PXE International
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Join us for the PXE Global Virtual Conference

The conference will be held November 5 to 7, 2021 and you can view the schedule on the website here.

We will have timezone specific schedules, clinicians and researchers from around the world - so that all timezones will be able to participate.

To register for the conference, click the button below!

2021 PXE Conference Registration

2021 ASHG Advocacy Award

We are excited to announce that our fearless leader, Sharon Terry, was honored at the American Society of Human Genetics meeting for her work in Advocacy.

She is recognized for her major contributions as an exemplary advocate at the Genetic Alliance on behalf of people with genetic diseases. Her work has improved access to information and representation for individuals, families, and communities from around the world. She cofounded PXE International, a research advocacy organization for the genetic condition pseudoxanthoma elasticum (PXE), in response to the diagnosis of PXE in her two children in 1994. In addition, she established the first ever lay managed biobank, registry, IRB, and research consortium, and as a result co-discovered the ABCC6 gene and has initiated clinical trials. For more than a decade, she led the coalition that was instrumental in getting the Genetic Information Nondiscrimination Act passed. Her TED talk has been viewed more than a million times.

The ASHG Advocacy Award recognizes individuals or groups who have exhibited excellence and achievement in applications of human genetics for the common good.

Click the button below to watch Sharon receive this prestigious award.

2021 Advocacy Award

Martha Rich
1923 to 2021

PXE International pays tribute to Martha, in great gratitude for all she gave us. Martha died this summer, at the age of 98. Her memorial was held this past week. I could not attend in person, due to the pandemic, but was able to participate in her truly moving memorial service at the First Congregational Church in Sharon, Massachusetts.

Martha Rich did not have PXE but was very important to PXE International's origins. When Elizabeth and Ian Terry were diagnosed with PXE, by our neighbor and dermatologist, Lionel Bercovitch (PXE International's medical director since the start), Martha lived across the street. We moved to Sharon when Elizabeth was a baby, and Ian wasn't even born yet. And so, we had been neighbors for about 6 years when this diagnosis radically altered our lives forever.

Martha was like a grandmother to the kids – showing them the various plants in her yard, informally teaching them a variety of "subjects" in their home-schooling retinue, and showering them with Christmas presents on Christmas morning, sitting around her tree with her family. And, as they got older and had sleep overs, sneaking cool late (9 PM) night TV show watching together became the norm.

When the kids were diagnosed with PXE, as you all have heard many times now, we went into overdrive to found PXE International and jump start research. One aspect of that research was to search for the gene. This required lab space – space we borrowed from Harvard – about 40 minutes from our home. And the only time we could go was after work, after the kids were done with their homeschooling day. And so many times a week, Martha came over after the kids were in bed, about 8:30 PM, and stayed with them until we returned about 2 AM. Night after night she was their quiet guardian as we jousted with the patriarchal biomedical research enterprise. We won – we discovered that the ABCC6 gene was associated with PXE. And all of us won, we now have several potential therapies that would not have been possible without our gene discovery, without Martha Rich.

Martha not only performed this amazing personal service, but also served on the first PXE International board of directors. While a board seat should have been hers for simply giving us this amazing attentive presence with our two most precious little ones, she was also more than qualified. You see, it was Martha who elevated school nurses to their rightful place as full partners in the lives of students, along with their teachers, founding the National Association of School Nurses (NASN). Martha was an early activist! She was a force of nature, and there were many days I sat having tea with her, while the kids discovered some new plant or insect in her backyard. From her, I received a jolt of energy and affirmation, and renewed, I could joust at the next wall, the next challenge. How well she knew what it meant to be a minority in a profession (school nurse in a sea of nurses, or a sea of teachers) and be smart enough to apply it to other situations.

Martha was both no nonsense and warm. She was super smart and very practical. She was strong willed and empathetic. Now that I think of it, Martha epitomized what it means to be both clear about the line, the boundary, and to also have heart, compassion, empathy. Bravo, Martha.

Martha, I am so grateful for the time we had together. I appreciate all the childcare and board work you did. Most of all, I am grateful for how you showed up day after day – present in this moment, ready for what came. I am glad that a few days before our first COVID lockdown I traveled to Sharon and was able to see you and Natalie.

My profound condolences to Brad, Natalie, Doug, Ann, Jon, and Bill and all of Martha’s many grandchildren and greatgrandchildren. And condolences to us, who have lost a great mother. PXE International says goodbye to one of its mothers.

Do you have a question about your life, symptoms, and PXE that you want answered?

Let's answer your questions!! To do that, we will capture the experience of thousands of PXEers. Your participation in the PXE International registry is the most effective way to help yourself and others with PXE.

Click the button below to register.

Already registered? Click the button to answer new questionnaires and connect your electronic health records.

Click here to share your experience

It's Marathon Time

For many years, a good friend of PXE International, Al Ditheridge, has been running and biking to raise funds to further PXE research. While a little last minute this year, we are happy to announce that he successfully completed the Manchester Marathon on October 10th and has raised $1050 for PXE International and two other fantastic local organizations. It's not too late to support Al and his fundraising. Click below to donate to Al's Manchester Marathon go fund me and read about why he runs.

Fundraising to tame PXE, help FOSP & support RTR

COVID-19 Vaccines Informational Webinar

We've received many questions about PXE and the COVID-19 vaccines and are grateful to Dr. Francis Collins for taking the time to talk to the rare disease community and address some of your questions.

In this webinar, Francis Collins, Director of the National Institutes of Health, shared with us why it is critical that individuals living with chronic diseases and conditions receive the coronavirus vaccine. He described the scientific evidence for the safety and efficacy of the vaccine and why it is important to get vaccinated when it is your turn. Click below to watch a recording of the webinar.

COVID Vaccines Informational Webinar

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