

Better understanding of ataxia could improve the lives of patients with the condition, says Mary-Claire Mason

FEAR OF FALLING

It is common for people who have ataxia to be accused of being drunk because symptoms include slurred speech and problems walking in a straight line.

More than 10,000 people in the UK are thought to be affected by a form of the condition, according to the charity Ataxia UK. However, there continue to be misunderstandings among healthcare professionals. They usually recognise ataxia as a symptom of conditions such as multiple sclerosis, but are much less familiar with cerebellar ataxias.

These are a complex group of complaints in which there are at least 60 different diseases, most hereditary and progressive, says Paola Giunti, consultant neurologist at the National Hospital for Neurology and Neurosurgery in London. The most common inherited ataxia is Friedreich's ataxia, and 32 types of spinocerebellar ataxia have so far been identified.

Cerebellar ataxia symptoms include slurred speech, difficulty swallowing, blurred and double vision, tremors and fatigue, and problems with walking that can result in unsteadiness and falls. Friedreich's ataxia can cause heart muscle thickening,

SUMMARY

The ataxias are a complex group of conditions for which there is no cure. But specialist care can help with the symptoms.

Keywords

Ataxia • Cerebellar ataxia • Disability • Disease management • Genetics • Isolation • Progressive disease • Support • Therapist

About ataxia

- ▶ Around 10,000 people in the UK have ataxia.
- ▶ Cerebellar ataxias are comprised of at least 60 different diseases.
- ▶ Most have a genetic component.
- ▶ Anyone of any age can get ataxia, but some types are more common in certain age groups.

Source: www.ataxia.org.uk

diabetes, hearing difficulties and scoliosis.

The age at which symptoms begin depends on the type of ataxia. Friedreich's ataxia, for example, often develops in childhood or teenage years, while the age of onset for Type 6 spinocerebellar ataxia is the late forties or early fifties. The severity of symptoms varies, but they are usually progressive.

Misdiagnosis

A lack of understanding about how various cerebellar ataxias present has sometimes resulted in misdiagnosis or underdiagnosis. An incorrect perception that little can be done for people with ataxia can mean patients are left to fend for themselves, compounding feelings of isolation and depression, says Dr Giunti.

Ataxia UK chief executive Sue Millman says the charity is working hard to change this. It first published best practice clinical guidelines for healthcare professionals two years ago. These now include a section on palliative care, as well as diagnosis, treatment and management information.

'Multidisciplinary management of these chronic conditions is paramount,' stresses Ms Millman. She adds that people need a

joined-up service where they see expert clinicians. To meet this need, the charity is setting up specialist ataxia centres around the UK in addition to the three already established.

Dr Giunti runs a centre that was set up in 2005. Patients at the diagnosis and treatment clinic see Dr Giunti, clinical nurse specialist Liz Redmond and an Ataxia UK volunteer.

Dr Giunti explains that it is important to diagnose the specific type of ataxia where possible. This gives the patient a sense of their prognosis, the likelihood of passing it on to any children and implications for other family members.

There is no cure, but symptoms can be managed. There are research trials investigating treatments for specific ataxias.

A second clinic has a multidisciplinary team, comprising Dr Giunti, an occupational therapist, physiotherapist and a speech and language therapist. 'The aim is to organise a tailor-made programme that the patient's primary care team can continue at home,' Dr Giunti says.

Ms Redmond says: 'My job is to act as the patient's advocate, to co-ordinate care and provide continuity of care. I support patients through the testing process and help them understand their chances of passing on the condition if a gene has been identified.'

'I make sure patients get the right medical care, therapy and support because that can



make all the difference to their quality of life.'

She helps people understand that there is life after an ataxia diagnosis. 'It can be devastating,' Ms Redmond says. 'Your whole world is turned upside down. You may feel trapped inside your home, scared to venture outside for fear of falling. You start to lose your independence and find it hard to do things you once took for granted, like talking on the telephone.'

'It is vital to keep an open mind about symptoms and not attribute everything to the cerebellar ataxia. We had one patient who had a persistent cough that was nothing to do with the disease. She was referred to, and successfully treated by, a chest physician.'

'It is also important to recognise that patients with ataxia need time to talk through their concerns and to understand that stress may aggravate problems such as slurred speech.'

'Emotional support is another part of the service I provide – listening to patients when they are going through dark times.'

Before receiving her diagnosis of cerebellar ataxia aged 19, Deanna Kaye was told she would grow out of her clumsiness and writing difficulties. Since then her condition has deteriorated gradually.

Specialist centre

'Over the years I have struggled with my balance, tripping and falling many times. My working life was difficult because of my physical limitations.'

'I did things more slowly than other people and my speech became more slurred when I was under pressure.'

Now 62, she still walks unaided. She adds: 'Every waking moment I have to concentrate hard on where to put my hands and feet to reduce the risk of falling.'

Ms Kaye has been referred to the London ataxia clinic for a variety of therapies. She says the centre is 'superb' because of its specialist nature. She finds the physiotherapy it provides especially useful because it has strengthened her abdominal and leg muscles.

Ms Kaye volunteers at the centre, offering support to people recently diagnosed with cerebellar ataxia. She says: 'You can feel isolated and frightened when you find out you have ataxia, which is a condition you may not have heard about before. Talking to someone with personal experience helps you realise that you are not alone' **NS**

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Clinical nurse specialist Liz Redmond (right) and senior physiotherapist Victoria Waddell (left) help patient Deanna Kaye

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