Ataxia UK's Research Strategy



ATAXIA

Ataxia UK

Foreword



Following extensive consultation and discussion, Ataxia UK recently launched a new overall Strategic Plan 2017 - 2020.

By 2020, we want:

people affected by ataxia to be able to say: “I feel supported and in control”

to consistently maximise the impact of ataxia research activity

to have fostered best practise in treatment and care

to have increased our funded research activity

Ataxia UK is an ambitious charity that exists to support people affected by ataxia. We invest in research to have maximum impact on the future development of treatments and cures.

Recognising the importance of working in partnership with organisations in the field of rare disease research, we are putting ourselves at the heart of numerous worldwide collaborations. With our input, significant progress is being made and we have real optimism that important new treatments will emerge during the lifetime of this strategy.

In years to come, people may look back at the early part of the 21st Century and be surprised at what health conditions are still a part of our daily lives - in the same way that we might look back today at myriad diseases which are now, largely, a thing of the past. The ataxias will be one such set of conditions. Exactly how many years this will take to happen is the key question. But happen it will.

Significant advances in new emerging technologies, such as gene-editing techniques, are taking place right now. As a result of these advances, treatments are being developed for such related, rare conditions as spinal muscular atrophy, where we have the first ever approved treatment that targets the underlying genetics of the condition. Similarly, the first gene-therapy treatment of an inherited disease was recently approved to treat a rare form of blindness. With similar approaches now being applied to the ataxias, the landscape for finding treatments for the ataxias in the future is very exciting indeed.

But the only way we will become free of ataxia is through on-going research - which is why this document exists.

The aims listed below underpin our new **Research Strategy**. Ataxia UK’s last Research Strategy was launched in 2012 and stated our **2020 vision: aiming for a treatment or cure for one or more of the ataxias by 2020**. Here, we reflect on many of the positive steps we’ve made since, our contribution to progress and the breadth of our involvement in the international research effort. We hope you will share our passion for advancing research for the ataxias and help us get closer to achieving the aim of developing the much needed treatments and cures for people with ataxia.

June 2018

Dr Harriet Bonney
Chair of Ataxia UK

Prof Barry Hunt
Scientific Advisor and Trustee of Ataxia UK

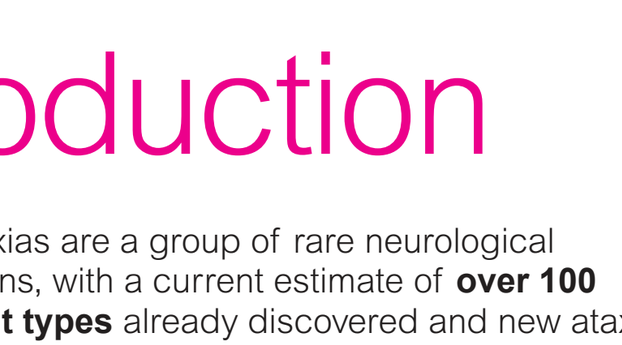
Introduction

The ataxias are a group of rare neurological conditions, with a current estimate of **over 100 different types** already discovered and new ataxias continuing to be identified.

Many are progressive and complex conditions for which there are no approved treatments. For the very few treatable types of ataxia, early diagnosis is of paramount importance. Many of the ataxias are inherited, and numerous different genes that cause ataxia have been identified, resulting in improvements in diagnosis.

This research strategy is a result of extensive consultation with key stakeholders, patients, researchers and healthcare professionals, including a large survey conducted among people affected by ataxia to identify **the needs of patients and their families** *(see '**What patients say**' below).*

Publishing our third edition of **Ataxia UK's Medical Guidelines** has also been instrumental in identifying gaps in the clinical management of the ataxias. We believe this has led us to the development of a robust Research Strategy that will direct our research activities over the next few years.

