16th Annual International Pachyonychia Congenita Consortium (IPCC) Symposium
May 7-8, 2019
just prior to the SID Annual meeting in Chicago, Illinois at the Hilton Chicago

The registration fee is waived with pre-registration; sessions & meals included.

**Schedule**

**Tuesday, May 7**
- 12 pm  Lunch and welcome
- 12:30-5:00 pm  Presentations
- 6:00 pm  Meet for offsite dinner cruise

**Wednesday, May 8**
- 7:30 am  Breakfast
- 8:30-12:30 pm  Presentations
- 12:30 pm  Lunch

**Invited Speakers and Attendees**

- Alain A. Hovnanian
- Albert A. Bravo
- Alex Hinbest
- Braham Shroot
- C. David Hansen
- Christopher Bunick
- David A. Giljohann
- David Kelsell
- Dennis R. Roop
- E. Birgitte Lane
- Edel A. O’Toole
- James Rittle
- John D. Doux
- Joyce M. Teng
- Liat Samuelov
- Mark P. de Souza
- Maria Morasso
- Michael Conneely
- Michael J. Caterina
- Michael J. Polydefkis
- Minh Ho
- Pierre A. Coulombe
- Rame Yousif
- Robyn P. Hickerson
- Roger L. Kaspar
- Thomas M. Magin
- Tracy L. Funk
- Vivien Chua
- Vu Van Quang
- Wesley Kaupinen
**EDITOR’S INTRODUCTION**  
*Edel O’Toole*

Spring is here and it is nice to wake up at 5:45 am in London to brightness and a new day. In this newsletter, we put the spotlight on Dr Hansen, a University of Utah dermatologist, who has dedicated a lot of time to genetic skin disorders and PC. Maria Morasso and Olivier Duverger from the NIH explain about their interesting work on keratin variants and teeth and what that might mean for our patients. In the USA, doctors and patients are about to start a clinical trial of topical rapamycin for PC. Over here in Europe, we are waiting for Brexit, but are hopeful a clinical trial of topical rapamycin will happen here soon too. We look forward to seeing as many of you as possible at the IPCC meeting in Chicago.

**IPCC SPOTLIGHT: C. DAVID HANSEN, MD**  
*University of Utah Dermatology. PC Project Board Member and International PC Research Registry PI*

After my residency training in dermatology at the University of Michigan, I returned to the University of Utah where I joined the clinical faculty in dermatology. My first project was as a sub-investigator using 13 cis-retinoid acid (later called Accutane or Isotretinoin) for the treatment of genetic keratinizing disorders like Ichthyosis, Darier’s disease, Punctate Keratoderma and others. We did not see or treat any individuals with PC at that time. The clinical response in this group of individuals was absolutely spectacular. As long as patient’s stayed on their medication, it made a dramatic change in their skin allowing them to function much better in society. This initial success has allowed me to continue to treat and follow this group of patients for 40 years – all continue on medication and are doing well.

Following our success with this oral “retinoid” we began to study a cousin medication, now called Acitretin, in the treatment of psoriasis. This proved to be helpful in a small number of patients but not as dramatic as the initial experience with the genetic skin disorders. We also participated in the early studies of “Accutane” for the treatment of severe cystic acne – again with striking success in a condition that had been very difficult to manage.

My exposure to genetic skin diseases and the remarkable response we found in this select group of conditions encouraged my interest in genetic keratinizing disorders. In 2004, I participated in the first PC symposium involving many of the scientists with whom we continue to work with at PC Project. I was delighted at that time to meet several individuals with Pachyonychia Congenita for the first time. My hope was that we could find a similar medication that would reverse some of the challenging skin changes found in PC.

Since that symposium I have been privileged to continue to associate with PC Project, helping with over 200 patient consultations, patient support meetings and sharing consultations with incredible scientists who are pushing the frontiers of therapy in Pachyonychia Congenita. For me this has opened a vision of the potential for diagnosis and management of PC along with other genetic conditions that involve changes in the way the skin functions. One of the great accomplishments of PC Project was to establish an international registry where we could document the specific mutations in the PC genes and establish the physical findings and challenges faced with this rare skin condition. Using the data from the registry, that was willingly provided by so many PC patients, we were able to publish articles helping doctors throughout the world to better understand and manage this difficult condition. Thanks to all the patients who participated in the questionnaire and support meetings – it has allowed us to make significant strides in understanding the potential for effective treatments.
At home I am married and have raised four children, one of whom is also a dermatologist at the University of Utah. As we continue to work together, I see great potential to modify the skin changes that produce the calluses, pain and other problems found in PC. This keeps me excited about continuing my association with PC Project – I see great things coming.

**WHAT ARE EPITHELIAL KERATINS DOING IN TEETH?**

By: Dr. Olivier Duverger and Dr. Maria I. Morasso

Concurrent with epidermal differentiation, during embryogenesis there is development of ectodermal appendages such as hair and teeth. Previously, the Morasso group showed that the epithelial hair keratin K75 is expressed in developing teeth and is an essential organic component of human mineralized tooth enamel.

The group recently published their research in PLoS Genetics (2018) and JID (2019) showing that K6a, K6b, K6c, K16 and K17, epithelial keratins that are mutated in patients with Pachyonychia Congenita (PC), are also produced in developing teeth and are essential organic components of the mineralized tooth enamel.

The expression of these keratins in dental tissues had not been described before, and dental manifestations in PC, a cutaneous disorder characterized by nail dystrophy and painful palmoplantar keratoderma, had not been reported. In these studies, using human genetic and intraoral examination data, the Morasso group identified several missense polymorphisms in the genes encoding K6a, K6b and K6c that lead to increased susceptibility to cavities, which supports a function of these keratins in the resistance to tooth decay (Duverger et al., PLoS Genetics, 2018).

Interestingly, their findings on K16 and K17 suggest a distinct function for these two keratins that exhibit a different distribution in the enamel when compared to the K6 family and seem to have a role in shock absorption and protection against cracking (Duverger et al., J. Invest. Dermatol., 2019).

These results raise numerous questions about the potential functions of these keratins besides intermediate filament components and provide a novel understanding of the genetics of susceptibility to tooth decay and other enamel defects. Furthermore, besides determining novel clinical features of PC, it has broad implications for personalized prevention risk assessment and dental care for patients with PC.

