

National Ataxia Foundation  
59<sup>th</sup> Annual Ataxia Conference

*Mike Parent, Executive Director*

Hosted by the SouthEast Region



THE MAGIC OF A CURE

Dream It.  
Hope It.

## Disclaimer

- ▶ The information provided by speakers in any presentation made as part of the 2016 NAF Annual Ataxia Conference is for informational use only.
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- ▶ or services mentioned during these presentations does not imply endorsement by NAF.

## Presenter Disclosures

- ▶ No relationships to disclose or list

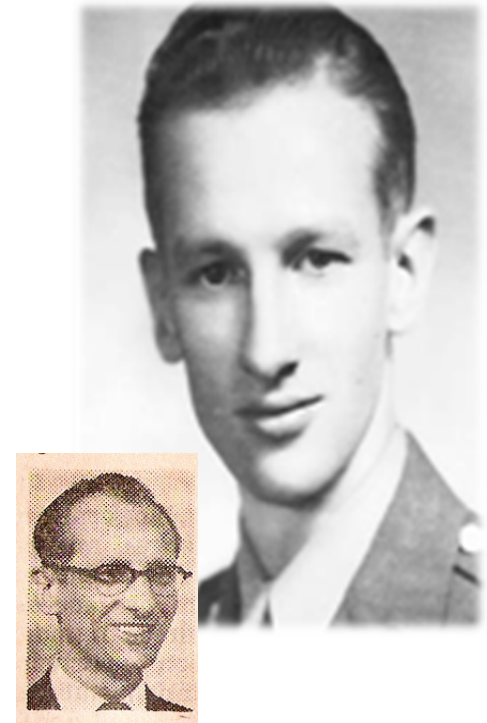
# ATAxia



is not a foreign cab

# The National Ataxia Foundation

The National Ataxia Foundation is entering its 59th year in serving ataxia families.



The main focus and purpose of the Foundation as envisioned by its founders remain the same : Fostering promising Ataxia research and helping those with Ataxia and their families.

# History

Many rare disease groups organizations are formed by those who have been impacted by a particular disease. The National Ataxia Foundation (NAF) was no exception and was established by a family affected by Ataxia. NAF was founded in 1957 by two brothers, Dr. John W. Schut and Henry Schut, along with other family members and friends, on the belief that by working together, ataxia families, clinicians, researchers, and others can accomplish great things.

Although the road the organization has traveled throughout its history has been challenging, its mission has always been clear.

The background features several flowing, wavy bands of color. At the top, a thick band of red and orange flows from left to right. At the bottom, there are more complex, layered waves in shades of red, orange, and yellow, creating a sense of movement and depth. The central area is a plain white background where the text is located.

**Here is our journey:**

1957

Thirteen attend  
the  
organizational  
meeting of the  
National Ataxia  
Foundation on  
April 8, 1957.

The first officers included  
President Ellis J. Sherman,  
Vice President and  
Secretary Henry Schut and  
Treasurer Robert Van  
Hauer.

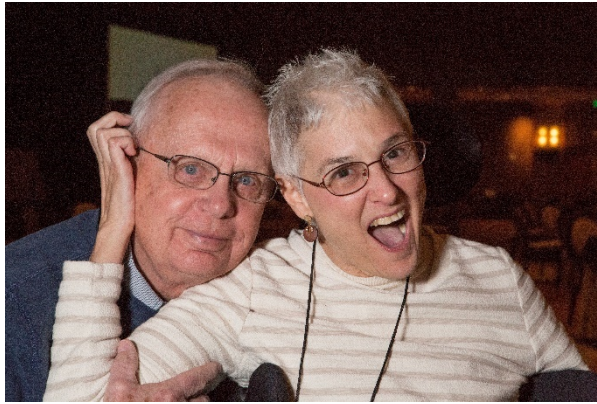
Dr, John W. Schut was  
appointed as the first  
Medical Director

1958

The 1<sup>st</sup> annual membership meeting was held April 16, 1958 in Minnesota. Nine of the thirteen members attended. Three more were elected to membership

When NAF first began, a membership was only given after the application was reviewed at the annual meeting. It was not until the 1970's when open memberships were accepted.