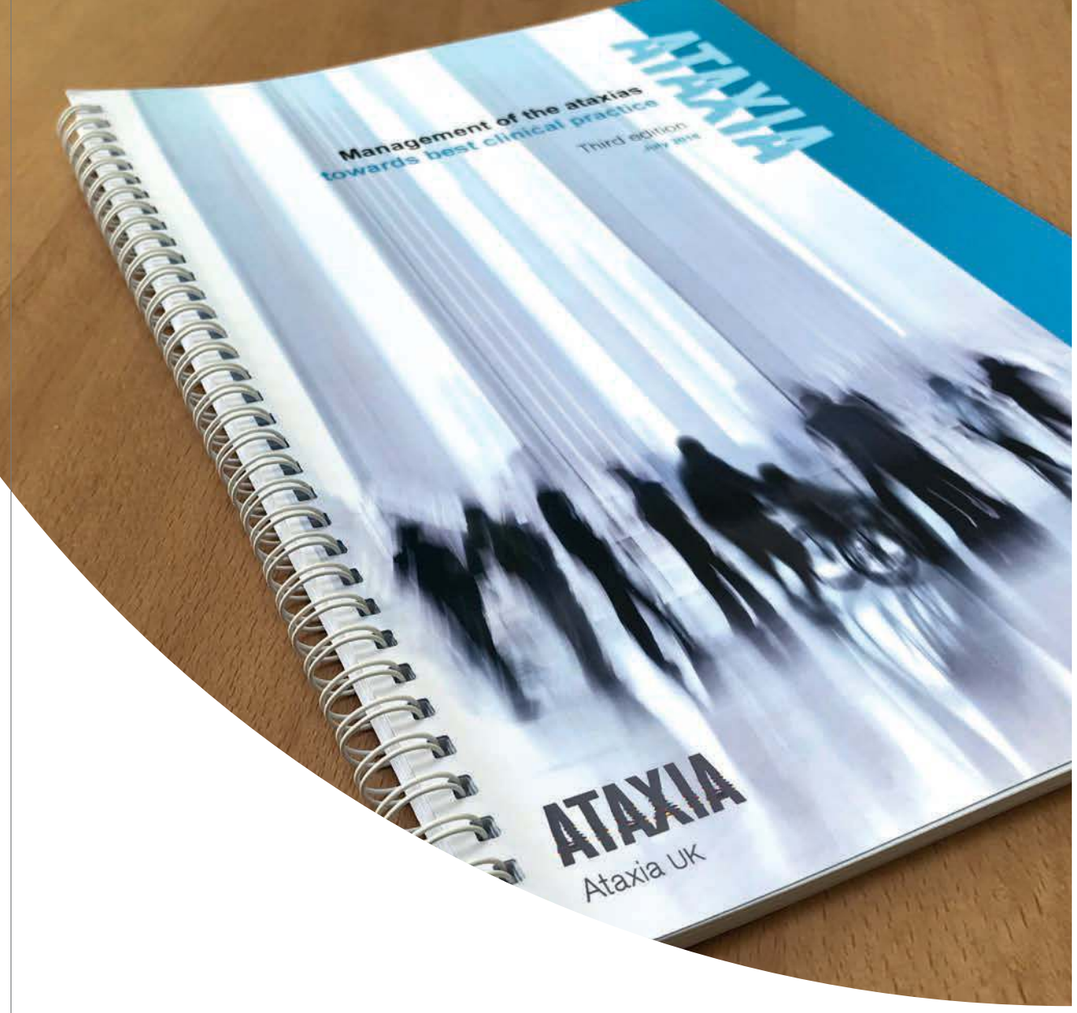


What patients say

Ataxia UK recently surveyed people with ataxia and their family members to better understand their needs and the impact ataxia has on their lives.

Data from **over 400 people** was collected and analysed. Results showed that the symptoms having the greatest negative impact were those related to **balance, coordination and speech**. Additionally, **fatigue** was identified as having an impact, followed by **bladder problems**. Unsurprisingly, a number of **mental health issues** were also highlighted.