**References:**


**RECENT PUBLICATIONS**


**WHY PC patients hide their PC?**

PC patients were asked the following question on a private PC Facebook chat group:

*Why do PC patients so often hide their symptoms or even the fact that they have PC?*

Katie: My daughter is only 11-years old and hides it as others don’t understand. She is always in pain but gets sick of telling people what’s wrong etc. I think it’s more to do with others not judging.

Ramon: Mostly, to avoid the long conversation: “Yes, my whole life. No, not contagious at all. Yes, it hurts. Yes, I’ve been to a doctor.” and the most annoying one: “Yes, I’ve tried that home remedy already.”

Lacey: Judgement. My whole life I’ve been judged. People thinking I’m contagious from the time I was a toddler and just wanted to play with the other kids but their parents wouldn’t let them, to even now just handing money to a cashier and getting a dirty look because my hands are different. I hide my disorder because the world is ignorant to what they don’t understand or care to even try to understand before judging.

Malgosia: Looking through my life I see a couple of reasons. First, the disease is rare and not typical, and people don’t understand what we really have. I usually say that I have problems with walking and standing. For some people it’s enough, some ask for more details, even doctors don’t want to know more. Second, usually it’s not visible, and people don’t believe that “healthy looking” people struggle. Third, shame to ask for help. I have to force myself to not overdo, and in my country, I don’t want to speak, to not have problems with work and study. It’s sad but the discrimination of disabled people, forces them to behave as normal to have a good education and job.
Tom: First, until a year ago, I thought I was the only person in the world that had this issue. I’ve seen what seems like a thousand doctors that just stared at me and shook their head. So because of that I “hid” my condition because even I didn’t understand. Second, I’ve dealt with the pain and if I told someone my feet hurt, they’d go into how they have a bunion or a blister from tight shoes etc., and tell me how to get rid of it. I’ve dealt with this so long, telling people I hurt, to me, sounded like I’m complaining...and I don’t want people to look at me like someone that just complains about everything.

Jason: I was recently bullied at work by someone who told me to go to a doctor for my “damned dirty nails”.

Laura: It’s embarrassing and ugly. I think when people see other disabilities, they’re more common so more accepted. I definitely don’t feel “pretty” or “womanly” with them. And that affects my confidence and self-esteem.

Marion: It is very hard on children - our son went through merciless teasing and bullying growing up when other kids saw his feet.

Nykole: About 8 years ago, I went to get my hair cut. The lady was combing my hair and said “You need to get this taken care of!” She threw the comb in the trash in front of me and didn’t even tell me she was not going to cut my hair. I sat in that seat for what seemed like hours with her and another stylist she called over. They were standing there glaring at me in disgust because my scalp had cysts on it. It was such an embarrassing and awkward moment.

Claire: I have the thick nails on my hands and feet and have more cysts than I’m able to count. My experience at school made me embarrassed by it. I vividly remember being called elephant girl and the constant ‘eeww’ if people saw my nails. This still pains me nearly 30 years later. This is why I hide my nails with false nails and don’t wear certain clothes as I like to have my bumps and scars covered.

Suzanne: I think it’s an instinctive need to fit in, to be accepted. We have no control over other people only control over ourselves and how we deal with differences. For me, the patient support meetings are the only place I feel accepted. I’d never experienced this before. I had 40 years of feeling I didn’t belong. I don’t think I could describe the feeling I had. I was allowed to be who I am. I have moments of “stuff it, I’m not going to hide” but then just one thoughtless comment sends me right back to that “head-down, stay-low” mindset.

BOSTON PC PATIENT SUPPORT MEETING
JUNE 20-22, 2019

All interested clinicians and scientists are welcome to attend. This is an excellent opportunity to observe and learn first-hand from the PC patient community. If interested please contact PC Project at info@pachyonychia.org or see meeting and registration details here: https://www.pachyonychia.org/2019psm/
PC Community Unites in Salt Lake City for Phase 2/3 Clinical Study Kickoff

By: Wes Kaupinen, CEO Palvella Therapeutics

Uplifting Kickoff Meeting held to discuss execution of study to evaluate Palvella Therapeutics’ Investigational Therapy for Pachyonychia Congenita

On a majestic January weekend in Salt Lake City, members of the PC community converged for the kickoff of the Phase 2/3 clinical study that PC Project and Palvella Therapeutics have been building towards together the last two years. The study plans to evaluate the use of PTX-022, a novel, targeted formulation of rapamycin developed leveraging Palvella’s QTORIN™ technology, in pachyonychia congenita. This meeting in Salt Lake City marked the culmination of many years of work by PC Project, TransDerm, and Palvella Therapeutics to advance this program towards what will be the first-ever Phase 2/3 study in pachyonychia congenita.

Inspiration and passion abounded at the meeting from the outset, beginning with Janice Schwartz captivating the audience by describing the determined efforts of PC Project to extend their love and care to PC patients worldwide while concurrently partnering with physicians and scientists to advance meaningful potential therapies to PC patients. Jan eloquently reminded all in attendance, some of whom were new to PC, that PC patients are chronically debilitated for life, live in a constant state of mobility-limiting pain, and are without a single FDA-approved therapy. Following Janice’s presentation, several physicians and scientists, including Dr. David Hansen (University of Utah), Dr. Joyce Teng (Stanford), Dr. Amy Paller (Northwestern), Dr. Sancy Leachman (Oregon Health and Sciences University), Dr. Tracy Funk (Oregon Health and Sciences University), Dr. Braham Shroot (Palvella Therapeutics), and James Valentine (Hyman, Phelps & McNamara) highlighted their work in PC, and more importantly, their underlying passion to strive towards improving the lives of PC patients.

The potential for the targeted delivery of rapamycin in pachyonychia congenita has its roots in the dedicated work of Dr. Roger Kaspar who utilized his background in gene expression to discover that the mutant keratin genes in PC could potentially be repressed through therapeutic intervention with rapamycin. Dr. Kaspar shared the story behind this original discovery more than a decade ago, as well as the work that immediately followed with Dr. Sancy Leachman conducting a study of oral rapamycin in pachyonychia congenita. Overall, while many key important themes emerged from the meeting, none were more powerful than “Love your PCer”, and meeting participants consistently offered a willingness throughout the meeting to deepen their commitment towards improving the lives of those affected by pachyonychia congenita.

