

The patient's journey: the progressive ataxias

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There are many different kinds of progressive ataxia. Although early symptoms tend to be similar, there is no "typical" patient. The speed at which the condition progresses varies between different types of ataxia and between individuals. The age when symptoms start also varies.

Overview of the progressive ataxias

Progressive ataxia describes a group of uncommon neurological disorders, characterised by problems with balance and coordination. Many general practitioners never come across these conditions during their working lives and are most familiar with ataxia due to cerebral palsy, multiple sclerosis, or stroke. The types of ataxia discussed here are chronic conditions that progress slowly and often result in people losing their ability to walk. Another common symptom is slurring of speech. Depending on the type of ataxia and its severity, many other symptoms can occur (box 1).

Many of the progressive ataxias are inherited conditions. The most common one in the United Kingdom is Friedreich's ataxia, which is inherited recessively, often coming into a family out of the blue when two carrier parents have a child who develops ataxia symptoms. Average age of onset is 15 years, but symptoms can start at an age as early as 2 years. Although rarer, in some people the condition is diagnosed much later in life (in their 40s and older). In general, the older age of onset, the less severe the symptoms.

In addition, there are many dominantly inherited cerebellar ataxias. To date 25 subtypes have been identified, known as spinocerebellar ataxia type 1, 2, 3, etc. Genetic testing is available for around 40% of these in some UK centres. Many of these dominantly inherited cerebellar ataxias are of later onset. Patients have often already had children before their own inherited condition is diagnosed. Tests are not available for all progressive cerebellar ataxias, and many people are therefore given a diagnosis of idiopathic cerebellar ataxia. The impact of the diagnosis of a hereditary condition reaches well beyond the individual.

Three patients' journeys

In this article, we focus on the lives of three people, attempting to illustrate the diversity of perspectives of parents of a child with ataxia, a mother of an adult with ataxia, and a working adult with ataxia. Their stories

reflect the variety of progressive ataxias in terms of severity of symptoms, and they also show people at different stages of their lives. We hope that this gives a broad view of the diversity of progressive ataxias, feelings at diagnosis, and the subsequent problems and solutions of everyday living.

Patient 1: Glen

Ellen Shorey tells the story of her son Glen, now 11 years old, who has Friedreich's ataxia.

"There was no history of ataxia in our family. I knew there was a problem with our younger son, Glen, when he was four. He didn't like going up and down stairs, and eating was a nightmare. He wouldn't chew anything with a texture. At school, they weren't happy with his handwriting; he wouldn't climb on the apparatus, but the school doctor was unconcerned.

"After I made a lot of fuss, he was diagnosed with dystaxia. When I insisted there was more to it, the paediatrician told me to see a psychiatrist. We did the rounds of various specialists. Eventually, a dietician identified scoliosis. More tests followed over many months, before a letter (which should have gone to our general practitioner) confirmed that Glen had a progressive ataxia.

"We had six more weeks of waiting to try to discover which ataxia Glen had. Our general practitioner finally told us Glen had Friedreich's ataxia.

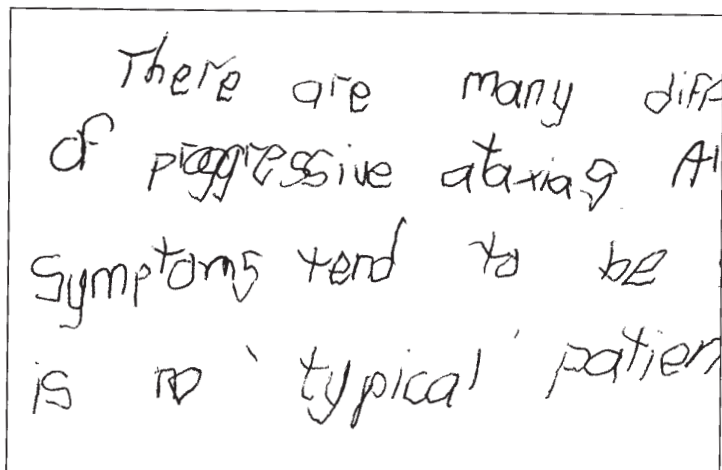
"At the time, Glen accepted it. His attitude was 'At least I can tell my friends I'm not stupid.' But two years later, he was in denial: 'I'm not going in a wheelchair.' Our family life has been torn apart. I feel I've neglected my other two children.

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Example of handwriting from a patient with cerebellar ataxia

"Glen is now at secondary school. The authorities say he doesn't need any extra help because he's got no mental problems. I keep pushing and pushing—I never stop. I wouldn't want another family to go through what we went through to get a diagnosis."