These findings were consistent with the information we gather from our Helpline; thus, as part of our new **Strategic Plan**, Ataxia UK aims to develop mental wellbeing activities for people affected by ataxia in collaboration with relevant organisations to try and help in this area.



Ataxia Medical Guidelines

In 2016, we published the third edition of our Medical Guidelines, **'Management of the ataxias: towards best clinical practice'**, a resource for healthcare professionals on the diagnosis and management of the progressive ataxias.

We worked with **30 healthcare professionals** with expertise in the ataxias to produce this comprehensive document. Whilst it is a useful resource, it also highlights the gaps in our knowledge and the need for more evidence-based research in many areas.

The lack of disease-modifying treatments for the majority of ataxias is well known, but there is also a need to develop drugs to alleviate symptoms. If a treatment could improve balance and coordination, it would have a huge impact on the lives of people with a range of ataxias.

Associated symptoms such as tremors and fatigue are also poorly managed, and there is little that can currently be done to assist people with vision and hearing problems.

Although there are a few pilot studies in physiotherapy, speech and language and occupational therapy, there is a lack of larger confirmatory trials. When the results of our patient survey are compared with the opinions of healthcare professionals who have an expertise in ataxia, there is much overlap in the needs identified.

Research progress

There have been **significant advances** in ataxia research in recent years.

We have seen developments in diagnosing the ataxias that are significant for not only **providing information about the condition**, but also for the **development of new approaches to treatment**. For example, in **SCA38** (a rare form of ataxia), the identification of the gene mutation which causes the ataxia has led to an understanding of how it could be treated. A trial showed positive results in treating SCA38 patients (who had this particular mutation) with a therapy that uses a **dietary fatty acid supplement (DHA)**. Overall, there have been an increasing number of **clinical trials** that test medications for a number of ataxias. This enhanced level of activity makes us hopeful that **successful treatments will emerge**.

In addition to a collaborative community of **university researchers** working on the ataxias, there are now numerous **biotech and pharmaceutical companies** also trying to develop treatments. **Gene therapy** and **genetic-editing** approaches are receiving much attention, especially following the successes of these approaches for treating other conditions.

Research progress in last 5 years - selected highlights

Researchers testing medications to improve **balance and coordination** potentially helping many ataxia conditions

Friedreich's ataxia treatment trials

10 trials testing different medications taken place or ongoing

8 new genes found causing spinocerebellar ataxias

2014: **SCA38**, SCA40

2015: SCA42

2016: SCA43

2017: SCA37, SCA44, SCA45, SCA46

Potential treatment found for a rare ataxia: SCA38

2014: Gene causing SCA38 identified. This gene encodes an enzyme involved in making a fatty acid called DHA and in SCA38 the enzyme's function is affected

2017: DHA (a dietary supplement) shows benefits in a trial of 10 people with SCA38

Widespread use of **ataxia gene panels** leading to **more people getting a diagnosis**

Encouraging results in mouse models of **Friedreich's ataxia** show **reversal of cardiac and neurological symptoms**, giving hope this may happen in humans



Ataxia UK's research staff at the **2015 International Ataxia Research Conference** in Windsor

Research strategy

Ataxia UK is committed to finding treatments and cures for the ataxias and in ensuring these are made available to patients.

To achieve these aims, we invest funds in our Research Department to manage the research activities of the charity. The Head of Research and the Research Officer have an understanding of the ataxia research landscape and keep abreast of developments, making them well placed to work with Trustees on **selecting research activities that promise to achieve the maximum impact** and to help us towards our goal of finding treatments and cures for the ataxias.

In addition, one of our Trustees is a pharmacologist and an ataxia parent, and acts as the scientific advisor to the Board. Our independent Scientific Advisory and Medical Advisory Committees also provide valuable advice and support to Ataxia UK.

