Clinical Trials for PC

Since 2004, the most important goal for PC Project has been ‘a cure/treatment for PC.’ Before a drug is approved, a clinical trial is needed. And, a clinical trial requires many partners working together—the scientists, the clinicians, the patients—and usually, a pharma company must join in the effort to provide the funding and expertise necessary for a successful clinical trial.

Before a treatment can be prescribed for patients, it must be approved by the governing agency (FDA in the USA) in each country. A key test to determine whether a treatment ‘passes’ and is approved is whether or not the drug results in something beneficial to patients. Before the clinical trial begins, ‘clinical end points’ must be set which determine the success or failure of the trial.

TransDerm, Inc. and Palvella Therapeutics are two companies working on treatments for PC. In the fall of 2017, over 200 PC patients responded to the Patient Survey on Clinical Trial Endpoints. This information is critically important in designing the next clinical trials for PC. We have a special message from James Valentine, the consultant working with Palvella, who helped with the survey: "A big thanks to all PCers and caregivers who took the time and energy to provide EXCELLENT insights on the survey we distributed. With your hugely valuable feedback now in hand, our collective mission of delivering a high impact therapy to all of you took a GIANT leap forward.

Here’s why: the data and insights you provided us will allow us to work more thoughtfully with researchers and biotech companies who are designing clinical studies of new therapies to treat PC. In order for those clinical studies to be viable, they must be able to measure the effect their drugs have on outcomes that are meaningful to PC patients, i.e. the things patients report are the most debilitating symptoms of having PC. The survey results provided by each of you clearly highlighted those outcomes most relevant to you or your loved ones, such as PCers’ desire to walk longer and have alleviation from the severe foot pain that is a hallmark of PC. Stay tuned for more news around upcoming clinical studies of new therapies for PC. There are many highly committed scientists, researchers, physicians, and biotech companies focused on helping you achieve better lives. More to follow soon. Until then, THANK YOU!"

NEWS ALERT!
Opportunity To Meet With the FDA

PC Project and dEBra of America have been asked to an in-person meeting with the FDA staff in Washington, DC. Sometime around the first week in April 2018 — PC patients, family, caregivers, researchers, investors and others interested in PC will be invited to meet with the FDA. While the FDA governs treatments in the US only, the FDA influences decisions in many/most other countries in the world.

This is an amazing opportunity! We need to have 50 to 100 participants at the meeting where the FDA will learn about PC in the morning and about EB and EBS in the afternoon.

If you and family members and caregivers can possibly attend, please immediately contact PC Project to let us know your interest and willingness to participate. In this meeting, there will be people from the FDA, who will in the future, be on committees that say ‘yes/no’ to drugs for PC and will be influential in many critical decisions.

It is so important that FDA leaders learn of the debilitating effects of PC. They must learn of the constant pain and of the many ways PC impacts your life. This is one place you are not going to want to hide your pain or hide the challenges PC brings to you and your family. Here, your story will be heard!

If you live in the USA, please come to Washington, DC and help us in this important effort to make the need for treatment known.

Please Tell Us You'll Participate
EMAIL: Info@Pachyonychia.org
A Bright Future—PC Project

In 2018, the PC Board of Trustees will focus on the work of PC Project to provide services to patients with pachyonychia congenita. The key programs of PC Project remain:

- **Annual PC Patient Support Meetings (PSM)** where patients meet, teach and support one another and effectively defeat the overwhelming loneliness caused by an ultra rare disease.

- **The IPCRR (patient registry)** founded in 2004 it is one of the most important achievements of PC Project and continues to grow. The IPCRR uses an on-line questionnaire to gather standard data which has changed the understanding of PC from a ‘nail disorder’ to a condition that causes a highly painful plantar keratoderma and other painful features as well as thickened nails.

- **Clinical Trials for PC.** In partnership with Palvella, efforts are moving forward to design a Phase 1b clinical trial for topical rapa for PC patients and TransDerm is working with PC Project to continue testing of TD101, the siRNA targeted at specific PC mutations.

- **The International PC Consortium (IPCC)** to encourage and support collaborative research efforts related to PC.

- **Newsletters** both for patients and for the IPCC members to inform and unite these communities.

- **Publications** to continue to bring attention to PC and explain the debilitating nature of this disorder, emphasize the need for treatment and highlight important research achievements.

- **Pachyonychia.org** website with accurate and current information on all things related to PC.

Giving Tuesday 2017

Thank you to all who raised awareness and/or donated to PC Project for the 2017 Giving Tuesday campaign. As you may have heard, we raised more than $90,000 which has been matched by an anonymous donor.

Your generosity makes all the difference in the lives of PC patients through research and patient support worldwide.

It is thanks to you that we can fight for a cure, connect and help patients, and empower scientific research. Thank you so much for being part of the PC Project family!

PC Pain Study Now Published


The study was conducted with PC patients at meetings in France, Scotland and USA. This is a major breakthrough in a recognized scientific journal to establish pain as a major feature of the PC syndrome. We will forward a copy of the article to each of the patient participants.

PC Project Board of Trustees

Janice Schwartz, Chair
Jack Padovano, Vice Chair
Barbara Feinstein, Secretary
C. David Hansen, Trustee
Jason Hunter, Treasurer

We express appreciation to Preston Cochrane for his service on the PC Project Board and wish him every success in his new position as CEO at a local charity focused on housing for the homeless in Utah.
PC Awareness and Fundraising

We always appreciate the many patients who continually work in their own communities to raise awareness of PC and to also raise funds. It is wonderful to share with you reports of these events.

We encourage you to find your own special project or way to educate friends, family, and medical experts about PC which is such an ultra rare disease few will ever know about PC unless each patient steps forward to carry the message.

How exciting it is this month to report this amazing effort by a patient in Switzerland — now for the 7th year. The funds raised and the information shared about PC are a major contribution to helping us achieve the goals of PC Project on behalf of PCers everywhere. What will you plan for 2018?

Marathon Spinning (Indoor Cycling)

Nathalie Kilchoer, of Switzerland, with her family and friends, held their Seventh Annual Spinning Marathon to raise funds for PC. She sent this report with wonderful photos.

“Dear PC Family:
Our spinning marathon took place on November 12, it was a success. We welcomed more than 220 people and raised CHF 6'500 ($6,774.71 US dollars) -- which we gave to the association "Le Coeur au Pied." (PC Project’s sister charity in France led by Sylvie Cierpucha.)

We are organizing our next spinning marathon on November 4, 2018.

With best regards,
Nathalie”
**A Special Heartfelt Offer from PC Project & Paolo Cognetti**

Dear Fellow PCers:

I have PC. But thanks to the PC Project, I also have hope — hope for a cure in my lifetime.

As a professional musician living with PC, composing music is not a problem. However, playing piano is a constant challenge. If I overdo it, my fingertips start hurting. I don’t stop playing, but I do pull back.

On the worst days, PC prevents me from being able to perform at my very best. As a result, I have to carefully plan my studio sessions, alternate demanding pieces with simpler ones, allow for frequent breaks, and vary my technique.

The same goes for public relations and "meet and greets," which are crucial in my line of work.

Promoting myself means a lot of walking and standing. When I have an appearance, I start getting ready at least two or three days in advance — stressing my feet only when necessary, inspecting venues online and asking for information in advance, and thinking of clever strategies to stay seated most of the time.

Few people in the audience know what it's like to live with the incredible challenge of PC. You do. That's why I'm asking you to make your most generous new-year contribution to the PC Project.

Fellow PCers—you know the daily struggles of living with PC. At the same time, I think you feel as I do ... that our unwavering support for the PC Project is the best hope for a cure.

There was a time I almost gave up playing music. Thankfully, I realized that music is my love. And since other people enjoy what I do, it's worth the pain and struggle!

The PC Project is a huge part of why I have hope. Today, I attend patient support meetings, serve as a PC advocate, and help raise funds so we can continue to move toward a cure.

Together, we are making life better for one another — and making real progress in the fight against PC.

Un abbraccio,
Paolo Cognetti
Composer & Pianist
www.paulocognetti.com

P.S. As part of my commitment to the PC Project, I'm honored to help make possible a special offer ...

February is the month of love and PC Project is ‘all about love.” So every donation over $25 received by the end of February will receive a complimentary copy of my new solo piano cd Paolo Cognetti: Rinascita and the donation will be matched by an anonymous donor.

Donate Now

Thank you in advance for your generosity! -PC

**PC Wiki—A Resource for PCers**

The [PC Wiki](http://www.pachyonychia.org/wiki) features information from patients with comments from PC physicians and scientists and includes tips, direct links to products and vendors. Choice of care techniques and products will vary depending on the specific type of PC, the gene and mutation and personal preferences. We will continue to update and add to the Wiki—continue to share your best tips and ideas. Send to info@pachyonychia.org.
In Loving Memory

Cameron Baker, 21, of Bar Harbor, Maine, died January 3, 2018 at his family home on Wilcox Place, Jamesville. Cam was born in Syracuse and resided most of his life in the Syracuse area. He was a 2014 graduate of the Big Picture School in LaFayette and attended the College of the Atlantic in Bar Harbor. Cam made his home in Bar Harbor for the past year.

Cam suffered from a rare genetic disease, Pachyonychia Congenita (PC), which is a painful condition that affects about 10,000 worldwide. Cam was active with the PC Project since its inception and attended the first national meeting of the PC Project in Kansas City. The PC Project is searching for treatment and a cure of this disease.

Although Cam suffered from crippling chronic pain, he would walk any distance and go to any lengths to help anyone who needed him. His heart, his pockets and his apartment were open to all his friends. He was an endlessly kind and sensitive soul who raged against injustice in any form.

Cam loved loud punk music, his mom, his friends and cats, especially Alice, a male cat he rescued in Bar Harbor.

Cam hated bigots, racists, facists, neo-nazis and Trump.

Surviving are his parents, Robert A. Baker and Marcia Hough; a sister, Alexis Baker of Yurihonjo, Japan; his maternal grandmother, Nancy Hough of Manlius; an aunt, Sandra Hough of Hampton, VA; a great-aunt, Virginia Baker of Westfield, MA; and his beloved Alice.

A celebration of Cam's life will be held after calling hours from 1-4 pm on Saturday, January 13 at Fairchild & Meech DeWitt Chapel, 3690 Erie Blvd East, DeWitt.

