



**Identification of a novel gene causing autosomal dominant ataxia and nystagmus
Researcher: Nicola Ragge, University of Oxford**

Scientific Abstract

We previously described a unique four generation family that develops ataxia and nystagmus in the first two years of life. We previously mapped the disease locus to chromosome 13q31-q33 with a LOD score of 6.322 at $\theta=0$ for D13S159. Haplotype analysis linked the disorder to an 11 Megabase region between D13S1300 and D13S158. We propose to screen all genes within this region for mutations in two affected and two unaffected individuals. If no mutations are found within this region, we also propose to screen two very good candidate genes (*FGF14* and *FARP1*) for deletions. If we uncover a mutation or deletion we propose to establish if it is pathogenic by screening all remaining individuals from the family, in addition to a control cohort of 192 healthy adults.

Lay Summary

We have identified a unique four generation family that develop ataxia and involuntary rapid movements of the eye (known as nystagmus) in the first two years of life. The disorder is dominantly inherited, which means that affected individuals have a 50% chance of transmitting the affected gene to their children. We have previously used genetic mapping to identify a region on chromosome 13 that contains the disease gene. We propose to screen a number of genes within this region for mutations or deletions of genetic material that might cause the disorder. Identification of a new ataxia gene is important to understand the mechanisms that cause ataxia and provide new targets for drug treatment. Additionally, there are a number of individuals and families with ataxia but no genetic diagnosis who can be screened for mutations in the new gene. This will provide affected individuals with more information about the expected course of the disease.

**For more support or information please contact: Ataxia UK, Winchester House,
Kennington Park, Cranmer Road. London SW9 6EJ**

Website: www.ataxia.org.uk.

Helpline: 0845 644 0606 Tel: +44 (0)20 7582 1444 Fax: +44 (0)20 7582 9444

Email: helpline@ataxia.org.uk.