**What is Spinocerebellar Ataxia (SCA)?**
Spinocerebellar Ataxia or SCA, in abbreviation, is a group of dominantly inherited ataxias. The name SCA is usually followed by a number that indicates the ataxia is caused by a specific genetic defect. The numbers are usually assigned in the order in which the genetic form was identified.

**What is Spinocerebellar Ataxia Type 14?**
Spinocerebellar ataxia type 14 (SCA14) is one of those types of hereditary cerebellar ataxias. The involved gene, discovered in 2003, is located on human chromosome 19 and encodes a protein called protein kinase C gamma (PKCγ, the gene is PRKCG). Inherited defects in this gene cause a slowly progressive degeneration of cells in the cerebellum that causes the neurologic phenotypes of ataxia. It is not yet known how abnormalities of this protein cause degeneration of the cerebellum.

**What are the symptoms of SCA14?**
The most common symptom of SCA14 is incoordination (ataxia) of walking (gait). Other symptoms may include poor coordination of speaking (dysarthria), and tremor of the hands when reaching for objects. Less common symptoms have included stiffness of the muscles (rigidity), muscular spasms (dystonia), and difficulty swallowing (dysphagia). A few persons with SCA14 have developed brief shaking episodes of the arms or body referred to as myoclonus. Cognitive deficits may be a part of SCA14.

**What is the prognosis of SCA14?**
The symptoms of SCA14 typically begin in mid-life (20s-40s), but childhood onset or later adult onset have also been reported. The condition is slowly progressive and may require a cane, walker, or rarely a wheelchair late in life. Life span is not shortened. Currently there is no cure for SCA14.

**How is SCA14 acquired?**
SCA14 is inherited as an autosomal dominant genetic disease. This means that each child of an affected parent has a 50% risk of inheriting the gene mutation and developing the disease. Males and females are both affected.

**How common is SCA14?**
SCA14 is not common. It represents less than 1% of all of the hereditary ataxias.

**How is the diagnosis made?**
A neurological evaluation by a physician makes the diagnosis of cerebellar ataxia. A CT or MRI scan of the brain may show atrophy of the cerebellum. However, a specific diagnosis of SCA14 can only be made by a genetic test done on a blood sample. This genetic test is sometimes clearly abnormal, but other times it is difficult to interpret. The test results often need to be evaluated by an expert in genetic medicine.
What kind of support is available after the diagnosis?
There is no cure or specific treatment for SCA14. Physical therapy and gait training may be of value. Genetic counseling is important for families with SCA14 and genetic counselors can be identified at www.nsgc.org, which is the website of the National Society of Genetic Counselors. Further information about SCA14 can be obtained from www.genereviews.org which is a website that includes expert-authored, peer-reviewed, current disease descriptions.

The National Ataxia Foundation provides a wide range of support materials and activities including the publication, Living with Ataxia: An Information and Resource Guide and a quarterly newsletter with the most up-to-date research and medical information on all the types of ataxia. NAF also provides many local support groups. NAF is active in social networking through Facebook and Twitter.

What research is taking place in SCA14?
Studies on cultured cells and a mouse model found that mutant PKCγ protein aggregates in the cytoplasm of cells and may alter intracellular biochemical activities and signaling pathway but the role of these aggregates in the development of SCA14 is not clearly known. Further investigation in cultured cells and animal models of the disease will shed light on SCA14-related pathogenesis and may suggest potential therapeutic interventions. PKCγ is a protein kinase – an enzyme involved in activating and inactivating other proteins. As has been found for other kinase genes it might be a feasible target for pharmacologic intervention.

What can be done to move research in SCA14 forward?
As ataxia research moves into the clinical phase, researchers will need to recruit patients to participate in clinical trials. Individuals with SCA14 or who are at-risk for SCA14 are encouraged to enroll in the CoRDS Ataxia Patient Registry. This can be done by going to the NAF website’s homepage and clicking on the “Ataxia Patient Registry” button. This is a secure site to complete the enrollment process in the patient registry.

The National Ataxia Foundation funds research studies around the world. Supporting NAF’s research funding efforts is another way that research in SCA14 and all the other forms of ataxia will move us closer to treatments and a cure for SCA14.

Who can I contact?
Contact the National Ataxia Foundation for a more complete listing of resources and support groups affiliated with the National Ataxia Foundation.

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