SCA27B (Spinocerebellar ataxia type 27B) - updated June 2025

What is SCA27B
SCA27B is a type of cerebellar ataxia. The cause of SCA27B was recently discovered in 2022, so researchers are still trying to understand the condition, including how common it is.

What are the symptoms of SCA27B?
Typically, the first symptoms of SCA27B are unsteady gait, stumbling, and imbalance. **[Chaar, WA. et al. Clinical, Radiological and Pathological Features of a Large American Cohort of Spinocerebellar Ataxia (SCA27B). Ann Neurol. 96: 1092-1103, 2024.]** Other common symptoms include vision problems (double vision, bouncy vision, or blurry vision), poor hand coordination, vertigo or dizziness, and difficulty speaking.

Less common symptoms include swallowing difficulties, tremors, urinary urgency, stiff or rigid muscles, and numbness or pain in your limbs.

In about 50% of cases, SCA27B symptoms begin as episodes. This is when people experience bouts of ataxia lasting from minutes to days but then regain their sense of balance and other symptoms go away. Over time, episodes may become more frequent until an individual experiences a more permanent form of ataxia, where symptoms are constant. Some factors that can trigger SCA27B ataxia episodes are alcohol, physical activity, and caffeine.

What causes SCA27B?
SCA27B is a genetic disorder caused by a change in the FGF14 gene. **[Pellerin, D. et al. Spinocerebellar ataxia 27B: A novel, frequent and potentially treatable ataxia. Clin Transl Med. 14: e1504, 2024].**

The change in the FGF14 gene that causes SCA27B is known as a repeat expansion, where a section of the letters that make up the genetic code is repeated lots of times. In SCA27B, the repeated section is a GAA section, and is known as a GAA repeat.

How is SCA27B inherited?

SCA27B is a genetic disorder which means that it is an inherited disease. SCA27B is an autosomal dominant disease which means that individuals of either sex are equally likely to inherit the gene and develop the disease. Each child of a person with SCA27B has a 50% chance of inheriting the SCA27B gene.

Healthy individuals will have fewer than 200 GAA repeats in the FGF14 gene. If a person has 200-249 GAA repeats, it is not clear what this number of repeats means, as scientists do not have enough data on what this number of repeats does in the body. So, with 200-249 repeats, some people may develop symptoms, whilst others may not. Similarly, if a person has 250-300 GAA Repeats, they may or may not develop symptoms. If someone has 300 GAA Repeats, they have reached the threshold to be diagnosed with SCA27B. Once a person reaches over 300 repeats, they will develop ataxia symptoms.

GAA repeat numbers can increase or decrease between generations. This can make it seem like ataxia ‘skips’ a generation, if the GAA repeat number decreases below the number needed to cause SCA27B. If in the following generation, the GAA repeat number reaches over 300 again, then individuals will develop symptoms. GAA repeat numbers are more likely to increase when passed to children by their mothers, whilst they are more likely to decrease when passed to children by their fathers.

For more information on inheritance see Ataxia UK's *'Ataxia: what's that'?'* leaflet.

When do symptoms start?
SCA27B is considered a late-onset condition, as age at onset of SCA27B symptoms can range from 50-80 years old. **[Pellerin, D. et al. Deep Intronic FGF14 GAA Repeat Expansion in Late-Onset Cerebellar Ataxia. N Eng J Med. 388: 128-141, 2023.]** However, symptoms can occur as early as 30 years old and as late as 90 years old. The severity of symptoms also varies considerably, even within families.

SCA27B generally progresses very slowly. Most people with SCA27B will eventually use walking aids such as a stick or walker. However, most do not require the use of wheelchairs. Lifespan generally is not shortened by the disease.

How is SCA27B diagnosed?
A neurologist will perform an examination to determine whether a person has symptoms of SCA27B. **[Chaar, WA. et al. Clinical, Radiological and Pathological Features of a Large American Cohort of Spinocerebellar Ataxia (SCA27B). Ann Neurol. 96: 1092-1103, 2024.]** This suspected diagnosis is then confirmed through brain imaging, such as MRI, and genetic testing to detect the presence of the abnormal gene (FGF14) that causes SCA27B.

The genetic test for SCA27B is not currently available on the NHS. Dr David Pellerin and Professor Henry Houlden at University College London are collaborating with the core genetic lab at Great Ormond Street Hospital in London to develop the diagnostic test, and expect to start offering the test in 2025.

If you have a genetic diagnosis of SCA27B, please let us at Ataxia UK know by emailing **sparr-reid@ataxia.org.uk**, so we can plan for future trials.

If your neurologist contacts researcher Dr David Pellerin, they can provide more information about genetic testing as part of their research project.

Contact details:

Dr David Pellerin and Professor Henry Houlden
Institute of Neurology, Queen Square, London WC1N 3BG
david.pellerin.21@ucl.ac.uk or h.houlden@nhs.net.

Patient referrals for assessment and testing can be sent to h.houlden@nhs.net.

How common is SCA27B?
So far, we know that SCA27B is a prevalent form of cerebellar ataxia, seen in lots of different countries, with many cases in the French-Canadian region. **[Pellerin, D. et al. Spinocerebellar ataxia 27B: A novel, frequent and potentially treatable ataxia. Clin Transl Med. 14: e1504, 2024].**

As more people are diagnosed with SCA27B and more research is completed, we will have a better understanding of the condition. This leaflet will be updated with new information as it becomes available.

Management of SCA27B
Whilst there is no cure for SCA27B, treatment focuses on management of symptoms such as through physiotherapy, occupational therapy, and speech and language therapy. **[Pellerin, D. et al. GAA-FGF14-Related Ataxia. 2024. https://www.ncbi.nlm.nih.gov/books/NBK599589/]**

Clinical trials for SCA27B

In 2025, the pharmaceutical company Solaxa announced that they will be conducting a phase 3 clinical trial of the repurposed drug 4-Aminopyridine in SCA27B, taking place in the US, with further sites to come. Read about the trial here: https://tinyurl.com/25tvybff.

Ataxia UK-funded Research on SCA27B

In 2024, Ataxia UK funded a research project looking at genetic modifiers of the age of onset of SCA27B. The project is being led by a collaborative group, including Professor Stephan Züchner, David Pellerin, Henry Houlden and Bernard Brais; with the lead site being at University of Miami Miller School of Medicine in the US.

Read about the project here: https://tinyurl.com/2tr2dwnd.

This information leaflet was written by Ataxia UK in collaboration with Professor Henry Houlden at the UCL Queen Square Institute of Neurology and The National Hospital for Neurology and Neurosurgery, London, UK.

June 2025

For more support or information please contact:
Ataxia UK, 12 Broadbent close, London N6 5JW
Tel: +44 (0)20 7582 1444
www.ataxia.org.uk