Contributions in Cam's name may be made to PC Project, PO Box 17850, Holladay, UT 84117.

Date of Death: January 3, 2018

To express sympathy, please visit fairchildmeech.com

In Loving Memory

We are saddened to report the death of one of our very loved PCers. The obituary of Cameron Baker beautifully captures the story of this very special young man who we have known and loved since the very beginnings of PC Project.

Cameron was only 7 years old when we met in 2004 at the very first PC Patient Support Meeting (PSM) in Kansas City. We remember Cameron with his Dad, Bob Baker (a PCer) and his Mom, Marcia Hough, and his sister, Alexis (just a little older than Cameron and also a PCer.) What fun we had at that meeting. With only 11 PCers known at that time, we definitely formed a lasting bond.

Cameron was part of many PSM through the years. We cheered as he grew through the challenging teen years and on to young adulthood. Our hearts go out to Bob, Marcia and Alexis at this tender time. We love you.
SAVE THE DATE! The FDA has set the morning of Friday, April 6, 2018 as the date for the Externally-led Patient-Focused Drug Development Meeting (EL-PFDD) for PC Project. This historic meeting in Washington DC is our chance to let our PC voices be heard by key FDA officials.

We have over 40 PCers and 20 family members registered to attend. We still have a few spots available for those willing to attend and participate in person.

Attend Remotely! For those of you who live further away, there will be a live webcast where you can participate in audience polling and submit your input in writing. More information will be provided closer to the date of April 6.

Why Should You Participate? The Patient-Focused Drug Development meeting is an opportunity to have Your Voices heard. The U.S. Food and Drug Administration, or FDA, approves all treatments for PC and needs to know what is important to you. This knowledge will impact their decision-making, and lead to better treatments and faster approvals for PC.

Why Now? The Patient’s Voice has historically been absent from the drug development process until a drug was approved by the FDA or when a clinical trial was failing and FDA wanted input from actual patients to understand the problem. Now, the FDA wants your Voices heard at the beginning of the process and throughout! Patient engagement is now a priority for the FDA.

What is the Purpose? Your experiences as an individual living with PC or as their caregiver or family member will educate the FDA about:

- What it is like to live with PC
- Your concerns and feelings about living with PC
- How you currently manage PC
- What meaningful treatments should look like

How Will I Be Able to Participate? Unlike other meetings you may have attended, this meeting is designed to hear from the experts in PC: You! There will be multiple sets of polling questions and extended periods of audience participation where you will be called upon to share your experiences and preferences. FDA officials, representatives from drug companies, and researchers will be there to listen and learn.

REMINDER: A SPECIAL HEARTFELT OFFER

Only a few weeks left in February! Every donation over $25 received by the end of February will receive a complimentary copy of PCer, Paolo Cognetti’s new solo piano CD *Rinascita* and the donation will be matched by an anonymous donor.

Donate at goo.gl/akZL5K

IN LOVING MEMORY

The Grandmother of Robert Hunt, one of our PCers, recently passed away. In lieu of flowers, people were asked to give donations to PC Project. Robert’s mother, Jane Hunt, said, “My mother’s funeral was only a small affair as she had outlived the rest of her family, but I know that all those remaining care deeply about Robert and his well-being. I was touched that they made donations.”

We also received many donations after Cameron Baker’s announcement. Thank you to the Hunt and Baker families for making a difference for others with PC, especially during this tender time for their families.
**SKY DIVE FOR PC PROJECT**

*Julie Peconi, PC Parent*

Chatting to my hair dresser one day in the summer about ways to raise money for PC Project, she suggested a sky dive. When I said no way was I up for that (!), she offered to do one on behalf of PC Project. Fast forward approximately 6 months and many cancellations of the jump due to bad weather, my hair dresser, Rachel Walters of Raw Hair Salon in Swansea, Wales finally completed her charity sky drive! She raised an amazing £701, mainly from her clients, for PC Project! But not only this, she also helped us raise the profile of PC in our area! I’d like to say a big THANK YOU to Rachel for her kindness and her bravery!

**PC MAILBOX**

*Question:* “We are a family with Pachyonychia Congenita PC-K6a and are registered with PC project.

We have a question about a specific symptom that our dermatologist hasn’t been able to help us with.

As you know, one of the symptoms of PC is extra sensitive skin that leads to callus, especially on the feet. Even if this is manage by cutting away the callus on a regular basis we will get small blood vessels (and nerves) that grows into the callus. Cutting in to these blood vessels/nerves will cause bleeding and extreme pain. The result is that we can’t cut away all the callus that we need to especially around cracks.

This is how it will look after a regular trim of the callus :

*Answer:* There is no proven method of how to deal with blood vessels in calluses, but we know many of you suffer with this. Do you have a way of managing that works for you? Let us know at info@pachyonychia.org.

Dr. Edel O’Toole: I have heard patients say that they pare down as far as the bleeding point and then stop. I have also seen patients try and pull out the nerve vessel bundle with they say relief, but looks very painful. In the UK we have lignocaine in a plaster (Versatis) and sometimes if a patient has one isolated extremely painful spot, sticking a little bit on the affected area helps. I have heard of people using silver nitrate and that usually does not hurt. It stops bleeding, sometimes helps pain and could kill cells superficially (eg blood vessels and nerves).

**2018 Patient Support Meeting**

*London, England UK*

This PSM meeting will be held in October 2018. Exact dates and information will be in the next PC News Brief.

Special thanks to Kate Fairbrother, Julie Peconi and Tom Baker for their help in planning and organizing this meeting.
Dr. Phil Gard: Similarly I’ve heard a few PCers say that they make a point of paring down the sites as close as possible, reported it as grim to begin with but a lot better with regular use.

Dr. C. David Hansen: We seem to encounter these commonly but have no specific suggestions for management. This will be a major consideration in a future study. I also wonder if a vascular laser may be helpful in destroying the vessels. I would suggest their dermatologist inquire about laser treatment of vascular lesions.

PC-K6a: I personally use either a ped egg or a rough pumice stone to whittle down the calluses. Since the blood vessels developed in my calluses, I have mostly given up razor blades/scalpels for trimming. Tools such as the ped egg make it easier for me to "feel" and control the depth while filing down, rather than make my calluses bleed with an accidental slice of the blade. Most of the calluses on my feet look like the ones the patient included in the pictures, with occasional bloody spots that one can't always spot while trimming - until the spot is hit.

However, my heel calluses have become quite covered with blood vessels in the past years. The calluses don't cover the bloody spots. They simply grow out together. So unless I want an extra thick mess of callus and blood vessels, I trim.

To stop the stinging/burning, I sometimes apply Americaine, a hemorrhoidal ointment. The active ingredient is 20% Benzocaine. The ointment is gentle and gets down into the bloody spots to numb the pain for a bit.

PC-K6a: I also have severe problems with blood vessels and nerves coming through the calluses. It's so painful! I unfortunately have found nothing to help that except cutting around that or enduring the pain. It would be awesome if you could also tell me what others said concerning this question. Thanks!!

PC-K6a: I cut these blood vessels every now and then (actually it happens more and more often as I grow older): in all cases except one I found that removing the callouses around the blood vessel and the part above it until you get really close to it (but without hitting it) leads to a general improvement, that is the blood vessel seems to pull back. Of course the statement is based merely on my observation and I have no idea whether the cause-effect relationship that I observe is valid...

In my experience I get better results (in terms of quality of life and blood vessels) with less deep trimming performed more often (every two weeks).

K16 patient: “At the moment, I can cut my skin quite low as I had an operation to remove all the callous in January. For a while after having this done, the skin is not so ‘wet’ underneath and also not so sensitive, and the blood vessels do not start to grow back into the callous for ages if you can keep the skin low. I have had a podiatrist take the skin too low before now and I have then had problems walking (because it becomes far too sensitive). I think it needs to be explained to them that getting the skin as low as possible is not always the best thing for PCers — it makes the pain worse, not better. I guess because they have been taught to remove as much callous as possible from all patients, then they feel this is what is best for everyone. I tell them to stop when I feel it is low enough — sometimes they protest but hey....they are my feet! I find the best thing for me is to trim them once a fortnight. Any longer than that and the pain can get worse, or more frequently than that and they can get too sensitive. A podiatrist once told me that it is better to rest your feet for a day after they have been trimmed to give them a chance to ‘heal.’ I find it really helps to do that if possible, and there is much less pain.”

PC-K6a: I wish I had an answer, but unfortunately do not. I suffer from the blood vessels/exposed nerve endings all the time. Yes, they are extremely painful. My podiatrist uses silver nitrate after trimming to try to stop the re-growth and exposure. I also just started using a hemp salve which does help. And I always use the good old stand-bys of Vaseline, Neosporin and Bactroban.

PC-K6b: This is a problem that I myself have, as well as my sisters and sons who have PC. What I do, which has been quite active, is after debridement of the lesions, use silver nitrate on them. This stops the bleeding and makes that much less sensitive, and tends to make them less prominent in the future. I would not necessarily recommend cutting them out, because you run the risk of developing scar tissue which can be painful. Would definitely try the silver nitrate first, as it has been quite helpful.
PC PATIENTS AND CAREGIVERS, WE NEED YOU!

To all PC patients and caregivers of patients, wherever you are in the world—The historic Externally-led Patient-Focused Drug Development Meeting with the FDA is less than a week away. The meeting will be broadcast live, April 6th, starting at 8am EDT near Washington DC, USA. We need your participation!

Please do the following:

1. Download the polling app either on your phone or computer. Do this BEFORE the meeting starts. You can do this today! Attached to this email are step-by-step instructions and links.

