PARTNERING FOR PROGRESS

Patients

Clinicians

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Pharma

2025

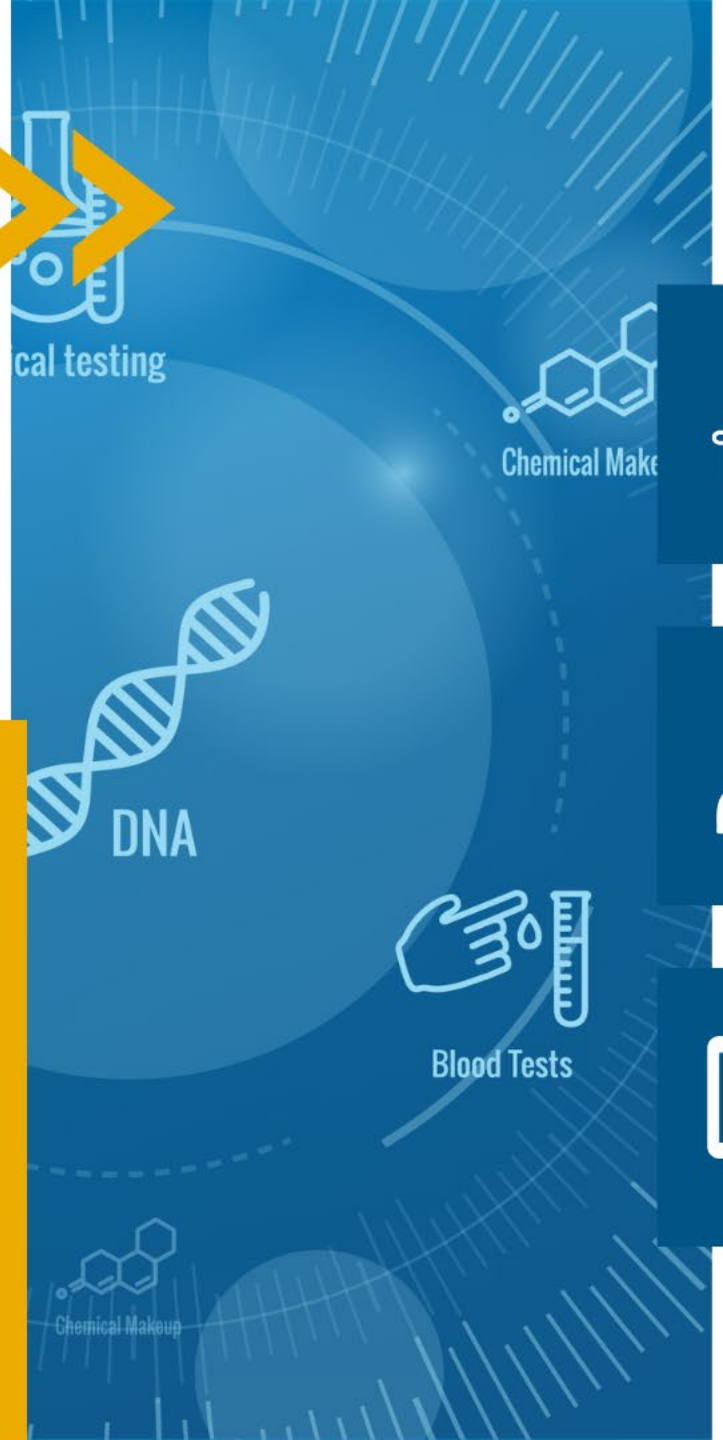
Annual Ataxia Conference

Genetic Testing: Arriving at the end of the Diagnostic Odyssey.