The aims of Ataxia UK are to maximise the impact of research in:

- **improving diagnosis**
- **developing treatments that are available to patients**

These aims will be achieved by Ataxia UK's involvement in various activities:

- **increasing research on ataxia**
- **translating research into clinical practice**
- **facilitating research studies**
- **disseminating research and facilitating networking amongst researchers**
- **initiating and coordinating collaborative projects**

Increasing research on ataxia

Funding research projects

Ataxia UK funds research projects that cover a **wide range of progressive ataxias**, mostly within universities and research institutes.

The charity's independent **Scientific Advisory Committee** advises our **Trustees** on which projects to fund and comprises of scientists with a wide range of expertise in relevant disciplines, along with lay people who have personal experience of ataxia.

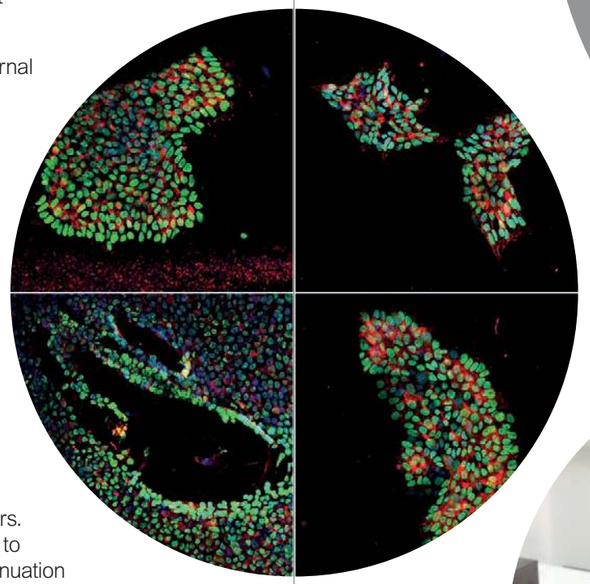
We award our grants to selected applications by considering advice from expert external peer reviewers. As a member of the **Association of Medical Research Charities (AMRC)**, Ataxia UK follows their good practice recommendations to ensure there is a robust selection process, and recently passed an AMRC's peer review audit. Membership of the AMRC also gives us access to its support, information and its member charities, connecting us to the medical charity research community.

Ataxia UK research staff administer and manage the research grant programme from every stage, including the selection phase, monitoring funded projects and assisting researchers in maximising the impact of the project, at all stages including the dissemination of results and follow-up of projects. We value and facilitate the input of people with ataxia in the design of research studies (such as clinical trials). The charity funds studies that have the maximum potential of making an impact. This includes studies that work towards developments in diagnosis, treatments and new scientific and clinical tools to assist in research.

When selecting projects to recommend for funding, the Scientific Advisory Committee considers the scientific quality, likelihood of success and potential benefit to people with ataxia as key parameters, and takes advice from external peer reviewers. Projects need to be based on strong preliminary evidence and show a clear pathway to maximising the impact of the outputs generated, and where appropriate, for the continuation of the studies at the end of the funded project. Innovative projects involving promising new ideas are encouraged, as are projects that are close to a potential treatment. Addressing the needs identified by people with ataxia, and the gaps in knowledge identified by ataxia healthcare professionals, are also important to Ataxia UK.

Ataxia UK will look favourably upon projects that:

- **have implications to more than one type of ataxia** *(in particular with rarer ataxias)*
- **have the potential to involve joint collaborative funding**
- **involve collaborations between research groups**
- **fund younger researchers who potentially may be developing a career in ataxia research** *(eg: PhD studentships)*
- **include public and patient involvement activities** *(this is considered essential for all clinical studies involving people with ataxia as participants)*