2. Watch the broadcast live

3. Refer to the schedule below to know when to participate in the polling questions.

EL-PFDD FDA Meeting Schedule – The times below are in Eastern Daylight Time. Please convert to your time zone. (i.e. 8:30am EDT=1:30pm UK)

<table>
<thead>
<tr>
<th>Time</th>
<th>Session</th>
</tr>
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<tbody>
<tr>
<td>8:30am</td>
<td>Opening Remarks</td>
</tr>
<tr>
<td></td>
<td>Janice Schwartz, Chair, Pachyonychia Congenita Board of Trustees</td>
</tr>
<tr>
<td>8:35am</td>
<td>Welcome Remarks</td>
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<tr>
<td></td>
<td>Julie Beitz, M.D., Director, Office of Drug Evaluation III</td>
</tr>
<tr>
<td>8:50am</td>
<td>Introduction and Meeting Overview</td>
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<tr>
<td></td>
<td>James Valentine, J.D., M.H.S., Meeting Moderator</td>
</tr>
<tr>
<td>8:55am</td>
<td>PC Clinical Overview</td>
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<tr>
<td></td>
<td>Anna Bruckner, M.D., University of Colorado</td>
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<tr>
<td></td>
<td>– Denver</td>
</tr>
<tr>
<td>9:05am</td>
<td>Audience &amp; Remote Demographic Polling</td>
</tr>
<tr>
<td>9:15am</td>
<td>Topic 1: Living with PC</td>
</tr>
<tr>
<td></td>
<td>• Panel discussion</td>
</tr>
<tr>
<td></td>
<td>• Audience &amp; remote polling</td>
</tr>
<tr>
<td></td>
<td>• Moderated audience discussion</td>
</tr>
<tr>
<td>10:35am</td>
<td>Break</td>
</tr>
<tr>
<td>10:45am</td>
<td>Topic 2: Current &amp; Future Treatments</td>
</tr>
<tr>
<td></td>
<td>• Panel discussion</td>
</tr>
<tr>
<td></td>
<td>• Audience &amp; remote polling</td>
</tr>
<tr>
<td></td>
<td>• Moderated audience discussion</td>
</tr>
<tr>
<td>11:55am</td>
<td>PC Session Summary</td>
</tr>
<tr>
<td></td>
<td>Kendall Marcus, M.D., Director, Division of Dermatology and Dental Products</td>
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</tbody>
</table>

SUCCESS ON A SPECIAL HEARTFELT OFFER!

Warmest thanks to Paolo for donating his talents and to all of you who donated to PC Project. You raised over $4,000.00 which was matched by an anonymous donor. We hope you enjoy the beautiful music from a famous PC composer, all the while knowing you’ve donated to an important cause.

SAVE the DATE

OCTOBER 19-21, 2018
LONDON, ENGLAND UK
PATIENT SUPPORT MEETING

Planning is well underway for the 2018 European Patient Support Meeting which will be held the weekend of October 19, 20, 21. Due to the high number of those suffering from PC living in England, we are breaking from tradition this year and will be holding the meeting in London!

To accommodate as many travellers as possible the meeting will take place in the vicinity of Heathrow Airport. We are excited to welcome all of you and your family and friends to this meeting.

More information will be available soon.
The PC Externally-led Patient Focused Drug Development (EL-PFDD) meeting with FDA officials on April 6, 2018 was a huge success!

With over 100 PC patients participating in person or via the live webcast, patients successfully communicated to key FDA officials the impact of PC on their lives, how they manage their PC and what a meaningful treatment will look like.

Patient input and experiences were expressed through panel presentations, live polling and audience participation. Patients who attended remotely submitted their answers to open-ended questions online. All answers will be incorporated into the official Voice of the Patient Report for FDA.

An impressive number of FDA officials attended the meeting - 24 in person, 18 via the webcast. This was a record amount of FDA officials to attend an EL-PFDD meeting.

Why was this meeting so important for PC patients, FDA officials, industry partners, scientists, and clinicians?

1) FDA officials who listened at the meeting will make critical decisions regarding future treatments for PC.

2) While FDA officials will seek out medical literature and input from clinical experts, most FDA reviewers will have little if any personal knowledge of PC from their time in practice. Neither the published literature, nor the clinical expertise can fully capture the experiences of what patients experience day-to-day. It is only patients (and their caregivers) who have that knowledge to share.

3) There isn’t always unity between patients and clinicians on what the most pressing unmet medical needs are, especially when a disease is as ultra-rare as PC. PC patients were able to express to FDA officials what those unmet needs are in a compelling way. The Voice of the Patient Report from this meeting will be used by FDA as a standard for PC.

4) FDA needs to know it is designing clinical trials that will generate empirical evidence on outcomes that are important to PCers. What a patient describes as the greatest burdens of a condition can lead to the appropriate selection of outcome (baseline) measures. Knowing about the day-to-day impact of PC can help determine when and how to implement outcome measures in clinical trials.

The most attractive thing about PC Project to scientists, clinicians and industry partners is our strong PC community. You have made our group robust by:

- Participating in the International PC Research Registry
- Advocating for PC through social media, volunteering, fundraising and awareness activities
- Responding to emails, surveys, and other calls for help including participating in meetings and studies

PC patients may be one in a million (literally) but united we are powerful!

The van der Laan-Kokx family raised $500 in donations to PC Project. $100 of this came from their local vegetable shop “De Pompoen” from Liempde.

Amanda Soderstrom created a Facebook Fundraiser for her birthday and raised $280 for PC Project. This was her fundraiser message: “For my birthday this year, I’m asking for donations to Pachyonychia Congenita Project. I’ve chosen this non-profit because I have PC and so few people know about it. Because so few people know about PC, there isn't much funding available. Their mission means a lot to me, and I hope you'll consider contributing as a way to celebrate with me. Every little bit will help me reach my goal and go towards finding a cure for this horribly painful disease.”

175 PCers or caregivers participated in the post-FDA Patient Focused Drug Development meeting poll. The results will be included in the official Voice of the Patient Report submitted to FDA. Your answers are already being shared among stakeholders interested in treatments for PC.

Dr. Anna Bruckner giving PC Clinical Overview at FDA Meeting
5) Patient input at the meeting also focused on patient preferences for future treatments. FDA recognizes that it is a judgement call as to whether the benefits of a drug outweigh the risks when it is making an approval decision. Thus, FDA heard the preferences of patients about future therapies; they can assign appropriate weight to those things that are most important to patients when balancing those benefits against the risks of a product.

6) Finally, FDA saw real patients, with real pain and challenges. FDA officials have learned not only about PC, but that PC is a disease that needs effective treatments to reduce or eliminate the debilitating, painful burdens PCers live with every single day.

PC Project appreciates the IPCC members who were able to attend the meeting in person – Drs. Anna Bruckner (who gave the PC clinical overview), C. David Hansen, Roger Kaspar, Sancy Leachman and Joyce Teng. PC Project also appreciates James Valentine (consultant and moderator) and the support of its industry partner, Palvella, including CEO Wes Kaupinen and members of his team who attended in person.

**Sampling of Polling Results**

Which PC conditions have impacted your life? Check all that apply.

<table>
<thead>
<tr>
<th>Value</th>
<th>Percent</th>
<th>Responses</th>
</tr>
</thead>
<tbody>
<tr>
<td>Thickened nails</td>
<td>80.1%</td>
<td>145</td>
</tr>
<tr>
<td>Painful calluses/blisters on the soles of your feet</td>
<td>97.2%</td>
<td>176</td>
</tr>
<tr>
<td>Painful calluses/blisters on hands</td>
<td>23.2%</td>
<td>42</td>
</tr>
<tr>
<td>Painful blood vessels/nerves in calluses</td>
<td>49.2%</td>
<td>89</td>
</tr>
<tr>
<td>Deep persistent itch in feet</td>
<td>43.6%</td>
<td>79</td>
</tr>
<tr>
<td>Infections in nails or feet</td>
<td>46.4%</td>
<td>84</td>
</tr>
<tr>
<td>P follicular hyperkeratosis (little bumps on waist, legs, arms, etc. that cause irritation)</td>
<td>43.6%</td>
<td>79</td>
</tr>
<tr>
<td>Leukokeratosis (white growth on tongue)</td>
<td>49.7%</td>
<td>90</td>
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<tr>
<td>Trouble feeding as a baby</td>
<td>18.8%</td>
<td>34</td>
</tr>
<tr>
<td>Other - Write in</td>
<td>17.7%</td>
<td>32</td>
</tr>
</tbody>
</table>

Of all the symptoms that you experience because of your PC, which 1-3 symptoms have the most significant impact on your life? This was an open ended question, so this word cloud shows the most used words. The bigger the word, the more it was used.

Overall, how would you characterize your PC disease severity or the severity of the person you care for compared to someone without PC?

How do your PC symptoms affect your daily life? Check all that apply. They:

<table>
<thead>
<tr>
<th>Value</th>
<th>Percent</th>
<th>Responses</th>
</tr>
</thead>
<tbody>
<tr>
<td>Limit my walking</td>
<td>90.9%</td>
<td>160</td>
</tr>
<tr>
<td>Limit my standing</td>
<td>77.3%</td>
<td>136</td>
</tr>
<tr>
<td>Limit my ability to work consistently and effectively</td>
<td>47.2%</td>
<td>83</td>
</tr>
<tr>
<td>Limit the types of jobs I can realistically perform</td>
<td>72.2%</td>
<td>127</td>
</tr>
<tr>
<td>Partially or completely limit my ability to participate in activities</td>
<td>76.1%</td>
<td>134</td>
</tr>
<tr>
<td>Cause depression or discouragement</td>
<td>46.0%</td>
<td>81</td>
</tr>
<tr>
<td>Cause difficulty sleeping</td>
<td>34.7%</td>
<td>61</td>
</tr>
<tr>
<td>Cause difficulty socializing</td>
<td>42.6%</td>
<td>75</td>
</tr>
<tr>
<td>Force me to hide my nails and bare feet</td>
<td>69.9%</td>
<td>123</td>
</tr>
<tr>
<td>Force me to make up stories about why I walk the way I do or why my nails and feet look the way they do, to avoid having to explain PC</td>
<td>39.8%</td>
<td>70</td>
</tr>
</tbody>
</table>

In the absence of a cure, a clinically meaningful treatment for PC would: Check all that apply.
FDA MEETING HIGHLIGHTS—PCers at PC Project Dinner April 5

Above: Some members of the PC Project Board of Trustees
EL-PFDD Meeting with FDA. Panel One: Living with PC
EL-PFDD Meeting with FDA. Panel Two: Managing PC & Future Treatments
For the past several years, June has been designated as Pachyonychia Congenita Awareness Month - and this year is no different. In fact, with the recent attention PC has received from PC Project’s meeting with the US Food and Drug Administration (FDA), this year it’s more important than ever that we join together to: (a) raise awareness of Pachyonychia Congenita and (b) raise funds for both research and patient support. By joining together from around the world to raise the profile of PC, we show the world, including the FDA, funding bodies, researchers and others, how much we want effective treatments and a cure for this condition!

