# Patient and/or Decision Supporter Information Leaflet

**Study title:** Research Use of Diagnostic Genomic Testing Data for Epilepsy

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| **Principal investigators names:** | **Professor Norman Delanty and Professor Gianpiero Cavalleri** |
| **Principal investigators titles:** | Professor of Neurology, Beaumont Hospital<br>Professor of Human Genetics, The Royal College of Surgeons in Ireland (RCSI) |
| **Telephone number of principal investigator:** | Prof Delanty 01 8092210<br>Prof Cavalleri 01 402 2146 |
| **Data Controller's/joint Controller's Identity:** | **Beaumont Hospital/RCSI** |

**Data Controller's/joint Controller's Contact Details:** dpo@beaumont.ie ; dataprotection@rcsi.ie

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| **Data Protection Officer's Identity:** | **Mr Mark Graham, Beaumont Hospital, Dublin 9**<br>**Tel: 01 8092162 or 01 8093392** |
| **Data Protection Officer's Contact Details:** | dpo@beaumont.ie; dataprotection@rcsi.ie |

**Introduction**

You are being invited to take part in a research study to be carried out at Beaumont Hospital by Professor Norman Delanty, Professor of Neurology, Beaumont Hospital and Professor Gianpiero Cavalleri, Professor of Human Genetics at the Royal College of Surgeons in Ireland.

Before you decide whether or not you wish to take part, you should read the information provided below carefully and, if you wish, discuss it with your family, friends or GP (doctor). Take time to ask questions – don't feel rushed and don't feel under pressure to make a quick decision.

You should clearly understand the risks and benefits of taking part in this study so that you can make a decision that is right for you. This process is known as 'Informed Consent'.

You don't have to take part in this study. If you decide not to take part it won't affect your future medical care.

You can change your mind about taking part in the study any time you like. Even if the study has started, you can still opt out. You don't have to give us a reason. If you do opt out, rest assured it won't affect the quality of treatment you get in the future.

## Why is this study being done?

We aim to understand which genes are causing epilepsy. We hope to describe common gene mutations we already know about and also discover new genes that might be causing disease.

As for any condition, our understanding of the genetic factors that contribute to epilepsy comes from the research domain. Such findings are translated to the clinical setting, to the point that today, genetic testing is a relatively standard diagnostic test in the epilepsy clinic. A result of genetics being used routinely in the clinic is that significant volumes of genetic data are now produced in the clinical setting. But such data is typically not used beyond the initial attempt to make a genetic diagnosis. This study proposes to bring those data in to the research domain, to enable further discovery which can then help clinicians to treat and inform their patients.

## Who is organising and funding this study?

This research is being conducted by Professor Norman Delanty of Beaumont Hospital and Professor Gianpiero Cavalleri of the Royal College of Surgeons in Ireland (RCSI). They are both investigators and collaborators of the FutureNeuro Research Centre (www.futureneurocentre.ie).

Established in 2017, and based in the Royal College of Surgeons in Ireland (RCSI), FutureNeuro is a world leading, Science Foundation Ireland (SFI) Research Centre which involves collaborations between academic partners (RCSI, Trinity College Dublin, Dublin City University, NUI Galway and University College Dublin.), as well as clinicians and industry.

The work is also funded from a variety of other sources including the European Union, local Irish Government agencies (Science Foundation Ireland and the Health Research Board) and charities (e.g. Epilepsy Ireland).

## Why am I being asked to take part?

You have been diagnosed with a form of epilepsy that is known or suspected to have a genetic cause.

## How will the study be carried out?

This study will take place in Beaumont Hospital beginning in June 2023. It will be ongoing long term research that will continue for many years. We don't have a specific end point. We are doing this because we believe that it will take many years to get a more comprehensive understanding of the genetic factors involved in epilepsy.

It may be recommended to you as part of your routine healthcare treatment that you give a blood sample to help investigate the causes of your epilepsy. If you agree to take part in this study then we will ask your permission to have access to your blood sample for research purposes. We will not ask you to provide a blood sample for research purposes only. We wish to gather the data that may already be generated as part of your medical care.

