

## **Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (ARSACS)**

What are the symptoms of ARSACS? How does the disorder progress?

ARSACS affects the spinal cord and the peripheral nerves; it has no incidence on the other parts of the body. The severity of symptoms differs slightly among people of specific age groups. Usually, there is no physical pain directly associated with the disorder.

Symptoms usually appear in early childhood, when children are able to walk unassisted. Due to a lack of balance or sometimes the presence of stiffness in the

legs, children living with ARSACS will show a tendency to fall more frequently, a warning sign that will bring parents to seek medical advice. Symptoms are generally rather mild until the teenage years. The disorder may even be unnoticed in some children. Frequent falls, an uncoordinated gait (ataxia), difficulty carrying out manual activities, and problems with writing are often observed during school age. Some children will also experience difficulties with sports such as bicycling, skating, or skiing.

During adolescence, teenagers will face increasing difficulty walking and a decrease in manual dexterity. Stiffness in the legs will often add to gait impairment. Young adults will often need the support of a cane or handrail to help with ambulation. Deformity of the feet (hollow foot) and hands is common. However, the ability to pursue study or work that does not require extensive standing positions or a high level of manual dexterity is maintained. Many people living with ARSACS are able to drive a car for several years. In some cases, the vehicle may need to be adapted to the driver's needs. At that stage, the

symptoms are such that the person might falsely appear under the influence of alcohol, and it would therefore be wise to have a medical certificate assessing their condition in order to prevent any potential troubles or prejudices.

### **What is Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (ARSACS)?**

Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (ARSACS) is a hereditary progressive neurological disorder that mainly affects people from the Saguenay-Lac-St-Jean (SLSJ) and Charlevoix regions as well as people whose ancestors are native to these regions. It is characterized by degeneration of the spinal cord and progressive damage of the peripheral nerves.

ARSACS can affect men and women alike. About 250 people of the SLSJ and Charlevoix regions have the disorder. And one (1) in 22 people living in these areas are carriers of the gene involved in ARSACS.

An adult living with the disorder usually uses a manual wheelchair for transportation outside the home. There is a need for special equipment to assist with bathing and getting up from the toilet seat. Many adults with the disorder have the necessary capacities to live on their own, but may need assistance with household maintenance and meal preparation. Some adults may experience difficulty with bladder control, and occasionally, with bowel control. However, most have no incontinence problems.

Older adults (between age 55 and 70) living with ARSACS may find mobility easier in a manual or electrical wheelchair. They often need help to transfer from one place to another. Severe dysarthria is often observed. Assistance with personal care, household maintenance, and meal preparation is often required.

ARSACS is a slowly progressive disorder that could remain stable for several years. It does not cause any intellectual barriers. Life expectancy of people with the condition is slightly shortened.

## What causes ARSACS?

ARSACS is caused by a gene mutation located on chromosome 13. This is an autosomal recessive gene. The term autosomal means that this gene is located on one of 22 pairs of chromosomes defining specific traits not related to sex characteristics. The gene responsible for ARSACS is recessive: This means that in order to have a child with the disorder, both parents must be carriers of the gene; when the two (2) parents are carriers, they have 1 in 4 (25%) chances to have a child with ARSACS with each pregnancy. (For a complete explanation of autosomal recessive genetics, see the INFO sheet "Genetics: What is Autosomal Recessive"). Carriers of the ARSACS gene show no symptoms of the disorder. One (1) person in 22 in the SLSJ and Charlevoix regions is a carrier of the ARSACS gene and has a chance to transmit it to their descendants. Most of these people do not know they are carriers.

A team of scientists from Quebec discovered the ARSACS gene in 2000. Two mutations in the gene have been identified and are present in 96.3% of cases; these mutations can be detected through genetic analysis of a blood sample. A very low ratio of gene carriers (3.7%) presents a mutation that is currently impossible to detect by genetic testing.

## What are the risks to have an affected child?

*When both parents are carriers of the ARSACS gene, at each pregnancy:*

- There is a 1 in 4 (25%) chance that the child will have the disorder.
- There is a 2 in 4 (50%) chance that the child will be a carrier of the gene.
- There is a 1 in 4 (25%) chance that the child will neither have disorder, nor be a carrier of the disorder gene.

