



MRC Prion Unit at UCL, Institute of Prion Diseases
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National Prion Clinic
Courtauld Building, 33 Cleveland Street, London W1W 7FF

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Director: John Collinge CBE, FRS

Diagnostic tests in prion disease and related disorders

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In order to help diagnose the cause of your illness, a blood sample can be taken to screen genes for abnormalities (mutations) that are associated with prion disease and related disorders. This routine diagnostic test may help to establish the precise cause of your symptoms. We would like to use this blood sample for research with your permission.

Genes, prion disease and related disorders

Most prion diseases are not caused by inherited abnormalities in genes and so do not 'run in families'. However, in a very small number of families, a change in a particular gene can make prion disease more likely. We know that some people can inherit an altered gene for a protein which means that they are at higher risk of prion disease. Analysis of your blood sample may determine whether you have an altered gene that could explain your symptoms.

Genetic tests for diagnosis

If you agree, the blood sample (approx. 10ml) will be taken and used to analyse genes for abnormalities (mutations) associated with prion disease and related disorders. The blood test is taken from a vein in your arm. This may cause some discomfort but the sample is usually taken quickly. Sometimes there can be some bruising after a blood test is taken.

The analysis of these genes in your blood sample is a complex process which takes time and it could be several months before a result is available. Your doctor will be told the result of the diagnostic test and he or she will communicate the result to you and your GP.

Potential benefits and disadvantages of genetic testing

- The main benefit of carrying out this gene analysis is that if an abnormality is found in one of your genes then your doctor may be able to make a definite diagnosis of the cause of your symptoms.
- If you find out that you have an abnormal gene then you will know that your children and other members of your family could also have the abnormal gene and have a higher risk of having similar disorders.
- At present, having the genetic tests will not affect your ability to obtain insurance or a mortgage. You should be aware that it is possible that in the future companies could ask whether you have had such tests.
- Before deciding to have gene analysis you need to decide for yourself whether the benefits of having this test outweigh any potential disadvantages to you.
- The collection and processing of personal data for clinical purposes will be done in accordance with the General Data Protection Regulation and UCLH policies: <https://uclh.nhs.uk/aboutus/Pages/Cookiepolicy.aspx>

Research into prion disease and related disorders

Please read the important information overleaf about how you could help with research into prion and other brain diseases.



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Using your sample for research

After diagnostic tests have been done your sample (or your child or relatives' sample) could, with your consent, be kept for use in research studies into prion and other brain diseases. Such research is of great value in understanding the causes of prion and other brain diseases. It is because of previous donations of samples for research that diagnostic tests for conditions have been developed. We hope that future research will help develop tests and treatments for many such brain diseases.

If you agree to donate your sample for research it could be used in studies that are being or will be performed into the causes of and possible treatments for prion and brain diseases, which may include genetic studies. These studies could be within the Institute of Prion Diseases or in other organisations.

All research projects are approved by a Research Ethics Committee before any samples are used. Your sample would be used anonymously and any research results that are published would not identify you in any way. You can request full details of our research projects from us at the address below or look at our [website: www.nationalprionclinic.org](http://www.nationalprionclinic.org)

We are studying which genes are involved in prion diseases and related disorders and how genes can affect the type and patterns of these diseases. This will help to determine how many people could be at risk of prion disease and related disorders. It is not yet known what effect some of these genes have on diseases and so we do not inform participants of individual results. Such research could ultimately lead to tests and treatments for disease being developed commercially for clinical use which would benefit future patients. Those donating samples would not benefit financially from such development. Samples such as these are a very useful way of studying brain disease and so your sample could be kept indefinitely and be used for future studies in the same area, which will always have approval from a Research Ethics Committee.

It is entirely up to you whether you agree to the sample you give being used for research and neither the diagnostic tests nor your medical care or legal rights will be affected by your decision about research. You can change your mind about taking part in this research at any time and ask that your sample be disposed of. **Please ask your doctor any further questions you may have.**

Information relating to research samples

In order to use your sample for research we need to keep some data about you on our research database, including your name, details of your illness and the result of the diagnostic tests. This information is stored on a confidential, secure database in the Institute of Prion Diseases. Professor Collinge is responsible for security and access to the database. Only authorised scientists, doctors and administrators directly involved in relevant research with ethical approval are allowed access to this data. To increase confidentiality, your sample will not have your name on it, simply an identifying number or codes. Under the Data Protection Act 2018 you are entitled to ask what information is on the database about you and also to request that that information is removed. UCL is the sole Data Controller for personal data processed for research purposes.