Today the NAF Annual Ataxia Conference attendance ranges between 400-700 people from the United States and other countries. The conference is now held in various cities throughout the United States.



## 1970

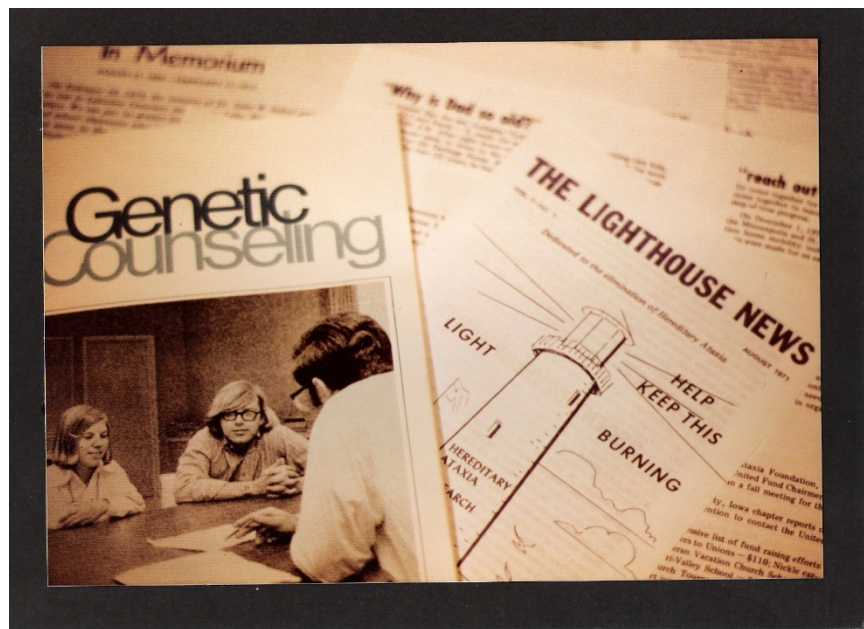
The National Ataxia Foundation struggled in the early years in terms of funding and ataxia awareness. However, in 1970 the NAF offered its first ataxia clinic. Drs. Roger Rosenberg, Larry Schut, and William Nyhan participated in this first ataxia clinic in Sioux Falls, South Dakota on December 13, 1970. There were 95 people attending this clinic and 33 were examined.

WCCO-TV, based in Minneapolis, Minnesota, covered the clinic and aired a story about ataxia. The three doctors were invited to New York City for a press conference which was attended by *Time*, *Life*, and *Associated Press*. *Time* published an article on January 25, 1971 which resulted in letters from ataxia families across the country. NAF continued to sponsored ataxia clinics, free of charge to patients, throughout the 1970 and 1980s. This multidisciplinary approach to ataxia clinics was a model for others to follow years later.



1971

There was a re-structuring and formation of the Medical and Research Advisory Board. This advisory board totaled 7 members. Today this international roster of world leading ataxia clinicians and scientists totals more than 25 members. NAF introduced its first newsletter, "Light House News" which became a quarterly newsletter in 1974. Today the four page "Light House News" has been replaced by the 48-page quarterly news publication "Generations." Also, in 1974, NAF offered its first brochure on ataxia. Today the Foundation is a world leader in providing current and accurate information through its many publications, books, and website.



## 1972

On February 19, 1972 the 15th Annual Membership Meeting was held in Minneapolis, Minnesota. There were 38 members who attended the annual membership meeting. Dr. John W. Schut's funeral was the same day. He was one of the original founders of the Foundation.

Dr. Schut was a brilliant neurologist and researcher who had also inherited the ataxia gene.

## 1975

NAF's first ataxia research symposium was held in Minneapolis on November 24, 1975, bringing researchers together to help accelerate ataxia research. These research symposiums continued throughout the 1970s, 80s and 90s and was the foundation for the establishment of NAF's International Ataxia Investigators Meetings (AIM) which are offered every other year and brings together 140 - 160 leading ataxia clinicians and scientists from around the world to help accelerate world-wide ataxia research.

