Happy New Year!

If you haven’t already, please make a note to update PC Project’s current contact information in your address books.

Phone: 801-987-8758
Mailing Address: PO Box 17850, Holladay, UT 84117
Email Address: info@pachyonychia.org

2019 PATIENT SUPPORT MEETING

The 2019 PC Patient Support Meeting will be held in Boston, MA on June 20-22nd. Please see the last page for more information and links to register.

PEDALING AWAY THE PAIN

For the past eight years, PC Patient, Nathalie Kilchoer, and her family have organized a spinning marathon to raise awareness and funds for PC.

Nathalie wrote, “My children (13 year-old Dylan, 9 year-old Jade) and myself are affected by the disease, so we try to do our best to be able to donate money to the association for research.

“We have just finished our 8th Spinning Marathon for PC with around 200 people. This one was a success like the others. Many people participated in this marathon and therefore supported us thoroughly. We have already reserved the room for the next marathon, which will take place on Sunday, November 17, 2019.”

Through this event, Nathalie and her family donated nearly 4,000 Swiss francs to PC Project, raised from the November 2018 Spinning Marathon. Thank you Kilchoer family and friends!
How some patients trim their PC Nails

Because the nails of PC patients can vary in the ways they grow, patients use a variety of tools to trim their nails. What works for one patient does not necessarily work for another. It is an individual choice of which tool to use, how often to trim, and whether the nails are wet and soft, or dry and hard.

Here is how one PCer, Roseann McGrath, has cared for her nails from childhood: “I am a 52-year-old female, spontaneous PC-K6a mutation, where my PC presented at birth on my hands and feet. I was thankfully diagnosed shortly after my birth with having PC. As early as 6 months old, my mother & father used a General Electric (G.E.) manicure set to drill my tiny finger and toe nails weekly. As I got older I started to do it myself. In my early teens I began to use a Sears Craftsman Dremel Drill every other day on my fingernails and weekly on my toenails.

“In 2014, a fellow PCer told me about the “Upower” drill, which is just as powerful, but much easier to hold and handle then the Dremel. It is made by Urawa Corporation Upower Nail Filing System UP200C Series Controller. To this day I drill my nails down to get them as flat as I can. I paint my fingernails with a light beige nail polish to cover the green hue in my PC nails. I cannot “trim” or “drill” too low because my nerves grow through my nails, thus when they start to bleed I know I’ve gone too low. Conversely, I cannot permit my nails to remain too high, because the nails are sensitive from the thickness. Therefore, I must find the right balance of how thick or how low my nails can be. With this said, I err on the side of drilling down as far as I can to hide how thick they are and to try to make them appear somewhat normal. I unfortunately cannot hide my calluses and blisters on each finger (and foot) at each pressure point like I can my nails.”

Here are some tips and tools from other PCers:

“Black & Decker drill with a fine sandpaper disk.”

“Large pointed nail clippers (I affectionately refer to them as “hedge trimmers”) which work best on wet/soaked nails, or a Dremel tool with a dimmer switch on dry nails/skin.”

“Nails clippers for large dogs, small pointed nail clippers, Dremel and Emery board. I grind them with an electric manicure device every three weeks.”

“I use this (see left) once a week for the very thick nails of my son. Our son watches television with a favorite snack during the weekly trimmings.”

“An Emery board to maintain my nails. I’ve only been able to tolerate the Dremel on my fingers in the last couple of years. As a child, my dad soaked and trimmed my nails with a razor blade. A Dremel still hurts when used on the fingernails.”

“These are a few of my trimming tools (see right).”

“Dremel on the worst nails, nail clippers and an Emery board on the other nails.”

“I use a thing called the PediNova by Medicool. It’s essentially a Dremel tool but they provide different bits specifically for nail grooming. They include a sandpaper barrel attachment, but I find that gets incredibly hot.”

“My nails are very thick, so when a podiatrist takes the clippers to them, it’s quite painful. I find that using a grinder is much more comfortable.”

“A spring-loaded toe nail clipper for the foot nails and a good nail file for the hand nails. Sometimes I use a single edge razor blade to help with toe nails.”
BOSTON PACHYONYCHIA CONGENITA PATIENT SUPPORT MEETING  
JUNE 20-22, 2019—HILTON BOSTON LOGAN AIRPORT HOTEL

Thursday, June 20th - welcome dinner starts at 6:30 pm
Friday, June 21st - all day presentations, discussions and meals
Saturday, June 22nd - half day meeting with the closing luncheon at 12:30 pm

REGISTER FOR MEETING at www.surveygizmo.com/s3/4788209/2019PSM
Please register as soon as possible so we can plan the event. There is no cost to register. Fees are paid separately and reduced fees apply until April 22, 2019.

