The National Ataxia Foundation's Drug Development Collaborative: Breaking Down Barriers to Clinical Trial Readiness for the Ataxias

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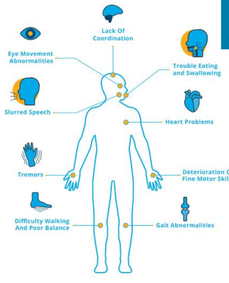
National Ataxia Foundation
www.ataxia.org

What is Ataxia?

Ataxia is a rare neurological disease.
It is progressive – affecting a person's ability to walk, talk, and use fine motor skills.

Between 15,000 - 20,000 people are estimated to have Spinocerebellar Ataxia (SCA) in the US.

Tens of thousands more are affected by recessive, sporadic, and as yet unknown forms of Ataxia.



- Ataxia is a group of rare neurological diseases that are estimated to affect nearly one million adults and children globally.
- Individuals with ataxia present with progressive, disabling impairment in balance and coordination due to damage to the cerebellum, typically leading to early disability and death.
- There remains no available treatment to slow or stop the progression of Ataxia.

Background

National Ataxia Foundation

Our Vision
- A world without Ataxia
- Ensuring no one experiences Ataxia alone, until no one experiences Ataxia, period.

Our Mission
- To accelerate the development of treatments and a cure while working to improve the lives of those living with Ataxia.

Established in 1957, NAF is the largest 501(c)(3) non-profit organization representing patients with all forms of Ataxia

11,123
NAF Members

Obstacles in Ataxia Research

Continually low rates of Ataxia diagnosis and patient registration → **Lack of adequate natural history data, biomarkers, and clinical outcome assessments** → **Need for increased patient education**

Improved Genetic Diagnosis Rates and Patient Registration

SCA1, SCA2, and SCA3 Genetic Counseling and Testing Program

A free program for participants who qualify

National Ataxia Foundation

Importance of Confirmed Genetic Diagnosis and Patient Registries for Individuals at Risk
- To assist in making informed decisions around family planning, healthcare/lifestyle choices, and professional/financial planning
- A confirmed genetic diagnosis if often a requirement for participation in clinical trials

Importance of Confirmed Genetic Diagnosis and Patient Registries for Research
- To better understand the prevalence of Ataxia
- To identify patients for clinical trials
- To connect newly diagnosed patients with resources to navigate their diagnosis

NAF's Genetic Counseling and Testing Initiative
- Free virtual genetic counseling
- Free diagnostic genetic testing for individuals who have a biological family member with a confirmed diagnosis of SCA1, SCA2, or SCA3
- Access to NAF newly diagnosed resources including information, connection to support groups, and more

Who is eligible for the program?
- Any individual who is at-risk for SCA1, SCA2, or SCA3 because they have a family member with a confirmed genetic diagnosis
- Participants must be 18 years of age or older
- NAF member
- Health insurance and/or a physician referral is not required

Rare Disease Patient Registries

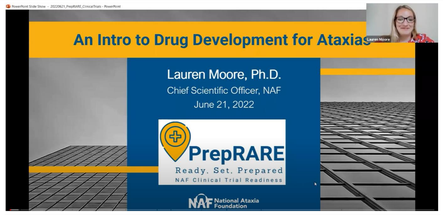
CoRDS Rare Disease Registry

The purpose of a patient registry is to advance the development of treatments and a cure for rare diseases by connecting those who are affected by the disease with researchers and industry.

NAF encourages all members to join the CoRDS Registry.

The CoRDS registry is used by NAF to assist in establishing an Ataxia database.

Education and Outreach through PrepRARE Initiative



DDC-driven initiative to educate and inform the Ataxia community on what is needed to become "Clinical Trial Ready".

A series of educational articles, infographics, videos, and webinars are released monthly to NAF members.