1976



**Dr. Robert Currier and his daughter  
Dr. Mary Currier 1983**

Funding challenges continued to face NAF with total contributions in 1976 of \$41,350 compared to \$6,537 in 1972. In 1978 the first ataxia research grant awarded by NAF was given to Dr. Robert Currier from Mississippi in the amount of \$5,000. Two research grants were awarded in 1979, one grant to Dr. Currier in the amount of \$3,250 and the other research grant to Dr. Kark in the amount of \$2,500. Today, the Foundation offers five research programs and has funded nearly \$3 million over the past 3 years to more than 70 promising ataxia research studies.

## 1982

On February 20, 1982, the 25th Annual Membership meeting was held in Jackson, Mississippi. This was the first time the meeting was held outside of Minnesota. Since that time, the annual membership meetings have been held in various cities throughout the United States.

The National Ataxia Foundation is able to continue to support promising ataxia research and offer meaningful programs and services for ataxia families because of the kindness and generosity of our donors and volunteers. We gratefully acknowledge their tremendous contributions to these efforts. The vision of our original founders has been our guiding path as we journey forward to end ataxia.

**1991**

The NAF was approved to be included in the Combined Federal Campaign (CFC). Since that time, government workers have donated more than \$500,000 to help support the NAF's mission.



**1995**

The NAF is on the internet for the first time.

## NAF Offers Five Research Programs



**Research Grant:** 1-year \$15,000-30,000 “seed-money” grants for early or pilot phases of studies and ongoing investigations



**Young Investigator Award:** 1-year \$35,000 grants to encourage young investigators to pursue a career in the field of any form of ataxia research



**Young Investigator Award for SCA Research:** 1-year grants of \$50,000 awarded to encourage young investigators to pursue a career in Spinocerebellar Ataxia (SCA) research



**Research Post-Doc Fellowship Award:** 1-year \$35,000 grants to post-docs who have shown a commitment to research in the field of ataxia



**Pioneer SCA Translational Research Award:** 1-year grants of \$100,000 focusing on research investigation that will facilitate the development of treatments for the Spinocerebellar Ataxias (SCAs)

## NAF Supported Research

- The first direct research funding made by NAF occurred in 1978 to Dr. Robert Carrier, University of Mississippi in the amount of \$5,000.
- In 1985, three researchers in the United States received funding totaling \$25,000.
- In 1995, six researchers from the United States received funding totaling \$40,500.
- In 2015, NAF received more than 100 research applications from around the world. Lead by Drs. Orr and Ranum, 62 peer reviewers were assigned selected research applications to review with a total of 212 individual reviews.
- In 2015, NAF supported 22 ataxia research studies in six countries with funding support of nearly \$1 million.

# Ataxia Investigators Meeting (AIM)

The 6th Ataxia Investigators Meeting (AIM) is being held this week. AIM 2016 focuses on the most recent scientific advances and emerging translational approaches toward therapy with four primary goals: 1) Identify common disease mechanisms; 2) Explore therapeutic strategies; 3) Help establish the future leaders of ataxia research; and 4) Bring trainees (graduate students and postdocs) into contact with Ataxia families.

This is the largest attended AIM with more than 160 ataxia investigators, industry, and patient advocacy groups attending this scientific conference on Ataxia.

The five prior AIM conferences have been instrumental in advancing ataxia research and in bringing young investigators into the field of ataxia research.

# The NAF's 4th Ataxia Investigators Meeting (AIM 2012-San Antonio)



# The NAF's 5th Ataxia Investigators Meeting (AIM 2014-Las Vegas)



**CoRDS–NAF Ataxia Patient Registry:** An International Disease-specific Registry for Individuals with Ataxia (Information provided by CoRDS)

### **What is the CoRDS–NAF Ataxia Patient Registry?**

- Collaboration between Sanford Research’s Coordination of Rare Disease at Sanford (CoRDS) and the NAF to create a registry that collects and organizes information from individuals with Ataxia (established 2013)
- Researchers are able to review de-identified data
- Participants may be notified of research studies for which they are eligible