If you have PC patients in your clinical practice that would be interested in participating in this future Phase 2/3 clinical study, please let us know at https://www.surveymonkey.com/s3/4854812/PCInterestSurveyPhase2-3

The clinical study plans to begin enrolling PC patients in the coming weeks, with clinical trial sites currently planned across the United States.
MESSAGE FROM THE EDITOR

Summer is here and the sun is shining in London. Despite the uncertainty around Brexit, there is a ‘feel good’ factor in the air as England just won the Cricket World Cup with an Irish man at the helm and Wimbledon has been as enthralling as ever.

In this newsletter, we congratulate Dr Michael Polydefkis on receiving the 10th Donlin M. Long Pain Service Award for his work on pain. His beautiful work illustrating how the skin can be used to look at various types of peripheral neuropathy is admired worldwide. We also provide updates on the recent IPCC meeting in Chicago, the Boston PC Patient Support meeting and the VALO study.

IPCC SPOTLIGHT: 
MICHAEL POLYDEFKIS, MD
Johns Hopkins Hospital, PC Project Medical & Scientific Advisory Board Member

On April 18th 2019, Dr. Michael Polydefkis, Professor of Neurology, Director of the Cutaneous Nerve Lab at Johns Hopkins School of Medicine and Director of the EMG Lab at Johns Hopkins Bayview, received the 10th Donlin M. Long Pain Service Award. This award, supported by the Blaustein Pain Research and Education Endowment, was created to honor an individual who has served the Hopkins community in advancing pain research and education. Dr. Polydefkis is being honored with this award based on his pioneering work in painful peripheral neuropathy.

Dr. Polydefkis is an Professor of Neurology at the Johns Hopkins Hospital. He grew up on the south side of Chicago and after graduating from Brown University attended medical school at Johns Hopkins. As a medical student he worked in Robert Siliciano’s laboratory as a Howard Hughes Medical Institute Fellow and studied pathways of antigen processing. Michael completed internal medicine, neurology residency and neuromuscular and clinical neurophysiology training at Johns Hopkins under the mentorship of Drs. John Griffin and Justin McArthur. He joined the Hopkins faculty in 2000.

Michael’s primary area of interest is neuromuscular and peripheral nerve disease. He is the Director of the Johns Hopkins Cutaneous Nerve Laboratory and also Director of the Johns Hopkins Bayview EMG Laboratory. His research focuses on developing novel measures of peripheral nerve disease and sensitive outcome measures for clinical trials. He has shown that epidermal innervation is affected early in diabetes and pre-diabetes and that novel measures of regeneration can be used in peripheral neuropathy trials. His capsacin model of denervation and re-innervation has been used in 5 multicenter diabetic neuropathy trials and has proven to be a robust and reproducible measure. These studies have been helpful in defining factors that influence human peripheral nerve regeneration. Michael has also been instrumental in defining the natural history of epidermal nerve fiber loss in diabetes and in developing punch skin biopsy as a biomarker of disease and drug effect. PC Project is grateful to work with Dr. Polydefkis on a number of projects.

WELCOME TO THE MSAB!

PC Project warmly welcomes Professor David Kelsell and Dr. Robyn Hickerson to the PC Medical and Scientific Advisory Board (MSAB) and appreciates their willingness to offer counsel and help for the PC cause. A list of PC Project MSAB members is available online at pachyonychia.org/about-pc/
PC Project hosted an impressive group of scientists, clinicians and drug developers from around the world at the International PC Consortium (IPCC) Symposium in Chicago, May 7-8, 2019 to present and collaborate on PC-related research. With 18 speakers and approximately 60 audience members attending at various times throughout the meeting, PC research continues to advance as a result of these dedicated professionals.

After the IPCC Symposium, the PC Medical and Scientific Advisory Board met to discuss projects, goals and next steps for PC research.

Following that meeting, PC Project manned a table at the larger Society of Investigative Dermatology Conference in order to raise awareness and make new connections.

On behalf of PC patients worldwide, PC Project thanks all who attended and participated in this special meeting.

The following is the list of presenters in alphabetical order and the title of their talks from the IPCC Symposium.

- **Christopher G. Bunick, MD, PhD** - Keratin 1/10 A11 heterotetramer crystal structures provide a molecular basis for a novel knob-pocket mechanism governing intermediate filament assembly
- **Michael J. Caterina, MD, PhD** - Pain mechanisms in palmoplantar keratodermas
- **Michael Conneely, PhD** - Opening a window into skin structure and function
- **Pierre A. Coulombe, PhD** - Novel roles for keratin in epidermal homeostasis and their significance for PC pathophysiology
- **Weston Daniel, PhD** - SNA-Based Therapeutics to Treat Diseases with Great Unmet Medical Need
- **Mark P. de Souza, PhD** - Developing mechanism-based therapies for rare skin diseases
- **John D. Doux, MD** - The Orphan Disease Business Model: One size does not fit all
- **C. David Hansen, MD** - Group Discussion: Moving Forward
- **Robyn P. Hickerson, PhD** - Next generation ASOs for treatment of PC
- **Alain Hovnanian, MD, PhD** - PERP, a novel gene causing Olmstead Syndrome
- **David P. Kelsell, PhD** - iRhom2 and the keratinocyte stress response
- **E. Birgitte Lane, PhD, FRSE, FMedSci** - Integrating the Keratinopathies
- **Thomas M. Magin, PhD** - Toward a compound-based approach for treatment of dominant keratin disorders
- **Edel O’Toole, MD, PhD, FRCP** - Quantifying plantar pain
PC Patient Support Meeting 2019

Sixty-three PC patients, plus family members, clinicians, scientists and drug developers attended the 18th annual PC Patient Support Meeting (PSM) in Boston, June 20-22.

One highlight of the meeting was when patients gathered into groups according to their affected PC genes and clinicians were able to visit each group and see a number of PC patients and their symptoms and clinical signs in one setting. Like a PC Grand Rounds experience, this was an opportunity for physicians to learn and observe first-hand the similarities and differences among those who share the same mutated gene.