## What Will Happen to My Blood Sample?

Your sample will be labelled with your name and patient identifier number and will then be sent to outside laboratories abroad where they will be processed and analysed. These may include commercial as well as research laboratories in some cases.

Samples will be destroyed after they have been processed by the laboratories they are sent to. Your sample and the information extracted from it will not be used by the lab it is sent to for any commercial purpose and they will not share it in any way.

The laboratory your sample will be sent to is:

CeGaT GmbH
Paul-Ehrlich Strasse 23
D-72076
Tuebingen
Germany

Email: info@cegat.de Website: www.cegat.com Tel: 0049 70715654456

## What will happen to me if I agree to take part?

If your medical team recommend that you have genetic testing as part of your routine healthcare for your epilepsy they will take a routine sample of your blood at one of your hospital visits. If you agree to take part in the study you are agreeing to the information gained from analysis of your blood sample to be shared with the research team. Taking part in the research does not require you to have any extra samples taken or to do anything else.

## How Will My Blood Sample be Used?

Your blood sample will be used to extract your DNA so that we can look at your genetic makeup. We will use your sample to study your genetic makeup using markers like DNA, RNA and proteins and use various techniques to identify genetic causes of epilepsy.

All the data is treated completely confidentially and none of the researchers will have any of your personal details. Only the principal investigators and a very small number of nurses, doctors working within the study team will be able to relate your clinical condition to the results of the laboratory research.

It is important to remember that the main purpose of taking a blood sample from you is to help your medical team decide on the most appropriate treatment for your epilepsy. While the research study in itself may not benefit you directly, we hope the information we obtain from this study will give us better understanding of why and how genetic factors affect epilepsy and how we may be able to better treat it.

## What Will Happen in Respect of my Results?

It is possible that our work will identify a genetic or molecular change that could explain why you have epilepsy or that might be useful in guiding the treatment of your epilepsy. It could also reveal potentially important clinical information which is unrelated to your epilepsy, but potentially related to the risk of another health condition as explained in the section in this leaflet on risks of genetic testing (p.6 below).

You are therefore given the option to receive results and the nature of the results you would like to receive from this study. Your options are to:

A. Receive results.

Or

B. Receive no results at all, even if clinically significant.

If you choose to receive results (i.e. option A), you then have the choice of receiving results regarding:

(i) your epilepsy

and/or

(ii) actionable secondary findings.

Should you have chosen to receive results, any important findings will be passed on to Prof. Delanty (or your referring physician). Should Prof. Delanty (or your referring physician) agree the finding is clinically significant, after doing further testing to confirm the result he will arrange a face-to-face meeting with you to explain the situation and offer any relevant medical referral. If we identify a result and you have chosen to receive results, to help us understand the result better, we may contact you about the possibility of testing your parents, where appropriate.

*Will researchers be looking at my medical records?*
Members of the research team will access details of your treatment and care through your clinical paper records, and electronic records at Beaumont Hospital. We may also wish to ask you a couple of questions in relation to your treatment response. In order to keep our records complete and up to date and/or to inform you about related projects, members of our team may wish to contact you in the future.

*Will my medical records be private?*
Prof Delanty will only give researchers access to clinically relevant details about you. Personal information that could identify you will not be given to the research team.

## Video/and or Audio recordings?

There will be no recordings made.

## What other treatments are available to me?

Taking part in the study will have no impact on your medical treatment.

## What are the benefits?

If your medical team recommend that you have a genetic test it is because they think there is the possibility that a genetic cause for your epilepsy may be found. If so, this may be of benefit in your future treatment. Genetics is an evolving science and new genes are being discovered all the time. Even if we do not discover a genetic cause for your epilepsy now, one may still be discovered in the future. Epilepsy is complex and there are many reasons for it to occur. By

contributing your genetic and health information you are helping researchers learn more about the causes of epilepsy.

## What are the risks?

### Are there any risks involved in giving a blood sample?

Whenever a blood sample is taken there may be a very small risk of mild discomfort, bruising, feeling faint or fainting, or, very rarely, infection at the blood collection site. However, taking blood samples is a very safe procedure and we do not anticipate any complications with this.

