# REQUISITION FORM FOR GENETIC DISEASES AND PHARMACOGENETICS

**Attachment III**

**LEAFLET / INFORMED CONSENT**

This document is divided into two parts:

- Leaflet: should be filled by your physician and it intends to explain this genetic test is being carried out, its risks and benefits and the importance of the results for you and your family,
- Informed consent: should be filled with your physician and you (or legal representative) after adequate genetic counseling.

**Package leaflet:**

Your physician prescribed the following genetic test (1):_____

To explain the following diagnostic/symptoms (2): _____

One **genetic test** allows the analysis of your genetic material using molecular genetic techniques. The genetic test to be realized (1) was selected by your physician because of its capacity to detect a specific disease (2).

The **methodology** used by this molecular genetic technique can be targeted or expanded, according to the genetic test selected by your physician. Usually, the selection is made accordingly the suspicion of a specific genetic disease or an extended group of diseases or symptoms. A genetic test to a specific genetic disease can be performed, for example, through the analysis of one gene (Sanger sequencing) one the other hand, to test an extended group of diseases or symptoms it is usual to analyze a gene panel through next-generation sequencing (NGS).

Usually, the **biological material** used for the genetic test is a sample of peripheral blood, collected through a venous puncture. Alternative tissues can be used, as buccal mucosa collected with a swab or skin sample through cutaneous biopsy with local anesthesia.

A genetic test can give different **results,** conclusive or inconclusive. It's considered inconclusive when the genetic test does not clarify the cause of the disease/symptoms, normally trough the identification of variants of uncertain (or unknown) clinical significance (VUS). The results are defined as being conclusive only when they explain the cause of the disease/symptoms, for example, when pathogenic variants are detected in a gene usually associated with the disease and compatible with the inheritance pattern of the disease.

A genetic test with a **conclusive result** may be used to make clinical decisions, like clinical management of the disease or for family planning. When results are **inconclusive** it may be necessary to continue analyzing through complementary analysis in the patients or their family and they should not be used for clinical decisions. A genetic test with **no alterations** does not exclude the possibility of having a genetic disease. This can be due to the lack of analysis of all the genes associated with the disease/symptoms or because the technique may have failed to detect variants in the analyzed genes.

Some genetic tests can give genetic information about **parenthood**, namely paternity and maternity information. Knowing the test results may cause **anxiety**; it is advised the discussion of the genetic test details, including possible results, with your physician.

Similarly, to other complementary diagnostic tests, a genetic test may obtain results that are not directly related to the reason why you realized the test, called **secondary findings**. When extended genetic testing are performed, like all disease gene analysis – clinical exome – the probability of finding variants not related to the suspicious disease increase, but those variants may have clinical importance for you and your family. These diseases may be relevant, like show an increased risk for cancer or cardiologic diseases. However, you can decide if you want to be, or not, informed about them. The decision of pursuing a genetic test is completely yours and that is why it is important to discuss it with your physician and to be aware of all the information that was given by him/her. It is also important to be able to discuss with your physician all the questions and doubts you may have.

You can revoke, completely or partially, your consent to the analysis/evaluation at any time, without the need to explain.

You have the right to not be informed of the test results, interrupt the analysis process at any time and to ask for the destruction of all the test results not delivered/disclosed.

**Physician (name):** _____

If you have any questions please do not hesitate on getting in touch.



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**Informed consent:**

By signing this consent, I recognize that (please read carefully):

1. I received all the appropriate explanations (by my physician) related with disease: _____ (to be filled by the physician).
2. I received, read and understood the explanations cited above on the package leaflet, related to the genetic test _____ (to be filled by the physician), including risks and benefits.
3. All my questions were answered and I had the time to analyze the situation.
4. The test sample and results will be used exclusively for this end. The results will also be used, if possible, to treatment decisions made by my physician.
5. According to the Portuguese law, n.º12/2005 from 26 of January, article 9º, "the communications of the genetic tests must be done exclusively to the patient, or, in diagnostic testing, to his legal representative or reference person, and only on a medical appointment."
6. I understand that I can revoke, completely or partially, my consent to this genetic test at any time, without justifying.
7. I understand that I have the right to not be informed of the test results, interrupt the analysis process at any time and to ask for the destruction of all the test results not delivered/disclosed.
8. I want ☐ **to be informed**/ ☐ **not be informed** [choose the preferred option] of the secondary findings not related to the requested genetic test.
9. I hereby declare that I have read the Privacy Policy of GenoMed® - Diagnósticos de Medicina Molecular, S.A., available at https://genomed.pt/en/privacy-and-cookies-policy/ and I give my consent to the processing of the personal data.

**Name (patient):** _____ or **Legal representative:** _____

**Signature:** _____ **Identification ID:** _____

**Date:** ____/____/_____

**Physician (signature):** _____ **Medical identification nº:** _____



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