PC Project also hosted a children’s program as well as special discussions for children and teenagers about PC and how they feel and deal with PC.

Some patient comments included:

♥ “This was one of the best experiences of my life to finally meet someone with the disease I have.”
♥ “You finally feel like you’re not different, find new ways to manage your PC, and learn about the research being done.”
♥ “Bring your family so they can go through a better understanding of your condition and the pain you are experiencing.”
♥ “It’s a worthwhile experience and really fulfils its name—Support.”
♥ “This is a great opportunity to meet people who can relate and learn new information that will help with the pain that comes with PC.”

PC Project is grateful for the IPCC physicians, scientists and industry professionals who participated at the meeting including Larry Bauer, Albert Bravo, Emily Cook, Ashlyn Downing, Robert Driscoll, Tracy Funk, David Hansen, Roger Kaspar, Wesley Kaupinen, Karan Lal, Evelyn Lilly, Kathleen McGowen and James Valentine. Members of the IPCC are always welcome guests at any PC Patient Support Meetings.
VALO STUDY
ACTIVELY ENROLLING PC PATIENTS

Recruitment has begun for the VALO Phase 2/3 study, sponsored by Palvella Therapeutics. PC patients have already expressed interest and given consent to have their information passed on to the study sites. In less than one month, more than 29 patients are enrolled, with all five sites across the US actively enrolling.

IMPORTANT: Because the study is still enrolling, physicians are encouraged to guide interested PC patients to fill out the short interest form at surveygizmo.com/s/4854812/Phase2-3

This clinical trial is a perfect example of how PC Project collaborates with industry and the medical and scientific communities. PC Project's International Pachyonychia Congenita Research Registry (IPCCR) is key in the collaboration in identifying patients with known mutations. PC Project is assisting with the trial by notifying the PC community about the study and by continuing to provide free genetic testing with detailed reports. For this trial, all participant need to have their own genetic testing reports. Therefore, PC Project is also providing Sanger sequencing confirmation tests for those with tested family members.

Additional clinical trial information is available at pachyonychia.org/valo/

RECENT PUBLICATIONS
pachyonychia.org/research-articles/


TOGETHER, WE ARE STRONGER THAN PC

June was PC Awareness Month. This year, the theme focused on “Together, we are stronger than PC.” PC Project is grateful to each of you for caring about PC and for supporting us. While patients were busy sharing their PC stories and raising funds last month, some of you posted on social media platforms.

Dr. Mark Field, formerly of Grunenthal, now the owner of Analgesic Innovation, a pharmaceutical company with the mission to “Help patients with chronic pain by increasing awareness and identifying new treatment solutions” created a LinkedIn post about PC and the need for more awareness and more research for patients with chronic pain.

Dr. Tycho Speaker shared, “The PC community has made extraordinary things happen, many seemingly by sheer force of will. The gains made in the science and diagnosis of this disorder over the last decade are nothing short of remarkable. I can’t help but think that this reflects the fierce determination that living with PC requires in the first place.”

Thank you, Drs. Field, Speaker and to all of you for being advocates for PC and for being friends to PC patients! Your support helps makes us Stronger Than PC.
MESSAGE FROM THE EDITOR

Greetings from London where we are still talking about Brexit. The European Society for Dermatological Research (ESDR) meeting in Bordeaux was very successful and PC Project was represented there. At the beginning of October I was back in France again at the European Reference Network for Rare Skin Diseases meeting. This has an ichthyosis and palmoplantar keratoderma subgroup (which includes PC) and aims to improve patient care throughout Europe. In this newsletter, we have updates from the VALO Clinical Study which is recruiting well in the USA, we congratulate Birgit Lane on her honorary membership of the ESDR and also report from the IPCC Symposium in Chicago earlier this year. Jack Padovano gives us some insights into living with PC. His comparison of living with PC to maintaining a bank account of steps is a great one and illustrates the thought that PC patients put into each step. The onus on all of us is to increase the number of steps that individuals with PC can take.

VALO PHASE 2/3 CLINICAL STUDY
By Emily Cook, Palvella Therapeutics

VALO recruitment continues to remain on track, with more than 60 PC patients having entered the study by Mid-October. Dr. David Hansen, Dr. Joyce Teng and Dr. Leonard Milstone have led the way by enrolling a significant number of PC patients at their respective institutions. Successfully recruiting a clinical trial at a site requires the investigator’s commitment to the study, engaging their clinical teams in meaningful ways and ultimately placing the patients at the center of the work. Palvella Therapeutics, and PC patients are fortunate to have these three individuals setting the pace and are deeply appreciative of their leadership. Several other investigators and clinical sites are actively enrolling into the study and Palvella is also very thankful for their investment in PC patients and the study. We look forward to spotlighting them in the future!

On the heels of the VALO study, Palvella Therapeutics is pleased to announce that an open-label extension study will be available for patients to continue taking investigational PTX-022 (QTORINTM rapamycin) for pachyonychia congenita. We look forward to updating you on this study status in the next newsletter!

For more information on VALO, Palvella Therapeutics’ Phase 2/3 study investigating PTX-022 (QTORINTM rapamycin) for pachyonychia congenita, please visit clinicaltrials.gov or pachyonychia.org/valo/

BIRGIT LANE HONORED WITH PRESTIGIOUS ESDR AWARD

Ellen Birgitte Lane (Birgit Lane) was presented with Honorary Membership of the ESDR, the ESDR’s highest honor, at the 2019 ESDR meeting. Edel O’Toole gave the citation and mentioned Birgit’s strong leadership in research, significant contributions to skin science including the discovery of many of the known keratins causing disease, mentorship of many successful individuals in epithelial biology and building partnerships between individuals, institutions and patient support groups. This prestigious award was presented by Edel O’Toole and David Kelsell at the 49th Annual ESDR meeting September 19, 2019 in Bordeaux, France. When she received this award Birgit said, “Thank you very much for this. I’m very honored and touched to get this award. And it’s especially nice to get an award for something you love doing.”

