Offering hope and changing lives through research

INGEMM Genetic Test Application Document 2: Informed Consent

European delegation of Dravet Syndrome Foundation

Copy for the INGEMM

INFORMED CONSENT

This informed consent form should be read by the requesting doctor and explained to the patient or her/his legal guardian. The patient or her/his legal guardian must sign two copies of this document, one copy will be sent with the Information Document to the INGEMM and the other copy should be kept by the patient or her/his legal guardian. Samples with no signed informed consent will not be accepted.

AIMS OF THE PROJECT

Mutations in genes that make up our DNA are responsible for various diseases, including the Dravet Syndrome and its other related spectrum disorders. The aim of this project is to determine if the patients have Dravet Syndrome or one of its related spectrum disorders by identifying the mutation responsible of these disorders. In order to achieve this objective a mutation screening of the six genes linked to date with the Dravet syndrome will be performed: SCN1A, PCDH19, GABRG2, SCN1B, SCN2A. Subsequently, a research to identify the mutations responsible for the disorder in those patients with no identified mutation in the previously reported genes will be carried out.

We also want to create a record of cases to support future research by any researcher interested in this disease.

PROJECT PROCEDURE

The procedure involves a DNA extraction from blood (or other tissues) in order tR perform a mutation screening of the genes linked to Dravet Syndrome, and associated spectra.
In order to identify new mutations that could be responsible for the Dravet Syndrome and/or its related spectrum disorders, the DNA sample obtained, the clinical and genetic data will be stored, coded and managed anonymously. However, the donor or her/his legal guardian could request the destruction of the stored sample at any time.
Access to the samples will be restricted to those responsible for the project : research department Dravet Syndrome Foundation , Institute of Molecular and Medical Genetics , University Hospital La Paz, Madrid (INGEMM) and other international records that meet criteria adequate custody) for an indefinite duration.
For any question regarding to the technical details of the project, your doctor may contact Dr. Eva Barroso, technical responsible of the project in the INGEMM, at the phone number: +0034.912071010-Ext.251., or by email to eva.barroso@dravetfoundation.eu.

TEST RESULTS AND INTERPRETATION

The results of the genetic test will be sent to the requesting doctor and will not be communicated to third parties without written consent. Any information obtained during this study will be handled with the utmost regards for confidentiality, in agreement with the Constitutional Personal Data Protection law 15/1999. To that effect, the samples will be codified and only the doctor responsible of the project will know the real identity of each codified sample.

The data will be held in a computer record adequately protected. Similarly , the data will only be made public in reports, scientific meetings , medical conferences or publications , reported as aggregated anonymous data , ie as percentages or numerical data without participant identification and never individually. The data will not be used for research other than Dravet Syndrome and associated spectra, unless you give your consent.

During the genetic testing, genes responsible for other diseases distinct from the Dravet syndrome and its related spectrum disorders will be analysed. Please mark which type of results should be informed to you:

___Only mutations associated with the Dravet syndrome and its related spectrums.
__ Any anomalies discovered

TEST LIMITATIONS

About 70-80% of the patients with Dravet syndrome present mutations in the SCN1A, PCDH19, GABRG2, SCN1B, and SCN2A genes. The margin for error in the methodology employed in the diagnostic test is 5%, and thus the capability of detection reduces to 65-75% of the Dravet syndrome cases.

EXPRESS CONSENT

I consent to those responsible for the registration or, where appropriate, especially experts appointed by the Research Committee of the Dravet Syndrome Foundation , to use my information , including personally identifiable information coded for (mark an X in the option with which you disagree) :

__ My disease research.

__ The related research on rare diseases, whose progress can benefit the knowledge of my disease.

I consent to the registration officials to facilitate my data, including personally identifiable information coded:

__other Researchers, provided they run an investigative approach for the study of my illness and have the approval of the Research Committee of the Dravet Syndrome Foundation.

SIGNATURE OF THE PATIENT OR HER/HIS LEGAL GUARDIAN

Through the information given by Dr._____, having read and understood this document I authorize the INGEMM to utilize the donated sample for diagnosis and research:
_____(City),_____(Date)

Signature of the patient or her/his legal guardian

_____

In the case of legal guardian please describe:
Relationship to the patient: _____
ID, passport or another identification document: _____

Copy for the patient or her/his legal guardian

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