“Just a quick email to say how awesome and inspiring I found Giving Tuesday to be!! It was amazing to watch the comments and the number of people sharing your posts! It really felt like everyone was pulling together! Thank you! I hope too that this was reflected in the donations!!”

We also were thrilled with the results of the ‘Giving Tuesday’ response. Jessica Martin headed this for PC Project with Jack Padovano working as a consultant. What a great team effort.

The following email to PC Project clearly shows why we raise funds, provide all services without charge — and why we have a PC community.

I’m tired of isolating myself from the PC Organization, and I want to contribute in any way that I can. Living with PC has taken both an emotional, and, of course, physical toll on me my entire life, and instead of reaching out, seeking help, and being an active member of the community. I’ve avoided this responsibility. My hopes are to be of any use I can be to finding a cure, so that way no one else had to live in this terrible way. I want to be able to walk without pain. I want to feel human. NOTE: If you’d like to reach out to this PCer, let us know as he would be glad to share his email address.
"Gene Therapy
Gene Correction
Gene Editing"

What do these terms mean? There is a lot of confusion about these terms. Recently, the National Academies of Science, Engineering and Medicine held “an international summit to convene global experts to discuss the scientific, ethical, and governance issues associated with human gene-editing research...”

Here is the link to the presentations given at the summit: http://nationalacademies.org/gene-editing/Gene-Edit-Summit/webcast/index.htm

Gene therapy (not gene editing) research efforts for Pachyonychia Congenita are focused on turning off the mutant gene or turning up the activity of the non-affected gene/genes to overcome the effect of the mutation. This type of gene therapy treatment must be repeated to continue to have the desired therapeutic effect and does not 'correct' or 'edit' the gene. With this therapy, the mutant gene may still be passed to offspring with the 50/50 ratio with each pregnancy. If effective, patients using this gene therapy will have relief from the effects of Pachyonychia Congenita but their genome is not altered. The science of the drug is solid, the ability to deliver the drug is not yet available.

On the Genetic Alliance Forum this interesting post was made following the gene editing summit: “Gene editing is so simple - in terms of the science. I think that’s a fair statement when compared to the ultimate complexities of the ethics of gene editing. While each of us with a genetic disease wish for our disease to be ‘cured’, and would be pretty happy if corrected genes could be given to our affected loved ones and in many cases not passed along to our yet to be born siblings or to their siblings by eliminating carrier status, we seldom pause to think that this same technology can be used to modify other ‘less important’ genes as... height, weight, looks, sex, brawn, etc.”

Dean Suhr, MLD Foundation

Frances Smith, PC Project Chief Scientific Officer, represented PC Project at these important meetings.

**FASTER CURES MEETING NOV 1-3, 2015 NEW YORK**

The idea behind faster Faster Cures is that it is an action tank to speed and improve the medical research system with the aim to bring together leaders from all sectors of medical research to find non traditional partners who can help them get things done - to search for new discoveries, turn these discoveries into therapies and ultimately bring therapies to patients.

This was a high energy meeting with lots of panel discussions between scientists, clinicians, patient advocacy groups, industry, members of NIH and the FDA. There were many breakout sessions in which to meet individually with potential partners. There was emphasis on the need for 'patients as partners' - to participate in research and shape the process.

**PEDRA (PEDIATRIC DERMATOLOGY RESEARCH ALLIANCE)
NOV 6-7, 2015 IRVING, TX**

PeDRA is run by leading dermatologists in the USA with the mission ‘To promote and facilitate high quality collaborative clinical, translational, educational, and basic science research in pediatric dermatology’. PeDRA’s vision is ‘To create sustainable, collaborative networks to better understand, prevent, treat and cure dermatologic diseases in children’. The 2015 meeting was attended by pediatric dermatologists, scientists and patient advocacy groups.

PC Project presented a poster at this meeting entitled ‘Correct Diagnosis and Care’ with the emphasis on diagnosis of babies with PC who often are misdiagnosed with thrush which may lead to failure to thrive in some cases. The poster also shared information about the Int’l PC Research Registry. Another section showed PC “Grand Rounds” The poster was well attended and provided new information to attendees, in particular to junior dermatologists.

Many liked the idea of Grand Rounds arranged by PC Project where a number of PCers attend. Please do attend if invited by PC Project. If invited by your physician to attend a Grand Rounds please let PC Project know. Attending with other PCers will have a much greater impact on the education of physicians which is the purpose of Grand Rounds.

It was apparent during the meeting that since the first PeDRA meeting 3 years ago, a number of strong collaborations between pediatric dermatologists, scientists and patient advocacy groups have been formed to create focused multicentre research groups. PeDRA strongly supports training and mentoring of junior pediatric dermatologists and encourages members to be involved in these clinical research programs.