#### **Currently the CoRDS-NAF registry includes:**

925 fully enrolled participants  
64 participants in screening

#### **Participants have enrolled from:**

49 United States  
28 Countries

# CoRDS-NAF Enrollment

## Gender Breakdown

Females	51%
Males	49%

## Age of Enrollment

81-90	9	1%
71-80	93	10.1%
61-70	222	24%
51-60	223	24.1%
41-50	163	17.6%
31-40	98	10.6%
21-30	50	5.4%
11-20	49	5.3%
<u>0-10</u>	<u>18</u>	<u>1.9%</u>
<b>Total:</b>	<b>925</b>	<b>100%</b>



## How Patients and Families Can Get Involved

- Participants that enroll can contribute information to the **CoRDS-NAF** registry by filling out a brief questionnaire
- Enrollment involves reading the consent form and completing the questionnaire
- Participants may learn about research opportunities for which they are eligible
- When notified about research opportunities, you have the ability to decide whether or not to participate

## Why Should You Participate?

- Provides participants an opportunity to be informed of research studies and clinical trials for which they are eligible
- Provides researchers with a central resource for the identification and more rapid recruitment of potential research participants
- Provides the potential to accelerate research into Ataxia
- No financial cost to you with easy and quick enrollment

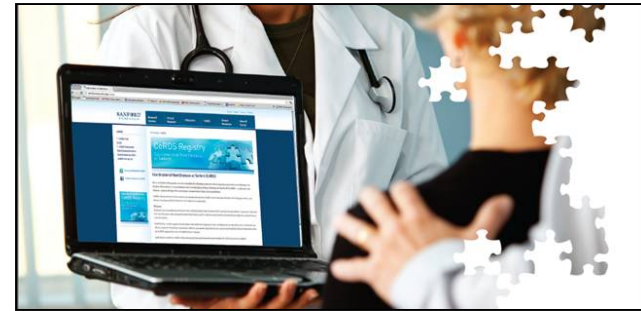


## Enroll at the Conference

- Informed Consent Form and **CoRDS-NAF** questionnaire available
- Read the Informed Consent & complete the questionnaire and return to **CoRDS** table
- The questionnaire is brief and takes about 30 minutes to complete

## Other Enrollment Options

- Online enrollment on the **CoRDS** website: [sanfordresearch.org/cords/](http://sanfordresearch.org/cords/)
- Mail-based enrollment
- In-person enrollment at the Conference



## Become Fully Enrolled

To be fully enrolled, you must fill out both the screening form AND the questionnaire. Speak to a **CoRDS** team member about finishing enrollment today!

REMEMBER

# Awareness

- In 1982, NAF and members wrote to Dear Abby asking her to print a letter about ataxia in her column. From those efforts, a letter in “Dear Abby” appeared resulting in over 3,400 letters to NAF.
- In 1994, ABC’s 20/20 aired a news program about ataxia and NAF that was filmed at the NAF offices.



The first International Ataxia Awareness Day (IAAD) occurred on September 25, 2000. This was the first time that ataxia organizations throughout the world declared and recognized September 25th as IAAD.



- In 2015, NAF's website received 200,995 visitors from 185 countries: 111,639 from a desktop, 62,765 from a mobile device, and 26,592 through tablets.
- NAF's E-Blast list has now grown to more than 8,500 subscribers. In 2015, 96 E-Blasts were sent out regarding events, research updates, support group meetings, and other activities.
- Through awareness events such as the Walk n' Rolls for Ataxia, media coverage, social media, government proclamations, information booths at Abilities Expos, CFC events, American Brain Foundation Brain Health Fair, as well as medical/research conferences, ataxia awareness events and NAF's website, the word "Ataxia" is getting out.

# Education

The National Ataxia Foundation now offers more than 20 *Frequently Asked Questions (FAQs) Fact Sheets* as well as the quarterly newsletter *Generations*. In addition, NAF offers books and other publications on ataxia. In 2015 fact sheets that were updated included *Episodic Ataxia*, *Gene Testing for Hereditary Ataxia*, *Sporadic and Multiple Systems Atrophy and Medications for Ataxia Symptoms*. In 2015 more than 5,000 paper copies of the NAF fact sheets were distributed and thousands more were downloaded. In addition, the second edition of “*Children with Ataxia*” was introduced.

