genetic testing of patients can reduce the risk of specific diseases but cannot eliminate the 3% - 4% residual risk for birth defects that all newborns have.

In autosomal recessive (AR) diseases, an individual must have mutations in both copies of the gene to manifest the disease. These mutated alleles come from their parents, who are usually healthy heterozygotes (they have one normal copy of the gene and one mutated one). If both members of a couple are healthy carriers, they have a 25% chance of having an affected child and this figure remains the same for each pregnancy. They are most frequently associated with consanguinity, ethnic groups with high endogamy, or geographic regions with little population migration.

Monogenic diseases linked to the X chromosome generally affect only men since they only have one of their daughters will be carriers. Men transmit the disease to 100% of their daughters, who will be carriers.

Exome analysis: Clinical exome sequencing consists of analyzing the coding region of approximately 6,500 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, small deletions or insertions in the exome, not large deletions or duplications. The latter must be confirmed using other techniques. Each person's genome has millions of genetic variants. However, the majority of these variants are not related to a disease, or have no defined relevance so far, or they may be associated with a disease, but unrelated 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 on rare occasions, even when our analysis is guided by the clinical picture and medical indication. In these cases, the result report will contain the information found. Variants of uncertain significance may be included in the result at the discretion of the Genetix medical team.

Dystrophinopathies including Duchenne and Becker muscular dystrophy and dilated cardiomyopathy analyzed by NGS detect deletions when >60% of an exon has a copy number abnormality. Inframe deletions are not detectable using this technique.

This test does not allow the analysis of triplet expansions such as fragile X syndrome, nor the analysis of gene or intragenic deletions or duplications for conditions such as Spinal Muscular Atrophy or Alpha Thalassemia.

Microarrays: The purpose of the microarray is to determine copy number change (CNV) due to high-resolution deletions and duplications, and it also allows the analysis of loss of heterozygosity and single nucleotide polymorphisms. More than 9,400 pathogenic variants associated with 600 autosomal recessive or X-linked conditions are analyzed.

This test does not allow the analysis of triplet expansions such as fragile X syndrome.

Risk interpretations and calculations are based on ethnic and clinical information along with family history provided, as well as understanding of current molecular genetics.

Clinical sensitivity and specificity vary for each disease and for each ethnic group.

The standard of care for carrier detection of Tay-Sachs disease in all ethnic groups is based on enzyme analysis (hexosaminidase A) in addition to DNA variant analysis.

The standard of care for determining carrier status for sickle cell anemia and other hemoglobinopathies is through hemoglobin electrophoresis and DNA testing.

The information contained in the result will necessarily be combined with the clinical history, physical examination and/or other diagnostic tests in defining the diagnosis and medical conduct.

Complications:

1. 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.

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

3. 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 to provide the indicated clinical relevance.

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

Benefits: Making the decision to undergo this analysis can directly benefit you and your family environment. The results of this genetic test may help you and your doctor make more informed decisions about your health care, such as screening tests, risk-reducing surgeries, and preventive medication strategies. Identifying a mutation(s) in a gene in a family allows other family members to determine whether or not they share the same risk of cancer or hereditary disease. If you get a positive result, during genetic counseling you can learn about how the specific disease is inherited and learn about the likelihood that your children and blood relatives may have inherited the same mutation (or mutations) in the gene or genes tested. The patient's closest relatives share genetic information, and this analysis may reveal it.

TEST RESULTS: The test results will be sent to the health care provider when referred by a specific entity, if you are an individual, you will use your username and password to download the result from the web, present the result to your treating doctor for give proper management. Genetix will not disclose personal information or test results to third parties unless required to do so 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.

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INFORMED CONSENT FOR REPRODUCTIVE GENETICS TESTING

LAG-FOR-60 V1; 19/12/2023

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.