The title of the study is "Identification of biomarkers that can be used to track the progression of Huntington's disease (HD)."

We would like to invite you 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 HD 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, urine, buccal smear (cheek) cells or cerebrospinal fluid (CSF) and used to track the progression of HD in presymptomatic and symptomatic individuals.

We are asking if you would donate a sample of blood for this purpose. We would need up to 100ml (approximately half a cup) of blood, which would be taken in the usual way from a vein in your arm. We would only take the maximum amount of 100mls if you do not have any other medical problems, such as anaemia or heart disease. There is a small risk of discomfort, bruising and bleeding with this procedure.

We also ask if you would donate 20ml of urine. Your blood and urine samples will help us to develop a test that can track the progression of disease in HD. Your blood and urine samples will be compared with those from people who have HD, or have had a positive gene test but no symptoms, to look for a difference.

We also ask for a ‘buccal smear’ which is a painless swab of the inside of the cheek, to collect cheek cells. The purpose of this is to see whether these cells, rather than cells extracted from blood, might be suitable for the kinds of biomarker test mentioned above.

This is a research study: the significance of any test results is not known, and you will not be told the results of your blood or urine test. We will collect the following data at the time of your sample collection – your name, clinical information about your disease, and the result of your
genetic testing. This data is necessary for us to be able to try and link up the stage of your disease with the presence or absence of a particular marker in blood or urine. The data will be stored at the Institute of Neurology, UCL by Professor Sarah Tabrizi who will be responsible for security and access to the data.

Some people will be invited to donate a sample of cerebrospinal fluid (CSF). If you are being invited to donate CSF, you will be given an additional information sheet and a full explanation of the procedure.

Your blood sample may also be used for other approved studies (UK and/or world-wide) that are looking at biomarkers of disease in related conditions to HD. No additional testing will be performed on your blood sample that you have not consented for. You should also understand that there is no personal financial gain to yourself 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.

DNA from your blood sample may be used for epigenetic analysis, looking at chemical modifications to the DNA that might be useful for monitoring disease progression. This may be performed in the UK or overseas. The analysis will have no clinical significance and will be returned to Professor Tabrizi but not reported to you.

Your 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 future medical care. Participation in this study will in no way affect your legal rights.

If you are harmed by taking part in this research project, there are no special compensation arrangements. If you are harmed due to someone’s negligence, then you may have grounds for a legal action but you may have to pay for it. Regardless of this, if you wish to complain, or have any concerns of this study, the normal National Health Service complaints mechanisms should be available to you.

Optional component: ECG and echocardiography

ECG recording is a painless procedure that involves applying adhesive electrodes to the skin and making electrical recordings of the activity of the heart. It takes about 5 minutes. Echocardiography is a painless procedure that allows live measurements of the functioning of the heart using ultrasound. It takes about 30 minutes. You may be invited to exercise on a treadmill before the echocardiogram. This will only be done when the risk is negligible. For ECG and echocardiography, you will need to remove the top half of your clothing. It’s possible that these investigations might reveal findings of clinical significance. If this happens, you will be informed and offered an NHS cardiology assessment.

Optional component: Optical Coherence Tomography (OCT)

OCT uses infrared light in a similar way to ultrasound to build up a picture of the nerve layer at the back of the eye. It is a harmless and painless eye test that is carried out in the Eye Department. It does not involve the use of radiation and takes five minutes to complete. We will also need to undertake a visual assessment including a taking a history of any eye problems and carrying out an examination which will comprise a test of near and distance vision, colour vision and the range of vision - the standard assessment you may be familiar with from opticians. If the initial data is promising we may invite you back for up to two further visits, likely to be 6 months and 1 year apart. We will store the information we collect safely and without any personal identifiers by assigning a nine digit number to the data. In the unlikely event that the pre-OCT test or OCT test shows something relevant to the health of your eyes, advice will be given, and with your permission we would pass on our observations to your GP and if necessary to Ophthalmologists, in line with standard clinical practice.
This research project has been reviewed by The National Hospital for Neurology and Neurosurgery and the Institute of Neurology Joint Research Ethics Committee (ref: 03/N008).

Thank you for taking the time to read this information sheet

Professor Sarah Tabrizi
Contact number – 0203 456 7890 Ext 723420