NATIONAL ATAXIA FOUNDATION  
FAQ  
FREQUENTLY ASKED QUESTIONS ABOUT...  
**Ataxia Classification**

Patients often ask the familiar question “What kind of ataxia do I have?”. This document is intended to provide patients, families, and their caregivers with basic information regarding how doctors classify the different forms of ataxia. Correctly defining the type of ataxia is an important step in a patient’s evaluation and can help guide their medical workup, suggest potential treatment strategies, and even indicate if appropriate clinical or research studies are available. Many different terms have been used to describe ataxia over the years and have fallen out of favor for various reasons. The terms listed here are ones currently used by many doctors who treat patients with ataxia.

Ataxia is a neurological symptom seen by your physician during a physical examination. The definition of ataxia is a specific problem with balance and coordination not due to muscular weakness. Ataxia can be caused by problems in the inner ear or nerve damage in the legs, but most often ataxia describes damage to a part of the brain called the **cerebellum**. Doctors may also refer to this problem as **cerebellar ataxia**.

Cerebellar ataxia is classified by its cause (see Figure 1). Unfortunately there are many causes and the

**Known Causes**

- Acquired (Traumatic Brain Injury, Stroke, Alcohol Abuse)
- Genetic (Spinocerebellar Ataxia (SCA), Friedreich Ataxia (FA), Dentatorubral Degen. (DRD), Ataxia Telangiectasia (AT))
- Idiopathic (Sporadic)

**Unknown Causes**

- Genetic (Spinocerebellar Ataxia (SCA), Friedreich Ataxia (FA), Dentatorubral Degen. (DRD), Ataxia Telangiectasia (AT))

terminology can become confusing. In general, there are two main classes of ataxia, **acquired** and **genetic**. Two additional classes of ataxia, **sporadic** and **unknown**, are also sometimes used. It is not uncommon for patients to undergo detailed medical evaluations before their cause is known and their ataxia can be properly classified.

**Acquired ataxia** has an external cause, meaning a person develops it because of something that happens during their life. This can include many different problems and events such as vitamin deficiencies, autoimmune conditions, some infections, exposures to toxic substances or drugs (especially alcohol), various cancers, and many many more. Acquired ataxia often appear “out of nowhere” and the medical term used for this is **sporadic**. Unfortunately, ataxia from all other categories can also occur sporadically, so this term is not useful to categorize the cause of the ataxia. Acquired ataxia are important to recognize early because, in some cases, they may be treatable.

**Genetic ataxia** has an internal cause, meaning it is due to inherited damage in a person’s DNA (their “genetic blueprint”). A person’s DNA is made up of 23 pairs of chromosomes (one pair each from their mother and their father) containing sets of genes which together code for all the information which makes a person who they are. People each have over 20,000 genes and we now know of many specific examples where damage to a certain gene causes cerebellar ataxia to develop. Genetic ataxia often run in families (some doctors may say it is a **hereditary** or **familial** ataxia when this occurs) but can also be sporadic without a known family history.

- If an ataxia passes from generation to generation it is said to be **dominant** and is due to a single copy of a defective gene passing from parent to child. Dominant ataxia are most often seen in adults and some can get worse each successive generation. The most well-known dominant ataxia are the spinocerebellar ataxia (or SCAs) such as SCA1, SCA2, SCA3, SCA6, and SCA7, which are each caused by mutations in different genes on the 15th chromosome.

Figure 1

NATIONAL ATAXIA FOUNDATION  
FAQ  
FREQUENTLY ASKED QUESTIONS ABOUT...  
**Gene Testing for Hereditary Ataxia**

This fact sheet provides an overview of gene testing for ataxia. It also addresses commonly asked questions about gene testing for ataxia, and offers general guidelines for testing of the hereditary ataxias. Keep in mind that the answers and information given here are general. Your physician and a genetic counselor should be consulted for more specific recommendations pertaining to your care and genetic testing.

**What is ataxia?**

Ataxia is defined as a lack of muscle coordination. It can affect almost all of your movements, including walking, speech, eye movements, and hand control. Ataxia is a symptom that can occur as the result of strokes, head injuries, illness, and hereditary disease.