For PC Awareness this month, we invite everyone to join. Participation this year is simple and is the same as it’s been in similar years. Here’s how to participate:

1. **CHOOSE your action.** Choose something you already know and like... or any action that will be fun for you, family and friends to help raise awareness and funds for PC. Your action may be LARGE or very small and may raise only $1 (or $100,000!) — the important thing is that with PC so rare, you are the ‘heart’ of PC action where you live. No one can take your place — you are the headquarters this June for PC Awareness in your local area.

2. **SHARE Your Event:** Let us at PC Project know what you are up to! Post on the PC Awareness Facebook event page at [facebook.com/events/211923368366107](http://facebook.com/events/211923368366107) or send an email to info@pachyonychia.org. We can provide PC flyers and other assistance as needed. Share your event with the PC Project story to your local news outlets and on social media. The many events around the world (large and small) create a Pachyonychia Congenita news story which will enable us to meet the goal of public awareness for PC.

3. **REPORT Your Success:** Please send us an email to info@pachyonychia.org telling us what you did, and how much you raised. We will share your stories, hopefully helping inspire others who may want to use your ideas in the future.

We appreciate all you do to help raise support for PC in your area and look forward to working with you for this PC Awareness Month, June 2018. That said, if you are unable to do an event in June please don’t panic. We welcome awareness and fund raising at any time of the year and are here to offer our support as needed! We are grateful for those who have already held events raising awareness and funds for PC.
MONTHLY SUSTAINING DONORS

PC Project’s logo has always had a heart in it. Why? Because, PC Project began with love.

One person’s love for her daughter-in-law and grandchildren affected with PC motivated her to do something to make a difference. And she did. Mary Schwartz, our beloved founder, brought together the hearts of PCers, scientists, physicians, and industry leaders to collaborate in finding treatments for this ultra-rare disorder. Her PC legacy of love is left to us to continue.

There is a special group of eight individuals in our PC community who have been consistently sharing their love by donating monthly to PC Project. We call these individuals our “PC Love Builders”. Their monthly support gives us encouragement and goes directly to our programs and services which fulfill our mission to help patients, empower research and ultimately find a cure.

THANK YOU to our monthly donors, our “PC Love Builders” for continuing the legacy of love.

In June we will introduce a special challenge for the rest of us, so please watch for those emails.

IPCC SPOTLIGHT: FRANCES J.D. SMITH, PhD

PC Project Chief Geneticist, IPCC Genetics Team Chair and Medical and Scientific Advisory Board Member. University of Dundee Associate Staff

After completing my undergraduate degree in Scotland, followed by a couple of years in a lab at the Royal Veterinary College, London, I returned to Scotland to take up a PhD in the Dermatology Department, University of Edinburgh, to investigate the cause of Epidermolysis Bullosa Simplex (EBS). This was an extremely exciting time in the keratin field as the genetic cause of various rare keratin disorders were being discovered. The first, epidermolysis bullosa simplex, due to mutations in KRT5 or KRT14, was elucidated by several labs worldwide, including Birgit Lane’s lab, University of Dundee, Scotland. Fortunately, I was able to move to Dundee to Birgit’s lab to perform genetic analysis of Scottish EBS lab to perform genetic analysis of Scottish EBS and that became the start of my ‘gene hunting’ career.

As a postdoc in Birgit’s lab, my interest in the molecular basis of human keratinizing disorders continued. By now the first mutations had just been identified in PC (in KRT16 and KRT17 by Irwin McLean, in Birgit’s lab & in KRT6A by Paul Bowden, Cardiff, UK) and so here began my first involvement with PC and the genetic analysis of subsequent cases.

Other projects included the ‘plectin adventure’ – plectin was a candidate gene for recessive epidermolysis bullosa with muscular dystrophy but required cloning of the gene before genetic analysis could begin. With strong competition from several other groups we teamed up with one, Jouni Uitto’s lab at Thomas Jefferson University, Philadelphia, USA resulting in several trips to Philadelphia in the space of a few months by myself and Irwin to successfully complete the project. Later that year we returned to Jouni’s department to set up a research lab where I continued ‘gene hunting’ for various rare genetic skin disorders, and during which time we identified the fourth causative keratin gene for PC, KRT6B.

On returning to Dundee at the end of 1998, the lab had established an international reputation for discovery and diagnostics of rare keratinizing disorders. My passion for tracking down causative genes continued in Irwin’s lab with the ‘filaggrin experience’ and our discovery of the first filaggrin mutations in ichthyosis vulgaris and atopic dermatitis.

A few years before, in 2004, I was invited to the first International PC Project Consortium Symposium, in Salt Lake City USA. This was a highly motivated meeting of clinicians and scientists to set the aims and goals of PC Project. Subsequently, I established the genetic testing service for PC Project in Dundee and a few years later, we discovered the fifth keratin gene associated with PC, KRT6C. Alongside the diagnostics, other projects were established in the lab to develop therapeutics for PC including siRNAs and drug screening.
PC Project and Irwin’s lab, co-hosted the first European PC Patient Support Meeting in Dundee in 2004 – with around 15 patients and their families attending. The meeting was hugely inspiring for all of us – highly emotional for many patients who had never met anyone else with PC, educational for clinicians who so rarely meet more than one or two PC patients in a lifetime, and impacting for lab members providing inspiration for their studies. The success of this meeting led to biennial European PC patient meetings in and around Dundee.

In Oct 2015, after many years of pipetting, PCR and sequencing! I joined the small team at PC Project, and work from Dundee. Although not in the lab I oversee the genetic testing so am still very much involved in the diagnostics and providing patients with a definitive answer for what is causing their condition, as well as being involved in many other aspects of PC Project towards the ultimate goal of a treatment.

To do this we need your help!! All information we receive will be reviewed by physicians but you are all experts on PC - how you care for it and how it affects your family. We need your input to determine what is important to you as PCers and what information should be included in these projects. For example, as a PCer what do you wish your parents would have known or done differently? As a parent of a child with PC what do you do that helps your child, or alternatively, what do you want to know?

Over the next few weeks we will be circulating a small questionnaire to help us collect the information that you find helpful and that you want to share. In the meantime, please start thinking! PC Advocate, Julie Peconi, is coordinating this project. She can be contacted via PC Project.

PCERS MAKING A DIFFERENCE

James Andrade and Sean Dempsy, two PC patients, participated in the Atlantic Dermatological Conference Grand Rounds in Providence, Rhode Island on May 12th. They educated 400 doctors about PC and gave informational handouts to each physician.

Adam Rocha participated in a local doctor training session in San Antonio, Texas on May 17th. He also educated doctors and distributed brochures about PC.

The efforts of these patients are vital in educating physicians about PC. If you are invited to participate in a similar event, please let us know. We can provide PC Project brochures and invite other PCers to attend with you.

JEANS FOR GENES GRANT

PC Project received a small grant from the British funding body Genetic Disorders UK under their Jeans for Genes fundraising. The aim of the grant is to improve the lives of children affected by a genetic disorder and with this valuable funding we plan on developing resources to help children with PC and their parents.

It is our aim to help empower children to manage and care for their condition at different ages, and to help them work through any feelings of isolation they may have as a result of their PC. For example, through the development of a child specific section on our website and an information booklet, we will look to include tips for how children can put PC into their own words, examples of how children around the world manage their PC, and case stories and pictures of PC children.

PC Project leaders met with key members of the International PC Project Consortium in Orlando, Florida May 16th (in conjunction with the International Investigative Dermatology Conference) for a strategic planning session targeted at moving PC Project and PC research forward.

IPCC STRATEGIC PLANNING MEETING

PC Project leaders met with key members of the International PC Project Consortium in Orlando, Florida May 16th (in conjunction with the International Investigative Dermatology Conference) for a strategic planning session targeted at moving PC Project and PC research forward.

Register Now

LONDON PATIENT SUPPORT MEETING

OCTOBER 19-21, 2018

PACHYONYCHIA.ORG/2018-LONDON-ENGLAND-PATIENT-SUPPORT-MEETING/
INTERNATIONAL PACHYONYCHIA CONGENITA RESEARCH REGISTRY (IPCRR) UPDATE

Joining the IPCRR is the most important action a patient can take. The clinical and molecular (genetic testing) data collected by the IPCRR is central to our success in developing therapies and a cure for PC.

- As of May 2018, the IPCRR has enrolled over 1,000 patients.
- Clinical data (IPCRR questionnaire and photos) and molecular data (genetic testing) has been completed by 814 PC patients (444 families) in over 48 countries.
- 116 different PC mutations have been identified.

Location of 814 individuals with genetically confirmed PC

<table>
<thead>
<tr>
<th>Country</th>
<th># Individuals</th>
</tr>
</thead>
<tbody>
<tr>
<td>Argentina</td>
<td>3</td>
</tr>
<tr>
<td>Australia</td>
<td>14</td>
</tr>
<tr>
<td>Austria</td>
<td>3</td>
</tr>
<tr>
<td>Belgium</td>
<td>4</td>
</tr>
<tr>
<td>Brazil</td>
<td>27</td>
</tr>
<tr>
<td>Bulgaria</td>
<td>1</td>
</tr>
<tr>
<td>Canada-Alberta</td>
<td>15</td>
</tr>
<tr>
<td>Canada-British Columbia</td>
<td>1</td>
</tr>
<tr>
<td>Canada-Ontario</td>
<td>11</td>
</tr>
<tr>
<td>Canada-Quebec</td>
<td>4</td>
</tr>
<tr>
<td>Canada-Saskatchewan</td>
<td>6</td>
</tr>
<tr>
<td>Chile</td>
<td>1</td>
</tr>
<tr>
<td>China</td>
<td>9</td>
</tr>
<tr>
<td>Colombia</td>
<td>3</td>
</tr>
<tr>
<td>Czech Republic</td>
<td>4</td>
</tr>
<tr>
<td>Denmark</td>
<td>6</td>
</tr>
<tr>
<td>Finland</td>
<td>6</td>
</tr>
<tr>
<td>France</td>
<td>57</td>
</tr>
<tr>
<td>Germany</td>
<td>15</td>
</tr>
<tr>
<td>Greece</td>
<td>1</td>
</tr>
<tr>
<td>Guatemala</td>
<td>1</td>
</tr>
<tr>
<td>Hungary</td>
<td>1</td>
</tr>
<tr>
<td>India</td>
<td>14</td>
</tr>
<tr>
<td>Iran</td>
<td>1</td>
</tr>
<tr>
<td>Ireland</td>
<td>6</td>
</tr>
<tr>
<td>Israel</td>
<td>9</td>
</tr>
<tr>
<td>Italy</td>
<td>7</td>
</tr>
<tr>
<td>Japan</td>
<td>11</td>
</tr>
<tr>
<td>Malaysia</td>
<td>3</td>
</tr>
<tr>
<td>Mexico</td>
<td>5</td>
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<tr>
<td>Netherlands</td>
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<td>New Zealand</td>
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<td>Norway</td>
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</tr>
<tr>
<td>Peru</td>
<td>1</td>
</tr>
<tr>
<td>Philippines</td>
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<td>Poland</td>
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<td>Portugal</td>
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<td>Puerto Rico</td>
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<tr>
<td>Romania</td>
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</tr>
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<td>Serbia</td>
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<tr>
<td>Slovenia</td>
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</tr>
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<td>South Africa</td>
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<td>South Korea</td>
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<td>Spain</td>
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<td>Switzerland</td>
<td>10</td>
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<tr>
<td>Turkey</td>
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<td>UK-England</td>
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<td>UK-Northern Ireland</td>
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<td>UK-Scotland</td>
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<tr>
<td>UK-Wales</td>
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<tr>
<td>United Arab Emirates</td>
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</tr>
<tr>
<td>USA</td>
<td>360</td>
</tr>
<tr>
<td>Venezuela</td>
<td>2</td>
</tr>
<tr>
<td>Vietnam</td>
<td>5</td>
</tr>
</tbody>
</table>

