



Ofreciendo esperanza y cambiando vidas  
a través de la **investigación**



**Offering hope and changing lives through research**

## **INGEMM Genetic Test Application Application Instructions**

**European delegation of Dravet Syndrome  
Foundation APPLICATION INSTRUCTIONS**

### **Introduction**

The Dravet Syndrome Foundation is sponsored economically by the BBK Foundation. They are offering a free genetic analysis for those with the diagnosis or tentative diagnosis of Dravet Syndrome and related spectrum disorders. The analysis will be carried out at the Institute of Medical and Molecular Genetics (INGEMM), Hospital La Paz, Madrid.

## Procedure

In order to send the sample to the INGEMM, the doctor must first submit an application that will be reviewed by the INGEMM and the Dravet Syndrome Foundation. **No sample will be accepted or analyzed without the confirmation of accepted application.**

The process to follow for submitting an application and mailing samples is detailed below:

1. Download Document 1 of Patient Clinical Information, and Document 2 of Informed Consent, posted on the Dravet Syndrome Foundation web page: [www.dravetfoundation.eu](http://www.dravetfoundation.eu)
2. E-mail completed Document 1 (without signing), to Dr. Eva Barroso, technical responsible of the project in the INGEMM: [genetictest@dravetfoundation.eu](mailto:genetictest@dravetfoundation.eu)
3. Wait for confirmation of accepted application.
4. Send peripheral blood or genomic DNA samples together with completed and signed Documents 1 and 2 to the INGEMM's postal address shown below – further information on required sample type is available at the Sample section.
5. In case that INGEMM has recommended the molecular analysis of index patient's relatives, in order to determine the putative co-segregation between the mutation detected and the pathological familial phenotype, the application for the performance of genetic study via email is not required, since it is directly accepted. However, shipping of Documents 1 and 2 is necessary, together with the samples of relatives.

## Mailing address

**Instituto de Genética Médica y Molecular (INGEMM)**  
**Edificio Quirúrgico**  
**Hospital Universitario La Paz**  
**Paseo de La Castellana, 261**  
**28046 MADRID**  
**ESPAÑA**

**NOTE:** The procedure and cost of shipping is to be covered by the applicant, and thus are not covered by the Dravet Syndrome Foundation. We will not accept any sample requiring any type of payment.

## Sample

The types of samples required are either blood or DNA. However, when sending samples from outside Spain it is recommended to choose DNA samples.

**Blood sample:** Peripheral blood (**Adult: 3-6 ml, Child: 1-3 ml, Infant: 1 ml**) in **EDTA** tubes clearly labelled. The blood samples should be sent in a temperature controlled container and should arrive at the INGEMM within a maximum of 24 hours post extraction. However, if the blood is maintained refrigerated or frozen, delivery time increases to a maximum of 72 hours.

**DNA sample:** Total amount of genomic DNA superior to 20µg and concentration superior to 50ng/µl. The patient's name should be clearly labelled on the tube.