PC Project is already working with a number of PeDRA members and hopes to have a formal PeDRA project focused on PC during 2016.
♦ **PC Natural History** is so important for clinical trial preparation. Please continue to update your information online using the IPCRR registry forms and including updated photos of PC.

♦ **PC patients are invited to attend the Patient Day reception in Manhattan, NY in June 2016.** This will be sponsored by Grunenthal Ltd. (a pharma company focused on pain.) If interested in attending and helping us spread awareness of PC to important researchers and executives, please contact PC Project.

♦ **FDA Awards NORD $250K.** To further the study of rare diseases, the U.S. Food and Drug Administration (FDA) has awarded a $250,000 grant to the National Organization for Rare Disorders. NORD will use the grant to develop 20 **natural history** studies for 20 rare diseases based on a lottery system. PC Project will apply and hope we win this ‘natural history’ lottery.

♦ **2016 PC Patient Support Meeting.** Edinburgh, Scotland. October 28-30, 2016. A flyer will be sent to all in Europe and Asia so you can register soon.

♦ **2017 PC Patient Support Meeting.** Salt Lake City, Utah in June 2017.

♦ **PC Patient Advocates** held their first webinar and a second session will be in mid-December.

♦ **The disease spotlight on the NORD website this week is Pachyonychia Congenita.**
We appreciate so much the wonderful photos you are sending. Please continue to send us photos of your adorable PC kids — and of wonderful PC adults, too. These photos send a message that no words could convey properly. The photos are so important in all of our work to help others learn about PC and how it affects lives. We want others to know the patients and not just the disease. Your photos are wonderful. Please continue to send photos showing fingers, toes, callus, etc. on beautiful pictures of you and your children showing them and their PC—with or without a sign. We want to find an effective treatment—a cure—for these adorable children.

Pachyonychia Congenita causes my nails to grow very thick.

Pachyonychia Congenita causes me to have blisters and painful callus on my feet.
We are preparing a ‘THANK YOU’ video for Jerry Seinfeld to express appreciation for his donation of the proceeds from his show in January in Salt Lake City to benefit PC research. We appreciate so much the wonderful photos you are sending.

For the “Thank You, Jerry” photos with thank you signs (like the ones on this page), the deadline is December 30, 2015.

For other photos, please continue to send those anytime. No deadline.
A DIFFERENT TYPE OF PLANNED GIVING. Planned Giving usually refers to patients and families who include PC Project in their Will and Estate planning. We hope many will do this. This past month, we have learned of another type of ‘planned giving.’ A patient contacted us to donate his body for PC research. We were not prepared to take advantage of this incredibly kind offer from a patient with terminal cancer. We are hopeful that he will respond to treatment and have months to live. However, we are actively learning how skin can be saved at death and how his desire can be fulfilled to make a difference in research. It is complex as skin ‘dies’ within minutes of death and we have to ‘plan ahead’ to be able to benefit from his gift. We thought it would be fun to show you a picture of this PC STAR back in the 1980’s — before chemotherapy begins to take his hair. Thank you Harry for your amazing donation.

THANK YOU FOR TESTING OUR SMARTPHONE PAIN APP—15 patients responded to our invitation to try out the Smart Phone Pain App that allows you to record your highest level of plantar pain each day. We’ve pulled together these responses in the table below. Of course, the responses from 15 patients isn’t enough to make definite conclusions, but some trends are evident. Here are a few initial thoughts. We are attaching the SmartPhone Pain App instructions to this NewsBrief—feel free to use it if you wish to help us gather data.

1. Those with the most pain are most likely to respond
2. Pain levels vary depending on the type of PC with PC-K6c and PC-K6b having significantly lower pain levels (again from only a few patients).
3. Pain levels vary for the same patient from day-to-day.

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Highest level by patient

We will begin an IRB-approved study in January 2016 using the Smart Phone Pain App and an activity...
USING THE PC PAIN SCALE: Please complete this pain scale about the same time each day.
There are only two questions to complete (a) your name (b) your highest pain for the previous 24 hours.

INSTALL THE PC PAIN SCALE to Android or iPhone/iPad

Android – add to home screen


2. Select the menu button and add the page to your bookmarks. In Chrome, just tap on the star and follow the prompt. Open your bookmarks and using the menu button, find the new bookmark you’ve just added.

3. Press down and hold the bookmark until you see the action menu appear. Select Add to your home screen. The bookmark will now be pinned to your home screen for quick access. You may arrange it into the place of your liking.

iPhone/iPad—add to home screen

1. Open Safari on your iOS device. Go to http://sgiz.mobi/s3/PC-Foot-Pain

2. Tap the share button on the menu bar (at top for iPad or at bottom for iPhone).

3. Tap on Add to Home Screen. You will then be shown a short cut name and confirm the web address. Then click Add and the icon will be on your home screen.