healthincode
> Oncology

INFORMED CONSENT for genetic testing by massively parallel sequencing (NGS)

I, Mr/Ms. ..... , with ID no. ....., have been

informed by Dr....., have

understood and agree with the performance of the following genetic test:

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Description

Genetic testing by massively parallel sequencing (NGS) allows simultaneously determining the DNA sequence of a variable number of genes, which is particularly useful for genetic diseases or disorders caused by multiple genes.

Mass sequencing is especially appropriate when no specific clinical guidance is available or when looking for specific molecular targets, since it allows studying a subset of genes (gene panel) or even all 22,000 genes through exome sequencing, with the purpose of identifying the altered molecular pathways to be targeted by the treatment.

The study is performed by analyzing DNA from a blood sample (10-20 ml) and/or from tumor tissue.

Blood samples are collected by venipuncture at the bend of the elbow or the back of the hand.

Tumor tissue samples can be submitted by the Anatomical Pathology service in your center from the biopsy used for diagnosis; usually, no new biopsy is required.

In some cases, collecting blood samples from parents or other relatives may be required in order to interpret results.

Occasionally, technical failures or insufficient quality of the biological sample could lead to the need for additional sample collection.

The estimated turnaround time varies depending on the type of test.

Benefits

Identifying the genetic alteration(s) associated with the type of cancer under study can, in some cases, improve diagnosis, prognosis, and treatment.

The results of this genetic test can help you and your physician make healthcare-related decisions about treatment selection, early detection tests, risk-reducing surgery, and other preventative or therapeutic strategies.

Detecting a pathological germline variant (in DNA extracted from blood) and adequate genetic counseling allow expanding the study to other relatives susceptible of carrying the variant, with the purpose of finding out whether they have an inherited genetic susceptibility.

Potential results

1. Detection of one or more alterations considered pathologic and related to the type of cancer or disease indicated by the study, which would confirm or explain the diagnosis. The genetic report will provide relevant information about the identified genetic variants with diagnostic, prognostic, and/or therapeutic interest. Upon assessing the report, your oncologist will discuss the scope and implications of its results with you. Your oncologist will make the final decision on the selection of the most suitable treatment within the context of your disease in order to maximize the benefits.
2. Detection of one or more variants of uncertain significance. In this case, it may be necessary to request additional examinations or to test other family members to confirm whether the findings are related to the type of cancer or disease that is the object of study.
3. Failing to detect alterations that can explain the disease.
4. Detection of incidental or casual findings. These are defined as incidentally detected alterations that are unrelated to the type of cancer that led to the study, but which could have relevant health implications for the patients and/or for their relatives.

Limitations and risks

NGS-based genetic studies generate large amounts of data, and it is essential to distinguish between those that could be relevant and those that are not. Moreover, understanding their technical limitations is necessary for the accurate interpretation of the results:

- Certain gene regions can be difficult to analyze. Whenever this occurs, it is notified in the report.
- If the study is limited to the analysis of a group of genes, or gene panel, only mutations in those genes can be detected. Mutations in other genes not included in the panel cannot be detected. The report will indicate the exact list of genes analyzed, as well as the design of the specific panel used for the test.
- The inclusion of a large number of genes in a panel increases the probability of an unexpected finding, i.e. identifying a pathogenic variant in a gene that is not

associated with the tumor(s) found in the patient or in the family. This result may or may not have implications for preventative clinical management and for providing counseling to other family members. Moreover, you should always keep in mind the possibility of the result being uncertain or not leading to changes to your clinical management.

The clinical interpretation of the results will be based on the currently available scientific information. As medical knowledge is expanded and new discoveries are made, the interpretation of your results could change. It is possible that, in the future, a new interpretation of your results could provide new information about your medical condition.

A new interpretation must be requested by your physician and could involve additional costs. In some cases, testing other family members could be necessary to obtain a conclusive result.

This test may provide genetic information about the studied individuals or their family members that is unrelated to the indication for this test (incidental findings).

I agree to receiving information about these findings: ☐ YES ☐ NO

In case of deciding not to receive such information, I understand that nevertheless, when this information is necessary to prevent great harm to the health of my biological relatives, the affected persons or their legally authorized representative may be informed at the discretion of the requesting physician. In any case, the information provided will be limited to the necessary data for these purposes. (Art.49.2 of the Spanish Law on Biomedical Research).

Confidentiality

All personal information (clinical, genetic, etc.) will be gathered and treated in a confidential manner, always following the basic ethical principles of research on biological samples and complying with the applicable laws (Spanish laws LGS 14/1986, LOPD 2018, LBRAP 41/2002, and LIB 14/2007).

Alternative tests

The proposed genetic test is considered the best diagnostic strategy currently available for this specific clinical case. However, new clinical findings or scientific discoveries could suggest the need for other complementary genetic techniques.

Biological samples

Once the genetic study has been completed, the remaining DNA sample will be stored at the laboratory following the established quality criteria. After anonymization, this sample can be used for the development and standardization of genetic tests and/or for research purposes (please let us know if you do not agree to their use for these purposes).

Withdrawing consent

This informed consent may be revoked at any time by means of a written notification addressed to IMEGEN (Instituto de Medicina Genómica).

Patient's statement

After having read and understood the information contained in this document and having had the chance to ask any potential questions, I hereby consent to the use of this sample for the above mentioned genetic study by IMEGEN (Instituto de Medicina Genómica) or by any center(s) designated by them:

Date: ..... Signature of the patient (or guardian):

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In the case of familial studies, a sheet will be sent for each family member that participates in the study.

In the case of minors, the signer that has been dully identified (name and ID) authorizes the minor's participation in the genetic study

Forfeit of the Right to Information

Based on what is stated in article 49 of the Law 14/2007, 3 of July, of Biomedical Research, you have the right to be or not to be informed.

☐ I declare that, for personal reasons, I forfeit the right to information that assists me as a patient, and I hereby state my current wish to not receive information about my disease, notwithstanding the possibility that I may consent to the performance of this intervention, as stated and signed in the above section.