You should understand that while it will not be possible to remove your individual data from previously analysed and reported results, your sample will not be used in any further analyses or studies. Requests for access to your information should be referred to: data-protection@ucl.ac.uk or the National Prion Clinic: www.nationalprionclinic.org.



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Request and consent for diagnostic prion and related disorders genotyping

All fields must be completed before sample(s) can be processed and accompanied by details of clinical information including the family mutation for predictive testing.
 Please also send contact details for return of results.
 Please give a copy of this consent form to the patient and insert a signed copy in the records.

1. Statement of health professional (with appropriate knowledge of tests and counselling)

- I have explained the procedure and nature of the requested tests to the patient **or**
- The patient lacks capacity and I consider the tests to be in his/her best interests

Signed..... Date.....
 Name (PRINT)..... Job Title.....
 Consultant (in charge)* Hospital.....
 *to whom results will be addressed

2. Consent of patient to routine diagnostic genetic tests

Please tick either diagnostic or predictive box

- Diagnostic tests:** I consent to the genetic tests above. I confirm that I understand the implications of these genetic tests
or
- Predictive tests:** I confirm that I have undergone genetic counselling and understand the implications for myself and my family
or
- Store** Store blood for possible future diagnostic genetic tests

Patient/Parent/Guardian/Nominated consultee/Witness signature.....
 Name (PRINT)..... Date.....

For those patients willing to donate their diagnostic sample for research, please complete the consent form overleaf.

PLEASE TURN OVER

patient label or details

Name.....
 Address.....

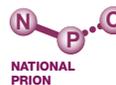
 DoB..... Hosp no.....

lab use only

Sample ID.....
 Specimen type: Blood DNA
 other.....
 Received: date /.... /..... time.....



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Research into prion disease and related disorders

I agree that in addition to routine diagnostic tests my blood sample may be used for research into these diseases and their genetic associations. I understand that this research will not necessarily provide information that will be of direct benefit to me. A relative, carer or nominated consultee can sign this form if the patient is unable to take their own decisions and if they do not believe that the patient would have objected to taking part in this research.

Please tick box to indicate agreement to each part:

- I have read and understood the information sheet (v2.5 08/10/18) about this research and have had the chance to ask questions about it.
- I understand that donating my sample for this research use is voluntary and that I can withdraw my permission at any time, without having to give a reason and this will not affect my medical care or legal rights in any way.
- I agree that my sample can be analysed for research into prion and related diseases using genetic or other tests. I understand that the results of such research will not be available to me on an individual basis and that any published results will not identify me.
- I agree that relevant sections of my medical records can be looked at by senior responsible individuals from the Institute of Prion Diseases or Regulatory Authorities. I understand this would only be done if it was relevant to the sample being used in research and also that my confidentiality will be respected.
- I agree that information about me that is relevant to the research may be held on a secure confidential database that is kept in accordance with the General Data Protection Regulation and that UCL is the Data Controller of my personal data. I understand that I can request to see that information if I wish, and that the sample I give for research could be used in the development of diagnostic tests and treatments with academic or commercial collaborators in the UK or worldwide, but that my personal details will not be provided to them. I understand that neither I, nor my legatees will benefit financially in this case, and that such research will always have approval from a Research Ethics Committee.
- I understand that my sample is a gift to the Institute of Prion Diseases that decisions about its future use will be made by senior members of staff and that my sample may be used in future studies subject to the same conditions as in the points above. After research studies have been completed, if the sample is not retained, the Institute will arrange lawful and respectful disposal of the sample.

I consent to the use of my sample for research and have read the information overleaf relating to this **or**
 The patient cannot give consent for themselves. I do not believe s/he would object to the sample being kept for research use

Signature.....Relationship to patient.....
 Name (PRINT).....Date.....
 Person taking consent.....Signature.....
 Position:.....Date.....

Statement of interpreter (if used)

I have interpreted the information given to the patient to the best of my ability and in a way which I believe s/he can understand

Signed.....Date.....
 Name (PRINT).....Contact details.....