

About Ataxia:

- The cerebellar ataxias are a group of rare neurological disorders, many of which are inherited.
- Inherited cerebellar ataxias are serious genetic conditions that progressively get worse. The rate of progression varies between individuals.
- Initial symptoms typically include poor co-ordination of the arms and legs and slurred speech. As the ataxia progresses people can lose the ability to walk and do other activities, sometimes leading to total physical dependency. There may be further complications such as problems with vision, hearing and swallowing.
- Symptoms begin at different ages for different types of ataxia and this also varies between individuals. Some ataxias are diagnosed in childhood, others later in life.
- Some of the inherited cerebellar ataxias are diagnosed with genetic tests, and these tests can also be offered to other family members.

Find out more

To find out more about ataxia, and for information and support contact:

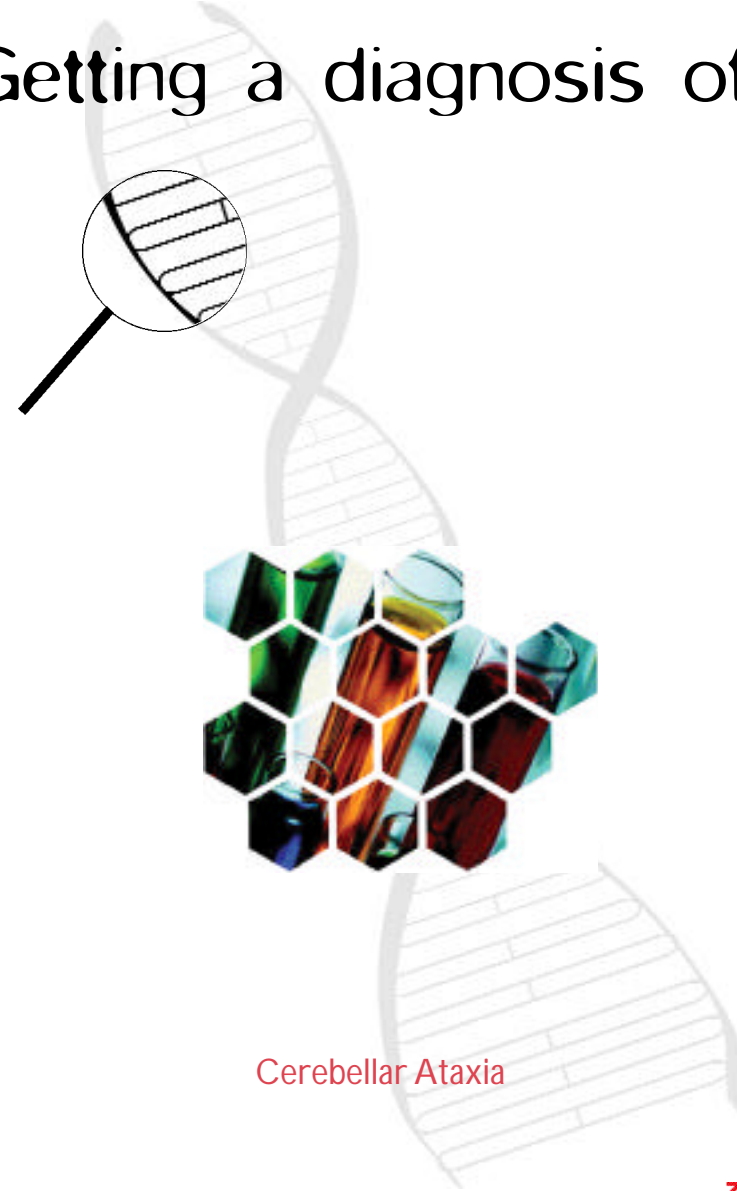


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Getting a diagnosis of...



Cerebellar Ataxia



Cerebellar Ataxia

Who should have a genetic test for cerebellar ataxia?

The cerebellar ataxias are a group of rare neurological disorders, many of which are inherited. Some are inherited in a dominant fashion, so if a person has ataxia there is a fifty percent chance it will be passed to each of their children. Examples of these are the spinocerebellar ataxias (type 1, 2, 3 etc.). Some are inherited recessively. This is when a child could inherit ataxia from parents who are carriers but do not have ataxia themselves. The most common of the ataxias inherited in this way is Friedreich's ataxia. There are also some rare ataxias that are inherited only from the maternal line (mitochondrial ataxias), or can be X-linked (only males are affected).

Anyone who has a diagnosis of cerebellar ataxia could have a genetic test to see whether they have one of the inherited ataxias for which tests are available. At the moment such tests are available for Friedreich's ataxia, spinocerebellar ataxia (SCA) 1, SCA2, SCA3, SCA6, SCA7, SCA10, SCA12, SCA14, SCA17 and DRPLA. Although a number of other ataxias have been identified, (for example 23 SCAs have been identified to date), the genes responsible for these ataxias have not been found, and there are therefore no genetic tests for them.

If someone has tested positive to a genetic test for an inherited ataxia it is possible to test other family members. In this case it is also possible to have a prenatal genetic test to determine whether an unborn baby will develop an inherited ataxia. These are complex issues and discussing them with a trained genetic counsellor may be helpful.



Ellen and Paul are the parents of Glen, who has Friedreich's ataxia. They have two other children.

'From the time he was about four and a half, I thought there was something wrong. Glen's handwriting was bad, he didn't like climbing on apparatus, he was wobbly when he walked.'

After badgering teachers and then the school doctor, Ellen was told that Glen had a problem with his balance and co-ordination.

'When I insisted there was more to it, I was told to see a psychiatrist.'

A paediatrician confirmed the original diagnosis. But physiotherapy and occupational therapy made no difference. Meanwhile, Glen had developed a lump on his shoulder blade, next to his spine.

'After more battles, I managed to get Glen seen by a consultant who specialised in nutrition. He immediately said, "Does your son always walk like that?" and identified scoliosis.'

After a series of tests, Ellen and Paul discovered that their son had something called 'progressive ataxia'.

'I'd never heard of it. I looked it up on the internet. I read about all the different ataxias – was Glen's a 'mild' one? Whatever it was, it was really bad.'

We went back for the final results and were told Glen had Friedreich's ataxia. We knew what it meant, because we'd read up on it. My husband started to cry. I'd never seen him cry in his life.'

At that time, Glen accepted the diagnosis. He said, "At least I can tell my friends I'm not just stupid."

Ellen and Paul also got in contact with Ataxia UK, who were able to put them in touch with other parents. Attending the Annual Meeting in 2003 was a daunting proposition, but they found it enlightening and inspiring. The family has taken part in Ataxia UK Touch Therapy trials, to discover if simple massage techniques can help children with ataxia and their parents.

Ellen and Paul are also energetic fundraisers. Glen's school and other local groups have raised an amazing amount of money for Ataxia UK.

'The diagnosis took three years – I hope no one else goes through that. I kept pushing and pushing and pushing, though people just thought I was a neurotic mum. If we'd known earlier, I could have started battling then for things he needs now, like help at secondary school.'

After Glen was diagnosed, most other family members were tested to see if they had the faulty gene that causes Friedreich's ataxia, or were carriers.

'I was terrified for my other children but I needed to know. My youngest son, who's five, doesn't have the faulty gene and he's not a carrier. Neither is my husband's daughter from his first marriage. She's now in her twenties and she wanted to know. My oldest son is 15 and decided not to take the test at the moment. He says when he is older, if he is in a stable relationship, he will want either himself or his girlfriend to be tested.'