### Are there any risks to genetic testing?

There are some risks to genetic testing in general. Because we are studying a larger number of your genes than would have been possible in the past through clinically indicated DNA sequencing (exome, whole genome etc., as indicated by your doctor), it is possible that our work will identify a genetic or molecular change (variation) that:

- Could explain why you have a particular medical condition or that might be useful in guiding the treatment of your medical condition.
- Could reveal potentially important clinical information unrelated to your medical condition, also known as 'secondary findings'.

'Secondary findings' are findings that have been interpreted as having significant implications, and for which treatments exist that could prevent development or progression of that condition. Examples of actionable secondary findings we could potentially detect in a person's genetic profile include hereditary breast and ovarian cancer. Where someone is at increased risk of hereditary breast cancer, they could consider a mastectomy to reduce that risk of cancer occurring. Another example is that it is possible that genetic testing may reveal that you are not related to one or more of your family members, for example we might discover that your father is not who you expected him to be.

Note these are just examples, we are not mentioning them because they are likely to happen.

As a participant in this study, you have the option whether or not to receive findings, if found. Further details of how these findings will be delivered back to you can be found on pages 4 and 5 above.

This information may be beneficial to you and your wider family's health and future treatment options. However, it may also bring an emotional bearing on both you and the wider family. As a study participant you need to weigh up these factors and decide wither or not to receive any

findings if they are found during the study. It is important that you fully understand what you are consenting to and you may wish to discuss this further with a family member, friend or GP.

**Social and economic risks**

There is also the possibility of social and economic disadvantages from genetic information being shared in the wrong way and it could affect you and your family, such as if an employer or insurance company was to obtain the information. This is termed 'genetic discrimination' which is illegal under Irish law.

We will do our utmost to guarantee complete confidentiality but, however unlikely it is, we are obliged to tell you that there is a theoretical risk of this information becoming available to others. We can, however, reassure you that we are compliant with all data protection rules and regulations and we use the most up to date IT methods and facilities to protect participants' data.

**Data Breach Risks**

As with any study involving health information, the risk of data loss or misplacement is always present. In this unlikely event of this occurring, it may be possible for participants to be identified. The project team are taking every step to reduce the likelihood of such an event occurring including the storing of data in a pseudonymised manner. This means that you will be given a project code as opposed to using your name. This ensures that your personal information, consent, health information and data cannot be traced back to any identifiable information such as your name. The keys to these codes are stored in a separate password protected file and **only the necessary** research staff have access to this. All research material will be kept in secure storage devices managed by RCSI. For further information, please refer to the *Data Protection section* on pages 8 and 9 below.

## What if something goes wrong when I'm taking part in this study?

This study involves analysing your blood sample in a laboratory. You will not undergo any procedures as part of this study. Your medical team will be in charge of your care at all times and will look after you in the unlikely event that you feel unwell when they take your blood sample.

If you have opted to be informed if an actionable secondary finding occurs, your consultant will also be contacted to liaise with and inform you about the nature of this finding. Further information on this is found on pages 4 an 5 above.

## Will it cost me anything to take part?

There will be no costs or payments involved.

**Is the study confidential?**

**Yes.** All information collected about you during the research study will be kept strictly confidential. -Your name, address, hospital number or date of birth will not be published or passed on in any way.

*Will you be contacting my GP or any other healthcare provider?*

We will not automatically tell your GP or any other healthcare provider about your participation in this study. However, we can do so if you wish.

*What information about me (personal data) will be used as part of this study? Will you be looking at my medical records?*

In line with the consent which you will provide to us if you wish to participate, we will be collecting a range of personal information about you, your medical condition and genetic profile.

Firstly, the researcher nurse, on behalf of Prof Delanty, will keep some information such as your name, email address, telephone number, postal address so that they can contact you over the course of the project to; (i) post/email study related materials, (ii) make arrangements for the return of results, if applicable, (iii) allow researchers to re-contact you as the research progresses.

