Information sheet for patient's relative/carer

Title of study - Identification of biomarkers that can be used to track the progression of Huntington’s disease (HD)

We would like to invite your relative to help us with a research project to try and find a blood or urine test that will help us track the progression of disease in HD. This is of great importance as therapies become available for testing in patients. This marker may let us know whether a possible therapy was being effective in a particular patient.

Our understanding of the causes of Huntington’s disease has increased dramatically over the past five years. In hand with this has been the development of a number of possible therapies for the disease. These therapies have been tested and developed in animal models of HD. Already, a number of compounds have shown to be effective at slowing the disease in mouse trials and we are entering a phase in which there will be many candidate drugs that could be tested in the clinic.

However it is very difficult to determine how effective a therapy is being over time because the bedside examination we use in patients is not sensitive enough to pick up significant changes over short time periods. Therefore the identification of biomarkers that could also be used to track disease progression would be invaluable.

In addition, markers capable of detecting disease related changes in otherwise presymptomatic individuals (these are patients who have had a positive gene test but do not yet show any symptoms of the disease) will be essential for the future detection and monitoring of treatments that can delay onset.

Therefore this study aims to identify biomarkers that can be detected in peripheral blood or urine and used to track the progression of HD in presymptomatic and symptomatic individuals. We have already found some potential candidates in a mouse model of HD and we want to see if these are present in patients.

We are asking if your relative would donate a sample of blood for this purpose. We would need 20-30 ml (4-6 teaspoonfuls) of blood, which would be taken in the usual way from a vein in your arm. There is a small risk of discomfort, bruising and bleeding with this procedure. We also ask if your relative would donate 50ml of urine. Their blood and urine samples will help us to develop a test that can track the progression of disease in HD. Their blood and urine samples will be compared with those from people who do not have HD to look for differences.
This is a research study, the significance of any test results is not known, and neither you, nor your relative, will be told the results of their blood or urine test. We will collect the following data at the time of their sample collection – your relative's name, clinical information about your relative's disease, and the result of their genetic testing. This data is necessary for us to be able to try and link up the stage of their disease with the presence of absence of a particular marker in blood or urine. The data will be stored at the Institute of Neurology, UCL by Dr Sarah Tabrizi who will be responsible for security and access to the data. Following the collection of sample data, their urine and blood samples will be anonymised, and will not be able to be linked to them.

Your relative's blood sample may also be used as a control sample for other studies that are looking at biomarkers of disease in related conditions to Huntington's disease. No additional testing will be performed on their blood sample that you have not consented for. You should also understand that there is no personal financial gain to yourself or your relative, now or in the future, should this research result in a biomarker being developed for use in HD therapy trials, even if this involves collaboration with a commercial Company.

Their participation is entirely voluntary. You are free to decline at any time without having to give a reason and this will in no way affect your relatives' future medical care. Participation in this study will in no way affect either yours' or your relatives' legal rights.

This research project has been reviewed by The National Hospital for Neurology and Neurosurgery and the Institute of Neurology Joint Research Ethics Committee.

Thank you

Dr Sarah Tabrizi
Professor Gillian Bates
Dr Elaine Holmes
Contact Number 020 7837 3611 Ext 3839