Demographics of 814 individuals with genetically confirmed PC

<table>
<thead>
<tr>
<th>PC-K6a</th>
<th>PC-K6b</th>
<th>PC-K6c</th>
<th>PC-K16</th>
<th>PC-K17</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td># Individuals</td>
<td>331</td>
<td>74</td>
<td>24</td>
<td>251</td>
<td>134</td>
</tr>
<tr>
<td>in # Families</td>
<td>208</td>
<td>30</td>
<td>9</td>
<td>124</td>
<td>73</td>
</tr>
<tr>
<td>Female</td>
<td>177</td>
<td>34</td>
<td>12</td>
<td>120</td>
<td>83</td>
</tr>
<tr>
<td>Male</td>
<td>154</td>
<td>40</td>
<td>12</td>
<td>131</td>
<td>51</td>
</tr>
<tr>
<td>In USA</td>
<td>134</td>
<td>24</td>
<td>12</td>
<td>126</td>
<td>69</td>
</tr>
<tr>
<td>Outside USA</td>
<td>197</td>
<td>50</td>
<td>12</td>
<td>126</td>
<td>69</td>
</tr>
<tr>
<td>Spontaneous</td>
<td>140</td>
<td>11</td>
<td>0</td>
<td>58</td>
<td>38</td>
</tr>
<tr>
<td>Familial</td>
<td>191</td>
<td>63</td>
<td>24</td>
<td>193</td>
<td>96</td>
</tr>
</tbody>
</table>

Distribution of mutations in PC genes in 814 individuals with genetically confirmed PC.

We are grateful for all of you who have joined the Registry. If you have family members with PC who have not registered with PC Project, please encourage them to do so. We are preparing for studies and clinical trials and the most attractive thing to researchers and industry leaders looking at PC and PC Project is the strength of our Registry.

The graphs and additional information are posted at pachyonychia.org/pc-data/
Thank you for participating in PC Awareness Month 2018! No event or effort is too big or too small. Here is a list of some of the thoughtful and clever events we know about:

**Birthday Fundraisers:** Nearly $700 was raised by PCers or their family members who had birthdays in June. They used the Facebook Birthday Fundraiser to raise awareness, funds and to celebrate through social media. Happy Birthday to each person and thank you for sharing your special day with us!

Holly Jones [facebook.com/donate/1986751198014686/](facebook.com/donate/1986751198014686/)
Kimberly Noemi [facebook.com/donate/185289528839072/](facebook.com/donate/185289528839072/)
Jennifer Rocha [facebook.com/donate/233470324084469/](facebook.com/donate/233470324084469/)

**Ice Cream Social:** The Block family held an ice cream social for PC Awareness month at the end of a bike riding event. This was a clever idea – a way to enjoy ice cream while raising PC awareness and receiving $650 in donations!

**Le Coeur Au Pied:** The wonderful French Group holds many fundraising and awareness events throughout the year. Above is a photo of Eric, Stephanie, Sylvie, Marie Jose and Valerie (taking photo) in Paris for the national annual day of Solhand.

**Social Media Posts:** PC patients and family members shared PC Project posts and videos or posted comments about PC in order to spread awareness to their own social media networks.

**Individual Donations:** Many generous individuals made a monetary donation to advance the cause of PC patients.

**Firewalk for PC:** Julie Peconi and Tom Baker along with their family and friends will be holding their second “Firewalk for PC” on September 7, 2018. “The fire walk is a technique for turning fear into power.” See more details at [facebook.com/events/1635709006536593/](facebook.com/events/1635709006536593/)

**In Memory of Cameron Baker:** Nora Baker created a GoFundMe page in memory of PCer Cameron Baker who passed away earlier this year. Nora raised nearly $700 for PC. You can read her story at [gofundme.com/in-memory-of-cameron-baker](gofundme.com/in-memory-of-cameron-baker)

**The McLean Family** specifically Bob, Redmond & Rosemary raised £750 for PC over the past year through various ways such as collecting old car batteries, scrap metal for recycling, afternoon teas and craft sales, then donated the money received.

If you have awareness activities or events you’d like to share for the next NewsBrief, please contact us at [info@pachyonychia.org](mailto:info@pachyonychia.org)
PC LOVE BUILDERS: MONTHLY SUSTAINING DONORS

The 6.20 Campaign was a huge success! We are extremely grateful to report that during the June 6, 2018 PC Project Giving Challenge 66 people donated to PC Project. We welcomed 53 monthly sustaining donors to the PC LOVE BUILDERS family and received 13 one-time donations!

An anonymous donor has agreed to match every dollar AND make an additional gift of $5,000 to PC Project. With the match and with the current monthly pledges, after a year our grand total will be over $55,000. THANK YOU!

Every donation adds up to sustain PC Project. At this time, PC Project is completely dependent upon individual donors. Please know that everything you give goes directly to this important cause of finding effective treatments to relieve the suffering of PC patients.

EMPLOYEE MATCH

Employee matching gift programs are corporate giving programs in which a company matches donations made by the employees to eligible nonprofit organizations. This is a fantastic way to double the funds and raise PC awareness. Requesting a matching gift is normally a five-minute process which must be initiated by the donor. You can typically do this by filling out and submitting a paper match form provided by your employer or through an electronic submission process.

Here is a list of some companies that offer Employee Matches. 1800runaway.org/wp-content/uploads/2016/08/corporate-match-directory.pdf

Please check with you employer to see if and how it matches charitable donations. Some of our PCers are already taking advantage of their employer’s matching giving program.

PATHWAY TO CLINICAL TRIALS OF TREATMENTS FOR PC

We are pleased to report that your efforts at the Eternally-led Patient Focused Drug Development meeting with FDA officials on April 6, 2018 are successfully paving the way for clinical trials of treatments for PC. In working with our various industry partners, we’ve been pleased to hear that FDA officials have indeed heard your voices and are working collaboratively to change the trajectory of no FDA approved treatments for PC. They know PC patients need an effective treatment and they need it now. Your efforts have already advanced these potential treatments towards clinical studies.

In addition, your efforts to the FDA have distinguished you in the minds of these officials. Your words, your voices through polling and testimonials, your hand-written thank you cards and pictures are absolutely making a difference! This is the collaborative PC community effort that sets our PC family apart from all other groups.

We will keep everyone posted on upcoming clinical studies...more to follow in the coming weeks and months!

RARE ON THE ROAD

PC Project participated June 30, 2018 in the “Rare on the Road” Rare Disease Leadership Tour. The meeting was sponsored by Global Genes and EveryLife Foundation for Rare Diseases. This was a day full of education and inspiration about navigating the world of rare diseases.
2018 LONDON PATIENT SUPPORT MEETING


We have 62 attendees registered for the patient support meeting in London including 36 PC patients. Meeting information and registration is available at pachyonychia.org/2018-london-england-patient-support-meeting/. Please register early.

WHY ATTEND A PATIENT SUPPORT MEETING
By Julie Peconi, PC Advocate

I remember the day my husband Tom, who has PC (K16), discovered there may be an official diagnosis for his callused, painful feet. He had been searching online trying to find answers and stumbled upon an academic publication by Frances Smith, PC Project's Chief Geneticist. Like so many others with PC, throughout his life Tom had been told his calluses were a result of palmoplantar keratoderma, which didn't identify the underlying condition causing the hard skin. He tried many different treatments from Chinese medicine to acupuncture to salicylic acid. At no point had any specialist mentioned Pachyonychia Congenita or that what he had was a genetic condition. You can imagine our hope when we got in touch with the PC Project and he was diagnosed properly!

It was not long afterwards that we, along with our two sons (one of whom inherited PC) attended our first Patient Support Meeting in Edinburgh and that hope continued to grow. We were nervous about what to expect but our nerves soon dissolved when we were so warmly welcomed by Holly Evans, Frances Smith and Irwin McLean. These were people who genuinely understood and cared about the pain that Tom and my son experienced daily.

We met others with PC. It was amazing how everyone walked in the same careful way! (I've now learned this is called the PC shuffle!!). Tom was able to share his tips for management while learning from others, and our son met other children with PC. For me however, what sticks in my mind are the talks and presentations. I was blown away by the commitment and dedication of the PC Project staff, the researchers, the scientists and those suffering with PC in putting themselves forward for research in order to find a cure.

With PC being so rare, it's easy to feel quite alone with what we're going through but attending the Patient Support Meetings made me realize that we are part of a wider community - a community that understands what it means to have PC, that feels our pain, but one that is also working hard to make life easier for all of us who suffer as result of PC!

I would encourage anyone who is considering attending the upcoming meeting in London to go for it. These meetings really are a great opportunity, both to connect with others and to learn about what's happening in the wider PC community.
2018 London Patient Support Meeting (PSM)

As a result of the work of these PCers, PC Project will soon be sending a survey to the larger PC patient population to further refine these efforts. Please watch for an email in the coming days with more information.

