

Consent Bioarray Carrier Panel

My signature at the end of this document indicates that I understood and accepted the information contained herein and that I had the opportunity to clarify all my questions. I give, therefore, consent to Bioarray S.L. to use these samples in the performance of the genetic study indicated in the attached form, as well as in other centers assigned by it when necessary.

WHAT IS THE CARRIER PANEL?

The carrier test developed by Bioarray is a genetic analysis that simultaneously studies, using mass sequencing techniques - NGS, mutations in 298 genes responsible for hereditary recessive pathologies.

Recessive hereditary diseases occur when 2 mutated copies of the same gene are inherited from the parents. Each copy (allele) is inherited from one of the parents (one from the father and one from the mother) who are carriers.

Patients carrying recessive mutations are healthy patients, in fact, all of us are carriers of some mutations in recessive genes.

If a couple is a carrier of mutations in the same gene, their offspring have a 25% risk of transmitting it and having an affected child, 50% of the offspring are carriers without the disease and another 25% are free of the mutation (non-carriers).

In X-linked recessive diseases, the causative genes are found on the female sex chromosome. Therefore, 50% of the offspring of a carrier mother will receive the mutated copy of the gene. This copy will affect a child differently. Males receiving this mutated copy of the gene will be affected and manifest the disease. In the case of girls, if they have only one mutated copy, they will be carriers but will not have clinical manifestation (although this cannot be completely ruled out due to the phenomenon of X chromosome inactivation).

The availability of this information helps the couple to make appropriate reproductive decisions and, if necessary, to use currently available techniques to reduce reproductive risks and decide on future actions.

With the BIOARRAY recessive disease carriers test we can find mutations with different implications for the patients and their relatives. We can include them in several groups:

- Mutations associated with serious diseases with a significant impact on health that can affect people's life expectancy and quality of life.
- Mutations associated with diseases of varying severity, which can affect to a more variable degree the quality of life of people, but not life expectancy
- Mutations associated with rare diseases, with a significant clinical effect, and which occur with a high incidence in specific populations.
- Mutations present in X-linked genes.

DESCRIPTION OF THE TEST

This test examines a specific number of genes of the patient in search of genetic alterations that help to determine if the patient is a carrier of mutations associated with a series of pathologies. Mass sequencing techniques are used to identify known mutations in specific genome genes. This technique, unlike others, allows us to analyze millions of DNA fragments at a time, allowing us to analyze a large number of genes in a single process.

This test cannot determine all types of mutations; especially, it does not detect alterations that affect genes not included in the test. Your doctor can provide you with information about the specific changes that the test detects.

The Spanish Law 14/2007 of July 3 of Biomedical Research (LIB), establishes regulation for the performance of genetic analysis with health purposes. In accordance with Law 41/2002 on Patient Autonomy and Law 3/2018 on the Protection of Personal Data, the applicant must have the patient's consent to carry out the diagnostic tests requested and to process his/her data. In this way, and as information to be provided to the patient, we must inform you that the data collected in this form will be included in a confidential automated file, duly registered in the Spanish Data Protection Agency, in accordance with the terms established in Law 3/2018, whose ownership corresponds to Bioarray, S.L., in order to manage the diagnostic study in the form described, the patient may exercise at any time the rights of access, rectification, cancellation or opposition, recognized by the aforementioned legislation on the protection of personal data, addressing the following address: Bioarray S.L., Parque Científico de la UMH. Edificio Quorum III 03202 Elche (Alicante), email: info@bioarray.es Ph: 966682500 Fax: 966682501

ABOUT THE TEST RESULTS

A positive test result indicates that a genetic alteration has been found. A negative result indicates that either no alterations have been found, or the alterations found are of unknown significance (variants that are not known if they are related to a disease).

Although this test detects a high number of mutations, it may not detect, due to technical limitations, the total number of mutations causing recessive diseases.

This test is not the only way to look for genetic alterations, so my doctor may recommend the test before or after other genetic tests.

The results of this test may have implications for me and my family.

It is advisable for the patient/partner to receive genetic counselling before and after the test is performed. Given the complexity of the genetic studies and their important implications, the results will always be communicated to me through a doctor or genetic expert of my choice, and always with the utmost confidentiality.

PROCEDURE

To test the DNA in your blood, your doctor will take a blood sample (between 5 and 7 mL, in a standard blood sample). The physical risk to the patient is minimal.

CONSERVATION OF SAMPLES

The remaining DNA will be retained for future validation studies. This will require the written consent of the patient at the end of this document. You have the right to refuse such consent at any time, without giving any explanation and without this having any impact on the quality of the treatment you require.

CONFIDENTIALITY

Only experienced personnel will have access to the blood sample and test information and results. All results will be kept confidential in accordance with applicable laws and guidelines. The results will only be disclosed to your doctor and the requesting health care institution.

Only the requested and authorized tests shall be carried out on the identified blood sample.

The results obtained may be used in scientific publications or presentations, but the identity of all persons studied will not be revealed at any time. The collection of the information obtained is part of a laboratory's standard practice for quality purposes, and is required by laboratory accreditation.

DATA PROTECTION

In accordance with data protection regulations, we provide you with the following treatment information:

Responsible party: BIOARRAY, S.L.

Rights that assist you: access, rectification, portability, deletion, limitation and opposition.

More treatment information: <http://bioarray.es/es/>

BIOARRAY S.L. is responsible for the processing of personal data of the Interested Party and informs that these data will be treated in accordance with the provisions of Regulation (EU) 2016/679 of April 27 (GDPR) and Organic Law 3/2018 of 5 December (LOPDGDD), so the following treatment information is provided:

Purposes and legitimation of the treatment

By the legitimate interest of the responsible party (GDPR, Article 6.1.f): maintaining a professional relationship, sending communications, analysing data and publishing scientific and informative articles.

By consent of the interested party (GDPR, article 6.1.a): sending communications, analysing data and publishing scientific and informative articles.

Data retention criteria: will be kept for no longer than necessary to maintain the end of the treatment and when it is no longer necessary for this purpose, they will be eliminated with adequate security measures to guarantee the pseudonymisation of the data or the total destruction thereof.

Communication of the data: the data will not be communicated to third parties, except legal obligation.

I give my consent for the storage and preservation of the samples for possible use in the research on genetic disease and I authorize the transfer of the results of the clinical studies in an anonymous form for the study and pharmacological development, the sending of communications, data analysis and publication of scientific and informative articles:

☐ Yes

☐ No

Informed person (Name and signature):

ID number (if available):

Relationship with the patient:

Physician (Name and signature):