Ataxia as a therapeutic marketplace opportunity

What is Ataxia?

The ataxias are a group of rare progressive neurological conditions. The majority have no cure or disease modifying treatments. As such, the ataxias represent **an untapped rare disease market opportunity**.

Ataxia means lack of coordination meaning people progressively lose the ability to coordinate their movements. Many people lose their ability to walk, and need to use wheelchairs. Problems with hearing and speech can cause communication difficulties that make people feel isolated, and people with ataxia often become reliant on others for their everyday needs. **They are serious chronic conditions and many types are life-limiting, reducing people's quality of life.**

Why consider ataxia?

There have been a number of advances in the last few years in the identification of new genes causing specific ataxias, largely due to recent developments in gene sequencing technologies. **Most ataxias are indeed genetic and caused by mutations in single genes**. Research is also progressing in understanding the basic biological mechanisms that underlie ataxias and many therapeutic targets have now been identified. This has led to pre-clinical studies of potential disease-modifying drugs in animal and cell models and, encouragingly, a number of clinical trials in people with ataxia are ongoing. There is an urgent need to develop disease modifying treatments that at the very least slow or stop the progression of each ataxia. But there is currently not even a treatment to relieve the symptoms of ataxia and help people coordinate their movements, whilst waiting for a cure.

How big is the market?

There are more than 10,000 people in the UK today with ataxia within a national population of upwards of 65 million, and as yet, there is no cure or effective treatment. With a population in the rest of Europe as a whole being upwards of 680 million, there could be more than 100,000 people with ataxia in Europe. Studies to date have collated the following statistics:

- The most common inherited ataxia in Europe is Friedreich's ataxia; the estimated disease incidence based on carrier frequency of **1 in 85 is 1:29,000**. In the UK estimates suggest around 2,000 people.
- The minimum prevalence of childhood ataxia in Europe was estimated at 26 in 100,000 in a recent systematic review. This included conditions such as ataxic cerebral palsy, thus the estimate for progressive inherited ataxias is **1:21692**.

Why talk to Ataxia UK?

Ataxia UK is a charity supporting people with ataxia, by providing support and information services and promoting research. We fund world-class medical research around the world. The networks of centres and expertise can be harnessed for collaborative partners, with an interest in bringing a treatment to market in this area. **We can offer a ready-made, fully-coordinated research programmes infrastructure around the globe**.

At Ataxia UK, our mission is to fund and promote research to develop treatments and cures for the ataxias. Our research reflects the wide spectrum of ataxias that affect the people we support and we believe in joining efforts and working collaboratively to accelerate progress in research. **We are committed to becoming your valued collaborative partner.** Ataxia UK can bring significant resources and infrastructure to your therapeutic development programme. Our established network of global research experts and easily-accessible patient populations will help you to cut preclinical and clinical development time to a minimum. This will enable you to get your products to market faster and at a lower overall cost.

What we can offer you

1. **A ready-made infrastructure for your clinical trials**
   Ataxia UK has helped develop and accredit Specialist Ataxia Centres within the National Health Service, which play a fundamental role in the translation of research into benefits for patients. We have two in the UK (London and Sheffield), plus an additional clinic in Oxford. These are centres of excellence for patient diagnosis, management, support and research. They provide infrastructure for trials and are currently involved in natural history studies, genetic screening for diagnosis, verification of rating scales and biomarker research for future therapeutic trials. These Centres have also run ataxia clinical trials, both as academic-run trials and commercially sponsored trials.

2. **A UK patient registry facilitating patient engagement and recruitment to studies**
   Ataxia UK supports research by helping to recruit participants for scientific research/healthcare studies. We have a registry of around 3,500 patients with a number of different types of cerebellar ataxias, including around 700 patients with Friedreich's ataxia.

3. **Ataxia UK Medical Guidelines**
   Ataxia UK has an understanding of patient pathways and patient needs. In fact, our charity is driven by the needs of people with ataxia and we believe in their involvement in all our activities, including helping to ensure that patient voices are heard in research. Our Medical Guidelines were created by ataxia specialists and respond to the needs of patients. Their value has been affirmed by the European Reference Network for Rare Neurological disorders (ERN-RD).

4. **European network of patient groups – Euro-ataxia**
   We are active members of Euro-ataxia, the umbrella organisation of ataxia patient groups and thus provide a direct link to other patient groups across Europe. Visit www.euroataxia.org

5. **Global networks for clinical translational research**
   Ataxia UK is actively working with European and global clinical networks, creating patient databases and supporting natural history and biomarker studies.

6. **An experienced collaborative partner**
   Ataxia UK works closely with many other charities and researchers worldwide and we believe that by working in partnership we will get the best results. Partnerships include jointly-funded projects, coordination of information exchange and new initiatives to drive research forward.

7. **International Ataxia Research Conferences**
   Ataxia UK works jointly with other ataxia charities in organising the International Ataxia Research Conferences on the Ataxias (IARC) and other scientific meetings. The next IARC will be held on 14-16 November 2019 in Washington, USA.

8. **An established research-funding body**
   We continue to fund a range of research projects into various aspects of the ataxias. This includes basic and applied research, long and short term projects into the gathering of preliminary data, speculative projects vs projects with clear potential benefits, as well as projects incorporating patients of all ages.

What research groundwork have we done so far?

Ataxia UK has invested in ataxia research since the charity was first established over 50 years ago and great progress has been made in the following areas:

- *Harnessing Genetics for Diagnosis* - Huge strides have been made in the last 20 years in the genetic etiology of the ataxias, leading to more people than ever getting a specific diagnosis.
- *Causes and Mechanisms* - We have pioneered advances in understanding the mechanisms and cellular dysfunction pathways in many different ataxias. This offers easier identification of promising targets for therapy.
- *Cell and Animal Models* - Ataxia UK has played a major role in developing a number of cell and animal models that are now in use by scientists around the world. With increased understanding we are now at a point where we have promising therapies being tested in human trials that we hope will lead to treatments.
- *Focus on Early Stage Work* – Developing collaborative research partners is key to meeting our vision for the ataxias. Our commitment to early stage work is vital in making ataxia an attractive research pathway for pharmaceutical partners. We are also dedicated to validating clinical outcome measures and biomarkers and have funded some pilot clinical studies.

Who to contact If you would like to know more or have an in-depth discussion, please contact Ataxia UK's Head of Research, Dr Julie Greenfield on 020 7582 1444, or by email at jgreenfield@ataxia.org.uk.
You can also find more information on our website: www.ataxia.org.uk