CONSENT FORM - GENE TESTING TO CONFIRM A DIAGNOSIS OF HUNTINGTON DISEASE

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I understand that it is possible to have a test to see whether or not my (my relative's) symptoms are due to the gene change which causes Huntington disease (HD), and I wish to proceed with this test. I understand that the test can have three possible outcomes:

1) The gene change is found, confirming a **diagnosis of Huntington Disease** and a 50% risk of passing the HD gene on to my (my relative's) children
2) The gene change is not found, **excluding a diagnosis of HD**
3) The result is **borderline** which is more difficult to interpret

NAME (**block capitals**)……..……

ADDRESS……..…..…… …

... … … … … … … … … … … DATE ……HOSPITAL or NHS NO ……

Signature of patient ……...
AND
Signature of spouse / partner ……. (not essential but preferred if applicable)
OR
Signature of next of kin……...…….
(if the patient is unable to give informed consent)

For medical staff:

I have explained the principles and implications of **diagnostic HD** testing to the above, as detailed in the supplied information sheet, and have sent a copy of this form to the DNA laboratory. **I confirm that the patient has motor symptoms and/or signs consistent with a diagnosis of HD, and that this is NOT a PRESYMPTOMATIC test.** (Presymptomatic testing of those at risk of HD but with no definite physical signs is governed by an internationally agreed protocol. It is offered only in Genetic centres where pre-test genetic counseling and long term supportive follow up is available.)

Name (in capitals) ……. Signature ……..…….

Specialty……. Hospital…….....

PLEASE SEND A COPY OF THIS FORM TO THE REGIONAL MOLECULAR GENETICS SERVICE AND PLEASE FILE THE ORIGINAL IN THE PATIENT'S NOTES.

INFORMATION SHEET - THE GENETIC TEST FOR CONFIRMING A DIAGNOSIS OF HUNTINGTON DISEASE

What is Huntington disease (HD)?

HD is an inherited neurological disorder which causes abnormal movements and difficulties with thinking and behavior. The children of someone with HD each have a 50% chance of inheriting the condition themselves. The age at which HD develops is very variable, even within members of the same family; symptoms usually start between the ages of 30 and 50, but can vary from teenage years to sixty or even later.

Most people with HD have other affected family members. However, we now know that some people can inherit HD even though they do not have an affected relative.

If your doctor thinks you may have HD, a genetic test can help to confirm or rule out this condition as the cause of your illness. An accurate diagnosis is important so that you can be given the appropriate treatment for your condition. It is also important because a diagnosis of HD affects your family, particularly your children, and they would probably like to know about this for their own benefit, particularly if they have children of their own or are thinking about starting a family.

What is the basis of the test?

The genetic abnormality which causes Huntington disease was identified in 1993. The HD gene contains a section of genetic code which varies in size from one person to another, and is longer in people who have HD than in people who do not. The laboratory can measure the size of this genetic segment in genetic material prepared from a blood sample.

Is the test reliable?

Yes. Most people get a result which is definitely normal or definitely abnormal. However, a small number of people fall into a "grey area" between the normal and the abnormal range. Some people with an HD gene in this intermediate range develop symptoms, while others do not. If your result falls in the intermediate range, it can be quite complicated to decide whether or not your symptoms are due to HD.

What happens about my children if I do have HD?

They will benefit from genetic counseling (if they wish), which can be arranged at St. Mary's Hospital or at another genetic centre if this is more convenient for them. The purpose of genetic counseling is to give them an opportunity to ask their own questions about the condition and the help available to them. It is also possible to test people at risk of HD (over the age of 18 years) to see if they have inherited the abnormal gene or not, because some people in this situation prefer to know rather than live with uncertainty. This is called Predictive or Presymptomatic testing. There is currently no treatment which prevents HD developing, or stops it getting worse, but there is a great deal of research going on in this area. It is also possible to choose not to pass on the abnormal gene by having tests during early pregnancy or by using IVF and PGD (Pre-implantation genetic diagnosis). The main reason for producing this information sheet is because the diagnosis of HD has a major impact on people who have the illness and on their families. For this reason, we would like to have your written consent to test your blood sample for the HD gene if you want to have the test, and will therefore ask you to sign a consent form.