Grant Applications for PC Project

PC Project has partnered with The Rayvan Group, www.therayvangroup.com to research and apply for grant money to help fund our programs like the Patient Support Meetings and International PC Research Registry (IPCRR). Grant applications also serve to build awareness for PC Project. We plan to apply for at least two grants per month for the next year. Two grants have been already submitted in July.

Networking makes grant awards more successful. If you, or someone you know, has contacts with a grant-giving foundation or company, please contact us at info@pachyonychia.org.

Learning from PCers

“How many of you have to sleep with your feet outside of the sheets?”

This picture and question was recently posted on the private PC Patient Chat Facebook group. Twenty-nine PCers commented that they sleep with their feet outside of the sheets and often with their feet hanging off the side of the bed so their feet don’t touch anything at all.

For those who aren’t on Facebook, here are some of the responses to that question:

“Me always. Otherwise the feet start sweating and the next day morning pain increases significantly.”

Pathway to Clinical Trials

The Value of the Patient Voice

PC patients are effectively using their voices to give valuable input in order to pave the way for clinical trials.

Last week, five PC patients participated in a special focus group aimed at helping to draft options for patient reported outcome measures to use in clinical trials. Special thanks to these patients for donating their time and expertise in this informal discussion to better understand how PCers describe their physical activity levels and how they might differentiate between the various levels.


If you have a specific topic or questions you would like discussed at the PSM, please email your inquiry to info@pachyonychia.org. We welcome your input!
“Always off the bed and outside of the sheets. The slightest friction hurts me or at best annoys me.”

“Wow it feels good to know I’m not the only one.”

“Yes, on my belly with my toes just over the edge of the bottom or side. If my feet are somehow not over the edge I’ll wake up because of the burning.”

We are learning that while PC can affect each person differently, we have many commonalities in our ways of coping!

**SOCIETY FOR PEDIATRIC DERMATOLOGY MEETING**

PC Project attended and participated with the Society for Pediatric Dermatology (SPD) at the annual meeting July 12-14, 2018 at Lake Tahoe. As the only patient advocacy group to be invited, this was a not-to-be-missed opportunity for PC Project to network and raise awareness among the 430 attendees. The goal of PC Project for this conference was for every single participant to learn about PC and PC Project, to join the IPCC (International PC Consortium) and to encourage every dermatologist to invite their PC patients to join the International PC Research Registry (IPCRR).

In addition to brochures, PC Project handed out small treats with poems to educate researchers and physicians about the painful nature of PC and to entice attendees to visit the PC Project table that was located directly outside of the main presentation room. Thanks to Nate Schwartz, a PC patient, for his limerick-writing abilities. The treats and the poems were enthusiastically received.

During the “Hurwitz Lecture—Lessons Learned from Great Thinkers,” Dr. Eulalia Baselga, the director of the Pediatric Dermatology unit at Hospital de la Santa Creu in Spain, who has referred patients to PC Project and who attended the 2016 Spain and 2016 Edinburgh Patient Support Meetings, shared her positive experiences from attending PC Patient Support Meetings. Dr. Baselga told the group how she learned about current research at the patient meeting and how the meeting helped her own PC patient. Dr. Baselga encouraged doctors to attend patient support meetings with their patients. In addition, she gave an enthusiastic shout-out to PC Project!

Dr. Tracy Funk, Assistant Professor, Dermatology and Pediatrics at Oregon Health & Science University, who works with Dr. Sancy Leachman who was influential in PC Project’s creation and a founding member of the International PC Consorium (IPCC), gave an informative 30-minute presentation called, “Update on Pachyonychia Congenita.” Dr. Funk also encouraged scientists and clinicians to learn more from PC Project.

Because there are many rare diseases, to have PC and PC Project talked about twice in one conference was extraordinary. PC Project met new researchers, physicians and medical students, and reunited with IPCC members and friends such as Anna Bruckner, Amy Paller and Joyce Teng. Thanks to the Society for Pediatric Dermatology for the invitation to share their annual conference with PC Project.
2018 LONDON PATIENT SUPPORT MEETING

Only a month and a half left until the London Patient Support Meeting! Already over 60 PC patients have registered. If you are interested in attending, please register right away. Registration will close October 4.

pachyonychia.org/2018-london-england-patient-support-meeting/.

PC MAILBOX—ADVICE FOR PARENTS

Parents of a newly diagnosed 11-year old girl with a spontaneous K6A mutation reached out to PC Project seeking support. Several PC Patient Advocates responded with informative, helpful responses. The following advice is from one of these advocates Katri-Anna, a PCer and a PC parent:

“I’m sure it is difficult for a parent of a spontaneous PCer to find ways to help their child. I have an 11-year-old girl with inherited PC-K16. We have a different mutation, but I hope I can be of some help.

I have bought my daughter the most comfortable shoes I have found, and seamless socks. I encourage her to use talcum powder to avoid sweating, and if necessary, Compeed plasters. She trims her own feet with a sharp knife and Knipex cutters little by little a few times a week. I have found that too much trimming causes me more pain – I let my callouses grow fairly much as they also protect the foot to a certain point. If I mess with my nails a lot, I will certainly manage to cause trauma and more pain also. Too much soaking is not good for my feet either. We are all different, of course. Avoid standing – it is easier said than done, I know. It is really important for a person of any age to remain active, and I am convinced being in good condition and being active also helps in managing the pain. Since my daughter was very young, I have tried to guide her towards activities that do not require being on her feet. She does cycling, horseback riding and swimming.

Only on special occasions would she take painkillers (ibuprofen). She and I both feel that with this condition, we have to find ways to avoid situations that cause pain, because it is not good for one’s body to take painkillers for years and years. With the help of painkillers we may also end up walking too much and will suffer the consequences later.

Having said that, she is 11 and refuses to use a wheelchair in public because “it's embarrassing”. It is her choice, of course. She sometimes uses it at home. We also got her a good bike she can use outdoors - we live in the country, so this is easy. I grew up in the city and just the walk to the bus stop was often too much. We drop her off at the entrance of wherever she is going and let her stay home or in the car if she feels like it.

We have told the school about the condition and they know she should go and take a seat if gym classes, for example, are too painful for her. But again, a young lady does not want to stand out, she wants to participate in most things like all the others. I think in time she will become more selfish and stop worrying about what others think. At that point, a wheelchair and other help will interest her more.

Also, if you have not yet been to a Patient Support Meeting, but would be able to come, it is my warmest recommendation that you come with your daughter. Remember that you are not alone. Lots of love, Katri-Anna Lehto.”
This past month, PC Patients have enthusiastically responded to three extremely important surveys designed by the bio-pharmaceutical company partnering with PC Project for a major clinical trial. Because of the time and service given by PC patients, successful endpoints for the trial are being developed.

Three days after the first Pathways to Clinical Trials Survey was sent to PC patients, PC Project had received over 100 responses. The bio-pharmaceutical company then worked with PC Project to review and analyze the answers. Because of the thoughtful and valuable feedback provided by PC patients, the company then fine-tuned the first survey. The second Pathways to Clinical Trials Survey was sent and three days later PC Project once again had over 100 responses to share with the company.

Finally, again based on what was learned from the responses from the PC patients, a third survey was given and once again PC patients responded like the champions they are.

The value of patient feedback cannot be overstated. At PC Project, the staff is small and consists of only one person with PC. However, the PC community is large and to have such an active and responsive community share their insights about their disease is an invaluable collaboration to industry partners. PC patients continue to demonstrate again and again that they will do whatever it takes to find an effective treatment for PC. The excitement continues to build as PC Project prepares to participate in the largest clinical trial ever targeted specifically for PC patients.

Please see the results of some of the surveys below and on the following page.
On an average day, how would you answer this question: Over the past 24 hours, please mark the response which best describes the difficulty you experienced, on average, in carrying out your daily activities involving you being on your feet — such as walking, standing, keeping your balance, etc.

- 1% Unable to do these activities
- 19% Much difficulty
- 13% Mild difficulty
- 6% No difficulty
- 43% Moderate difficulty

In a given week, how much variation would you expect in your response to the question and scale on the left?

- 16% High variation (+/- 3+ levels)
- 9% No variation (+/-0 levels)
- 44% Moderate variation (+/- 2 levels)
- 32% Limited variation (+/- 1 levels)
- 50% Moderate variation (+/- 2 levels)

Thinking about the answer provided regarding the difficulty you experience on an average day in carrying out daily activities that involved being on your feet (walking, standing, keeping your balance, etc.). Which responses best describes the level of difficulty you experienced?

PC Survey 2

On an average day, how would you answer this question: Please think about any difficulty you may have had over the past 24 hours in carrying out daily activities that involved being on your feet (walking, standing, keeping your balance, etc.).

- 1% Extreme difficulty
- 5% 4-point improvement
- 4% N/A
- 17% 2-point improvement
- 18% 5-point improvement
- 14% 1-point improvement
- 9% 3-point improvement
- 5% 2-point improvement
- 10% 0-point improvement
- 4% 1-point improvement

In a given week, how much variation would you expect in your response to the question and scale on the left?

- 20% High variation (+/- 3+ levels)
- 9% No variation (+/-0 levels)
- 21% Limited variation (+/- 1 levels)
- 50% Moderate variation (+/- 2 levels)

Would a 1-point improvement on the scale be meaningful to you in improving your quality of life in living with PC? 58% Yes and 42% No

Would a 2-point improvement on the scale be meaningful to you in improving your quality of life in living with PC? 77% Yes and 23% No

Those who answered ‘No’ to the questions on the left, please select the minimal level of improvement that would be meaningful to improving your quality of life in living with PC. 23% 3-point improvement; 23% 4-point improvement; 19% 5-point improvement; 15% 7-point improvement and 19% N/A a 1 or 2-point improvement would be meaningful.
On Friday September 7, here in Swansea, Wales, UK we held our second Firewalk Challenge and BBQ social evening for PC! As there are only four people genetically confirmed to have PC in Wales, and only 2 of them in our community, Tom (my husband) and Timothy (my son), we wanted to raise awareness and funds for PC by running an event that is entirely relevant to the condition: walking on something that is perceived to be painful! Even though walking on fire doesn’t hurt, we wanted those attending to understand some of the challenges of living with PC. We had over 30 people sign up this year, from some of the teachers at our son’s school, to football coaches, to colleagues from work and friends.