Patient 2: Rachel

Liz Harrison, a general practitioner, has an oldest daughter, Rachel, who has Friedreich's ataxia.

"Rachel was bullied at school, but so are a lot of children. At about 15, she became terribly tired at school and extraordinarily wobbly. So we went to see her general practitioner. I have never in my working life seen another case of Friedreich's ataxia. So I didn't know what we were dealing with.

"After nerve conduction tests the diagnosis was peripheral sensory neuropathy. But in the two years between 15 and 17, she realised that something was really wrong. When she was 17, her neurologist suggested a genetic test. It was fairly new, but Rachel was old enough, and very sensible, and able to make her own choice. We didn't make the choice for her. After four months, Friedreich's ataxia was confirmed.

"But when the result came back, for me it was the most enormous relief because suddenly we had a definite diagnosis, and we found it opened doors. It makes life a lot easier once you have a label. A lot of people never get a diagnosis and have to deal in that grey area for years.

"Rachel went off to university to read maths. It was a courageous decision because, although she's good at maths, she's not brilliant, but it doesn't involve a lot of writing. In her second year she went 'off her legs' and started using a wheelchair. She says she cried a lot that year, and she felt very angry and alone. I was angry too.

"It doesn't stop; there's always something else. Rachel can't drive. And she never will. We have two younger children who seem to be perfectly normal, and they've had to watch their eldest sister go off her legs. They were in their early teens. They've also wondered, 'Oh, I could have it too.'

"How do you cope? We coped by not making ataxia the focus of our lives. It's part of our lives, but we all have other things that we're doing. We deal with one thing at a time and don't try to do too much. And we try to anticipate what is going to be necessary, because there is a lot of help, but the system is enormous and sluggish. We've had to kind of flag up what we'll need in six months' time.

Box 1: Some symptoms associated with progressive ataxia

- Balance problems
- Coordination problems (leading to difficulties in writing, as well as in many other daily activities)
- Slurred speech
- Hearing problems
- Vision problems
- Weakening of the heart
- Diabetes
- Scoliosis
- Pes cavus (overarched foot)
- Swallowing problems

Box 2: Ataxia with vitamin E deficiency—a treatable form of ataxia

Ataxia with vitamin E deficiency presents with many similar symptoms to Friedreich's ataxia. People have been misdiagnosed as having Friedreich's ataxia instead of ataxia with vitamin E deficiency. This form of ataxia responds to treatment with vitamin E supplements.

"Many years ago (I am now 46 years old) my parents were told I had Friedreich's ataxia. I always refuted that diagnosis and kept trying to find out what my real problem was. This time last year I went for computed tomography scans, a spinal tap, and numerous blood counts. Only then did my doctor diagnose a deficiency in vitamin E. I, of course, have been 'gobsmacked' at such a simple sounding problem." (Dianne Fraser, personal communication, 2003.)

It is important to test for vitamin E measurements when diagnosing the symptoms of Friedreich's ataxia. People who had a diagnosis of Friedreich's ataxia years ago, who have not had a genetic test to confirm this, should also have vitamin E concentrations tested.

"Since university, Rachel has been teaching A level maths, one to one, at a local sixth form college. She has a full life, living independently, with good friends and family support."

Patient 3: Harriet

Harriet Bonney has cerebellar ataxia and is a doctor.

"I'd always wanted to study medicine and qualified from Glasgow Medical School in 2001. I'm now a senior house officer in psychiatry, sometimes working weeks of 90 hours or more.

"When I was 15 I was diagnosed with Friedreich's ataxia. However, when a genetic test became available some six years later, I discovered that the original diagnosis was incorrect and that I have an unidentified cerebellar ataxia.

"I am independently mobile, but when I'm tired or stressed my ataxia gets worse. I get comments from colleagues—usually in jest—about my walking. It's hard to hear, but I've just had to get used to it. Some people presume I've been drinking, even in the morning. That's an experience shared by many people with ataxia because of the combination of unsteady walking, poor coordination, and slurred speech.

"In the 10 years of training and working as a doctor I have met only two patients with a hereditary ataxia. It's not surprising that doctors just don't know about it, but it's important that general practitioners are aware of the cerebellar ataxias. Early diagnosis is so important for patients. They can access appropriate help earlier in the course of their disease. It also means there are larger cohorts for research projects."