REGISTER FOR HOTEL at www.hilton.com/en/hi/groups/personalized/B/BOSLHHH-PCP-20190620/index.jhtml or by phone 1-617-568-6700. Be sure to mention the meeting code PCP. The meeting discounted room rates per night are $189 single/ $189 double/ $209 triple/ $229 quad. Please book now. You are not charged until you check in at the hotel.

PAY MEETING FEES at www.paypal.com/us/webapps/mpp/search-cause?charityId=80948&s=3
Meeting Fees will be waived for PC Patients attending the meeting for the first time and one family member. PC Project pays 80% of the meeting costs which include the meeting room, equipment rental fees and food including Thursday dinner, Friday breakfast, lunch and dinner, and Saturday breakfast and lunch as well as break food.
Pay before April 22, 2019: $130/person 15 years or over; $75/child ages 2-15.
Pay after April 22, 2019: $150/person 15 years or over; $80/child ages 2-15.
Children two and under are free.

APPLY FOR SCHOLARSHIP FUNDING at www.surveygizmo.com/s3/4788439/PSMScholarship
After you have registered for the meeting and have your hotel reservation, you can apply for a scholarship as needed to cover meeting fees, travel and/or hotel costs. Scholarship Application Deadline is March 15th, 2019 and awards will be announced by April 1, 2019. Scholarship funds are based on donations and the number of scholarships will depend on funds received.
IT’S ALL ABOUT LOVE

Have you noticed all PC Project’s logos have hearts? That’s because everything about this work is based on the love we have for, and the love we feel from, the PC community.

PC Project was founded on love. Over 15 years ago, Mary Schwartz was determined to do something about the daily suffering of her daughter-in-law and two grandchildren who have PC. Inspired by love, in 2003 Mary established PC Project. Her love soon grew to include all PC patients worldwide as she worked tirelessly for years to build this exceptional charity.

This foundation of love has been strengthened by many others including:

♥ Physicians and scientists in the IPCC (International PC Consortium) who lovingly volunteer their time and expertise in helping our cause and serving individual patients. Many have dedicated large chunks of their careers and their time to help patients and/or work on PC-related research.

♥ Industry partners who have caught the PC vision and who are highly motivated to see patients receive effective treatments for PC.

♥ Patient Advocates around the globe who lovingly answer our calls for help, lead at Patient Support Meetings, help us with projects, and give assistance and advice to other PCers.

♥ A small staff of dedicated personnel at PC Project who genuinely care for PC patients. They work long hours, and at all times of the day and sometimes night. As we discuss cases, work on projects, serve patients, assist physicians, researchers and industry representatives, their commitment and love for PC patients is evident.

♥ A working PC Project Board of Trustees who volunteer their time and resources to further the cause. In some ways these individuals are like part time staff members for all the work they do for PC Project - all because of the love in their hearts for PC patients.

♥ PC patients, family members and other cheerleaders for PC Project, who donate their time and resources to further our mission. We’ve seen patients rise to our calls for help numerous times over the years. They’ve answered surveys, participated in studies, offered advice, attended meetings, raised awareness, held fundraising events, shared social media posts, and have even donated their skin.

♥ Individual donors who contribute to this cause. As you may know, PC Project is almost solely reliant on individual donations. We cannot express enough our thanks to each of you for your gifts. You are the reason we are still here. Your love, given through donations, large or small, is the reason we are able to continue.

What Mary started 15 years ago, all because of love, continues on today. Everything we do to accomplish our mission is motivated by love. It always has been and it always will be. #itsallaboutlove.
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 are interested in participating in this future Phase 2/3 clinical study as a PC patient or have PC patients in your clinical practice that would be interested, please let us know at https://www.surveygizmo.com/s3/4854812/PCInterestSurveyPhase2-3

The clinical study plans to begin enrolling PC patients in the coming weeks, with clinical trial sites currently planned for sites across the United States.
A ‘SCARY’ BUT EFFECTIVE FACEBOOK BIRTHDAY FUNDRAISER

Edward Pyner, a PC patient, recently created a Facebook Fundraiser to support PC Project. In addition to using the suggested words by Facebook, Edward wrote the following to accompany his fundraiser:

“I thought I would support this charity as I suffer from it. This is why I walk strangely. I have pain on my feet. I have good and bad days.”

