

# RAF Newsletter Vol.3

## March 2018



### RAF celebrates March Madness



Come join the board of directors from the Rochester Ataxia Foundation at our Fourth Annual March Madness Fundraiser at Jeremiah's Tavern @ 2200 Buffalo Road in Gates.

March 24, 2018 5-9p.m.

For \$40 enjoy all you can eat appetizers and all you can drink beer, wine, and well drinks. There will be a basket raffle, a 50/50 raffle, and March Madness basketball squares. So bring your family and friends to watch the games and support a great cause. You can pre-pay online by visiting our website—<http://www.rochesterataxia.org/>

### Knock Out Ataxia submitted by Jesse Diehl



Last year, I approached a physical therapist to develop a program utilizing boxing as a treatment for Ataxia. My goals were to increase Ataxia awareness as well as to learn what works and is helpful for me in managing my condition. I have taken this first step so that I may encourage others affected by the symptoms of Ataxia.

The program is called Rock Steady Boxing.

<http://rochester.rsbaaffiliate.com/>

It was developed for the Parkinson community and those affected by its symptoms. Ataxia has affected me with similar symptoms, so I had a vision that this type of core, balance and strength training could be beneficial. I have had weekly personal training sessions using this program since last December.

My Physical Therapist, Lindsay Perez, and I are seeing positive signs. I will continue to participate in this program in 2018. My hope is that it will be a step in the direction of finding a treatment for the symptoms of Ataxia. I'm interested in forming a group which will reduce the individual cost or it may be covered by some health insurance plans. Currently, it is a one on one training session for which I pay out-of-pocket.

If you are interested in participating, or would like more information, see their website or contact me at [jesse.diehl61@gmail.com](mailto:jesse.diehl61@gmail.com).



Jesse

**UPCOMING FUNDRAISER—  
SAVE THE DATE**

**June 3, 2018**

**Golf Tournament**

**Webster Golf Club**

## NAF 2018 Annual Ataxia Conference Philadelphia, PA

Thursday, April 5th—Friday, April 6th with an Ataxia Investigator's Meeting Poster Session on Wednesday, April 4th @ 5p.m. Visit the NAF website for further information.

[ataxia.org/annual-ataxia-conference](http://ataxia.org/annual-ataxia-conference)

AAC is the largest gathering of those affected by all forms of hereditary and sporadic Ataxia in the world. At AAC, attendees will have two days to meet and learn from world-leading Ataxia researchers and clinicians, visit vendor exhibits, and network with others. Sessions at the conference will include topics that help a person with Ataxia manage their symptoms, presentations from some of the top Ataxia researchers and clinicians, small groups that are disease-specific, and updates about the latest in Ataxia research. This year's conference will also include a poster session from the Ataxia Investigators.

### Ataxia research update from Dr. Fogel @ UCLA

1) UCLA Neurologist Dr. Brent Fogel and his laboratory completed the latest round of analysis on several hundred ataxia patients, including samples from all undiagnosed Rochester Ataxia Foundation patients. They have some potential candidate genes under evaluation and, if these hold up, they may be contacting families for updates in the coming months.

2) Key publications from 2017

The following article is a multi-center and international collaboration led by Dr. Fogel at UCLA studying the contribution of SCA36 to genetic ataxias in the US.

Valera JM, Diaz T, Petty LE, Quintáns B, Yáñez Z, Boerwinkle E, Muzny D, Akhmedov D, Berdeaux R, Sobrido MJ, Gibbs R, Lupski JR, Geschwind DH, Perlman S, Below JE, Fogel BL. Prevalence of spinocerebellar ataxia 36 in a US population. *Neurol Genet.* 2017 Aug;3(4):e174

The following paper is a multi-center and international collaboration better defining the features of SCA29, in the largest population of patients reported to date. The Fogel Laboratory provided approximately 20% of the subjects to the study.

Zambonin JL, Bellomo A, Ben-Pazi H, Everman DB, Frazer LM, Geraghty MT, Harper AD, Jones JR, Kamien B, Kernohan K, Koenig MK, Lines M, Palmer EE, Richardson R, Segel R, Tarnopolsky M, Vanstone JR, Gibbons M, Collins A, Fogel BL, Care4Rare Canada Consortium, Dudding-Byth T, Boycott KM. Spinocerebellar ataxia type 29 due to mutations in ITPR1: a case series and review of this emerging congenital ataxia. *Orphanet J Rare Dis.* 2017 Jun 28;12(1):121

The following is an opinion article discussing how advances in precision medicine must be directed to benefit adults with chronic disability such as cerebellar ataxia. Precision medicine is an approach for disease treatment and prevention that takes into account individual variability in genes, environment, and lifestyle for each person.

Wolf SM, Thyagarajan B, Fogel BL. The need to develop a patient-centered precision medicine model for adults with chronic disability. *Expert Rev Mol Diagn.* 2017 May;17(5):415-418

**As always, thank you for your continued support. You may contact us at:**

**Rochester Ataxia Foundation P.O. Box 16560, Rochester, NY 14616**

**Email—[rochesterataxia@gmail.com](mailto:rochesterataxia@gmail.com)**

