IMPROVING RARE DISEASE CARE ACROSS THE UK

Lessons from ataxia

Across the UK an estimated 3.5 million people are living with a rare disease, and 1 in 17 people will be affected by a rare disease at some point in their life.[1] In 2021, the UK Rare Diseases Framework was published, which aims to ensure that the lives of people living with rare diseases continues to improve.[2] At Biogen and Ataxia UK, we welcome the progress made since the Framework was introduced. However, the current Framework is due to expire in 2026 and more still needs to be done to support people with rare diseases, including ataxia.

Around 10,500 people in the UK have a type of ataxia;[3] a group of rare progressive neurological disorders that can affect every area of a person's life – from co-ordination and balance to speech.[4] In March 2025 and in collaboration with Ataxia UK, we held a virtual workshop with 8 people in the UK over the age of 18 who are living with, or caring for someone with, ataxia. During the discussion, attendees highlighted challenges including slow diagnosis, a lack of awareness from healthcare professionals regarding their specific needs, and delayed and poorly co-ordinated care. These challenges align closely to the priorities set out in the UK Rare Diseases Framework and underscore the importance that a new Framework is committed to as a matter of urgency.

This briefing summarises the discussion from the March 2025 workshop and makes a series of policy recommendations for improving care for people with ataxia, which could also benefit people with rare diseases across the UK.

This report was commissioned and funded by Biogen.

Document number: Biogen-269623 | Date of prep: January 2026

RECOMMENDATIONS

1 **RENEW THE UK RARE DISEASES FRAMEWORK:** The UK Government and the devolved administrations should publish a new Rare Disease Framework when it expires in 2027, and ensure that the new Framework includes refreshed targets for the diagnosis of rare diseases, more closely aligned to NHS constitutional standards.

2 **IMPROVE EDUCATION AND TRAINING ON RARE DISEASES:** The NHS should continue working to ensure that healthcare professionals receive education and training on rare diseases, including ataxia, so they are able to quickly refer people onto specialists, where necessary. They should work with patient organisations and industry to ensure that healthcare professionals have the most up-to-date resources to support people with rare diseases.

3 **DEVELOP NEW PATHWAYS AND SERVICES TO BRING CARE CLOSER TO HOME:** In England, the Rare Disease Collaborative Network (RDCN) for rare ataxias should lead on the development of new pathways and services which bring care closer to home with input from the patient, industry and clinical community.

4 **SET UP SPECIALIST ATAXIA CENTRES:** The devolved administrations should work with the NHS in each of their countries to set up specialist ataxia centres, so that people living outside of England are able to access specialist care closer to home. In addition to the establishment of specialist centres in the devolved nations, clinicians around the UK are encouraged to engage with the RDCN for rare ataxias.

5 **RECOGNISE THE IMPORTANCE OF WRAP-AROUND CARE SERVICES:** The UK Government and the devolved administrations should recognise the importance of wrap-around care services, such as genetic counselling, physiotherapy, hydrotherapy and psychological support, in any future version of the UK Rare Diseases Framework.

6 **EXPAND THE WRAP-AROUND CARE IN THE COMMUNITY:** The NHS should explore ways to expand the wrap-around care that is offered in the community, potentially as part of the development of neighbourhood health services, so that people with rare diseases no longer need to travel long distances to access support.

WHAT IS ATAXIA?

Ataxia is an umbrella term for a group of rare neurological disorders that affect co-ordination, balance and speech.[4] **An estimated 10,500 people in the UK are living with ataxia – and in most cases there is no cure for the disease.[3]**

Common symptoms include:[5]

COORDINATION AND BALANCE PROBLEMS	**SLURRED SPEECH**	**DIFFICULTY SWALLOWING**
TREMORS OR SHAKING	**FATIGUE OR TIREDNESS**	**PROBLEMS WITH VISION**

Most ataxias are progressive, meaning symptoms worsen over time, although the speed at which this happens will vary depending on the type of ataxia a person has.[4] Likewise, the prognosis for people with ataxia varies considerably depending on the type of ataxia. For people with hereditary ataxia (where the faulty gene that causes ataxia has been passed on from parent to child), life expectancy is generally shorter than normal, although some people can live into their sixties and beyond. In more severe cases, the condition can be fatal in childhood or early adulthood.[4]

HOW CAN WE IMPROVE THE CARE OF PEOPLE WITH ATAXIA?

DIAGNOSIS

The UK Rare Diseases Framework rightly commits to helping patients get a final diagnosis faster.[2] However, many people with a rare disease continue to face long waiting times and need to attend multiple appointments with different specialists before they are diagnosed. On average, people with a rare disease have to wait 5.6 years before they receive an accurate diagnosis.[10]

During our workshop, people with ataxia described the process of receiving a diagnosis as difficult and time consuming.[11] On average, people are experiencing delays of between six months to over five years before they are referred to a neurologist.[12] During this time, diagnosis is delayed and symptoms may worsen, as people are unable to access appropriate care that can help them manage their condition as much as possible.

People with ataxia have also reported that healthcare professionals often lack knowledge of ataxia and that when they are diagnosed, they are not given suitable information about the condition.[2] This is despite the fact that the UK Rare Diseases Framework commits to raising awareness of rare diseases among healthcare professionals.[2] A 2021 survey of people living with ataxia and carers found that:[13]

73.7% of respondents felt that GPs and other primary care professionals **lacked knowledge** about ataxia

43.2% of respondents felt that there was a **lack of suitable information** about ataxia and relevant symptoms

We welcome the commitment from policymakers to drive improvements in this area, including in England through the Department of Health and Social Care's (DHSC's) commissioning of research to measure the time that it takes for people with rare diseases to receive a diagnosis.[2] However, the slow progress of this work has been a source of frustration. More needs to be done to ensure that people with rare diseases, including ataxia, can receive a timely and accurate diagnosis – enabling them to access treatment that could improve their outcomes earlier.