**What is hereditary ataxia?**

Ataxia is one of the symptoms in more than 300 hereditary conditions. However, inherited conditions where ataxia is the primary symptom are much less common. These inherited forms of ataxia are often referred to as “hereditary ataxias.” Hereditary ataxia means the condition is caused by changes in genes that are inherited from parents and may be passed to children.

**What is genetic information?**

We are all born with genetic instructions of how to grow and develop. We inherit our genetic information through chromosomes passed to us from our parents. We have 23 pairs of chromosomes because we receive one copy of each chromosome from our mom and one from our dad. 22 pairs are the same in males and females and are called autosomal chromosomes. The 23rd pair differs between males and females and the chromosomes in this pair are called the sex chromosomes. There are a number of different

genes that can cause ataxia and at this time most are located on autosomal chromosomes. Chromosomes are made up of substances known as nucleotides. These nucleotides, identified by letters, are linked together in chains. A group of nucleotides together form genes. There are thousands of genes located on each chromosome.

Many of the genes that cause dominant forms of ataxia have a mutation resulting from expanded sections in these nucleotide chains called “trinucleotide repeat expansions.” For instance, a mutation in the SCA1 gene on the 15th chromosome results in extra copies of a series of nucleotides identified by the letters C-A-G. In some conditions, the number of trinucleotide repeats is associated with the severity of the disease and the age of onset.

**For which of the hereditary ataxias is gene testing available?**

Discovery of specific ataxia genes makes it possible to develop blood tests to facilitate the diagnosis of both symptomatic individuals and at risk family members. In 1993, the first ataxia gene was identified by a research team led by Drs. Harry Orr and Heidi Zoghbi. This gene is responsible for spinocerebellar ataxia type 1 (SCA1). Its discovery paved the way for the identification of many additional ataxia genes. The list continues to grow as we discover new forms of ataxia.

As of 2015, genetic testing is available for dozens of forms of ataxia including dominant ataxias (e.g. SCA1, SCA2, SCA3, SCA6 and others), recessive ataxias (e.g. Friedreich ataxia, ADAM, AGAL and others), X-linked ataxias (e.g. Fragile X tremor ataxia syndrome), and mitochondrial ataxias (e.g. MELAS). New genetic testing technologies have enabled the discovery of many new ataxia genes and the list of available tests is constantly growing.

In 2007, Walk n' Rolls for Ataxia first began adding another layer of ataxia awareness.



The first NAF Walk n' Roll for Ataxia was held in September, 2007 in San Diego, California hosted by the San Diego Ataxia Support Group. Since that time, NAF Walk n' Roll for Ataxia have continued to grow nationwide from California to Georgia from Massachusetts to Denver from Minnesota to Utah and beyond.



In 2015, twelve Walk n' Rolls for Ataxia were held throughout the United States raising more than \$285,000 with more than 1,600 participants and tens of thousands more being informed about Ataxia.

## Why Walk n' Rolls?

Walk n' Roll for Ataxia's primary objectives are to have FUN, raise awareness and funds to support the mission of NAF. Families, friends, co-workers, neighbors, and communities come together each year to honor those with Ataxia. Each Walk n' Roll is unique, but all provide a setting of fun, sharing, and caring.

## Get Involved!

Support groups, families, and individuals have all conducted Walk n' Rolls for Ataxia. Join them in hosting your own NAF Walk n' Roll for Ataxia or participate in a Walk n' Roll near you. For more information or to start a Walk n' Roll in your community, please contact NAF. Together, we can continue to grow this event!

A heartfelt thank you to the organizers, participants, donors, sponsors, and volunteers...you are all amazing!!!