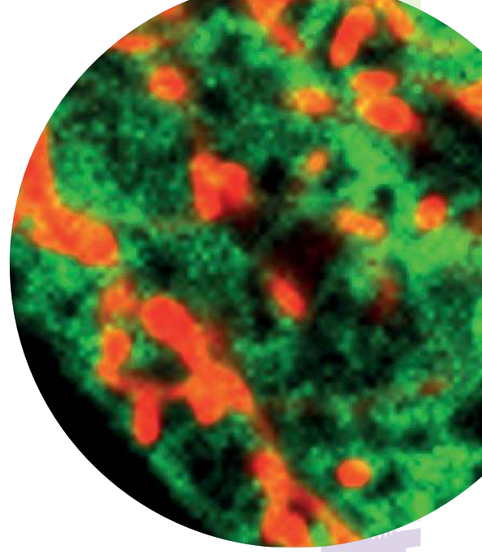
researchfish®

Case study

Ataxia UK funded research makes an impact

Recent data on the outcomes of Ataxia-UK funded research has been extremely positive, as demonstrated by our use of **Researchfish**, an online tool commonly used by UK medical research charities. There has been a substantial increase in ataxia research as a result of our funding; indeed, the data shows that for every pound invested by Ataxia UK, an additional **£3 of further funding to ataxia research** is generated. In terms of the dissemination of research, **27 funded projects** generated **39 research papers** and **56 further engagement activities**. In addition, our funded researchers produced **25 new research tools or methods** that can be of use to the wider research community.





Ataxia UK funds research studies that:

- cover a wide range of progressive ataxias
- work towards developments in diagnosis, treatments and new tools to assist in research
- are recognised to have high scientific quality
- are innovative projects involving promising new ideas
- are relevant to patients' needs

A detailed guidance document for researchers wishing to apply for funding can be found on the Ataxia UK website: ***www.ataxia.org.uk/news/apply-for-funding***

Fostering collaborations with industry

As the ataxias are rare conditions, they have not received much attention from the pharmaceutical industry: hence the lack of approved treatment for the majority of ataxias.

However, this is changing; it is now recognised that these conditions offer opportunities for pharmaceutical and biotech companies to make a huge impact by being the first to develop an approved treatment. Ataxia UK will highlight the opportunities that are available to pharmaceutical and biotech companies if they enter the ataxia field. In addition, Ataxia UK will continue to play a pivotal role in assisting researchers in academia and industry by:

- **providing information and advice on the research landscape**
- **facilitating meetings and introductions**
- **explaining the impact ataxia has on people with the condition**

This will be achieved by Ataxia UK research staff attending and presenting at relevant conferences, and working in partnership with existing and new contacts. A new **Pharma Advisory Committee** has also been established to help us in this area of activity.