Special Categories of personal data includes sensitive data about you. For this study, we will be collecting the following special categories of data: gender, ethnicity, genetic data, and data concerning health. Data concerning health may include; name of Consultant, medical record number (MRN), condition, medications information, intervention history (e.g. surgeries, implant etc.), any known drug allergies, comorbidities, other physical and mental health conditions and medical imaging such as MRI, or CT scans.

The collection and use of personal data will go no further than is necessary for us to achieve the goals of this project, as outlined on pages 1 and 2. **All information handled in the course of this project is strictly confidential and will not be made available to anyone other than those directly involved in this research project.** Any clinical details coming to the attention of the researcher about you or your treatment will be managed in strictest confidence by members of the research team. The researchers themselves will not be able to identify you from your genetic information alone.

*What will happen to my personal data?*

All research work will be pseudonymised. A special code linking your name to the sample will be held by Prof. Delanty and RCSI Biobanking Service to allow updating of clinical information only in respect of the current research project. It will not be used for identification for any other purpose.

In recent years, large databases of genetic sequences have been set up to help researchers. For some of our collaborating projects, the funding bodies prefer that de-identified genetic data obtained be shared with other researchers to bring greater benefits to society. In some cases, de-identified genetic information from your DNA samples may be made available to researchers working separately from us at other hospitals, universities or companies for use in research investigating the genetic cause of epilepsy and related disorders. This is unlikely to be of any direct benefit to you. Your age, gender and brief information about your seizures may be included with the genetic data, but no identifying information will be included. However, genetic information is unique to each individual and, therefore, there is some inherent risk of identification. For example, people not involved in our study who have information about your DNA could potentially identify the participant by comparing your genetic DNA to the information we make available. We believe this is unlikely to happen. By consenting to the research, you acknowledge that you are aware that such an event could occur.

*How long will you be keep the information about me?*

Your sample will be retained until we are satisfied that adequate genetic data has been obtained from them, after which point the sample will be destroyed.

Your data from this specific project will be retained indefinitely, or until the study is deemed complete. But we would like to use anonymized information resulting from your participation again for other, future ethically-approved research.

*Where will you be sending my sample and information about me?*

As part of this study we will send your identifiable blood sample for analysis to another institution with whom we have a formal agreement in place outlining how the sample and data is to be securely stored, for what specific project it is to be used and how the sample is to be returned or destroyed. Such an institution could be overseas and could be in industry. We will not be sending identifiable information about you anywhere else. The data generated from analysis of your sample will be stored within RCSI secure computer servers indefinitely, or until the study has been deemed complete.

*Will you be publishing the results of this study in medical journals?*

The results of our study may be published at a later date, your name or number will not appear in the publication. Unless you have chosen to receive results (see above) you will not be informed of individual results from this study.

*Will you be presenting the results of this study at medical conferences?*

The results of our study may be presented at medical conferences, your name or number will not be presented at the conferences. Unless you have chosen to receive results (see above) you will not be informed of individual results from this study.

The results of these studies may be published at a later date, but individual patients will not be identified in any publication.

**Future Research Studies**

You will notice on the consent form that we are giving you some options about how you would like our findings about you used in the future. We would like to keep open the possibility that we could use what we learn in future studies, both those that are related directly to finding genetic causes of epilepsy as well as those that have a different focus (for example, therapies or treatments). If you agree to future uses then we will not have to seek your consent again unless you wish us to, however all future research studies would have to go through a strict ethics approval process.

*Results of the Study*

The results of this study will be used as a basis for future research. If we find things that are deemed interesting by researchers from other hospitals and universities, the results may be published in a scientific journal. If this happens, no one will be able to identify you as having taken part in the research, as all information about patients will be anonymous.

We are also committed the principle that useful data should be shared for research purposes so that the research community can continue to learn and make scientific and medical advances that can improve people's lives in the future. Therefore at the end of the project we plan to put anonymous information in an appropriate, cost-free data repository, whose security policy has been written according to best practices, for indefinite data retention and to ensure that the research community has long-term access to the data.

**Data Protection**

1. The data will be processed for the purposes of scientific research (in accordance with the European General Data Protection Regulation (GDPR) and the Irish Data Protection Act.

