



PXE International
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eNewsletter
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Join us for the 2020 Member Conference!

From the comfort of your own home!

We are excited to announce that we will hold the PXE International Conference October 16 -18, 2020. We will use Zoom to present the talks and enable you to meet one another from all around the world. We realize this is a challenge given all of the timezones we must cover, something we don't have to accommodate when we are in Philadelphia.

This means you will be registering for sessions that work in your timezone, and we will repeat them for other timezones. If, because of presenters' timezones, we cannot offer everything twice, then there will be recorded sessions with live questions and answers.

Stay tuned for more information and registration shortly!

Interns helping out this summer

Heather Chou

I am a rising second year at the University of California, Los Angeles (UCLA). For my project, I will look at two different ophthalmologic aspects of PXE. First, I plan to explore how PXE treatment approaches and patient experiences differ.

Because patients' responses or experiences to the injections used to treat retinal bleeding can differ, I will examine the medical records that so many of you have donated and search for clues that point toward ways to mitigate the pain associated with these injections.

My second project will look at the stories we have heard that many people affected by PXE report going to the retinologist with visual distortion, and the ophthalmologist says they cannot see any issues. I will try to discover if there is a discrepancy between what the retinologist can see and what the patient experiences.

Thus, in exploring these two issues further, I hope to bridge the gap between PXE

patient experiences with PXE symptoms in the retina and clinical treatments, records, or diagnosis done by ophthalmologists. Thanks for sending in your ophthalmology records for these two studies!

Jessica Young

I'm a Masters of Genetic Counseling student at Northwestern University. I'm currently working from home in Vancouver, British Columbia, Canada. I have begun to look at how we can return research results of sequencing the ABCC6 gene to PXEers. We will ask people to take a pre- and post-survey before we return the research results. Research results are not as reliable as clinical results and we want to be sure we are clear about that.

Eventually, when we have a treatment for PXE, it might make a difference to know which mutations in ABCC6 cause your PXE. Right now this information is not yet useful, but we like to plan ahead!

Studies on Hold

Some of you graciously replied to our request to participate in a study that was supposed to take place in Philadelphia in March and April. And then, COVID-19 came and shut down all studies across the globe. We are in the process of re-planning this study, to do it in people's homes, rather than have people come to Philly. This is something that Sharon Terry, in her role as CEO of Genetic Alliance, has been working on for years, and so she is excited to see that PXE International might lead the way. Stay tuned for more information.

Webinar Available

PXEers often ask us about the Magnesium Study, this was a popular webinar in 2019, if you missed it you can find it [here](#). It is labeled PXE and the Eye with Dr. Fuchs. you can get an update on the Study at the beginning of the webinar and stay for the rest and learn about the Eye and PXE.

Have an idea for a webinar you'd like to see? Let us know!

If you have an idea for a webinar you'd like to present yourself, or a topic you'd like to see discussed, please email [Sharon](#).

Mindfulness Through Gestalt Awareness Practice

In response to the stress many PXEers reported feeling because of the pandemic, Sharon offered three online experiences of [Gestalt Awareness Practice](#), something she has studied for a little over four years. In these short practice experiences, Sharon helps open a space for you to feel support moment to moment. She offers tools to help ground you, even in a time of challenge. This practice can lead to self-support and openness to what is. Recordings of these guided practices for PXEers and others are [available here](#).

[Listen Here!](#)