Facilitating research studies

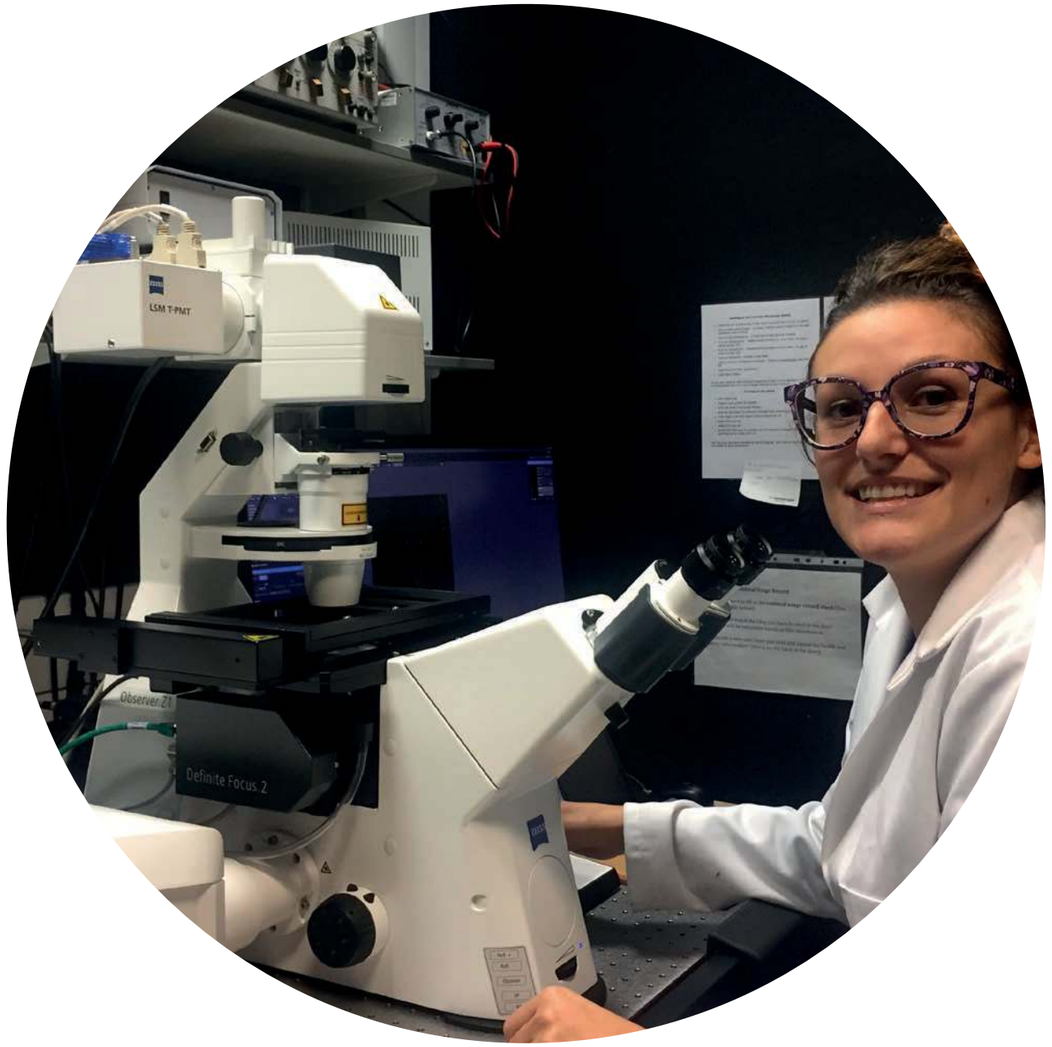
Ataxia UK has a large registry of people with ataxia, and the charity is therefore in the unique position to act as a link between researchers and patients.

We provide assistance to researchers by publicising and helping with recruitment for research studies. This helps to reduce the timeframes for recruiting for studies, and their completion.

The impact of involving people affected by ataxia in research studies cannot be underestimated and Ataxia UK continuously seeks ways of maximising patient engagement. For example, we have been able to improve the relevance of this strategy for people affected by ataxia as a direct result of our recent patient survey. The membership of people affected by ataxia in our **Scientific Advisory Committee** and **Board of Trustees** ensures that all major decisions take the views of people with ataxia into consideration. Ataxia UK also engages people affected by ataxia in individual research studies, including their input in the design of clinical studies.

Carla and Adam, parents to Eva, four years old: Eva took part in a next generation sequencing research project. "Thank you Ataxia UK! Your funding has meant we have successfully navigated the first step on our long and unknown journey. You have made a direct difference to our lives and for that we will always be grateful. Keep doing what you do; your funding makes a massive difference to ordinary families catapulted into an extraordinary life!"

Translating research into clinical practice



Translating research findings into clinical practice is the ultimate important step to ensuring that patients are benefiting from research advances.

As more trials are taking place to test treatments for the ataxias, translating research requires the increasing involvement of Ataxia UK research staff. Much of the infrastructure is already in place to facilitate this translational medicine process.

The **Specialist Ataxia Centres**, clinics in the **NHS** established in partnership with Ataxia UK, and our engagement with clinicians interested in ataxia via our **Medical Advisory Panel**, will assist us with this task in the UK.