*When a person affected with ARSACS has children:*

- If his/her partner also has the disorder, then all of their children will also have the disorder.

## How can I know whether I am a carrier of the ARSACS gene?

Parents of a person affected with ARSACS are almost always carriers. The carrier status of the ARSACS gene can be confirmed through genetic testing. This test is available to members of families affected with the disorder and their close relatives. This test may potentially be offered in communities where the risk of being carrier of the ARSACS gene is high.

- If his/her partner does not have the disorder, but is a carrier of the ARSACS gene, they will have a 1 in 2 (50%) chance of having a child with the disorder with each pregnancy. The remaining children will be carriers.
- If his/her partner neither has the disorder, nor is a carrier of the gene, all their children will be carriers of the ARSACS gene, but will not have the disorder.

If you have a family history of ARSACS, you might wish to consult a geneticist or a genetic counselor. These health professionals will diagnose your chances to have a child affected with ARSACS, suggest you to take a genetic test, or discuss with you the family planning alternatives you might wish to consider.

## The physical rehabilitation process throughout different stages of life

Once the diagnosis is confirmed, children may be assessed in physiotherapy in order to evaluate their joint mobility, test their facility to move from one position to another, test their ability to walk, and to evaluate their capacities in activities involving gross motor skills (jumping, running, or climbing stairs). A regular or control monitoring will be proposed, depending on each child's needs, completed with a program of activities to practice at home. This program is aimed at strengthening or maintaining range of motion, coordination and balance.

The physiotherapist will continue monitoring the people living with ARSACS during adolescence and the beginning of adulthood. They may recommend any special equipment needed, such as an orthosis or a walking aid. As part of this monitoring process, they may also suggest appropriate recreational activities offered in the community.

Once the diagnosis is confirmed, children may also be assessed in occupational therapy. The role of the occupational therapist is to foster fine motor skills development and preschool aptitudes of each child, and lessen the impact of the disorder on the lifestyle. Children and teens with ARSACS attend the regular school system. Accessible school transportation is often needed for several of them. Physical education classes have to be adjusted to each child capacities as early as primary school. Therefore, the occupational therapist and the physiotherapist should be available to the day care and school staff to assist with integration of these kids.

Adults living with the disorder rely on the occupational therapist who may assess the need for specialized equipment that can ease mobility and daily living activities such as wheelchairs, scooters, or self-care equipment. Various government or other assistance programs available (such as Muscular Dystrophy Canada) may help support or refer sources of funding for this equipment. An occupational therapist may also assess driving abilities and recommend specific accommodations to bring to the vehicle and home, if necessary.

A neurologist may also monitor people living with ARSACS. This specialist may prescribe specific drugs that will help reduce spasticity or stiffness affecting the

## How is ARSACS diagnosed?

The disorder is usually diagnosed between the ages 2 to 5; diagnosis of ARSACS requires a neurological examination, specific neuropsychological assessments and a genetic analysis. The neurologist is usually the medical specialist who will make the diagnosis. If both parents are carriers of the gene, the disorder may also be diagnosed in the fetus during the pregnancy with a genetic testing made on fetal cells collected either through chorion biopsy or amniocentesis procedures.

lower limbs, or prevent incontinence problems. A neurologist can respond to questions asked by the parents or the individual regarding the course of their disorder.

The orthopedic surgeon is another member of the medical team a person living with ARSACS will be monitored by. They may delay and reduce certain foot deformities by prescribing the use of orthosis, a series of correcting casts treatment, or surgery. Lengthening of the Achilles tendon and ankle fixation (arthrodesis) are the main surgical procedures performed in order to facilitate ambulation and transfer.

The nurse may provide much additional information about the disorder and available services and resources. They may offer support to help the individual and family members cope with the disorder, provide guidance with the administrative processes when required, and liaise with the various health care professionals from the clinic and other external services (CLSC and others).

The genetic counselor can inform the person living with the disorder and their family members about the inheritance processes involved in ARSACS, the genetic testing available to carriers of the disorder's gene, as well as family planning alternatives they may wish to consider.