Edward’s words are simple but powerful. While it can be scary to share words about our PC with others (especially when many patients hide their PC), those who make their fundraisers personal are often the most successful. People like to support people. When a person writes that they suffer from PC, or their child suffers from PC, and then shares a little about the impact of PC or what PC Project has done for them, their friends and family want to support them, not just the charity.

Thank you Edward for sharing a vulnerable part of yourself with others. And happy, happy birthday!

PC FAMILY MEMBER LOVE

While PC patients accomplish much on their own, they are often supported in many ways by their loved ones. After a non-PC related surgery, one PC patient couldn’t trim one of his feet. Here is what his wife shared about his recovery:

"He can do most things (since the surgery) except trim his left foot. For the first time in 53 years, he had me dremel it. With his guidance, I was able to complete the task. His faith in me was extraordinary. He guided me to "how" and told me the "whys" on each individual callus.

“I have such a greater appreciation for our PC family and friends, plus my sons and husband. I stand in awe for each of you PCers. What courage, strength, determination and joy you all have. I am also amazed at those wonderful healers, scientists, etc. that have heard the cries of those in despair, and risen to the challenges to offer assistance.”

JETBLUE EMPLOYEE DONATION

Recently, a JetBlue crewmember, Rebecca Wells, earned four (4) one-way ticket vouchers through volunteer service and selected Pachyonychia Congenita Project as the recipient. Rebecca had met a PC patient several years ago and was so impressed by the young man’s character, she chose PC Project as the beneficiary. These certificates will pay for travel for two PC Scholarship applicants to attend the upcoming patient support meeting.

Here is the response from Rebecca when PC Project wrote to thank her and tell her more about PC:

"Please believe me when I say it’s an honor to have PC Project receive travel certificates. I read your email with tears screaming down my cheeks. My friend did share a little about PC and I wanted to be a part of giving to your organization. I’m happy to hear the tickets will be used to help encourage others with this disease.

JetBlue encourages crew members like myself to volunteer. We track our hours and turn them into confirm seats on JetBlue flights for an organization of our choice to use as they need/want.

I’d like to thank you for creating the PC community and getting others involved to find a cure. I hope one day scientists will find a cure for PC so no one will be in pain.”

Thank you, Rebecca, for your service and thank you to the PC patient, who set a good example. This is another reminder that people donate because of people they know and to causes they trust.
Boston Pachyonychia Congenita Patient Support Meeting
June 20-22, 2019—Hilton Boston Logan Airport Hotel

Thursday, June 20th - welcome dinner starts at 6:30 pm
Friday, June 21st - all day presentations, discussions and meals
Saturday, June 22nd - half day meeting with the closing luncheon at 12:30 pm

Register for Meeting at www.surveygizmo.com/s3/4788209/2019PSM
Please register as soon as possible so we can plan the event. There is no cost to register. Fees are paid separately and reduced fees apply until April 22, 2019.

Register for Hotel at www.hilton.com/en/hi/groups/personalized/B/BOSLHHH-PCP-20190620/index.jhtml or by phone 1-617-568-6700. Be sure to mention the meeting code PCP. The meeting discounted room rates per night are $189 single/ $189 double/ $209 triple/ $229 quad. Please book now. You are not charged until you check in at the hotel.

Pay Meeting Fees at www.paypal.com/us/webapps/mpp/search-cause?charityId=8094&s=3
Meeting Fees will be waived for PC Patients attending the meeting for the first time and one family member. PC Project pays 80% of the meeting costs which include the meeting room, equipment rental fees and food including Thursday dinner, Friday breakfast, lunch and dinner, and Saturday breakfast and lunch as well as break food.
Pay before April 22, 2019: $130/person 15 years or over; $75/child ages 2-15.
Pay after April 22, 2019: $150/person 15 years or over; $80/child ages 2-15.
Children two and under are free.

Apply for Scholarship Funding at www.surveygizmo.com/s3/4788439/PSMScholarship
After you have registered for the meeting and have your hotel reservation, you can apply for a scholarship as needed to cover meeting fees, travel and/or hotel costs. Scholarship Application Deadline is March 15th, 2019 and awards will be announced by April 1, 2019. Scholarship funds are based on donations and the number of scholarships will depend on funds received.
BOSTON PC PATIENT SUPPORT MEETING
JUNE 20-22, 2019

Don’t forget to register for the PC Patient Support Meeting (PSM) at the Hilton Boston Logan Hotel.