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SPECIALIST CARE

Improving access to specialist care, treatment and drugs is also listed as one of the key priorities of the UK Rare Diseases Framework.[2] The Framework correctly acknowledges that the complex nature of rare diseases means that patients often require the expertise of multiple specialists, who could be spread across different hospitals, requiring people to travel long distances to access care.[2]

Some people with ataxia receive treatment at a specialist ataxia centre (SAC). These centres are dedicated to providing a co-ordinated service for people with ataxia, combining diagnosis, treatment, support and research. A recent survey found that 96.8% of people attending a SAC gave positive feedback on the role of the centre in understanding their condition, and 77% gave positive feedback on the SAC's ability to manage symptoms.

Despite this, there are very few SACs in the UK, and lack of access to a SAC is particularly a problem in the devolved nations.[14] Only one third of people living with ataxia in the UK have access to a SAC, and attendees at the workshop listed long travel times as one of the main reasons that they were unable to access a centre.[12] In England, the UK Government has set out the importance of care moving closer to home as part of their three shifts.[16] Yet for many people with ataxia, and other rare diseases, this is not an option. New models of care need to be considered that balance the need for people to access specialist care with the need to access care in their local community. For example, in England, there is a Rare Disease Collaborative Network (RDCN) for rare ataxias which should take a greater role in leading the development of pathways and redesigning services, closer to home.[17] There is an opportunity to build on existing guidance, such as the optimal clinical pathway for movement disorders developed by the National Neurosciences Advisory Group.[18]

Another important source of support for people with ataxia is specialist ataxia nurses.[19] Specialist nurses are the central point of contact for people with ataxia and are often the first port of call for any questions that people have about their care.[19] However, specialist nurses are only available to people who receive care through a SAC – the majority of people are unable to access support in this way.[15] While it may not be practical, or desirable to have a specialist ataxia nurse based in every hospital, it is important that every person with ataxia has access to a specialist nurse who is familiar with the neurological symptoms associated with ataxia and so can provide them with support that is tailored to their needs.

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4 **SET UP SPECIALIST ATAXIA CENTRES:** The devolved administrations should work with the NHS in each of their countries to set up specialist ataxia centres, so that people living outside of England are able to access specialist care closer to home. In addition to the establishment of specialist centres in the devolved nations, clinicians around the UK are encouraged to engage with the RDCN for rare ataxias.

GENETIC COUNSELLING AND WRAP-AROUND CARE

Access to additional services such as genetic counselling and wrap-around care can help to improve quality of life for people with rare diseases. The type of wrap-around care that works will depend on the individual, but can include services such as voice banking, speech therapy, physiotherapy, hydrotherapy, and counselling or other psychological support.[11]

Given most ataxias are hereditary, genetic counselling can be a particularly invaluable tool to help people, and their families come to terms with their diagnosis.[11] A genetic counsellor can help to explain the results of any genetic tests which have been carried out and give people the knowledge that they need to move forward with their lives. However, there are wide inequalities in access to wrap-around care and genetic counselling across the country, which are delaying people from accessing this vital support.[8]

During our workshop, many participants reported that they have been unable to access the wrap-around care or counselling that they need, with long travel times to access wrap-around care cited as a particular problem. They also said that healthcare professionals are often unsure of the type of wrap-around care that would be most beneficial to them. When people are referred, they often face long waiting lists and travel times.

A recent survey of people living with ataxia and their carers found that:[13]

- **While 86.6% of people had accessed physiotherapy at some point, only 51.8% reported that it met most of their needs**
- **56.7% of people had accessed speech therapy, and 48.9% reported that it met most of their needs**
- **Only 16.6% of people had accessed counselling – and 38.7% of people reported that they could not access counselling or that the counselling support that they did receive met none of their needs**

The UK Rare Diseases Framework does not currently recognise the importance of wrap-around care or genetic counselling for people with rare diseases. Wrap-around care and counselling should be included in any future iterations of the Framework, and it should set clear targets to ensure that people are able to access the appropriate care, closer to home. New ways of expanding wrap-around care into the community should also be considered, potentially through the development of neighbourhood health services, to reduce the distances that people have to travel to access support and allow them to access the care they need, closer to home.

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If you would like to discuss the recommendations contained in this briefing or how you could support people living with ataxia across the UK, please contact Nisha Tailor, External Affairs Lead for Rare Disease at Biogen UK and Ireland, at **nisha.tailor@biogen.com**.

About Biogen

Founded in 1978, Biogen is a leading biotechnology company that pioneers innovative science to deliver new medicines to transform patients' lives and to create value for shareholders and our communities. We apply deep understanding of human biology and leverage different modalities with aspirations to advance first-in-class treatments or therapies that deliver superior outcomes. Our approach is to take bold risks, balanced with return on investment to deliver long-term growth.

To learn more, please visit **www.biogen-uk-ie.com**.

About Ataxia UK

Ataxia is the umbrella term for a group of neurological conditions and Ataxia UK is the UK's leading ataxia support and research-active charity. Our Services team is dedicated to supporting everyone affected by ataxia in the UK to live their best possible life. We offer a Helpline and Advocacy service that provides trusted information, tailored advice, and one-to-one support to help people access their rights. Ataxia UK also promotes and facilitates research activities to drive research forwards for the ataxias. Alongside this, we deliver a range of activities, engagement opportunities, and volunteer-led initiatives designed to bring the ataxia community together and reduce feelings of isolation.

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