The final stages

For some patients, the last stages of their illness will require continuing care by professionals in a care home. People with cerebellar ataxia have been in the news recently, intimately involved on both sides of the debate about the right to live or die. It should be emphasised, however, that total physical dependency is not the inevitable outcome for all people with the disorder. We are aware of people with ataxia who are in

their 60s and older and, although they may be in a wheelchair, take an active and joyous part in family life.

Although the progressive ataxias are often life shortening, it is impossible to predict life expectancy. This is because there are many different progressive ataxias and variation between individuals is great. For example, we know of a baby diagnosed with ataxia who died under the age of 1, and at the other end of the spectrum there are people with ataxia in their 90s.

Companions on the journey: medical professionals

As with other chronic conditions, a multidisciplinary approach is essential to manage the ataxias successfully. A neurologist is a key companion, involved in the initial diagnosis and ongoing monitoring. A clinical geneticist and genetic counsellor will also be relevant in certain cases. Physiotherapists, speech and language therapists (for advice on speech and at later stages to help with swallowing and choking problems), occupational therapists (for adaptations in the house and walking aids), and social workers are all closely involved. Referral to a wheelchair clinic for specialist seating advice is also important at the appropriate stage of the condition. Regular monitoring by a cardiologist is important for people with Friedreich's ataxia if they develop cardiomyopathy, as well as referral to an orthopaedic surgeon for their scoliosis or pes cavus (overarched foot). Box 2 gives an example of a treatable form of ataxia.

Finding help along the way

People have also identified the following as helpful:

- getting a diagnosis (see Liz's story about Rachel)
- sensitive health professionals
- contact with others in a similar position (for example, of a similar age with ataxia; other parents; local support)
- carrying a card explaining the effects of ataxia (to explain that people are not necessarily drunk if they are unsteady on their feet and slur their words)
- going into a wheelchair; after accepting the inevitable, many people enjoy increased mobility and independence.

Box 3 shows what made a difference for Glen's father.

Box 3: What makes a difference?

Glen's father, Paul, recommends counselling. "When our doctor first suggested counselling I was totally opposed to the idea. Then I thought about it, and decided to give it a try.

The sessions are held at our local surgery, and they're pretty informal. It's helped us to realise that my wife and I need to take some time for each other, and that our youngest gets left out because we focus so much on his brother. He doesn't get involved in the counselling directly, but the counselling has helped think about his needs.

It's been a help in practical ways too. Because the doctor is so experienced he gives us a fresh approach about how to tackle the different health departments. I think everyone in this situation should jump at the chance. I've been bereaved, but coming to terms with ataxia is another thing altogether."

Additional resources for patients and professionals

Ataxia UK—Provides support services and funding for medical research and information on the ataxias for both lay people and doctors, including personal experiences, local branches, and research (www.ataxia.org.uk; tel 0207 582 1444)

Geneclinics—American website funded by the National Institutes of Health (NIH). GeneClinics is expert authored and peer reviewed; includes information on genetic testing, has clinical descriptions of inherited conditions (including the ataxias), and information on research (www.geneclinics.org)

Euroataxia—International organisation formed by national ataxia groups throughout Europe. Publish a comprehensive newsletter focusing on research (www.euro-ataxia.org)

Friedreich's Ataxia Research Alliance—American organisation focusing on Friedreich's ataxia (www.faresearchalliance.org/)

Conclusions

Supportive, informed companions are essential for people with a diagnosis of ataxia. Some feel that meeting others with ataxia helps prevent isolation, and as the ataxias are uncommon it is difficult to do this without support groups. The internet is also a hugely helpful companion for people with mobility and speech problems. An understanding of ataxia by friends, relatives, and healthcare professionals makes the patient's journey less difficult.

Current research

Research is ongoing and has resulted in improvements in diagnosis, with the recent discovery of many new genes that cause ataxia. Research has also led to the discovery of a potentially treatable form of cerebellar ataxia caused by sensitivity to gluten. Trials are under way for Friedreich's ataxia, testing the effect of antioxidants, with initial promising results. Also the recently discovered ataxia with coenzyme Q10 (CoQ10) deficiency responds to treatment with CoQ10.

Communication is key

Communication between the patient's doctor, other professionals, and the patient is vital. At recent consultation meetings organised by Ataxia UK, this was highlighted by patients and neurologists alike. A consensus was reached at the consultation meetings for the development of specialist ataxia centres of excellence throughout the UK which would be given Ataxia UK accreditation if they meet specific criteria. The first two Ataxia Centres, in London and Belfast, have been established. For more information on these centres contact Ataxia UK.

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