This year we also had so many children wanting to take part, so we added on a Lego Walk to the event. This involved walking over two metres of thousands of Lego bricks - and this as you can imagine was uncomfortable!! Despite this we had over 50 children take part in the Lego Challenge!! Although Timothy didn’t take part in this because of his PC, he was able to watch his friends taking part in a fundraising challenge to raise money for research into his condition!

We still have sponsorships coming in but it looks we have raised over £3,500 for Pachyonychia Congenita and we were in the top 5% of fundraisers for the month of September on the online donation service Justgiving! We also raised the profile of PC in our community and received such fabulous support. All in all, the evening was a great success with everyone enjoying whilst raising awareness and crucial funds for Pachyonychia Congenita (PC) Project Europe.

It’s not too late to sponsor the brave walkers! If you would like to give, please go to: justgiving.com/fundraising/firewalkforpc2018
**FACEBOOK BIRTHDAY FUNDRAISERS**

Thank you to those who raised money for PC by creating a Facebook Fundraiser for their birthdays: Here are the descriptions they gave for their fundraisers:

**Amy Alphin** - For my birthday this year, I'm asking for donations to Pachyonychia Congenita Project. I've chosen this nonprofit because their mission means a lot to me, and I hope you'll consider contributing as a way to celebrate with me. Every little bit will help me reach my goal.

**Briannan Buchta** - For my birthday this year, I'm asking for donations to Pachyonychia Congenita Project. I've chosen this nonprofit because their mission means a lot to me, being that myself and family members suffer from this. I hope you'll consider contributing as a way to celebrate with me. We have about a month before my birthday and every little bit will help me reach my goal. Even if you can't donate please share to help spread awareness of PC so one day if you come across one of these rare individuals we won’t feel so left out - Searching for a Cure.

**Isabella Mortier** - For those who do not know what Pachyonychia Congenita is, it is a very rare condition that primarily affects the nails and skin. It may be seen on several parts of the body including palms of your hands, soles of your feet, fingernails/toenails, your tongue, and your cheeks. This condition causes the affected area to grow thick, abnormally-shaped callus which often cracks on the soles of the feet making it extremely painful and nearly impossible to walk. My affected family members continue to struggle with this side affect every day of their life. It is very hard to see anyone, let alone my own blood, continue to have to fight their way through life and not experience it to the fullest, because of this condition. Dermatologists can only do so much to help with the little bit of understanding that they have of this condition, being that it is so rare. Conventions are being held around the world to unite people who have been diagnosed with Pachyonychia Congenita in attempt to create a better understanding of the different types of the condition, let people know they are not alone, and work together to make a change. With the help of the PC Project and donations, we may one day discover a cure for future generations and find relief for those who continue to struggle with it present-day.

Happy Birthday to all three of you and thank you for making a difference in the lives of PC Patients!

**IN LOVING MEMORY**

We express our sincere condolences to the family members of Silvia Meister at the passing of their beloved mother and grandmother. Silvia's young granddaughter, Ela Kislal, has PC. Silvia wished for Ela's pain to end. For this purpose, donations were given to PC Project in Silvia's honor. Our greatest hope is that Silvia's wish for her granddaughter will be realized sooner than later.
Living with PC: A Patient’s Experience

Hello, my name is Jack Padovano. I’m 56, live in Phoenix Arizona and my genetic mutation is on keratin gene 16.

My PC shows up with thick calluses on over 50% of the bottom of both feet, cracks and occasional blisters along the middle and sides of most calluses, and thickened nails on 100% of my fingers and toes.

Like most people, I have a bank account. But not the kind you’re probably thinking. This one isn’t filled with money, but instead, it’s filled with the number of steps I can physically walk each day before tremendous pain sets in.

Just like a checking account filled with money, I spend very wisely, or try my best. Each withdrawal, or step I take is mentally recorded and physically felt, right down to my bones. Overnight while I sleep, the bank account is refilled before I wake up. The amount of the refill varies. If I overdrew from the account the day before by walking too much, I have fewer steps in the account. If I got a good night’s rest, and monitored my walking, the account is completely filled up.

On my best day, I can walk down a long city block without thinking once about the pain. On my worst day, I simply refuse to walk. Period. Unfortunately, there are few best days. If I’m lucky, I get one per month. Most days I think about the pain with each and every step, including standing in place.

PC hurts both physically and emotionally. PC pain feels like someone is sticking pins and needles in the bottom of my feet. It’s a deep ache that cuts all the way to the bone. I treat the pain with hot water soaks, cold-water soaks, elevating my feet, rubbing creams, massage, Vaseline baths, Advil, and a lot biting, mostly under my breath. I treat my PC by pairing down the calluses once a week trying to navigate those pesky blood vessels and nerve endings that get cut and inflamed in the process. Nothing really works. The pain is constant and often makes me grouchy, sometimes to the point of lashing out to the people I love, work with, and even total strangers. I even think it contributes to my struggle with depression.

PC makes my fingernails ugly, so ugly that growing up other kids made fun of me. PC makes me walk “weird”, something we PCer’s affectionately call the “PC Walk”. But, kids being kids, they didn’t see any humor or have any compassion for my walk. I was just different and that made me a target for bullying. The really mean kids took to stomping on my feet, so hard I would fall to the ground and writher in pain. And as those with PC can attest, the last thing we PCer’s need is more trauma to the feet.

PC also significantly impacted my parents. After my diagnosis at three years old my parents had a name for my condition. But that’s all they had…no treatment, no answer why, or especially, no cure. In fact, they were told the condition would most likely worsen to the point where I couldn’t walk. My mother was certain it was her fault. She would often say, maybe if I smoked less, ate differently, didn’t take aspirin, etc. etc. Today, we know none of that mattered. I’m a spontaneous case, meaning that my PC is not inherited.

As an adult, the bullying has stopped but is replaced by questions, mostly thoughtful and kind, but sometimes not. Questions I really don’t like to answer because the answers are never simple one-word answers.

PC changed my life, or better said, it IS my life. And because of its’ enormous power, I respect it and have learned to live side-by-side with it. It’s made me a stronger person and taught me how to be courageous by exposing it to doctors, strangers, and people who love me…not an easy task. It’s taught me how to stand up to bullies who have no interest in learning anything about PC except as way to call me out as different. Good life lessons.

Recently, I learned that the average person walks 10,000 steps per day, or 5 miles. I’m envious. That’s a big bank account. For me, I’m lucky to get a quarter mile, or 250 steps under my belt before the pain sets in. So, while my account may not be as rich as yours, I treasure every step I take.

Future forward, I worry that my condition will worsen as I get older. I know my pain has gotten progressively worse every year, particularly in the last 20 years or so. I can see it in my walk and feel it in my bones. My wish is simple. I want to stand and walk without excruciating pain. I hope and pray that’s not too much to ask.
PC Project and Palvella have been working very closely over the last several months to prepare for the imminent initiation of the Phase 2/3 study evaluating PTX-022 (novel, high-strength topical rapamycin, optimized for dermal targeting) for the treatment of PC. Significant progress has been made in recent months, the most important of which is: i) The selection of a final formulation to be evaluated in the Phase 2/3 clinical study; ii) Decisions around which efficacy endpoints to prioritize for the study. Notably, the final formulation has been architected specifically for the PC patient population, and it was selected after a rigorous testing program that included more than 20 prototype formulations evaluated by Palvella and their formulation partner MedPharm, a leading global topical formulation development company based in London, UK.

With regards to efficacy endpoints, as many of you are aware, hundreds of PC patients have willingly participated in qualitative and quantitative surveys to help narrow to specific efficacy endpoints. PC is truly a condition where the patients are the experts in the disease, and hence, the 'voice of the patient' will play a central role in the Phase 2/3 study. In addition to the tremendous response from PC patients, a Global Pachyonychia Congenita Working Group comprised of clinicians with expertise in PC as well as rare disease regulatory experts was assembled more than 16 months ago, and that group has worked diligently to iteratively narrow to efficacy endpoints for the study. Overall, the meaningful progress made these last few months is a testament to the tremendous collaboration that defines PC Project and the PC community worldwide. More frequent updates on the upcoming Phase 2/3 clinical study will follow in the coming weeks.

FDA has granted Fast Track Designation to Palvella for the advancement of PTX-022 (novel, high-strength rapamycin topical formulation, optimized for dermal targeting) for the treatment of PC. This milestone has its roots in the transformation that occurred at the FDA on April 6, 2018. The FDA’s views on, and understanding of, PC forever changed that day as the team at PC Project led PC patients from all across the country in passionately and courageously sharing their personal stories of living with PC with 42 FDA officials.

Further instrumental to this milestone has been the unwavering clinical and scientific support from all of you, over many years, who have thoughtfully guided the TransDerm and Palvella teams in navigating through the significant, but surmountable, challenges that present when developing a therapy for a serious rare disease. A special thanks to the dynamic duo of James Valentine and Frank Sasinowski who have worked tirelessly to engage the senior ranks of the FDA in an effort to ensure that the voices of PC patients are heard at the FDA.

While we have a long ways to go to deliver on our shared objective of introducing safe, effective, and FDA approved treatments for PC, this recognition by the FDA nevertheless sets the stage for the acceleration of the PTX-022 program and other therapies with potential to significantly improve the lives of individuals with PC. Thanks to all of you for your continued personal and professional friendship to me and the Palvella team every step of the way.

Celebrated on the Tuesday following the US Thanksgiving holiday and the shopping events of Black Friday and Cyber Monday, Giving Tuesday is our day to show gratitude through giving to those causes we care about. YOU are the cause we care about.

Please join us November 27, 2018 for our largest fundraising event of the year. Watch for emails and our PC social media pages during the month of November for more information or donate now.

Every dollar donated furthers our mission to help those suffering from PC.

50TH ANNIVERSARY DONATIONS FOR PC

Happy 50th Wedding Anniversary to Floris and Martheke van der Laan, the Grandparents of Ian van der Laan-Kokx, a PC Patient.

Ian’s mother, Elise wrote,

“My parents-in-law have been married for 50 years on December 12. On September 22 they were given a party to celebrate this beautiful event. Because my parents-in-law have everything they could wish for, they decided to raise money for Pachyonychia.

They came up with this idea because their only grandchild, Ian, suffers from this condition and they see that he has to live with the limitations of this disease.

