

RAF Newsletter Vol. 8 Spring 2021



Greetings from the RAF as we recover from the COVID-19 Pandemic

First and foremost we hope that you have all stayed safe and healthy during the pandemic! Like so many other non-profits, the Board of Directors have been meeting via Zoom.

We are all doing as well as expected working from home, going into work, parenting at home, home-schooling, trying to exercise in our basements, and of course enjoying the outdoors!

I'm sure you will all agree, it has been a challenge!

We are optimistic that we are beginning to return to some sort of normalcy with more and more people being vaccinated against the COVID 19 virus. Also with the return of the summer weather, we can gather and dine outdoors again!

As many of you know, we did one online fundraiser—March Madness squares which was very successful. Currently, we are in the midst of our virtual 5K, participate at your convenience during the first full week in June. We are planning to move forward with an in person golf tournament towards the end of the summer. Thank you all for participating in these events.

In addition to the update in this newsletter, we will be providing information on our website related to Dr. Fogel's research @ UCLA as well as information related to the URMC Ataxia clinic.

Thank you to all who have continued to make donations to the RAF and made it possible to continue our work to find—A Cause and a Cure!



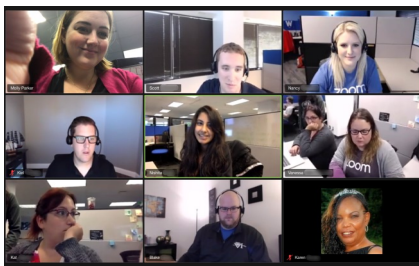
UPCOMING EVENT:

**Summer 2021
SAVE THE DATE—**

**August 1, 2021 @
Victor Hills Golf Course**

More information coming soon!

WNY Ataxia support group goes virtual



ZOOM meetings are increasing in popularity!

The Western NY Ataxia support group goes virtual! Due to the pandemic the western New York Ataxia support group is no longer meeting in person. Instead they meet on ZOOM. Several of the participants said that they enjoyed the new format. No need to get in the car and drive but enjoy the meeting in the comfort of your own home. The group is enjoying the participation of new members from all over the world as a result!

Please contact jesse.diehl61@gmail.com or 585-315-1578 to join and the link will be sent.

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1st Annual RAF Virtual 5k, how it works.....

We invite you to join us for a Virtual Walk-Run-Bike to support the Rochester Ataxia Foundation (RAF)!

Between the dates of Sunday June 6th - Saturday June 12th, 2021, walk, run or bike the distance of your choosing (5K to marathon!). Walk with your friends and family around your neighborhood or gather a team for a socially distanced stroll through a park. No matter your route, distance or group know that you are supporting the RAF in raising funds to find a cause and a cure for Ataxia. A t-shirt will be provided to all participants.

We hope this virtual event will inspire and energize you while we raise funds and awareness for the Rochester Ataxia Foundation.



Registration is on the website!



Thank you to **John Soldi** for 10 years of service on the board of directors. We appreciate all of the time and talent that you have given to this mission over those years and we wish you all the best!

Thank You

Updates from Dr. Fogel @ UCLA

Hello and thank you for the opportunity today to provide a brief update on how the generous support from the Rochester Ataxia Foundation has continued to advance our work at UCLA to improve the understanding of the genetic mutations that can lead to the diagnosis of ataxia.

Despite the complications of the COVID-19 pandemic, 2020 was a productive year for our group, contributing to 17 publications including the first study on the prevalence of a new repeat expansion disorder, RFC1 ataxia, suggesting it is the 2nd most common recessive disorder in the U.S. after Friedreich’s ataxia. We also did work expanding our knowledge on how clinical exome sequencing can be better applied to diagnose up to 50% of patients who are currently undiagnosed.



Brent L. Fogel, MD, PhD UCLA

However, new methods are still needed to diagnose those who still remain undiagnosed after that. In addition, we participated in several smaller studies improving our clinical understanding of several rare ataxias including SCA21, SPG7, and others. I would also like to mention that your contributions helped support the education of future ataxia investigators. Currently, I have one graduate student, 3 undergraduate students, and 2 high school students actively performing research on these projects in my group.

Our goals for 2021 include continuing on our on-going work toward identifying novel ataxia genes, conducting additional studies using genome sequencing to improve diagnosis,

and further our studies in understanding the reasons why specific gene mutations lead to ataxia in the first place.

Thank you all for your generous support. We couldn’t have done any of this without you. Thank you.

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