PrepRARE
Ready, Set, Prepared
NAF Clinical Trial Readiness

There is an urgent need for the Ataxia community to be "clinical trial ready" because many therapies are in the drug development pipeline. To successfully bring a treatment to the market, patient involvement is crucial. The Ataxia community is needed to participate in research studies and clinical trials, but that is not their only role in drug development. Drug developers need to understand how Ataxia symptoms impact a person's daily life in order to find meaningful solutions. NAF developed PrepRARE as a program to educate members about all stages of the clinical trial process. Check out our series of webinars, articles, and other information to make sure that you are ready to make informed decisions about participation when a new clinical trial begins recruitment.

Tackling Obstacles in Ataxia Treatment through Industry Partnership

The past decade has seen a significant rise in drug development efforts in Ataxia.

Pharmaceutical companies began approaching clinical stages in their Ataxia therapeutic programs, it became clear that there remain significant gaps hindering clinical trial readiness in the Ataxias.

In response, the NAF partnered with key industry leaders to found the Drug Development Collaborative (DDC) in late 2020, a pre-competitive pharmaceutical consortium with the primary objective of accelerating the development of treatments for ataxia.

The DDC works serves as a bridge between the Ataxia community, academic researchers, and industry to tackle obstacles in Ataxia research.

- DDC provides monetary support for NAF initiatives including the CRC-SCA and the Genetic Testing and Counseling Initiative.
- NAF hosts quarterly advisory meetings seeking input on NAF initiatives from DDC members to further future partnership
- Monthly roundtable meetings are held where recent Ataxia research, patient panels, and updates on NAF initiatives is presented

Expanding Natural History Data & Identifying Improved Biomarkers

CRC-SCA
Clinical Research Consortium for the Study of Cerebellar Ataxia

OUR MISSION
To better understand the factors that determine disease progression among Cerebellar Ataxia with the goal of improving the understanding of the disease process, current treatments, and development of disease-modifying therapies

The CRC-SCA has 14 sites across the United States.

In July 2022, the CRC-SCA launched its first international site in Canada.

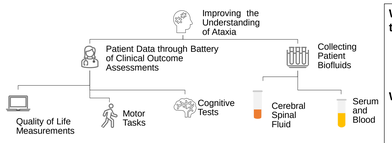
Cooperative Ataxia Group
Establishment of a group of clinical researchers that would develop rating scales for the ataxias, design the ataxia database and create an ataxia patient registry. Overall goal was to improve treatments of the ataxias.

RDCRN
The National Institutes of Health Rare Diseases Clinical Research Network (RDCRN) awarded 2 years of funding to 8 CRC-SCA sites. Dr. Ashizawa was the Principal Investigator (PI) for the Clinical Research Consortium for Spinocerebellar ataxias (CRC-SCA). NAF's involvement was a requirement of the grant.

NAF Grant
Lead PI applied for and was awarded a $100,000 one-year grant from NAF for a Pioneer SCA Translational Research Award to continue the work of the CRC-SCA. 4 sites joined consortium without funding.

Private Donor/NAF
A major part of NAF's strategic plan was to have the SCA research community become clinical trial ready.
A working group of clinicians wrote and submitted a proposal requesting $248,000/year to fund 14 sites. The Macklin Foundation and NAF co-funded the CRC-SCA proposal.

Drug Development Collaborative
DDC funds utilized to revitalize the CRC-SCA with a new payment structure and milestones. 15 sites, biofluid collection added, and QC/analysis of data emphasized.



Who is eligible to participate in the CRC-SCA?
- Individuals who have a confirmed diagnosis of SCA 1, 2, 3, 6, 7, 8 or 10

What is natural history data?
- A collection of data on how a specific disease progresses in individuals over time

Recent Publications

Lin, C. C., Ashizawa, T., & Kuo, S. H. (2020). Collaborative Efforts for Spinocerebellar Ataxia Research in the United States: CRC-SCA and READISCA. *Frontiers in neurology*, *11*, 902. https://doi.org/10.3389/fneur.2020.00902

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