Meron Azage, MS, LCGC and Mary Hogan, MS, MPH

NAF
National Ataxia Foundation

March 27-29, 2025
Planet Hollywood
Las Vegas, NV



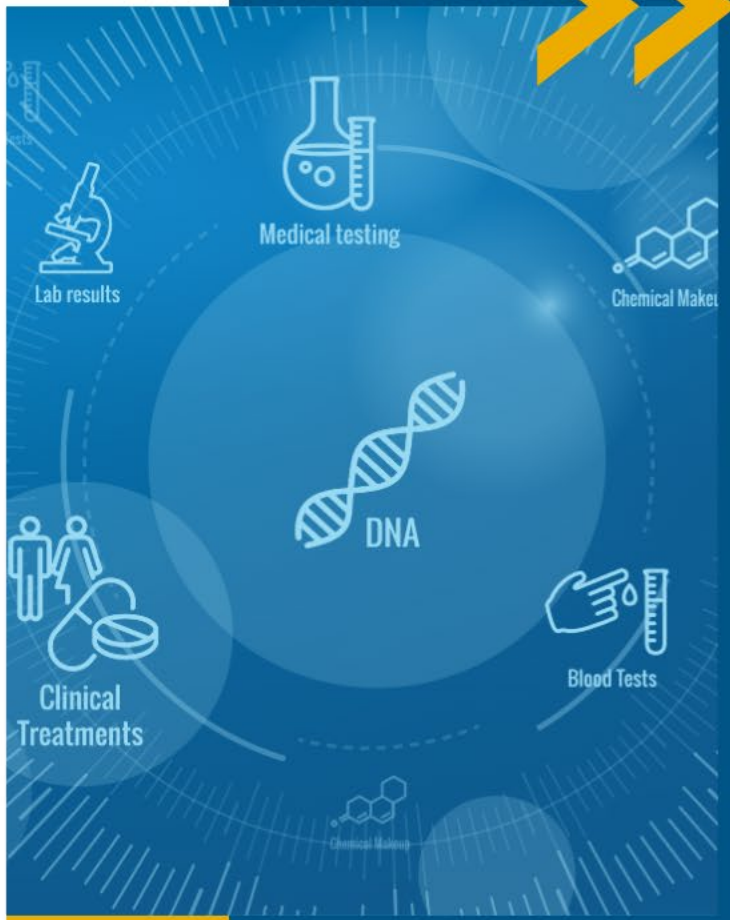
DISCLAIMER

The information speakers provide in any presentation made as part of the 2025 NAF Annual Ataxia Conference is for informational use only.

NAF encourages all attendees to consult with their primary care provider, neurologist, or other healthcare provider about any advice, exercise, therapies, medication, treatment, nutritional supplement, or regimen that may have been mentioned as part of any session.

Products or series mentioned during these sessions do not imply endorsement by NAF.

PRESENTER DISCLOSURES



Meron Azage

The following personal financial relationships with commercial interests' relevant to this presentation existed during the past 12 months:

No relationships to disclose or list

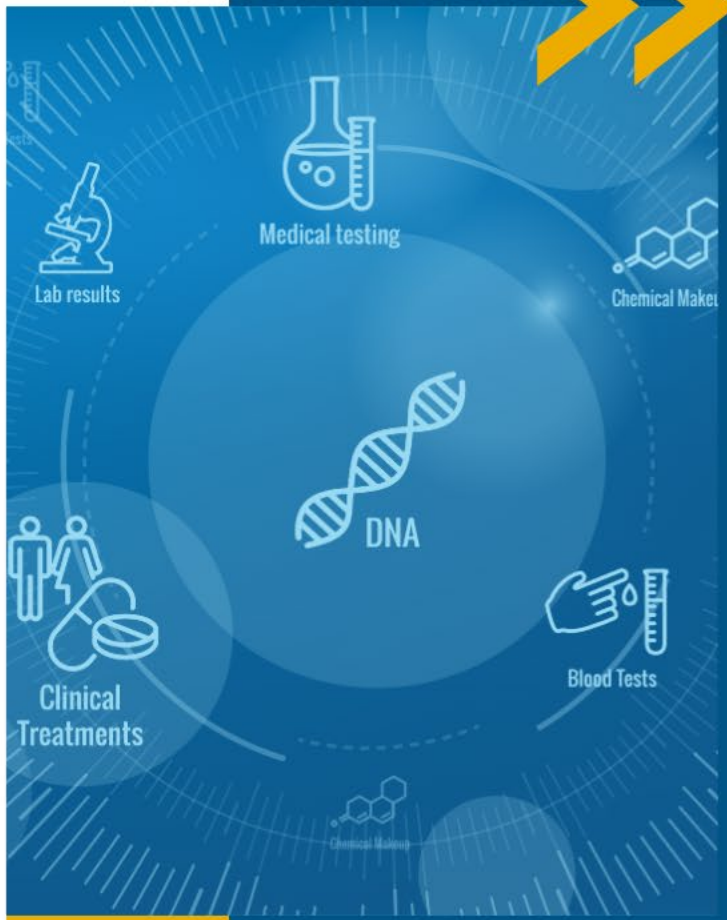
PRESENTER DISCLOSURES

Mary Hogan

The following personal financial relationships with commercial interests' relevant to this presentation existed during the past 12 months:

Relevant financial disclosure:

- Contracted to Solaxa Inc. as Director of Patient Advocacy and Scientific Affairs. Solaxa is a clinical-stage biopharmaceutical company developing treatments for rare neurodegenerative diseases and nerve injury.



Reproductive Medicine Statement

NAF believes that access to comprehensive reproductive healthcare and genetic testing information promotes informed decision-making. We are committed to providing patients with medical resources on these topics, as well as amplifying the voices of ataxia community members to share their stories surrounding reproductive healthcare. We are also committed to supporting genetic counselors in providing comprehensive, unbiased genetic counseling about reproductive options. We acknowledge that the issues surrounding genetic testing and reproductive healthcare are complex. As an Ataxia patient organization, we are committed to transparency in our discussions and respect for all who are considering these options for themselves and their families.

