



## **The Genetic and Functional Characterisation of Spinocerebellar Ataxia type 11**

**Principal Researchers:** Dr Henry Houlden, Dr Paola Giunti, Professor Nicholas Wood, Institute of Neurology, University College London

Researchers at the Institute of Neurology recently identified the gene causing spinocerebellar ataxia type 11. This same team now have funding from Ataxia UK to study this gene further (see details of project in the section on Research funded by Ataxia UK). Part of the project involves screening patients to try and find more individuals whose ataxia is caused by defects in the SCA11 gene. They are therefore seeking individuals who have been diagnosed with cerebellar ataxia of unknown cause who would be willing to be tested for SCA11.

### **Who can take part in this study?**

Any individual who has been diagnosed with cerebellar ataxia and the cause is unknown (probably meaning you had a negative result when tested for other spinocerebellar ataxias). The researchers are particularly interested in hearing from individuals with ataxia who also have relatives with ataxia (eg: parents, siblings).

### **What is involved?**

First of all, you will need to contact Dr Houlden (contact information below) to express an interest in participating in this project. Then you will need to ask your GP to send a blood sample and consent (consent form and patient information sheet) to have your DNA examined for genes that are associated with spinocerebellar ataxia or familial cerebellar ataxia.

As a result of taking part in this study you may get a specific diagnosis. The testing is currently done on a research basis and not as a diagnostic service. In all cases we screen the SCA11 hotspot where the known mutations are found in some familial cases we screen the whole gene. A diagnosis would only happen if you are found to have SCA11. If you are negative to SCA11 you will also be told through your doctor.

### **Contact details:**

If you would like to find out more about this project or take part please contact: Dr Henry Houlden, Institute of Neurology, Queen Square, London WC1N 3BG. Telephone: 0207 837 3611 x4391