Working internationally is especially important in research and healthcare for rare diseases, where small patient numbers in each country means it is often necessary to do trials and other studies collaboratively and internationally in order to involve sufficient patients to be of significance.

Our active involvement in **Euroataxia**, the federation of 20 European ataxia charities, helps us to share best practice/experiences, collaborate in funding research and campaign jointly for access to treatments.



The development of the **European Reference Networks for rare neurological diseases** (which includes the ataxias), also should assist in translating research into clinical practice both in the UK and across Europe.

Ataxia UK also collaborates with ataxia charities worldwide, having particularly close partnerships with two main US ataxia charities.

Collaboration with the pharmaceutical industry will also play a major role in getting medicines approved and delivered to people with ataxia.

Ataxia UK works closely with a number of companies already, and plans to increase the activities in this area, ensuring patient perspectives are taken into account at all stages.

Working with regulators and health service providers is also essential to succeed in making treatments available.

Ataxia UK works with policy organisations to campaign for better healthcare services for people with ataxia. This involves ensuring research developments are made available to patients.

We are active members of the **Neurological Alliance**, the organisation that campaigns for people with neurological conditions.

Ataxia UK is also a member and works closely with **Genetic Alliance UK** and **Rare Disease UK**, the umbrella organisations working for people with genetic conditions and rare conditions respectively.

An active involvement of Ataxia UK's CEO in **National Health Service Boards** also helps us to shape government policy.

Initiating and coordinating collaborations

Research conferences are hugely beneficial in pushing research forwards; providing a platform for dissemination of research ideas and findings; networking, and the creation of new collaborations.

We believe that by pooling resources and working jointly with other ataxia charities to organise the **International Ataxia Research Conferences**, we play a significant role in driving research forwards.

We also maintain registries of health professionals and researchers, to whom we regularly disseminate information about relevant events, funding opportunities and research findings.

We ensure that people affected by ataxia are involved in research and kept informed of research developments through Ataxia UK events; our quarterly magazine; monthly e-newsletter; Helpline and social media channels.





Case study

International Ataxia Research Conference 2017

Ataxia UK partnered with two other ataxia charities (**FARA** and **GoFAR**) to organise a hugely successful ataxia research conference in September 2017. With over **400 attendees** from **22 countries** including scientists and clinicians from universities, industry representatives and patient groups, it was **the biggest ataxia conference to date**. The theme of patient perspectives was at the forefront of this conference, as we held an interesting roundtable discussion with patients and carers to hear about the impact of ataxia and what patients want from treatments.

Case study

European Friedreich's ataxia consortium for translational research

A group of ataxia charities in Europe, under the auspices of **Euroataxia**, have been funding and supporting a natural history study involving a number of hospital sites in European countries, including the UK. Ataxia UK has played a pivotal role in organising this collaboration between academia and patient groups, and continues to have a place on the **Consortium's Steering Committee**. The importance of the data generated from this research in assisting the design of new clinical trials is highlighted by the involvement of the pharma industry in funding this work.

Working in partnership

As demonstrated above, Ataxia UK firmly believes that working in partnership with organisations that have the common interest of ataxia enables us to achieve more than we could do alone.

We already have many close links with ataxia patient groups around the world, and our **International Ataxia Research Conferences** are now being organised in partnership with other ataxia charities. The funding of research projects often takes place in partnership with ataxia charities worldwide, UK charities, universities (eg: co-funded PhD studentship schemes) and, more recently, pharmaceutical companies. Collaborative global initiatives such as patient registries and natural history studies are also underway, with Ataxia UK taking a prominent role in promoting and supporting them *(see case study above)*. Charity-pharma partnerships are also an important aspect of Ataxia UK's work.

Final thoughts

Our ambition is to develop **safe and effective treatments** for the ataxias. Our **Research Strategy** is an important plank in this aspiration. We are always keen to discuss our work and receive feedback. Please get in touch if you would like more information or to discuss our work.

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