ARSACS STUDY CALL FOR PARTICIPANTS

Volunteers needed to take part in autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS) and other ataxias using optical coherence tomography (OCT).

What is the purpose of this study?

The first aim of this project is to investigate whether OCT could be used as a screening test to detect people with ARSACS, to decide whether to send them for a formal genetic test. We have studied a family with a genetic diagnosis of ARSACS in which some of the unaffected carriers also had changes seen on OCT. It is therefore vital to know whether these retinal changes are a reliable diagnostic indicator amongst the population of people who are routinely assessed in an Ataxia Clinic. It is therefore important to study patients with ARSACS and the unaffected family members of patients with ARSACS who might be carriers of the gene. We will perform the same test on patients with other (usually genetic) types of ataxia (such as Friedreich's ataxia or spinocerebellar ataxia) in order to test the sensitivity of OCT in detecting cases of ARSACS.

The second aim of the project is to understand better the clinical and other features of people with ARSACS, so that these can be recognized in the clinic leading to a more accurate diagnosis. To this end, we will ask you to have a neurological examination. We may also ask you to have an MRI scan of the head and neck, and/or a test of memory and thinking called neuropsychometry.

The third aim of the project is to study the retinal anatomy of patients with other causes of ataxia using OCT, and to link these to the features seen on routine ophthalmological and neurological examination.

What will happen to me if I take part?

If you do decide to take part you will be asked to attend a consultation at the National Hospital for Neurology and Neurosurgery where you can ask further questions and where consent will be sought for participation in the study. In the same consultation, participants will undergo a medical check-up including the Scale for Assessment and Rating Ataxia (SARA) score, the Inventory of Non-Ataxic Symptoms (INAS), 9-Hole Pegboard (9HPB) and 8-metre Timed Walk (8mTW), and a series of questionnaires will be filled out.

We will perform an ophthalmological examination including assessment of visual acuity using a Snellen chart, assessment of colour vision using Ishihara colour plates, visual examination of the back of the eye using an ophthalmoscope, examination of the visual fields by Goldmann perimetry, photography of the back of the eye, and assessment of the retinal nerve fibre layer using OCT.

If a genetic diagnosis or carrier status for the ARSCAS gene is not known, you will be asked to give a small blood sample for genetic testing (approximately 6ml). If we perform a genetic test, you will be asked if you want to know the result.

We may also, either at the same visit or on a subsequent occasion, ask you to have an MRI scan of the head and neck.

Do I have to take part in all parts of the study?

No, it may well be that some people just have one or other parts of the study (e.g. OCT, MRI, neuropsychometric assessment), although we would obviously like to have as much of a comprehensive picture of ARSACS as possible. However, not taking part in one or more parts does not debar you from participating in the study.

What are the possible benefits?

You will be fully assessed by both a neurologist and an ophthalmologist, who can advise you on the neurological and ophthalmological aspects of your diagnosis. We hope the study will help improve the diagnostic process for patients with ARSACS and other related causes of ataxia, and should provide additional information on how these conditions affect the eye.

Will my clinical details be kept confidential?

Yes. Medical notes will be examined and assessments carried out during this study. All information that is gathered about you during the course of the research will be kept strictly confidential. Personal identifiable information will only be seen by members of the research team. All use of information within the United Kingdom is protected by the Data Protection Act 1998.

What will happen to the results of the study?

Results of the study will be published in peer-reviewed academic journals in an anonymous form. No patient-identifiable data will be published.

If you would like to participate in this study or know of any individuals who may be interested, and would like some more information, please don't hesitate to get in touch:

Prof Paola Giunti
Principal Clinical Research Associate and Honorary Consultant Neurologist
UCL Institute of Neurology
Queen Square House
Queen Square
London WC1N 3BG
Email: p.giunti@ucl.ac.uk

Nita Solanky
UCL Institute of Neurology
Queen Square House
Queen Square
London WC1N 3BG
Email: n.solanky@nhs.net
Tel: 0203 448 4130