ARSACS (Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay)

ARSACS

Autosomal recessive spastic ataxia of Charlevoix-Saguenay, more commonly known as ARSACS, is a type of ataxia associated with spasticity (increased muscle tone) particularly in the legs. This was first described in people of the Charlevoix-Saguenay region of Quebec, Canada [1]. Cases of ARSACS have now been seen in a number of countries.

How common is ARSACS?

The incidence of ARSACS in the Charlevoix-Saguenay region of Quebec, where it is more common, is estimated to be 1 in 1,932 [2]. Outside of Quebec the incidence is unknown. Although ARSACS is rare, it is now considered one of the more common recessive ataxias worldwide, with cases described in several countries including the UK.

What are the symptoms of ARSACS?

The most common symptoms of ARSACS include stiffness in the legs, balance problems, difficulty walking, reduced manual dexterity, incoordination of the arms and speech difficulties. As the disease is associated with peripheral neuropathy (damage to the peripheral nerves) patients may experience sensory loss in their legs and arms, sometimes associated with cold extremities and discoloration of the feet. Each person is unique and may not be affected the same way or at the same level as another person.

When do symptoms start?

Symptoms generally appear before the age of 10 (but can start any time between 0 and 40 years). The symptoms progress throughout adolescence and adulthood.

What causes ARSACS?

Mutations (abnormalities) in the SACS gene cause ARSACS [1]. The SACS gene provides instructions for producing a protein called sacsin. Sacsin is found in the brain, skin cells, muscles used for movement (skeletal muscles), and at low levels in the pancreas, but the specific function of the protein is unknown [3]. Mutations in the SACS gene cause the production of an unstable sacsin protein that does not function normally. It is unclear how the abnormal sacsin protein results in the signs and symptoms of ARSACS.

How is ARSACS inherited?

ARSACS is inherited through what is known as an 'autosomal recessive' inheritance pattern. This means that one copy of the abnormal (also referred to as mutated) SACS gene needs to be inherited from each parent for a person to have the condition. Therefore, if both parents are carriers of the abnormal SACS gene, then each child has a 25% chance of developing the condition. For more information on inheritance see Ataxia UK's '***Ataxia: what's that'?'*** leaflet.

How is ARSACS diagnosed?

ARSACS is diagnosed through genetic testing. A genetic test involves taking a blood sample to detect the abnormal gene. Of interest is the fact that there are a lot of different mutations on the ARSACS gene that can cause the disease. These mutations may differ in different countries. For example, the common mutations seen in Quebec, Canada are different to what is seen in the UK. However whole genome sequencing which is the genetic test done to look for genetic causes of ataxia, is able to identify all the described genetic mutations.

Management of ARSACS

Some of the symptoms associated with ARSACS can be treated, such as with anti-spasticity medication, and medication to treat neuropathic pain. As with other ataxias, physiotherapy and speech and language therapy can be helpful. A visit by an occupational therapist will be useful in order to assess the need for items such as walking aids, or for adaptations to the home. It is important to see a neurologist, who will monitor the condition, on a regular basis.

This information leaflet was written by Ataxia UK in collaboration with Professor Marios Hadjivassiliou, Consultant Neurologist at the Sheffield Ataxia Centre.

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References

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