

RAF Newsletter Vol. 9 Fall 2022



Updates from the RAF!

First and foremost we hope that you have all stayed safe and healthy during the pandemic! Many of us learned all about Zoom during the height of the pandemic and we continue to meet via Zoom with occasional in-person gatherings.

Like many non-profits, the RAF has been experiencing the ups and downs of recovering from the pandemic. After the loss of a few of our board members, we are in the process of re-organizing the board and recruiting new members. We are hopeful that we will weather the changes and emerge as strong as ever!

As a result of these changes, we decided that we would be unable to host a successful golf tournament in 2022. We are going to spend the next few months regrouping, planning, and revamping the Rochester Ataxia Foundation Board of Directors and the RAF website.

We have been fortunate to access the technical knowledge and talent of a Simon Business School graduate, Shah Baig. Shah is volunteering to organize and update the website. The website will soon be easier to navigate and will showcase resources for our members and families affected by Ataxia. Pages devoted to the latest Ataxia research and resources will be added.

We are excited to plan for the future and will keep all of you informed as we continue on this journey!

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

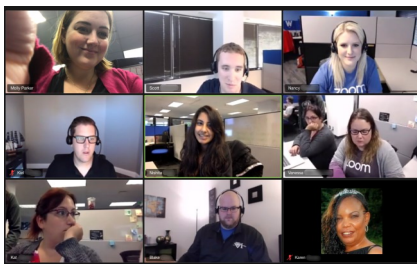


UPCOMING EVENTS:

Winter fundraiser—Online event—Superbowl squares

March Madness—stay tuned for further information as we plan to be back in person for this fun fundraising event

WNY Ataxia support group continues on Zoom!



ZOOM meetings are increasing in popularity!
(*generic stock photo)

The Western NY Ataxia support group remains 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 prefer the new, on-line format. No need to get in the car and drive but enjoy the meeting in the comfort of your own home. The group now includes local participants as well as people from all over the US, Canada, and the world! Please contact - jesse.diehl61@gmail.com or 585-315-1578 to join and the link will be sent to you.

Inside this issue:

Updates from RAF 1
Save the Date

WNY support group 1

Thank you to retiring board members 2

Update from Dr. Fogel @ UCLA 2

Thank you!

Thanks to all of our board members who have “retired” and those who continue to serve.

Thank you to all of our supporters who continue to make donations to the RAF!

Thank you to those of you who answer the call to join our Board!



Updates from Dr. Fogel @ UCLA

The generous support of the Rochester Ataxia Foundation (RAF) has continued to advance our work at UCLA to improve the understanding of the genetic mutations that can lead to ataxia.

In 2021-2022 our group contributed to 29 publications on ataxia, other neurodegenerations, and various genetic conditions. Two key publications focused on the use of molecular biomarkers to improve diagnosis of rare ataxias. One study utilized a complex bioinformatic analysis of changes in gene expression in patient blood to identify disease-specific molecular signatures. Patients who were suspected to have the disease in question could be assessed for the presence of this signature. In this way, our group identified a brand new form of a neuromuscular disorder caused by a well-known ataxia gene. Another study isolated aggregated proteins in blood derived from dying neurons and assessed whether these could be used to identify patients with multiple system atrophy, the most common non-genetic cause of sporadic ataxia in older adults. Additional publications focused on the genetic contributions to conditions that are often confused with or comorbid with ataxia, including Parkinson’s and Alzheimer’s disease, or addressed other genetic conditions that may present with ataxia in some patients, such as leukodystrophies.

RAF contributions have helped support the education of future ataxia investigators and clinicians in the lab. Currently, I have one graduate student and two undergraduate students actively performing research on these projects. In the past year, two undergraduate students from my group have graduated, with one heading to graduate school for a Ph.D. in neuroscience and the other applying to medical school. A recent alumni from my group in 2020 has joined the UCLA David Geffen School of Medicine Class of 2026 to become a future physician.

Our goals for 2023 include continuing on our on-going work on identifying novel ataxia genes and new mutations, understanding how breakdown of specific molecular pathways in the cerebellum lead to ataxia, and searching for clues that could lead to better future therapies. We thank the members of the RAF for your generous support. We couldn’t have done any of this without you.



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