National Ataxia Foundation
Connecting Ataxia families, researchers, clinicians and the community

Sporadic Ataxia and Multiple System Atrophy (MSA)

What is Sporadic Ataxia?

Sporadic Ataxia is a term to designate a group of diseases of the central nervous system that occur without any evidence that they were inherited, from a family member. The term refers to changes in the structures of the brain that result from loss of nerve cells in these brain sites: the inferior olive, the pons, and the cerebellum. This disease is termed "neurodegenerative" because the nerve cells in these structures (and often several other structures) gradually disappear over time without a known cause. Physicians often use various terms when they make a diagnosis of sporadic Ataxia. Some of those terms include:

- Olivopontocerebellar atrophy (OPCA) or Olivopontocerebellar degeneration
- Idiopathic late onset cerebellar atrophy or degeneration (ILOCA or ILOCD)

What are the symptoms of Sporadic Ataxia?

Difficulty with balance and incoordination (Ataxia) of the legs and arms are usually the first symptoms of sporadic Ataxia. Many people also develop thick or slurred speech (dysarthria). If the Ataxia becomes severe, the symptoms can make it difficult for a person to work at a job, walk independently, dress, bath, write, eat, and drink. These symptoms progress gradually over time.

What is Multiple System Atrophy (MSA)?

In some people, the symptoms of Sporadic Ataxia are a prelude to the development of Multiple System Atrophy (MSA), which includes Ataxia, Parkinsonian features (such as rigidity and slowness of movement), and difficulty with the autonomic nervous system. The autonomic nervous system controls the automatic functioning of the body, including regulation of blood pressure, digestion, bladder and bowel function, some sexual functions, and sweating.

What are the symptoms of MSA?

When Sporadic Ataxia develops into MSA, those affected frequently experience faintness or lightheadedness when moving from lying down to the standing up position, and they may experience urinary urgency, frequency, and, later, incontinence. In some people, the urinary difficulty arises first and postural lightheadedness occurs later. Men often develop erectile dysfunction several years in advance of other symptoms, including Ataxia. Some people become constipated, and the rare person can become incontinent of stool. People with MSA, whose symptoms begin with sporadic Ataxia, may develop Parkinsonian symptoms of slowness and stiffness of movement along with difficulty turning over in bed and rising from a soft chair.

Are sleep disturbances common?

Sleep disturbances are common in Sporadic Ataxia and MSA. The disorders include rapid eye movement (REM) sleep behavior disorder, a condition in which people act out their dreams with active body movements, sometimes shouting or speaking, and often thrashing in bed and striking the bed partner. People with Sporadic Ataxia and MSA also frequently snore and have obstructive sleep apnea, a disorder in which breathing stops for a few seconds to a minute or so because of airway obstruction.

When do symptoms of Sporadic Ataxia and MSA appear?

The symptoms of Sporadic Ataxia usually occur in middle to older adults and progress over several years. There have been cases of Sporadic Ataxia beginning in childhood, adolescence, and young adulthood, although these cases are unusual. Men and women are equally likely to develop this disorder. Those who have Ataxia and no other symptoms usually have slower progression over the course of the disease than those who develop MSA. There may be some difficulty with judgement and insight as the disease moves along, but this also is unusual. Psychological disturbances, particularly depression, occur frequently in both Sporadic Ataxia and MSA.

How Common is Sporadic Ataxia and MSA?

Sporadic Ataxia is a rare disease affecting about 1 in 100,000 people. MSA affects 4 in 100,000 people, but this includes people who initially develop Parkinsonian symptoms followed by autonomic failure and do not experience Ataxia. MSA affects both men and women and all racial groups.

What causes Sporadic Ataxia and MSA?

Sporadic Ataxia is a diagnosis that encompasses a number of different disorders that can not be defined more fully. We suspect that many cases are due to the inheritance of multiple, different genes that have not as yet been identified, and these genes together with an environmental trigger may bring on the disease. It is unclear why some people with sporadic Ataxia progress to develop MSA whereas others do not.

How is the diagnosis made?

