

Frequently Asked Questions about the SCA 1, 2, 3, 6 Natural History Study

What is the Rare Diseases Clinical Research Network?

The Rare Diseases Clinical Research Network (RDCRN) is made up of 19 distinctive consortia that are working in concert to improve availability of rare disease information, treatment, clinical studies, and general awareness for both patients and the medical community. RDCRN was the first specialized infrastructure at NIH to support the study of groups of related diseases rather than individual diseases. RDCRN also is unique in that it emphasizes partnership of scientific researchers with patient organizations which includes the National Ataxia Foundation.

What is the Clinical Research Consortium for Spinocerebellar Ataxia?

The Clinical Research Consortium for Spinocerebellar Ataxia (CRC-SCA) is a consortium of physician scientists, and clinical research resources dedicated to conducting clinical research in Spinocerebellar Ataxia. Currently ataxia researchers and clinicians are recruiting for the Natural History of and Genetic Modifiers in Spinocerebellar Ataxias (SCAs).

What is the purpose of the Natural History Study?

The purpose of this research study is to bring together a group of experts in the field of SCA for the purpose of learning more about the disease. The research questions are:

- How does your disease progress over time?
- What are the best ways to measure the progression?
- Do some genes, other than the gene that is abnormal in your disease, have any effect on the way the disease behaves?

How many Patients are being recruited for this study?

This is a nationwide study and we expect that close to 800 patients will participate all over the USA. The participants will be in the study for 2 years and have a total of 4 study related visits done every 6 months. The very first patient was enrolled in the study on April 27, 2010.

However, more patients are needed.

What will happen at each visit during the study period?

- Medical History
- Physical exam
- A neurological examination that will help your doctor calculate a score that will reflect how severe your disease is using the Scale for Assessment and Rating of Ataxia (SARA).
- A series of other tests and questionnaires to complete
- Disease stage estimation
- Demographics and disease-related information (ie. age, sex, race, age of disease onset, disease duration, genetic testing results)

In some cases, patients may be asked to participate in two tasks done at home.

1. Wear an electronic monitor in the form of a light ankle bracelet for a period of 8 days
2. Also take a computer key board to home with you. You (or someone that can help you) will have to install that on your computer for an arm movement test.

In addition, your blood sample will be collected at least once during the entire study to extract your DNA. The sample will be sent to the research laboratory of Dr Stefan Pulst at University of Utah.

Who is eligible to participate in this study?

To be eligible to participate, you must:

- Have symptomatic ataxic disease
- Have a definite molecular diagnosis of SCA 1, 2,3,or 6 either in you or another affected family member
- Be willing to participate in the study and able to give informed consent.
- Be 6 years of age or above

You are not eligible to participate if:

- You have been diagnosed with a known recessive, X-linked or Mitochondrial Ataxia.
- The diagnosis of SCA 1, 2 and 3 has already been excluded by previous DNA testing.
- You have a lack of willingness to participate in this study.

Who do I contact to see if I am eligible or if I have questions?

In order to participate in a study, you must personally contact the study coordinator of any of the participating institutions by phone or by e-mail. Please use the information on the next page to inquire about participation.

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What role does the National Ataxia Foundation play in the RDCRN?

The National Ataxia Foundation is a member of the Coalition of Patient Advocacy Groups (CPAG) within the RDCRN.

What is CPAG?

One impressive feature of the Rare Diseases Clinical Research Network is the direct involvement of supporting patient advocacy groups in network operations, activities, and strategy. Each consortium in the network includes relevant patient advocacy groups in the consortium membership and activities. These patient advocacy group representatives serve in an advisory capacity within their own consortia.

Collectively, the Coalition of Patient Advocacy Groups (CPAG) represents the perspective and interests of all patient advocacy organizations associated with the clinical research consortia. The CPAG group meets frequently throughout the year via teleconference and face-to-face meetings.

CPAG members influence the direction of the Rare Diseases Clinical Research Network as a whole. They participate in network-level discussions and meetings. The CPAG coalition is a voting member of the RDCRN Steering Committee.

What are the CPAG vision and mission statements?

CPAG Vision

Through collaboration, patient advocacy groups and researchers can make faster progress toward new treatment options and cures, which can improve the lives of all persons and families affected by a rare disease.

CPAG Mission

The Coalition for Patient Advocacy Groups will promote collaboration between rare disease advocacy organizations and the Rare Diseases Clinical Research Network in order to facilitate better access to, and earlier benefit from, research conducted on rare diseases. As the patient advocacy arm of the Rare Diseases Clinical Research Network, CPAG members will use their position to advance the cause of rare disease research and improved patient outcomes through the network.

What is the National Ataxia Foundation?

The National Ataxia Foundation is committed to education about ataxia, service to individuals affected by sporadic and hereditary ataxia and promoting and funding research to find the causes, better treatments and a cure for ataxia. NAF can help by providing information for you, your family and your health care providers about ataxia. The Foundation facilitates local support groups and hosts an Annual Membership Meeting with world-leading ataxia researchers and clinicians present. For more information visit www.ataxia.org.

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