Birgit obtained her BSc and PhD in Zoology from UCL and after post-doctoral work at Cold Spring Harbour and Imperial Cancer Research Fund, she started her own research group. She was the Cox Chair of Anatomy and Cell Biology in Dundee from 1991-2009. In 2005, she went on sabbatical to Singapore and founded the A-Star Institute of Medical Biology and was the inaugural director from 2007 until 2018. She founded the Skin Research Institute in Singapore in 2013 and is now it’s
Chief Scientist. In Singapore over the last 14 years, she has championed skin research, building partnerships with other institutions worldwide and working with patient support groups including PC Project and Debra Singapore. She currently works on understanding how keratin filaments function, what are the consequences of their failure, and how such knowledge can lead to strategies for therapy.

**PC Project at ESDR**

French PC patient, Marie Jose Billeau, and her daughter, Alice, represented Pachyonychia Congenita at the European Society for Dermatological Research (ESDR) annual meeting in Bordeaux, France on September 19-20, 2019. Marie and Alice shared materials about PC in French and in English with scientists from around the world and met with members of PC Project’s Medical and Scientific Advisory Board. In addition, they received contact information of new researchers who are interested in being connected to PC Project.

Thanks to attending IPCC members who visited with Marie and Alice, including Alain Hovnanian and David Kelsell, Birgit Lane and Edel O’Toole.

**Global Genes Features PC**

Global Genes, a worldwide organization that connects, empowers and inspires the rare disease communities, recently interviewed Janice Schwartz, PC Project’s executive director, in order to bring awareness to Pachyonychia Congenita and PC Project.

When asked what makes her hopeful, Janice replied, “All the advances in science that have potential applications now and in the future for PC make me hopeful. If I didn’t believe that one day, we will have effective treatments for PC, I would not be dedicating my time this organization. I’m also energized by the hope of others—not just the patients, but all the people who donate their wisdom and resources to this cause. We see lots of little miracles all the time. Sometimes I worry and wonder, “How is this going to work out?” And then somehow, things do work out, usually because of the goodness of people.”


**Pachyonychia Congenita Voice of the Patient Report now on FDA website**

Last year, PC Project submitted the Voice of the Patient Report to FDA from the Externally-led Patient Focused Drug Development (EL-PFDD) Meeting. This report is now on the FDA website. The definitive voices of real patients about the impact of PC on
their lives are official. (You may need to click the Meeting Reports tab to see the report.)

fda.gov/drugs/development-approval-process-drugs/external-resources-or-information-related-patients-experience

Or visit https://www.pachyonychia.org/externally-led-patient-focused-drug-development-meeting-el-pfdd-with-fda/

**Recent Publications**

pachyonychia.org/research-articles/


PC Viewpoint: A Bank Account of Steps

My name is Jack Padovano. I’m 56, live in San Diego and my genetic mutation is in keratin 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 of bitching, 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 writhe 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 500 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.
The International Pachyonychia Congenita Consortium (IPCC) is a global group of scientists and physicians working to develop therapies for pachyonychia congenita, a rare genetic skin disorder. The research reported at the 16th Annual Research Symposium of the IPCC, held on 7-8 May 2019 in Chicago, IL, U.S.A is summarised here.

**Summary**

The International Pachyonychia Congenita Consortium (IPCC) is a global group of scientists and physicians working to develop therapies for pachyonychia congenita, a rare genetic skin disorder. The research reported at the 16th Annual Research Symposium of the IPCC, held on 7-8 May 2019 in Chicago, IL, U.S.A is summarised here.

**What’s already known about this topic?**

Pachyonychia congenita (PC) is a rare genetic skin condition caused by a mutation in any one of five keratin genes (KRT6A, KRT6B, KRT6C, KRT16, or KRT17). PC is characterized clinically by the triad of plantar keratoderma, plantar pain and variable nail dystrophy. Patients may also have other findings such as oral leukokeratosis, follicular hyperkeratosis and/or cutaneous cysts. Plantar pain is severe and is out of proportion to that of non-syndromic callus formation, and it is the major symptom of concern for PC patients.

**What does this study add?**

We report on progress by scientists and clinicians from around the world who are working to understand the pathophysiology of PC and other keratinopathies. We discuss ongoing scientific efforts to understand the role of inflammation in PC and work to translate that information into therapeutic treatments for PC. We provide an update on the most recent advances in gene suppression or the modification of genetic expression as a treatment approach in PC. We discuss the status of current therapeutic trials in PC as well as the need for collaboration between scientists, clinicians, patients, and industry to further advance PC treatment.

The 16th Annual Research Symposium of the International Pachyonychia Congenita Consortium (IPCC) was held in Chicago, IL, U.S.A. on 7–8 May 2019. The meeting was attended by approximately 60 scientists, physicians, and industry leaders from around the world who are devoted to understanding the pathogenesis and developing therapies for pachyonychia congenita (PC). PC is a rare genetic condition caused by the inheritance of a mutation in one of five known keratin genes (KRT6A, KRT6B, KRT6C, KRT16, and KRT17). The condition is characterized by palmoplantar keratoderma, severe and debilitating plantar pain, and often nail dystrophy. Patients variably also have oral leukokeratosis, follicular hyperkeratosis and cutaneous cysts.

**Day 1: afternoon session – Building the Foundation for Successful PC Studies**

Janice Schwartz, Executive Director of the PC Project (Salt Lake City, UT, U.S.A.), opened the meeting with an announcement that the International Pachyonychia Congenita Research Registry (IPCRR) has now collected data from over 2,000 patients. The registry is an ongoing effort to identify, genetically test, and register patients with pachyonychia congenita. This process empowers patients and helps to advance scientific understanding and treatment of pachyonychia congenita. The registry is readily accessible and takes patient self-referrals or referrals from clinicians.

The first session of the meeting was chaired by Edel O'Toole (Queen Mary University of London, London, UK) and was entitled “Building the foundation for successful PC studies.” The session focused on cutting-edge basic science research that contributes to our understanding of PC and other keratinopathies, and that points to potential therapeutic targets for PC patients.