## Social Media

NAF continues to reach the ataxia community through various social media platforms:

	<u>2014</u>	<u>2015</u>
NAF Bulletin Board - Members	68,388	71,213
Forum Discussions	9	9
Topics Posted	2,202	3,162
NAF Facebook Group - Members	4,443	5,112
NAF Facebook Page - Likes	3,137	3,755
NAF Twitter Page - Follows	886	1,132
- Tweets	951	1,335
YouTube Channel - Subscribers	98	152
Video Views	8,127	28,246



# Sources of Support

*Chapters/Support Groups*

*Combined Federal Campaign*

*Corporate Support*

*Deferred Giving/Estates*

*Employer Matching Gifts*

*Endowment Funds*

*Family Fund Raisers*

*Foundation Support*

*Fund Raising Events*

*Gifts of Stock/Assets*

*Group/Family/Individuals*

*IAAD Events*

*Membership Support*

*Memorials/Honor Of*

*Online Shopping Portals*

*Partnerships*

*Planned Giving*

*Recurring Gifts*

*Research Drive*


*Social Networking*

*Special Projects*

*United Ways*

*Vehicle Donations*

*Walk n' Rolls*




The National Ataxia Foundation gratefully acknowledges each of our donors. Every donor plays an essential role in fulfilling our mission to serve ataxia families. In addition, our amazing volunteers not only give of their precious time and extraordinary talents, but also significantly help in reducing the cost of NAF expenses, so more funds may be used to support promising research and important programs.

## How are Funds Spent?

In 2015, Charity Navigator, America's premier charity evaluator, gave the National Ataxia Foundation a four out of four star rating. This is the third consecutive year NAF has received this four-star rating. Charity Navigator has awarded this to only **14%** of their charities.

According to Charity Navigator, "Receiving four out of a possible four stars indicates that your organization adheres to good governance and other best practices that minimize the chance of unethical activities and consistently executes its mission in a fiscally responsible way."



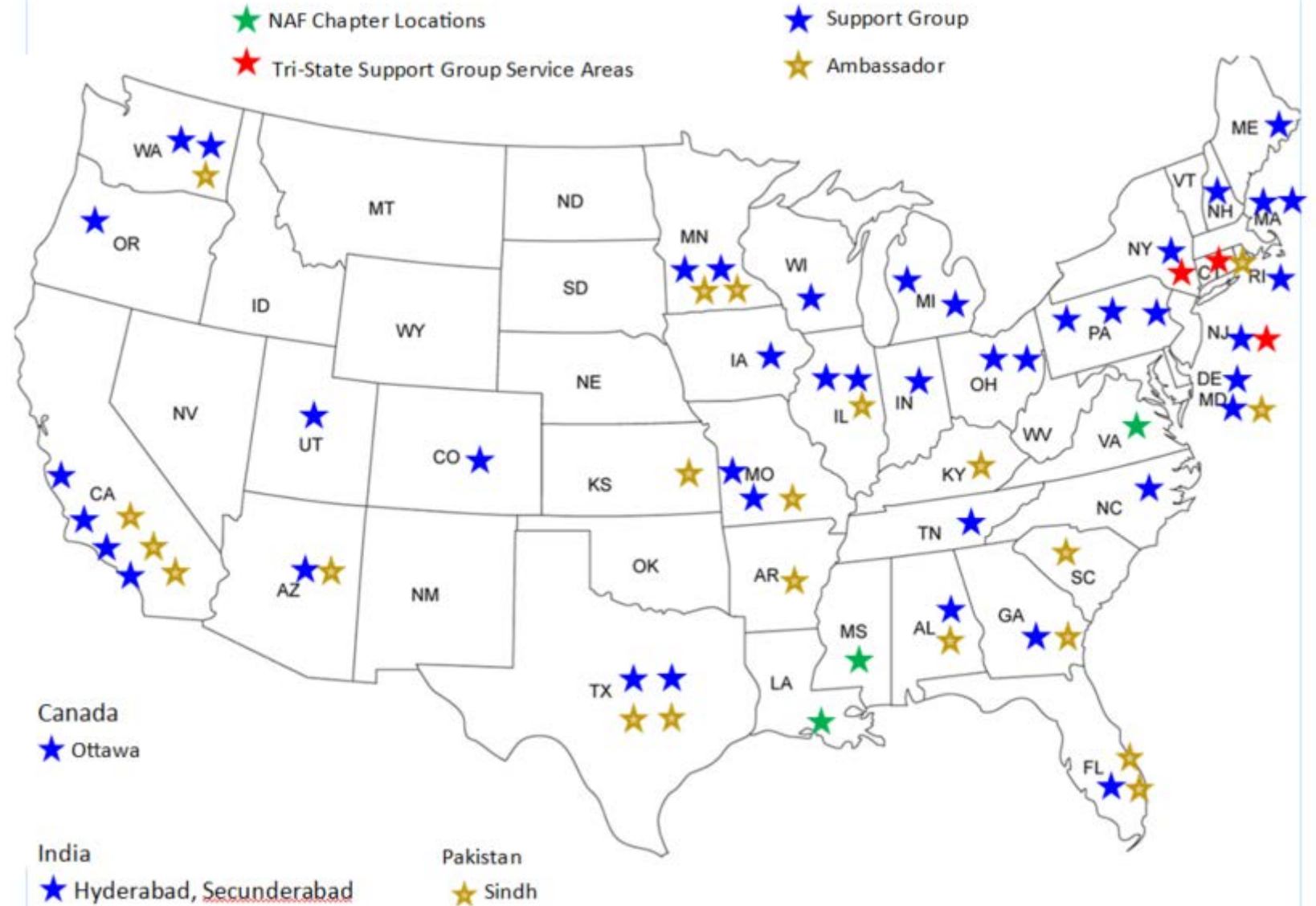
While the 2015 annual audited financial statements will be soon available, the 2014 CPA annual audit reflects fundraising costs were 3% of total expenditures and administrative costs were 5% of total expenditures. Program Services for 2014 totaled 92% of total expenditures. In other words, 92 cents out of every dollar spent was used to support research and programs. In the Program Services area, the highest expenditure was research totaling 61.5% of all expenditures for fiscal year 2014.

