Together towards the diagnosis

PROGRAM TO SUPPORT THE DIAGNOSIS OF RARE DISEASES WITH GENETIC TESTING

If your child is under 2 years old and suffers from epileptic seizures and developmental delay... we want to help you.

What is it about?

Together towards the diagnosis is a program designed to speed up the diagnosis of rare epilepsy-related diseases. From Foundation 29 we make available genetic tests for free, while we support and guide you to get a diagnosis.

Dx29, our artificial intelligence tool, facilitates the process by combining genetic information with symptoms, helping the doctor in the search for a diagnosis. Register at www.dx29.ai.

What are the requirements to participate?

- ✓ Be 2 years old or younger.
- ✓ Have epileptic seizures.
- ✓ Have developmental delay.

How does the program work?

The steps to follow are very simple and Foundation 29 will help you in all of them.

If you are a **patient** this is what you will have to do:

1. Fill your details at the form at www.juntoshaciaeldiagnostico.org.
2. Send us a medical report by email to confirm the data on the form.
3. Foundation 29 reviews all the information you sent in order to approve your application.
4. Talk to your doctor for genetic counseling and for a prescription for a genetic test, if you have not already done so. We will send you documents to make it easier for both of you. If you have difficulties completing this step, talk to us and we will try to help you.
5. We will order your genetic test together and take care of the cost.
6. You will receive a kit to take a saliva sample. Follow the instructions and send it to us. Don't worry, we know how to do this and we will help you.
7. Contact your doctor to have your data analyzed. Dx29 will help you both in your search for a diagnosis.

If you are a **physician**:

1. Refer patients who meet the criteria and can benefit from this program to sign up. To speed up the steps, provide them with genetic counseling and give them the prescription and a medical report containing the symptoms.
2. The patient will follow the process as described above.
3. When s/he has the sequencing data, it will be shared with you through Dx29. Dx29 will support you with genetic and phenotypic data analysis algorithms, comparing them with known diseases, helping you in the search for a diagnosis.
4. If you have doubts, you can share the patient's data (after patient approval) with another specialist through Dx29, or analyze it by other means.

Since the number of tests is limited, Foundation 29 will conduct a selection of applications based on medical criteria.

Why a genetic test?

The path to diagnosis of a genetic disease is complex as there are unfortunately many diseases that cannot yet be diagnosed.

In the diagnosis of rare diseases, it is very important to know if the disease has a genetic origin. Having a genetic origin does not always mean that the parents are transmitters of the disease, because there are diseases that appear spontaneously.

The genetic test offered in this program is a sequencing of the whole genome based on a saliva sample. You take the sample at home with the help of a kit.

The sequencing of all the DNA allows you to start this journey, but the answers are obtained from the subsequent analysis of this information. Despite knowing your child's DNA, the answers you are looking for cannot always be found simply because there is still not enough knowledge to do so. However, it is the best chance to get a diagnosis for your child. You can read more about this in the genetic counseling document.

Who are we?

We are Foundation 29, a non-profit organization whose objective is to care for and guide patients in need of a diagnosis, with a special focus on rare diseases.

We build tools that help you make the best decisions for your health and at the same time we want you to be the owner of all your data. We innovate in the standardization and exchange of data in a secure way, in order to help the maximum number of patients.

What is Dx29?

Dx29 is a website that allows patients to provide information about their symptoms and their genetic information and to share it safely with physicians, in order to - together - advance on the path to diagnosis.

In Dx29 you can upload medical reports (in Word, pdf or directly from a picture of the report) and automatically find the symptoms mentioned in them. Then, it compares those symptoms with the genetic information provided by the patient.

It allows doctors to receive high quality information from families and, if they have permission, share it with other colleagues and analyze all that information with artificial intelligence tools to help the diagnosis.

In addition, patients with a diagnosis can privately compare their data with other patients with the same diagnosis.

Dx29 is free and the data uploaded is not shared with third parties: it belongs to the patient.

Do you have further questions?

Contact us via email at juntoshaciaeldiagnostico@foundation29.org for questions about this program or via info@foundation29.org for questions about Foundation 29.