



Research project on Genetic Testing in Ataxias – recruiting now

Researchers in Oxford are recruiting volunteers for a new research project aimed at developing new genetic tests for the ataxias, especially for the autosomal recessive ataxias.

Background

Genetic testing is an essential part of clinical practice. It can aid diagnosis, allow patients to assess the risk to other family members, help people to plan families and help identify people who may be suitable for new treatments or trials as they become available. However, many patients with ataxia do not know what the underlying cause of their condition is, although their doctor may suspect that it is a genetic problem.

New genes causing ataxia are identified every year. However, genetic testing for the majority of these genes is not available, as genetic tests are expensive to develop and introduce into the NHS. Furthermore, the number of people who would benefit from such tests is unknown and therefore funding for them is limited. Additionally, the genes are large and complicated to analyse; this is especially true for the autosomal recessive ataxias. (For more information on autosomal recessive ataxias see Ataxia UK's leaflet on 'Genetics and ataxia' on the website. Generally if you have an autosomal recessive ataxia your parents do not have ataxia but they are carriers for the condition.)

New technology, known as 'high throughput sequencing', is now available; this can help to analyse large quantities of genetic material much more quickly. The John Radcliffe Hospital in partnership with Oxford University has recently been funded by the Department of Health to install two of these new machines as a result of becoming one of 5 UK Biomedical Research Centres. A grant from Ataxia UK means that there will be a dedicated Scientist working full time for two and a half years to use the new machines. The aim is to test genes causing ataxia which are not currently available on the NHS to work out which of these tests should be available and also find out whether the new high throughput sequencers are the best way to do the analysis. So taking part in this project may result in you getting a specific diagnosis of which type of ataxia you have.

Who can take part?

Anyone with an undiagnosed ataxia which developed under the age of 50 can take part, whether they have affected family members or not. The researchers are particularly keen to see people whose ataxia is likely to be genetic and inherited in an autosomal recessive way – which means there may be affected brothers, sisters, or cousins, but not affected parents. They are particularly interested to hear from people with childhood onset, teenage onset or young adult onset cerebellar ataxia which may have been thought to be Friedreich's ataxia but turned out not to be on

genetic testing. The researchers are also interested in analysing samples from patients with a condition called Joubert syndrome, which is a very rare cause of ataxia in children.

If your ataxia started when you were over the age of 50, the researchers still invite you to get in touch with them. If the new machines prove to be useful in clinical practice, they will start testing other genes that are inherited in different ways and will contact you to offer you these further tests.

The aim is to test as many genes as possible during the project so the researchers will be happy to hear from anyone who is interested in taking part. However, this project is not relevant for people with a confirmed diagnosis of Friedreich's ataxia.

What do you if you want to take part?

Your GP, Neurologist or other doctor will be able to tell you if your ataxia, or your child's ataxia is likely to be autosomal recessive, or is suspected to be, or if it is likely to be inherited in some other way.

If you are willing to travel to Oxford you can ask your GP to refer you to the Ataxia clinic and send any clinical information including details of any genetic or other tests you or your child has already had. An Information Sheet will be sent to you and an appointment will be arranged to see you or your child and take a small blood test which can be used to extract some genetic material to analyse.

If you are unable to travel to Oxford then you can ask your GP, Neurologist, Geneticist, Paediatrician or other doctor to contact the researchers at the address below. Patient Information Sheets and relevant forms can be sent to you, which will allow blood to be taken at your own clinic and sent to the laboratory in Oxford.

Any blood samples will be sent to the Oxford Regional Genetics Laboratory at the Churchill Hospital, in Oxford. From there the DNA is extracted, the sample is given a numbered code and the sample will be transferred to the Wellcome Trust Centre for Human Genetics where the laboratory work will take place.

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