## What are the psychological effects of ARSACS?

Living with ARSACS is a daunting challenge for the individuals and their families. Parents sometimes go through a great deal of psychological distress when faced with the diagnosis. Preschool children are not usually really aware of their illness and therefore it is very important to reinforce the positive aspects of each child's personality so that they can develop a positive self-esteem.

Learning problems at school are observed in nearly 50% of the adolescents living with ARSACS. Kids with ARSACS sometimes lack motivation at school; a psychological follow-up can help them cope with their disorder and regain confidence in themselves as well as an interest for school learning. A neuropsychological assessment may highlight cognitive issues that can sometimes be corrected.

As a teenager, individuals want to be like their friends and usually avoid talking about a disorder. They may sometimes show a certain denial of their health situation and be unwilling to mix with other kids facing the same challenges. Teenagers may also feel rejected by their friends when it comes to activities requiring good physical skills. People with ataxia can have a normal sexual and marital life, and have children.

## What are the treatments for the disorder?

There are currently no treatments available to cure people with ARSACS. However, several health-care professional may work together to help people living with this disorder to maintain or strengthen their physical abilities. These professionals work in the following medical fields: physical rehabilitation (occupational therapist and physiotherapist), medicine (neurologist and orthopedic surgeon), nursing care, and genetic counseling. They frequently work in a specialized clinic where they monitor people affected with neuromuscular disorders.

## What about research on ARSACS?

Several research teams, particularly in the province of Quebec, are investigating this disorder. Identification of the SACS gene, the gene involved in the disorder, is an important step towards a possible cure. This gene produces a protein known as saccin. While the role of this protein is still unknown, scientists have already formulated interesting hypotheses about its function. Some researchers are also focusing their efforts in order to better understand the disorder evolution process and find more effective ways of treatment, especially treatments that will lessen stiffness in the legs. Finally, several studies have been and are being conducted in the neuropsychological field to further understand the causes of learning problems experienced by children and teenagers affected with ARSACS.

Over the course of their lives, people living with ARSACS will be faced with the challenging process of adapting to their physical condition. Many may develop a strong moral strength and positive attitude towards life in general.

## Acknowledgements

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## How can I help?

Muscular Dystrophy Canada conducts year-round fund raising campaigns to support our diverse programs. Your gift will help the Association provide the dollars necessary to assist individuals living with neuromuscular disorders, and fund much needed medical research and educational information. Please make a gift through our National office or any Regional or Community Muscular Dystrophy Canada office.

**All Muscular Dystrophy Canada Information Sheets are available on our website:**  
[www.muscle.ca](http://www.muscle.ca)

**Ce feuillet d'information est aussi disponible en français.**

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What are the main resources available to people living with ARSACS and their families in regions showing high incidence of the disorder?

**Clinique des maladies neuromusculaires du Saguenay-Lac St-Jean (Saguenay-Lac-St-Jean Neuromuscular Disorders Clinic)** Carrefour de la Santé de Jonquière 2230 rue de l'Hôpital, C.P. 15 Jonquière, QC G7X 7X2 (418) 695-7777

**Clinique des maladies neuromusculaires de Charlevoix (Charlevoix Neuromuscular Disorders Clinic)** Centre Hospitalier de Charlevoix 74, rue Ambroise- Fafard Baie Saint-Paul, QC G3Z 2J6 (418) 435-5150, ext. 2086 and 2087

**Bureau régional de la Dystrophie musculaire Canada (DMC) (Muscular Dystrophy Canada [MDC] Regional Office)** \*Fonds ARSACS de l'ACDM 2230, rue de l'Hôpital, C.P. 15, Jonquière, QC G7X 7X2 (418) 695-7760  
\*A MDC Funds mainly devoted to research on ARSACS.

**Service de conseil génétique (Genetic Counselling Service)** Complexe Hospitalier de la Sagamie 305, St- Vallier C.P. 67 Chicoutimi, QC G7H 5H6 (418) 541-1234, ext. 2153 or 2081

**Bureau provincial de la Dystrophie musculaire Canada (Muscular Dystrophy Association of Canada Provincial Office)**  
1425, boul. René-Lévesque Ouest, bureau 506 Montréal, QC H3G 1T7