As the first speaker of the meeting, Birgit Lane (Skin Research Institute of Singapore) presented “Integrating the keratinopathies.” Dr. Lane’s team works on understanding how keratin filaments function and what the consequences are when they fail. She discussed the idea that keratinopathies, such as epidermolysis bullosa simplex (EBS), pachyonychia congenita (PC), and others might be productively grouped and studied together since the underlying similarities in the
pathogenesis of the disorders suggest that similar approaches could be taken to discover novel therapeutics. Dr. Lane discussed models that are being used to screen for compounds that reduce keratin aggregation with promising results. Her work demonstrates the need to better understand keratin biology in order to inform the development of targeted therapeutic modalities.

Furthering the discussion of keratin research, Pierre A. Coulombe (University of Michigan, Ann Arbor, MI, U.S.A.) presented his talk entitled “Novel roles for keratin in epidermal homeostasis and their significance for PC pathophysiology.” Dr. Coulombe’s team studies the genetic basis and mechanistic underpinnings of keratin mutation-based disorders, with a focus on EBS and PC. He reviewed what we have learned thus far from a Krt16 null mouse model for palmoplantar keratodermas (PPKs). The Krt 16 null mouse develops footpad lesions that mimic callus formation in PC patients, and the model has been shown through murine and human comparative gene expression surveys to be an appropriate model for the study of plantar keratoderma in PC patients. Based on Dr. Coulombe’s discoveries, the development of calluses occurs in three stages: pre-PPK, PPK onset, and active PPK. In the pre-PPK stage, keratinocytes in the palmoplantar skin of the mice exhibit both defective terminal differentiation and a dramatic loss of keratin 9 expression. This downregulation of Krt9 expression in the Krt16 null mouse footpad is likely to be important, since Krt9 null mice develop PPK. A possible therapeutic approach in PC might be targeted upregulation of keratin 9. During the PPK onset stage in the Krt16 null mouse model, increased oxidative stress, including hypoxia, Keap1-Nrf2 signaling, occurs and there is subsequently a lack of ability to return to normal homeostasis during the active PPK phase. Calluses in patients with PC-16 also show evidence of reduced Nrf2 activity, highlighting the inflammatory cascade in PPK as a target for future therapeutic developments for PC.

The inflammatory response was also noted as important by Dr. David P. Kelsell (Queen Mary University of London Whitechapel, London, UK), who described his work on iRhom2 and the keratinocyte stress response in tylosis, a syndrome characterized by PPK and a high risk of developing oesophageal squamous cell carcinoma. Based on multiple lines of evidence generated in his laboratory, he showed that iRhom2 functions as a major regulator of the response to cellular stress and disease, particularly of keratin dynamics in response to stress and in p63-mediated signaling pathways.

Christopher G. Bunick (Yale University, New Haven, CT, U.S.A.) studies the biochemistry of intermediate filament assembly to understand how keratins function in PC. In his presentation “Keratin 1/10 A11 heterotetramer crystal structures provide a molecular basis for a novel knob-pocket mechanism governing intermediate filament assembly,” he discussed the importance of atomic resolution structures in understanding disease using ‘genotype-structurotype-phenotype’ models, highlighting the critical knob-pocket assembly mechanism within the K1 helix 1B domain, which is conserved among all type II keratins. Dr. Bunick’s work represents the first crystal structure of any keratin mutation associated with human skin disease and sets a precedent for future atomic resolution characterization of pathogenic PC mutants in intermediate filaments.

Thomas M. Magin (University of Leipzig, Leipzig, Germany) presented “Toward a compound-based approach for treatment of dominant keratin disorders.” His team has established assays to assess restoration of a functional keratin cytoskeleton in an effort to discover chemical compounds for the treatment of EBS and PC that prevent or revert collapse of mutant keratin cytoskeletons into protein aggregates. Such compounds would be applied locally or given systemically to patients. Compounds that modify keratin post-translationally are a focus. He has already identified a lead compound that decreased aggregation of keratin in cells with the EBS-associated K14R125C mutation by 50%. A screen of 5,000 bioactive compounds is currently underway to identify additional compounds that restore the keratin cytoskeleton.

The field of nucleic acid delivery as a potential treatment strategy for PC was another major theme at this year’s IPCC meeting. Robyn P. Hickerson (University of Dundee, Dundee, Scotland, UK) discussed the use of small interfering RNAs (siRNAs) to target mutant keratin expression, highlighting that intraleisional injection has been required in the past for delivery, given that the epidermal barrier precludes penetration of siRNA. However, pain with the injectable delivery method is a major problem in PC. Her IPCC presentation titled “Next generation ASOs (antisense oligonucleotides) for the treatment of PC” described her more recent work on nucleic acid-based therapeutics and strategies for delivery of these therapeutics. Her team, including speaker Michael Conneely (University of Dundee, Dundee, Scotland, UK), has developed novel ex vivo human skin models to evaluate delivery and efficacy of knockdown. Dr. Hickerson described ongoing research in collaboration with WAVE Life Sciences (Cambridge, MA, U.S.A.) that has increased the specificity of ASOs towards targeting at a single nucleotide mutation level, which previously required siRNA, and her goal of bringing these nucleic acid molecules to the clinic within the next few years.

Michael J. Caterina (Johns Hopkins, Baltimore, MD, U.S.A.) presented “Pain mechanisms in palmoplantar keratodermas.” Dr. Caterina’s team at Johns Hopkins is focused on the molecular and cellular mechanisms underlying neuropathic and inflammatory pain sensation. His recent work has been focused on mechanisms that contribute to pain in mouse models of hereditary palmoplantar keratodermas. Dr. Caterina discussed new research on specific neuroanatomical changes that might be associated with pain and itch in some, but not all, patients with palmoplantar keratoderm. 
Alain Hovnanian (Imagine Institute and Necker Hospital, Paris, France) focuses on the study and development of new treatments for EB, Netherton syndrome and severe palmoplantar keratodermas such as PC and Olmsted syndrome. Dr. Hovnanian presented the final talk of the first session with “PERP, a novel gene causing Olmsted Syndrome” and discussed his co-discovery with Dr. Keith Choate of Yale University that variants in the PERP gene can lead to many of the features seen in Olmsted syndrome, including PPK. PERP codes for a component of desmosomes and is a downstream target of p63\(^1\) which participates in epidermal cell adhesion. This discovery contributes to our understanding of the biological pathways involved in the development of severe palmoplantar keratoderma.