My in-laws made a letter explaining what the association does and what it means for Ian to deal with Pachyonychia. Of course, most of the people at the party knew Ian already, but despite that we were gratefully moved by the generosity of the people. There were about 40 people at the party and together they gave an amount of 2,500 euros.”

The van der Laan family have been PC fundraising stars for a long time. Now we know it runs in the family. Thank you and congratulations!
LONDON PATIENT SUPPORT MEETING

On October 19-21, 2018, at the Park Inn by Radisson London Heathrow, 140 Pachyonychia Congenita patients, caregivers, scientists, clinicians and representatives from industry partners, Palvella, gathered to share knowledge, encouragement and love. Thank you to the MANY individuals who made this PC Patient Support Meeting a success, including our local organizers, Kate Fairbrother and Julie Peconi and additional patient advocates, Tom Baker, Kieren Eyles, Philip Gard, Melanie Hettler, Pamela Ibanez Triguero and Katri-Anna Lehto.

This meeting was sponsored by PC Project, Palvella Therapeutics and by a grant from Novartis Pharmaceuticals UK Limited.

We asked two of the attendees to write a brief report of the meeting.

PSM Report By Gary Millis

This year I attended the PC meeting with my wife Sally, along with my eldest daughter Laura and grandson Cian, who both suffer from PC. It was wonderful to see so many familiar faces and for this support meeting, as many new faces. It was so nice to hear everyone saying how good it was to all see each other again and exchange our latest news and realize how much we had developed a very close friendship with so many other PCers.

We all really feel that these meetings are so beneficial to everyone, especially to be close to others that feel and experience the same thoughts, difficulties and constant obstacles on a day to day basis. It’s great to talk to experts and give them an understanding of how we all feel and it’s also good to talk to each other to discuss what we think works for us. Knowing that there may someday be a cure gives us all hope for the future.

The presentations this year by the physicians, scientists, discussion leaders and presenters, were as always really interesting and informative and we look forward to seeing you all again at a future PC meeting.

I would just like to add that Cian was a bit apprehensive about attending the meeting, but after only one session in the children’s programme couldn’t stop talking about the amazing experiments he’d done and was really enthusiastic about attending the afternoon session. By the end of the day he’d made a lot of new friends! Needless to say he’s looking forward to seeing them again.

I have attached a photo of Cian with Braham, Roger and Wes, who are Cian’s hero’s! (and ours too), although I think they and Alain, may regret sitting on our table? At the raffle they very kindly handed all their prizes over to Cian, but he did give them the chocolates in return, but only the ones with nuts in, as he has a nut allergy!

Well done everyone who makes this event possible, we are so grateful to you all.
The PC Patient Support Meeting was fantastic. As a 26 year old with a spontaneous mutation of the K16 gene, it has taken 26 years and the amazing efforts of PC Project team for me finally to meet another person who shares the PC condition. The experience was emotional and wonderful for me and I recommend to anyone that hasn’t yet attended to go.

The scientists are dedicated, interesting and engaging individuals who truly care about your pain. Having a new language to explain to friends and colleagues about the physical manifestations of the disease as well as the chronic pain experienced is something I wish I’d had my entire life.

For example, when my parents/teachers/friends would tell me to stop ‘dragging my feet’, I’ll now say actually I have an ultra-rare autosomal dominant disorder called pachyonychia congenita brought on by a spontaneous mutation of my K16 Keratin gene which affects 1955 people across the world and is characterized by chronic pain in my feet. So, I’ll walk however I want, thanks (or words to that effect!).

I met some incredible people—big shout out to Jan, Kate, Frances and Holly and everyone else involved in organizing and also lots of friends. One of the lovely ladies with PC who also lives in London cooked me a vegan dinner last week, I met her beautiful cat Bella and we went for a cycle in the country. I know she’ll be a friend for life and that’s all down to the PC Project.

Ultimately whilst PC is debilitating and frustrating at times, I often feel blessed for having the disease. It has taught me strength and empathy. It has given me the power to look past the surface of someone’s physical appearance, words or actions and understand that often people are masking a pain that others can’t possible contemplate so to treat every human with kindness, dignity and respect. This has made me a better business leader, partner and friend. It drives my interactions with others and often means my relationships are more meaningful. So thank you PC and thank you PC Project. I can’t wait for the next one.

PCers can do amazing things! We strengthen each other and help reach new heights. This is one of Siobhan’s adventures.
2018 LONDON PSM PHOTO COLLAGE


**Giving Tuesday Success - Thanks to You!**

Because of you, and with the help of over 350 donors, we surpassed our Giving Tuesday goal and raised more than $80,000 for PC patients and research. This amount was matched by a generous donor and all contributions will continue to be matched for the rest of 2018. Your support is proof that combined efforts, big or small, equal impressive results.

On Giving Tuesday, as we saw your donations, along with your emails, personal fundraisers and shared social media posts, we were moved to tears by your goodness, generosity and love. Please accept our sincerest thanks. And please share our gratitude with any of your friends and family members who support PC through you.

As we continue to work together, we expect our hopes to be realized, and that one day all PC patients will enjoy pain free skin.

This was the meme most shared on social media on Giving Tuesday:

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**Facebook Birthday Fundraisers**

In addition to the many Giving Tuesday Facebook fundraisers, the following people created PC Facebook fundraisers to celebrate their birthdays:

- **Maria Ahlander**: In connection with my birthday this year, I ask for contributions to the Pachyonychia Congenita Project. I've chosen this organization because their work means a lot to me, and your donation is a way to celebrate with me.

- **Michelle Evans**: PC Project is Amazing!! They are near and dear to me because they completely covered Allie's genetic testing at no cost to us. This gave us valuable information that I am so grateful for!! They are doing further research on this very rare condition, trying to find a cure, or at least treatments to help. They are also a great resource for information on this condition and have the most recent research studies. They even have support groups since this is a lifelong condition. They can only continue to help other children/people like Allie however, through support/donations.

- **Buff Farrow**: Want to join me in supporting a good cause? This #GivingTuesday (which also happens to be my birthday!) I'm raising money for Pachyonychia Congenita Project and your contribution will make an impact, whether you donate $5 or $500. Every little bit helps.

- **Steve Pereira**: For my birthday this year, I am asking for donations to Pachyonychia Congenita Project. I have chosen this non-profit because their mission means a lot to me.

- **Stephen Webb**: For my birthday this year, I'm asking for donations to Pachyonychia Congenita Project. I hope that you'll consider contributing as a way of celebrating with me.

Happy Birthday to each one of you! And thank you for thinking of PC patients on your special day!
PC patient Featured on SkinSerious

One of our very own PC patients, Buff Farrow, was recently featured in the SkinSerious program of American Academy of Dermatology Association. SkinSerious highlights positive relationships between patients and their dermatologists. Because PC is ultra-rare, having a doctor who is willing to learn about and understand PC is valuable for any patient. Thanks to Buff for representing PC and PC Project so well. Below is a copy of the published article. Click here to see online version.

Buff Farrow’s Story:

“I live with a rare genetic condition called pachyonychia congenita, which makes my toenails and fingernails grow abnormally thick and causes painful callouses and blisters on my feet. I need to limit my walking, because taking even an extra 20 steps a day causes increased pain and can cause blisters to form.

I was diagnosed at age 16 by Dr. Jack Lesher, who also diagnosed my father, then in his 40s, with pachyonychia congenita. My son was diagnosed at birth. For almost 30 years now, our family has been treated by our dermatologists, Dr. Lesher and Dr. Loretta Davis.

While there is no cure for pachyonychia congenita, Drs. Davis and Lesher have helped us manage it as best we can, from trying new treatments to helping us get handicapped parking and other special accommodations. Throughout it all, they’ve always put our family first, treating us kindly and with thoroughness. They’ve also helped build awareness of pachyonychia congenita and invited us to present as case studies at dermatology conferences, so other dermatologists can better recognize and manage pachyonychia congenita in their patients. Growing up with such a rare condition is challenging. I went through childhood without ever meeting someone else with pachyonychia congenita, other than my father, which really takes a toll — you feel different and out of place. The physical effects can be debilitating, but the psychological aspects of it are just as bad. Fortunately, we eventually connected with the Pachyonychia Congenita (PC) Project, a support group for people with pachyonychia congenita.

Since connecting with the PC Project, our family has been looped into an incredible community of people like us who can understand and empathize with our lives, as well as researchers who are trying hard to find better treatments and ultimately a cure. Pachyonychia congenita is rare and challenging, but with Dr. Davis and Dr. Lesher, we’ve never felt overlooked.”

The Dermatologist’s Perspective

“It is an honor to treat Buff, her father and her son. The doctor-patient relationship is always special, but it is especially rewarding to care for multiple generations of such an inspiring family. After 25 years of caring for them, I find it is almost like caring for my own family. While there is no cure for pachyonychia congenita, we have worked as a team to help manage their condition and limit its effect on their daily lives. I am proud of how far we have come. Buff and her family are very generous with sharing their story. Our dermatology division has learned just as much from Buff’s family as they have from us. They have helped an entire generation of doctors improve their care of pachyonychia congenita.”

– Loretta Davis, MD, Medical College of Georgia, Augusta University

2019 Patient Support Meeting—USA

We are beginning to plan for the 2019 PSM. Watch for more information early next year regarding the date and location.
‘COOL’ SHOES

At the 2018 London Patient Support Meeting, Alexia Chatzitheodorou and Federica Chiti, physics students at the University of Dundee in collaboration with Dr. Michael Conneely and Dr. Robyn Hickerson, presented their research on cooling footwear technologies. Because many PCers hurt more when their feet are hot, the thought is these ‘cool’ shoes might help relieve pain.

The physics students looked at three different approaches to keep feet cool: The Peltier Effect (electronic cooling system), motion powered air circulation, and phase change materials. The phase change cold packs that were shown at the meeting can be found here: https://glaciertek.com/pare-chillydog-cooling-pack-set/

In the new year, the Dundee lab hopes to continue to incorporate this type of phase change material into an insole that can be put into a shoe. They are also trying to contact other companies that microencapsulate these materials so that they are more malleable and comfortable.

The shoes in the presentation that had the motion powered air circulation can be found here: https://www.clarksoutlet.co.uk/p/35384010. These shoes have ActiveAir Vent technology but it is still unsure how effective it will be for cooling patients’ feet. Further tests will be done with this technology in the new year as well.

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HAPPY KWANZAA!

Merry Christmas
Happy Hanukkah
Happy Holidays