SCA27b Ataxia Foundation

Director - Mary Barile Hogan, MS, MPH

sca27b.org

Gliding into the future to beat Ataxia

Our mission is to improve the quality of life of individuals and families affected by SCA27b while promoting research and treatments for Ataxia.

Overview

- Discovery of SCA27B
- Family Story
- Creation of SCA27b Ataxia Foundation
- Projects & Collaborations
- Future Goals
- Challenges & Take Aways

SCA27B

- 2 years ago - SCA27B named
- Intronic GAA repeat expansion in FGF14
- Highest prevalence in French Canadians
- Likely most prevalent SCA of Euro descent
- Late onset, may begin episodically
- Downbeat Nystagmus
- Favorable response to 4-aminopyridine

Family Story

- History of Ataxia in my family since 1777
- The Darby Glide
- Research studies, 1980 – present
- Connected to other families
- Next generations

June 2023 – Nye/Darby Family Reunion

27b Foundation

- Founded May 2023
- June 2023 – first event
- Educational website – sca27b.org
- Coordinate genetic testing
- Zoom & Facebook support groups
- Research recruitment & surveys
- 100% volunteer effort

Projects & Collaboration

- Awareness campaign – NAF & Bill Nye
- Redenlab speech tests
- St Jude – cell line for FA/SCA27B therapy
- Genetic study – U Michigan
- Fundraising for animal model, U Miami
- SCA27b Study Group – steering committee
- Solaxa – Clinical trial, 4-aminopyridine

June 2023

Family/Research Reunion

- Structure Films
- NAF
- Redenlab speech tests
- U Michigan blood collection

Future Goals

- Launch multi-lingual website
- Increase mental health support
- ✓ Spring 2025 – Social Work intern
- Fundraising intern
- Create in person SCA27B support groups
- More webinars

Challenges

- Fundraising
- Healthcare & insurance challenges
- Access to genetic testing

Takeaways

- Collaborate!
- Share resources
- Be open to new ideas
- Set the tone - positive & hopeful

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sca27b@gmail.com