

FREQUENTLY ASKED QUESTIONS ABOUT... **Sporadic Ataxia and Multiple System Atrophy (MSA)**

What is sporadic ataxia?

Sporadic ataxia is a term designating a group of diseases of the central nervous system that occur without evidence that they are inherited, that is, no other person in the affected individual's family has ever had the same disorder. The term refers to changes in the structures of the brain that result from loss of nerve cells in these particular brain sites: the inferior olive, the pons, and the cerebellum. This group of diseases is termed "neurodegenerative" because the nerve cells in these structures (and often several other structures) gradually disappear over time without known causes. Physicians often use various terms when they make a diagnosis of sporadic ataxia. Some of these 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 of the legs and arms (ataxia) 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, bathe, write, eat, and drink. These symptoms progress gradually over time. In some people the disorder remains an ataxia with gradual progression of symptoms, and in some people the disorder can stop progressing spontaneously, but usually only after the disorder has interfered considerably with activities of daily living.

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, the people affected frequently experience faintness or lightheadedness when moving from the 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, often later 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 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 adult life 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. It is distinctly unusual for people with sporadic ataxia or MSA to develop dementia (loss of cognitive functioning). There may be some difficulty with judgment 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 probably encompasses a number of different disorders that cannot 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 cause the disease to begin. It is unclear why some people with sporadic ataxia progress to develop MSA whereas others do not. Many people with adult onset cerebellar degeneration may have the dominantly inherited form, which passes down in the generations genetically from parent to child. Making the diagnosis of dominantly inherited ataxia, (which is called spinocerebellar ataxia or SCA), is straightforward if a parent has ataxia.

What about genetics?

Genetic testing is now available for many of the SCAs, including SCA types 1, 2, 3, 5, 6, 7, 8, 10, 12, 13, 14, 17 and 28. Genetic testing is also available for Dentatorubropallidoluysian atrophy (DRPLA), Fragile X Tremor/Ataxia Syndrome (FXTAS), and for Friedreich's ataxia, a recessively inherited ataxia. A total of 36 dominantly inherited SCAs have been identified, however, there are many that cannot be detected with genetic testing. Often, obtaining a clear family history is a challenging task because the affected person may be adopted without information about the birth parents; the affected parent may have died before the disease became apparent; or the history of a parent developing the disorder is vague. There are many causes of gait and speech disorders, and, unless the parent's history closely approximates that of the currently affected person, there may be uncertainty about diagnosis. People with sporadic ataxia should discuss genetic questions with a physician or a genetic counselor who knows the individual best because answers may be different for various individuals or families.

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 not entirely certain for many years.

Many medical and neurologic 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, particularly cancers of the lung and ovary; 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 effects 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, imaging the brain with MRI scans 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 sporadic cerebellar degeneration receives a diagnosis of a treatable cause, such as vitamin B12 deficiency, thiamine deficiency, or a cancer affecting an organ of the body with cerebellar degeneration, 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 and 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 or special diets to help avoid choking. Others may need to adapt their residence to accommodate wheelchairs or walkers. Families, friends, or aides can often help those who 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 as soon as possible and are as comfortable as possible with their physician. It is best, when faced with a chronic neurologic disorder, 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, including a speech pathologist, genetic counselor, physician specializing in physical medicine and rehabilitation, physical therapist, occupational therapist, 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 research is being done on sporadic ataxia and MSA?

Both scientific and medical research is ongoing in the United States and many other countries to determine the cause of ataxia and devise better methods to diagnose and to treat the ataxias. The research includes both the hereditary and the sporadic forms of ataxia. Research in MSA is very active now. A North American MSA Study Group currently is conducting research into the disorder with participating investigators located in multiple universities throughout the United States. There is also a very active European MSA Study Group that includes representatives from multiple European countries who interact frequently and meet on a regular basis to discuss their progress.

What can a person with sporadic ataxia or MSA do to help with research?

An essential component for ataxia disease research is the availability of people with sporadic ataxia and MSA to participate in drug trials, natural history studies and other research studies. A patient registry is a key resource to facilitate and speed up clinical research in rare disorders such as sporadic ataxia and MSA by allowing for rapid contact between researchers performing clinical studies and those patients who may be eligible and willing to participate in such research. You are encouraged to sign-up on the web-based, secure ataxia patient registry which can be accessed by going to www.ataxia.org and selecting “Ataxia Patient Registry” on the home page.

What kind of support is available for people with sporadic ataxia or MSA and their families?

Psychological counseling or participation in support groups often helps affected persons and family members. There are numerous ataxia support groups throughout the United States.

People with sporadic ataxia or MSA are welcome to participate in any of the support groups affiliated with the National Ataxia Foundation (NAF). Refer to the contact information at the end of this information sheet to request a list of ataxia support groups throughout the United States. A list of support groups and online support, such as chat groups and social networks, is available through the NAF website at www.ataxia.org. The National Ataxia Foundation hosts an annual membership meeting each year. At these meetings the foremost authorities in the research and treatments for ataxias present their latest findings. These meetings also provide an opportunity for those with ataxia to meet others and often form lasting friendships. For more information, you are invited to contact the National Ataxia Foundation.