**DAY 2: morning session – PC and the Therapeutic Horizon**

The second session of the IPCC meeting, chaired by Amy Paller (Northwestern University, Chicago, IL, U.S.A.) and Roger Kaspar (Santa Cruz, CA, U.S.A) was titled “PC and the Therapeutic Horizon” and focused on the status of current therapeutic trials in PC as well as the need for collaboration between scientists, clinicians, patients, and industry to further advance PC treatment.

Dennis R. Roop (University of Colorado, Aurora, CO, U.S.A) presented “An alternative method for delivering keratinocytes derived from patient-specific gene-edited iPSCs generated from patients with inherited epidermal fragility disorders.” He discussed work with colleagues, Drs. Ganna Bilousova and Igor Kogut, on the development of induced pluripotent stem cells (iPSCs) as a treatment for recessive dystrophic epidermolysis bullosa (RDEB). He noted that reprogramming fibroblasts with modified mRNA and miRNA is a non-integrative method of introducing genetic modifications in patient’s fibroblasts\(^14\). He then discussed two means by which to deliver genetically modified cells into a patient. He is partnering with Avita Medical (Valencia, California, U.S.A), a company that manufactures a method for spraying dissociated cells onto patients. This method has been used effectively to treat burn patients\(^15\) and may represent a plausible delivery mechanism for keratinocytes derived from gene-edited iPSCs that would obviate the need to develop sheets of skin for grafting. Moreover, spray delivery may be preferable in body locations that are traditionally difficult to graft, including the palms, soles, and oral cavity. Dr. Roop also discussed studies to increase the homing of modified mesenchymal stem cells, delivered intravenously, to areas of trauma or injury in internal epithelia.

Weston Daniel (Exicure, Skokie, IL, U.S.A) presented “Topical application of DNA for dermatology”. He spoke about the properties of spherical nucleic acids (SNAs), nanoparticle constructs that are dense and radial arrangements of oligonucleotides. Dr. David Giljohann, founder of Exicure (Skokie, IL, U.S.A), together with Dr Amy Paller (Northwestern University, Chicago, IL, U.S.A), discovered that spherical nucleic acids readily penetrate epidermal cells in vitro and intact human skin in vivo\(^16\). The oligonucleotide component of SNAs can suppress or modify disease gene expression, suggesting promise in the treatment of genetic skin diseases. Topical SNA-based therapies are currently in clinical trials for psoriasis with a goal to develop drugs for the treatment of rare genetic conditions.

Mark P. deSouza (FIBRX Derm, Inc, Berkeley, CA, U.S.A) presented ‘developing mechanism-based therapies for rare skin disease.’ He discussed lessons that he has learned over time regarding what makes for a good potential therapeutic drug, including a formulation that is stable, penetrates into the skin, is safe, and well tolerated. Eugene J. Sullivan (Palvella Therapeutics, Wayne, PA, U.S.A) reviewed the unique challenges that are faced by drug developers in rare diseases. John D. Doux (Palo Alto Investors, Palo Alto, CA, U.S.A) shared his experience as an investor and discussed what investors look for in drug development. Braham Shroot, PhD (Palvella Therapeutics, Wayne, PA, U.S.A) discussed the development of a promising new high strength rapamycin formulation ‘PTX-022’, which is currently entering clinical trials for PC.

**Conclusions**

This year’s Annual IPCC Research Symposium reminded us of the ongoing efforts of the PC project and the IPCRR, which now includes data from over 2,000 patient participants and is a vast repository of important disease-specific information. This data is readily available to physicians and scientists who are working to further the understanding of PC pathophysiology and treatment for PC patients. The IPCC Symposium highlighted the progress in the understanding of PC pathophysiology and advancements in the translation of this scientific knowledge into therapeutic options for PC patients. Highlighted research with the potential for therapeutic development include targeting the inflammatory response in callos development and suppression or modification of genetic expression via nucleic acid delivery or introduction of induced pluripotent stem cells. These potential therapeutic options move us closer to mitigating the debilitating pain that PC patients experience.

**References**


Happy Holidays

MESSAGE FROM THE EDITOR

In London, it is starting to get colder and yesterday it rained heavily for most of the day. Now that the UK General Election is over, everyone is looking forward to Christmas and the holiday season. The PC Project Medical and Scientific Advisory Board have been busy thinking about interesting topics to discuss at the IPCC Symposium in May 2020 in Scottsdale, Arizona and will be coming up with a complete programme soon. We are grateful to those who contributed original research articles, research letters, reviews, commentaries and editorials which will be published in a Rare Disease issue by the British Journal of Dermatology in March 2020. Seasons greetings to everyone and wishing you and your family and friends lots of joy and happiness in 2020.

VALO Phase 2/3 clinical study... Phase 3 has begun!
By Emily Cook, Palvella VP of Clinical Operations

PC patients have now entered Phase 3 of the VALO clinical study! Interest in the study from the PC community continues to remain strong with more than 75 patients having entered the study as of mid-December and 9 clinical study sites actively enrolling. In addition to the investigators that were spotlighted in the previous newsletter, Dr. Amy Paller, Dr. Tracy Funk, Dr. Jennifer Parish and Dr. Steven Kempers have all made significant contributions to the study. We are very thankful and appreciative for the intense engagement and collaboration from all clinical study investigators, PC Project and of course PC patients.

The open-label extension study is on track to open for enrollment in Q1 of 2020 and a sub-study is now planned to include KRT6C and KRT17 patients.

For more information on VALO, Palvella Therapeutics’ Phase 2/3 study investigating PTX-022 (QTORINTM rapamycin) for pachyonychia congenita, please visit clinicaltrials.gov or pachyonychia.org/valo.

IPCC Symposium, May 12-13, 2020, Scottsdale, Arizona

The 17th Annual International Pachyonychia Congenita Consortium (IPCC) Symposium will be held May 12-13, 2020 in Scottsdale, AZ, USA. All interested researchers, clinicians and drug developers are welcome. Details and information is found at pachyonychia.org/2020ipcc/.

PC Patient Support Meeting June 4-6, 2020, Paris, France

Patient support meetings (PSM) are an excellent way for professionals to see a large group of PC patients in one setting. Clinicians, researchers and industry representatives are warmly invited. Visit pachyonychia.org/2020psm/
**New Data on PC Findings**

Two issues that PC patients deal with that aren’t typically associated with PC are **deep itching** and **neurovascular-like structures** (as seen in photos of two PC patients) in the calluses.