Background

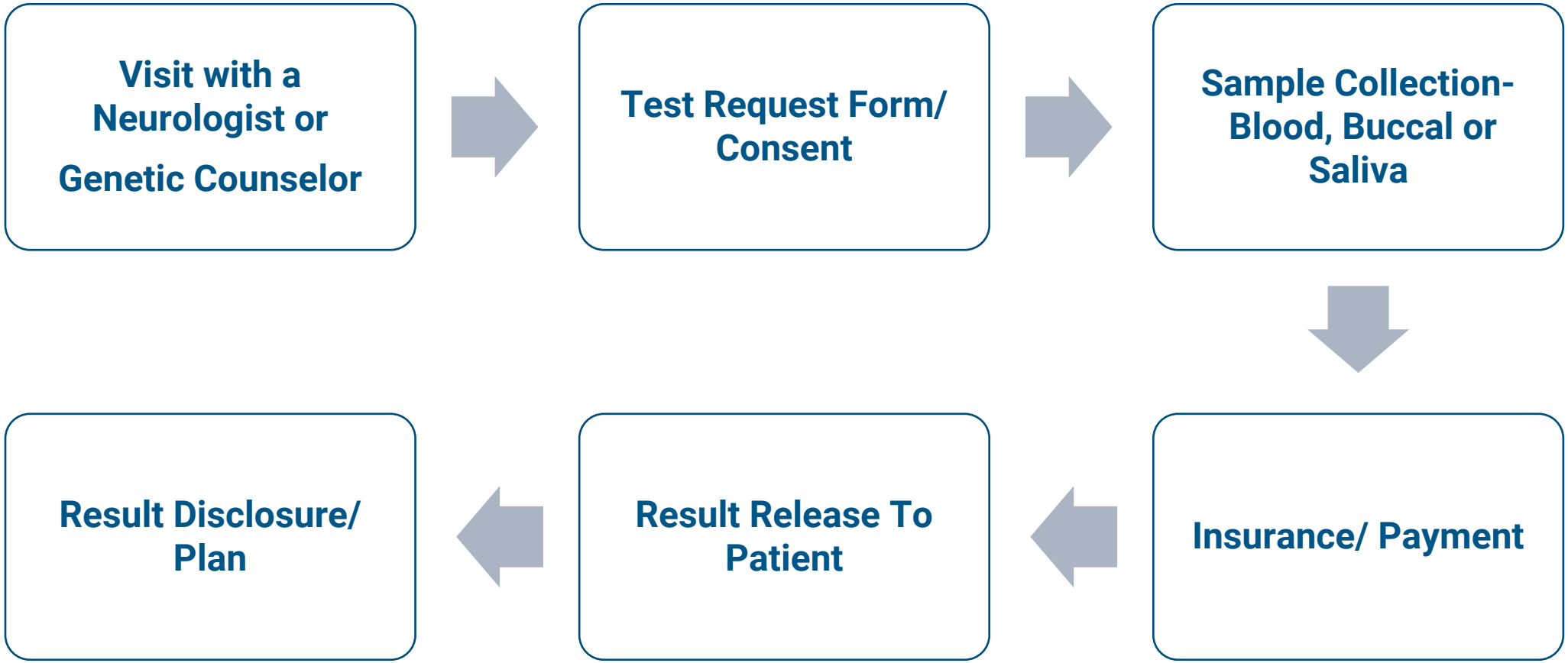
Spinocerebellar Ataxia 27B

- Common adult-onset form of hereditary ataxia
- About half of patients present with episodic features
  - Triggers: Alcohol etc
- Slow progressive with variable age of onset (usually ~age 60)
  - Unsteady gait, stumbling, and imbalance
  - Double, bouncy, or blurry vision
  - Vertigo
  - Cerebellar atrophy
  - Speech changes
  - Others