# NAF Chapters, Support Groups, and Ambassadors

Through NAF Chapters, Support Groups, and Ambassadors, NAF has a network of volunteers who bring local ataxia families together to share, learn, and network. Support Group Leaders, Chapter Presidents, and Ambassadors help identify local resources and offer support and information to local ataxia families. These leaders also help coordinate Ataxia awareness campaigns and events in their area.

To help leaders with their important work, a regional concept is being developed in which a Regional Coordinator would be assigned a region to work with leaders in that area to enhance communications and heighten support for Ambassadors, Support Groups and Chapters located within their region. These Regional Coordinators would be a focal point within each region for communications, fund raising and improved interaction.

In 2015, there were 3 NAF Chapters, 41 Support Groups, and 23 Ambassadors serving their local ataxia communities. These all give local ataxia families hope and understanding. Thank you for all that you do!




# Strategic Plan

## NAF's Strategic Goals

The Board of Directors of the National Ataxia Foundation held a two-day strategic planning session in November, 2015. A major outcome of this meeting was the establishment of five bold goals which provide the foundation for a formal strategic plan that will act as NAF's roadmap for the next three to five years.

## NAF's Five Bold Goals

- **Increasing revenues** to fuel the organization's growth and enable it to build on its history and past successes to become a stronger, more vital organization.
- **Increasing awareness and recognition** of Ataxia and NAF to strengthen and grow support for NAF's mission.
- **Becoming "clinical trial research ready"** thus enhancing NAF's role within the Ataxia community.
- **Building internal capacity through technology** to increase NAF's organizational efficiency and effectiveness as well as create new opportunities.
- Understanding of the **importance of having a strategic plan** not only to guide the organization, but to provide a mechanism by which NAF's progress and success may be measured and evaluated.



We expect that the strategic plan to be completed and in place by mid-2016. A very special thank you to all the NAF members who have participated in this strategic planning process. Your input has been invaluable!

# The Future

A promise for a brighter future

- Enhancing of fundraising capacity
- Accelerating ataxia awareness campaigns
- Strengthening clinical readiness
- Expanding worldwide ataxia research
- Collaborating with industry and ataxia organizations

**With the dedication and commitment of NAF volunteers, donors and our partners, together we can end Ataxia.**

The background features several flowing, ribbon-like shapes in shades of red, orange, and yellow, creating a sense of movement and energy. The ribbons are layered and overlap, with some appearing more prominent than others. The overall color palette is warm and vibrant.

**THANK YOU!**