As new patients join the registry and share their experiences, PC Project is ever-learning about PC and how it affects patients.

In October 2019, PC Project surveyed its genetically confirmed patient community with an IRB approved registry addendum. With 350 responses, **69% of patients deal with deep itching**, an itch that is under the calluses that is difficult to reach.

Furthermore, **62% of patients have neurovascular-like structures**. In truth, what they are exactly is not known. What is known is they significantly increase pain and make it difficult for patients to trim their calluses.

These two symptoms of PC are not typically talked about in PC discussions but are clearly a part of life for a significant portion of the PC population. Data from the series of questions about these issues will be studied and published.

**PC Project at PeDRA**

PC Project participated at the Pediatric Dermatology Research Alliance (PeDRA) Annual Conference in Chicago, November 14-16, 2019.

IPCC members Anna Bruckner, Amy Paller, Keith Choate and Joyce Teng (pictured below) had major roles at this meeting.

PC advocate and patient, Jim Rittle, attended the patient track, along with his daughter, Kaelyn who also has PC. Janice Schwartz, PC Project’s director spoke on how the April 2018 Externally-led Patient Focused Drug Development (EL-PFDD) Meeting with FDA has benefitted PC Project.

The PeDRA conference gave PC Project an excellent platform to share with all attendees the strength of the registry and the commitment of PC patients and their families to finding treatments for PC. Janice also participated on a panel with three other patient advocate leaders.
PC Project presented a poster (see image below) and had a display table. These encouraged networking and provided opportunities to discuss PC with many clinicians, researchers and representatives of drug companies. The work to educate others about PC will always be a priority for PC Project. The abstract for the poster “Critical Functions of the International PC Research Registry” was printed in the meeting program.

1. The PC registry empowers patients who typically live for years in isolation, either undiagnosed or misdiagnosed. By putting a name on the disease, providing care and management services, and connecting patients with others who share the disorder, the registry can be life-changing for patients who navigate a life with nearly constant pain.

2. Through the data of genetically confirmed patients, the registry helps facilitate PC-
related research and publications and enhances best practices for clinicians. The data is literally changing what is known about PC. For example, PC was once thought to primarily be a nail disorder. Now it’s known that palmoplantar keratoderma pain is the most significant symptom of PC. The data now shows additional symptoms of PC yet to be researched and published.

3. Through the registry, PC Project knows who the PC patients are, where they are located and what exact mutation they have. With a body of data from genetically confirmed patients, the registry provides opportunities for learning, advancing research, collaboration with scientists and drug companies, and the advantage of having a readily accessible patient community available for studies and clinical trials.

The International PC Research Registry is an IRB approved patient registry (WIRB Pro Num: 20040468) and is constantly growing with approximately four new patients each week. Most patients find the registry by searching online, but some are referred by physicians or family members. Those who wish to enroll complete an online questionnaire, consent form and send photos of their symptoms. Free genetic testing and physician consultations are available to those who join the registry.

Currently, the registry houses over 2050 participants, 950 who are genetically confirmed to have PC, in over 60 countries. Forty percent of those are PC-K6a, 32% PC-K16, 16% PC-K17, 9% PC-K6b and 3% PC-K6c. Of the last 200 publications on PC, PC Project collaborated on over half. In 2008, PC Project sponsored the first -in-human mutation-targeted siRNA phase 1b trial for PC and since then has assisted in many additional PC-related studies. Currently, PC Project is collaborating with Palvella Therapeutics on the phase 2/3 study to evaluate the use of PTX-022, a novel, targeted formulation of rapamycin. The use of the patient registry is critical to this clinical trial. PC Project welcomes all interested researchers and drug developers to collaborate and utilize the registry in order to help those suffering with PC and similar rare skin diseases.

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**Recent Publications**

[pachyonychia.org/research-articles/](pachyonychia.org/research-articles/)


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**Giving Tuesday 2019 Success!**

Because of you, and with the help of over 380 donors, Giving Tuesday was truly a day of awareness and giving for the PC community. Each contribution made that day, as well as any contributions made from now through
December 31, 2019 will be matched 2:1. Those who are looking to make a year end donation can give at pachyonychia.org/donate-help/

Please accept our heartfelt thanks for your support now and always. You are the reason PC Project is thriving and PC patients have hope for a less painful life.

From the bottoms of our feet, we thank you.

PC Project Spotlight

Many of you have either met or connected with Holly Evans via email or phone. As the International Patient Support Officer for PC Project, Holly fields many phone calls and emails from patients and physicians. She has worked for PC Project in numerous roles since August 2007.

Holly is the youngest of 14 children. She has been an aunt since age 5. Currently she has 42 nieces and nephews and 19 great nieces and nephews. Family is a huge part of her life. Many of Holly’s uncles were doctors with a family clinic called Evans, Evans, and Evans. She also has a few cousins who are doctors. So the medical community has always played a part in her family. In August 2007, Holly returned home to Utah from serving an 18 month mission for the Church of Jesus Christ of Latter-day Saints in South Carolina and was looking for what the next path for her should be. A long-time friend of the family, Robyn Hickerson, PhD, came to visit. After staying up much of the night talking and watching a movie trilogy, Holly took Robyn to the airport and on the way, made a stop to see someone Robyn knew, a woman who ran a charity who needed help. Well that stop turned into a very weird and unique interview and at the end, the woman, the founder of PC Project, Mary Schwartz, said, “Okay we will do a two week trial. Go take Robyn to the airport then come back. We will start today.”

Twelve years later and the PC theme “It’s all about love” is still the foundation of PC Project. For Holly, her roles have been a roller-coaster ride, with many successes and many lessons learned through PC Project, but it was the people, the doctors, scientists, patients, caregivers and other professionals who stole Holly’s heart and who continue to inspire PC Project’s mission.

While working at PC Project, Holly continued going to school part time and graduated from the University of Utah Business School in Information Systems and the Non-profit Academy of Excellence. Holly hopes that our wonderful PC family will be able to work together to find effective treatments to relieve the excruciating pain that PCers suffer daily.