

What you need to know before sequencing your genome

Genetic counseling for parents and guardians - This document has been created to inform you of the importance and nature of the data obtained from a full genome sequencing, as well as the risks associated with its analysis.

The importance of genome sequencing

A very important factor in the diagnosis of rare diseases is to know if the disease has a specific genetic origin. Having a genetic origin does not always mean that the disease is inherited from the parents, because there are diseases that appear spontaneously without the parents being carriers.

The path to the diagnosis of a genetic disease is complex, since unfortunately there are many diseases that cannot yet be diagnosed. The sequencing of all DNA is highly recommended to guide us on this journey, but in itself it will not provide you with answers. The important thing to find answers is the analysis of this information.

In order to give a diagnosis to your child, we look for characteristics in his/her DNA that are related to a known disease. Unfortunately, in 60-70% of cases, this is not possible as there is still not enough information available. It is important that the information in your child's DNA is analyzed for new developments in your child and periodically, since new diseases are found every day. To be able to do this, it is necessary that after the sequencing the data is delivered to you so that you can manage it. Consult your doctor about alternatives to perform the reanalysis.

The possible consequences of genome sequencing

The analysis of DNA data can have different implications and it is important that you understand them. These are some of the possible consequences:

1. Genetic characteristics may be found that explain what is happening to your child. In other words, a diagnosis could be made.
2. Genetic characteristics may be found for which an interpretation is currently not known or are not completely conclusive. In these cases, and depending on different factors, complementary tests could be performed to understand the meaning of these characteristics. Furthermore, reanalyzing the data in the future could solve this uncertainty, as science gradually learns their meaning.
3. On the other hand, casual or incidental findings could be encountered. Since your child's entire genome is analyzed, unintended alterations could be found, such as genetic characteristics of diseases other than the one being studied. It is important that every time the genetic data is to be analyzed you make sure to talk about this point. You have the right to decide what you are going to be informed about, so please make sure you also discuss this issue before each analysis. Not all analyses can lead to such conclusions.
4. Finally, these data not only have information about your child, but they also have a percentage of information about each member of his or her biological family.

Therefore, some conclusions that can be drawn may have implications for some immediate family members. You have the right to decide what you will be informed about, so please make sure you also address this issue before each analysis. Not all analyses can lead to such conclusions.

It is important that you understand the points made in this information sheet. If you do, please read carefully the footer of this page and sign this document. If you have any doubts regarding the information contained in this document, please consult your doctor.

Section for Parent or Guardian Signature

I confirm that I have had the opportunity to comment on this document and ask questions about its contents and that I am satisfied with the answers and explanations provided.

I mark the following points as a statement that I understand their meaning:

- ☐ I understand the importance and sensitivity of data resulting from a full genome sequencing.
- ☐ I understand the risks associated with each analysis of the data resulting from a full genome sequencing.

Mr/Ms/Mrs.

as the child's parent or guardian

....., is informed of the possible risks that may arise from the analysis of the data obtained.

There is a legal presumption that parental authority is exercised for the benefit of the children and, therefore, that the parent who acts without the other parent with his or her child does so for the benefit of the child. Therefore, with respect to third parties in good faith, each parent shall be presumed to act in the ordinary exercise of parental authority with the consent of the other.

.....

(Signature)

In..... On the.....

(Fill in location and date)