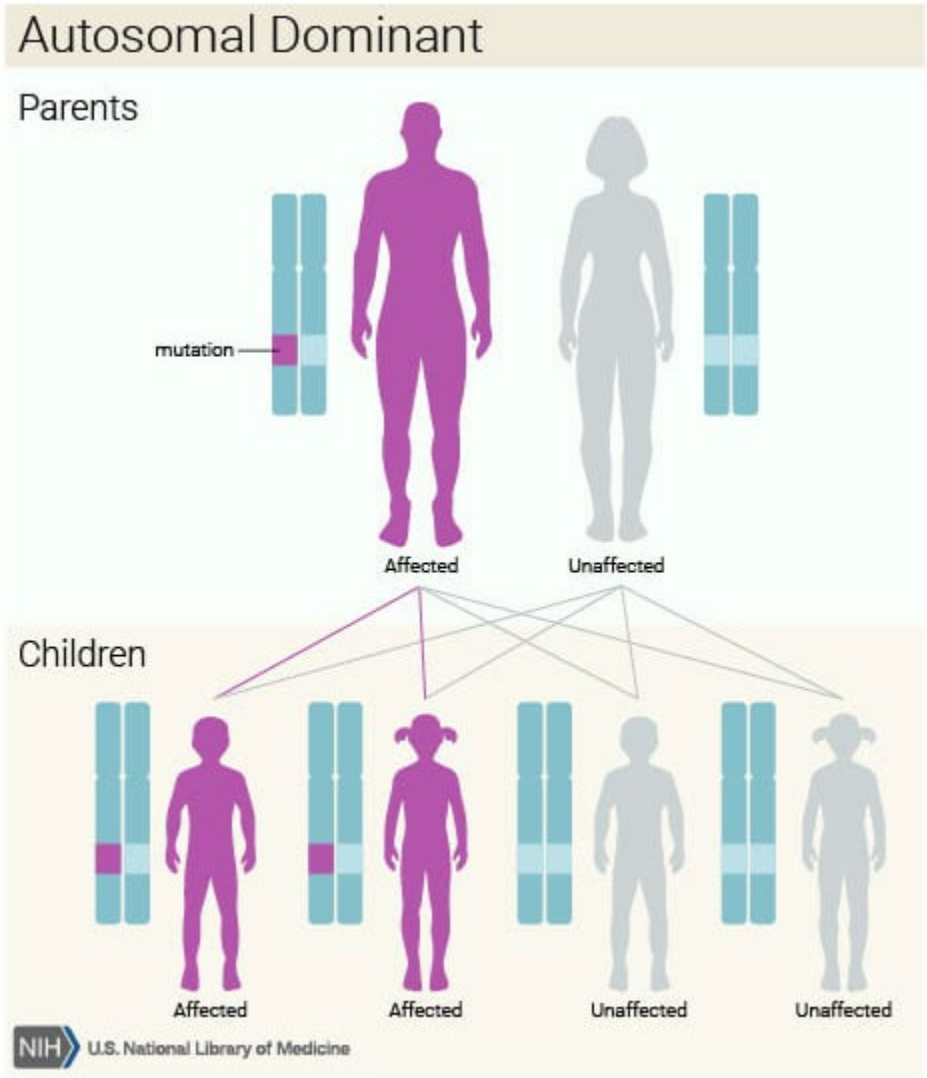
FGF14 Gene: Described in 2022

- Normal GAA repeats: <230
- Gray Zone: 230-249
- Incomplete penetrance: 250-300
- Positive/ Expanded: >300

Genetic Testing Process



Intergenerational Instability



Resources

SCA27b Ataxia Foundation

www.sca27b.org

Genetic Counselor Directory

www.findageneticcounselor.nsgc.org

Family Planning and Financial Considerations Toolkit

https://globalgenes.org/toolkit/2022-fair-toolkit-family-planning-and-financial-considerations/

Planning for Pregnancy When You or Your Child has a Rare Genetic Disease — Know Rare

https://www.knowrare.com/blog-v2/rare-disease-family-planning

https://www.ataxia.org/genetics/

Family Planning

NEW NAF Webpage

https://www.ataxia.org/family-planning/

Deciding to have children is one of the biggest decisions that someone will ever make. This is an incredibly personal choice. Choosing to start, or expand, a family is filled with hope, longing, and emotion. There are many things to consider, particularly for people living with ataxia and those who are carriers for Ataxia conditions.

Families affected by Ataxia may have questions about whether others in the family will develop the disease. There are types of Ataxias that are hereditary (genetic) and types of Ataxias that are not hereditary. Hereditary diseases have different inheritance patterns, or ways they are passed within a family. Knowing the inheritance pattern for the particular disease allows a prediction of the chances the next child will have the condition.

Webinars:
Family Planning / Genetic Testing / Embryo Donation for Research

Resources
Pregnancy Issues / Preimplantation Genetic Testing / Family Planning

NAF SCA1, 2, 3
Free Genetic Counseling and Testing Program

- Launched in 2022
- Free virtual genetic counseling & testing for individuals at-risk for SCA1, SCA2, or SCA3.
- Program has facilitated ~400 Pre-test Genetic Counseling sessions & returned ~300 genetic test results.
- Towards end of 2024, program reached quota of available counseling sessions & tests.
- Reopened March 2025 with a limited capacity of sessions available per month.

Qualifications

- 18 years or older
- Live in USA
- Family history of SCA1, 2, or 3
- NAF member

See if you qualify by answering questions on this webpage:
https://forms.office.com/r/eLpepTDpCW

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