2. We will rely on Article 6.1(e) 'public interest' and Art 9.2 (j) 'scientific research purposes' of the General Data Protection Regulations to process your personal data.

3. Your data will be shared with: your medical team, a team of researchers from The Royal College of Surgeons in Ireland, the RCSI Biobanking Service, and the specialist laboratory we use to do the analysis and extract the genetic information. The name of the laboratory is: CeGaT GmbH, Paul-Ehrlich Strasse 23D-72076, Tuebingen, Germany.

   Email: info@cegat.de Website: www.cegat.com Tel: 0049 7071565445

4. Your data will be stored for an indefinite period of time. We intend to keep all our data indefinitely because we believe that this is the best way we can build a comprehensive database about the genetic causes of epilepsy.

5. While the strict measures are put in place to protect data relating to you, the handling of your data may result in a data breach. This could cause you harm by impacting on your right to privacy. Precautions and safety measures have been put in place to protect your privacy as described above.

6. You (the data subject) have the right to withdraw consent. Participation in this research project is entirely voluntary and you may refuse to take part now or at a later stage without any effect on your current or future treatment. If you wish to have your information deleted, the study team will do so. You will be able to do this by contacting Professor Gianpiero Cavalleri at FutureNeuro, The Royal College of Surgeons in Ireland, (gcavalleri@rcsi.ie or 01 4022146).

7. You have the right to lodge a complaint with the Data Protection Commission, 21 Fitzwilliam Square South, Dublin 2, Ireland. www.dataprotection.ie.

8. You have the right to request access to your data and a copy of it, unless your request would make it impossible or make it very difficult to conduct the research.

9. You have the right to restrict or object to processing of your data, unless the request would make it impossible or make it very difficult to conduct the research.

10. You have the right to have any inaccurate information about you corrected or deleted, unless the request would make it impossible or make it very difficult to conduct the research.

11. You have the right to have your personal data deleted, unless the request would make it impossible or make it very difficult to conduct the research (for example, if you request to delete data at the end of a research project just before it is due to be published).

12. You have the right to data portability, meaning they have a right to move the data from one controller to another in a readable format.

13. This research does not involve any automated decision making or profiling.

14. The results will be used as a basis for future research and they may be published in a scientific journals. If this happens, no one will be able to identify you as having taken part in the research, as all information about patients will be anonymous.

15. Your data may be transferred to a country outside of the EU for further analysis. It will be coded and anonymous and data sharing agreements will be put in place to protect

your privacy prior to any transfer of information.

**Consent to Future Uses**

It is possible that future studies of epilepsy not described in this leaflet may be done. These will only be done after review and approval by an ethics committee. Consent to use your data in related future research is voluntary and you have the right to opt out of your data being used for future research.

In the consent form you will be given a number of options about whether to receive future contact from the research team. You will be asked whether you would like to be contacted about future research that is related and unrelated to this study. You may or may not want to receive more information from the research team, or you may decide that you would like to keep informed about further epilepsy studies. Take time to think about these options and we will be happy to answer any questions you may have.

The consent form will give you another set of options, this time about whether you would like your data to be stored with or without us contacting you for further consent. You may or may not want us to contact you again looking for consent. Please be assured that whether or not you choose to be contacted by us, Beaumont Hospital Ethics Committee must give approval for any other research using your data.

The consent form will also bring to your attention that some future research projects may be carried out by researchers working for commercial/pharmaceutical companies. You may or may not want your data being used by a commercial company and you are free to disagree to this. If you are happy to have a commercial company use your data for epilepsy research, please note that there will not be any financial compensation for this.

**Where can I get further information?**

Thank you very much for taking the time to read this lengthy information form. We really do appreciate your interest and involvement.

If you need any further information now or at any time in the future, please contact:


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| Name | Professor Norman Delanty |
| Address | Department of Neurology, Beaumont Hospital |
| Phone No | 01 8092210 (office hours) |

**Thank you for taking the time to read this leaflet. You can keep this information sheet and a copy of the consent form.**