# Spinocerebellar ataxia

**What is Spinocerebellar ataxia?**

Spinocerebellar ataxia (SCA) is a term referring to a group of hereditary ataxias that are characterized by degenerative changes in the part of the brain related to the movement control (cerebellum), and sometimes in the spinal cord.[1] There are many different types of SCA, and they are classified according to the mutated (altered) gene responsible for the specific type of SCA. The types are described using "SCA" followed by a number, according to their order of identification: SCA1 through SCA40 (and the number continues to grow).

**What are the symptoms of Spinocerebellar ataxia?**

There are many different types of spinocerebellar ataxia (SCA) and each may have unique signs and symptoms. However, in general, it is difficult to differentiate among the different types, and all are characterized by problems with movement that tend to get worse over time. Affected people may experience the following:

- Problems with coordination and balance (ataxia)
- Uncoordinated walk
- Poor hand-eye coordination
- Abnormal speech (dysarthria)
- Involuntary eye movement
- Vision problems
- Difficulty processing, learning, and remembering information

Depending on the type of SCA, signs and symptoms can develop anytime from childhood to late adulthood. SCA3, also known as Machado-Joseph disease, is the most common type of SCA. SCA types 9 through 36 are rare and less well characterized.

## How is Spinocerebellar ataxia diagnosed?

Genetic testing is available for many different genes known to cause spinocerebellar ataxia (SCA).[3] Carrier testing for at-risk relatives and prenatal testing are possible if the disease-causing mutations in the family are known.

For some types of SCA, the genetic cause is still unknown.[3] Genetic testing is not available for families with these types of SCA.

A diagnosis of spinocerebellar ataxia (SCA) is often suspected when certain signs and symptoms, such as a poorly coordinated gait (walk) and uncoordinated hand/finger movements, are present. Genetic testing is the best way to confirm SCA and identify the specific type, especially when a person also has family members with similar features. However, this is only an option if the disease-causing gene for that particular type of SCA has been identified. At this time, the genetic cause of some of the types is currently unknown; in these cases, imaging studies such as computed tomography (CT scan) and/or magnetic resonance imaging (MRI scan) may be necessary to establish a diagnosis. A CT scan is an imaging method that uses x-rays to create pictures of cross-sections of the body, while an MRI scan uses powerful magnets and radio waves to create pictures of the brain and surrounding nerve tissues. Both of these imaging methods can be used to identify brain abnormalities found in people with SCA.

## Treatment options for Spinocerebellar ataxia

There is no known cure for spinocerebellar ataxia (SCA). The best treatment options for SCA vary by type and often depend on the signs and symptoms present in each person. The most common symptom of SCA is ataxia (a condition in which coordination and balance are affected). Physical therapy can help strengthen muscles, while special devices (e.g., cane, crutches, walker, or wheelchair) can assist in mobility and other activities of daily life. Many people with SCA have other symptoms in addition to the ataxia such as tremors, stiffness, muscle spasms, and sleep disorders; medications or other therapies may be suggested for some of these symptoms. One report described some improvement in the symptoms with zolpidem 10 mg in four out of five family members with SCA type 2, and a trial of 20 patients with SCA3 found that varenicline led to improvement in some, but not all of the symptoms.

**Long-term complications of Spinocerebellar ataxia**
The long-term outlook (prognosis) for people with spinocerebellar ataxia (SCA) varies. Disease progression and severity often depend on the type of SCA.

Most available information on the prognosis of SCA is based on the four most common types: SCA1, SCA2, SCA3 and SCA6. People affected by one of these types of SCA usually require a wheelchair by 10-15 years after the onset of symptoms. Many will eventually need assistance to perform daily tasks.

**Resources**

- **Genetic and Rare Diseases Information Center (GARD) -** https://rarediseases.info.nih.gov/diseases/10748/spinocerebellar-ataxia
- **Spinocerebellar Ataxia: Making an Informed Choice About Genetic Testing** - http://ataxia.org/wp-content/uploads/2017/07/SCA-Making_an_Informed_Choice_About_Genetic_Testing.pdf
- **The Genetic Testing Registry (GTR)** - www.ncbi.nlm.nih.gov/gtr/conditions/?term=spinocerebellar+ataxia