People with Sporadic Ataxia can be difficult for physicians to diagnose correctly because there are many acquired and hereditary causes of Ataxia that must be ruled out before diagnosis of Sporadic Ataxia can be made with any confidence. No test can confirm the diagnosis accurately; rather, the diagnosis is made only after other conditions have been ruled out. Many people with Sporadic Ataxia consult a number of physicians before they receive a diagnosis, and sometimes the diagnosis is made for many years.

Many medical and neurological diseases can be associated with a disorder that appears to be a Sporadic Ataxia and need to be ruled out. These include the remote effects of a cancer; deficiency of vitamin B12, thiamine, or Vitamin E; severe chronic alcoholism with malnutrition and multiple vitamin deficiencies; hypothyroidism; normal pressure hydrocephalus; collagen-vascular disorders such as lupus erythematosus; residual effect of encephalitis; exposure to certain toxins such as heavy metals (lead and thallium); many rare enzyme or metabolic disorders; multiple sclerosis; and multiple types of peripheral neuropathy.

A number of medical tests need to be completed before the diagnosis of Sporadic Ataxia can be made. These include multiple blood tests to look for the disorders identified in the previous paragraph, MRI imaging of the brain to look for degenerative changes in the brainstem (inferior olive and pons) and cerebellum, often an EMG to examine the electrical activity of the muscles and nerves, and, in some cases, spinal fluid examination.

What happens after the diagnosis?

If the diagnosis of a treatable sporadic cerebellar degeneration is received a diagnosis of treatable cause, prompt treatment is absolutely essential to stop the progression of the disorder and, in some cases, to restore normal neurological function. If the cause of the progressive Ataxia cannot be discovered after a thorough investigation, then, by default, the diagnosis becomes Sporadic Ataxia. In people with this disorder, avoiding falls and injuries with appropriate mobility aids becomes essential. A physical therapist may be helpful in establishing exercise, stretching, and gait retraining. Therapy frequently proves helpful for people with incoordination. If there is muscle stiffness or muscle spasm, some medications may help. There are now methods to study sleep disorders and provide specific treatment for rapid eye movement sleep behavior disorder as well as obstructive sleep apnea.

What happens as these diseases progress?

With progression of symptoms, each person with Sporadic Ataxia may have unique needs. Some people need devices to assist them with eating as well as special diets to help avoid choking. Others may need to adapt their residence to accommodate wheelchairs or walkers and most will need assistance with bathing, dressing, and other activities of daily life. Speech pathology can provide helpful means of improving speech and of learning techniques to swallow safely. It is important that people with Sporadic Ataxia are certain of their diagnosis and are comfortable with their physician. It is best to have a neurologist who knows you well and with whom you feel comfortable discussing new problems as they arise.

The referring physician or neurologist may refer you to other specialists, which include a genetic counselor, a physician specializing in physical medicine and rehabilitation, an occupational therapist, a social worker, and/or a psychologist. It is important to learn about the disease so that you know what to expect now and in the future. People with Sporadic Ataxia and their families should plan for the future so that medical or financial crises can be avoided.

What kind of support is available for people with Sporadic Ataxia or MSA and their families?

The National Ataxia Foundation (NAF) is committed to providing information and education about Ataxia, support groups for those affected by Ataxia, and promoting and funding research to find the cause for the various forms of Ataxia, better treatments, and, hopefully someday, a cure. NAF has been at the forefront funding promising worldwide research to find answers.

Ataxia research has moved into the clinical phase, and pharmaceutical companies have begun recruiting participants for clinical trials. Individuals with Ataxia or who are at-risk for Ataxia are encouraged to enroll in the CoRDS Ataxia Patient Registry. To access the Registry, go to NAF's website www.ataxia.org and click on the "Enroll in the Patient Registry" tab and follow the directions on the CoRDS website.

NAF provides accurate information for you, your family, and your physician about Ataxia. Please visit the NAF website at www.ataxia.org for additional information, including a listing of ataxia support groups, physicians who treat Ataxia, social networks, and more. For questions, contact the NAF directly at 763/553-0020 or naf@ataxia.org.

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