What is Spinocerebellar Ataxia Type 14?
Spinocerebellar Ataxia type 14 (SCA14) is one type of Ataxia among a group of inherited diseases of the central nervous system. 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) or 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 (20's - 40's), 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 later 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 the hereditary Ataxias.

How is the diagnosis made?
A neurological evaluation by a physician makes the diagnosis a 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?
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.