Find meeting and registration details here: https://www.pachyonychia.org/2019psm/

Quotes from the 2018 PC PSM:

• “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.”

• “I knew very little about PC before the event, so was impressed and awed by the depth of research presented.”

• “It’s a worthwhile experience and really fulfils its name—Support.”

FROM THE PC MAILBOX:
A LETTER TO ALL PCERS

This week, we received a letter addressed to PC patients. As you’ll read from this individual who does not have PC, you are an inspiration to others by the courageous way you live your lives.

Dear PCers,

I don’t know why I am writing except that I feel compelled to reach out in a spirit of friendship and respect. This week, I was privileged to celebrate women on International Women’s Day in a media forum. While there, I was asked if there was a woman who I believed every women should know about. I mentioned my dear friend who has PC. Her courage and positive example in the midst of constant pain, has inspired and motivated me. Her vulnerability in sharing her journey with PC has helped me to understand a little of what each of you might experience on a daily basis; walking on your knees to brush your teeth; having people judge your nails or cysts; determining the amount of steps needed to accomplish your tasks; the torture of travel; infections, wheelchairs, and especially pain—burning, itching, throbbing, pain.

To all of you living with this disability, I want to say, “You matter!!” Every time, you show up to work, help your children, make a cup of coffee, you are doing remarkable things and I am inspired by you! I have written articles about my friend, where individuals wept and were inspired by her story. In a world where there seems to be so much judgment and prejudice, I want to be a voice of encouragement. I want to express my admiration and respect for you. I honor you for the daily battles
that you wage. I admire you, not because you have PC but because in spite of your PC you are choosing to live productive, beautiful, lives. You may say, "Teresa, you don't know me, some days I am lucky to get out of bed." I will let you know a secret. I hate pain. Any kind of pain. I am a wimp. So you see, even without knowing you, I can admire you because your daily realities would be too much for me.

Sometimes, in my life, I have felt lost, alone and weak. I wondered if any of you have felt that way? Recently, I saw a video clip from the movie, "The Greatest Showman." It inspired me to step into my own power. For any of you out there that may be feeling discouraged, alone or criticized, I hope you will feel of my respect and maybe this clip will give you some encouragement.

https://youtu.be/XLFEvHWD_NE

Thank you for inspiring those around you and thank you for inspiring me!

God Bless,
Teresa Burrell

FROM PC SOCIAL MEDIA:
WHY DO 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. I think it’s more to do with others not judging.

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

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. I wanted to play with the other kids but their parents wouldn’t let them. 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.

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.

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

Nancy: As a youngster, I always tried to tuck in my fingers... As an old (69) lady, I am more open to explaining to people, but still have some of my earlier insecurities.

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. 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, it sounded like I’m complaining...and I don’t want people to look at me like someone that just complains about everything.

Linda: I have it and I am not bothered by other people’s view. At least I get a clear space on the beach and plenty of room where naked feet are on show.

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.

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.

Malgosia: 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. 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...the discrimination of disabled people forces them to behave as normal to have a good education and job.

Andy: I would physically hide my feet so people don’t see their appearance...But anymore I’ll tell anybody that will listen. I’m more self-conscious of them wondering what’s wrong with me so I’m actually quite proud of it now—to tell them exactly what I’ve got and exactly what it does to me.

More responses can be found on the private Facebook PC chat page.

Facebook Birthday Fundraisers

Happy Birthday to Amanda Soderstrom, Rebecca Wells and Jamie Callahan-Simpson who created Birthday Facebook Fundraisers to support PC Project this month.

In addition, Jamie hosted a Pampered Chef party on Facebook where 25% of the sales are to be donated to PC Project.
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.

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 of you, we were able to publish articles helping doctors throughout the world to better understand and manage this difficult condition. Thanks to all of you who participated in the registry 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.

**Clinical Trial Update**

Much has been announced over the past year about the upcoming clinical trial to be held in the USA for PC. This will be the largest clinical trial ever designed specifically for PC patients. PC Project continues to collaborate with Palvela Therapeutics, whose team is preparing for the study. Information will be shared with the PC community as soon as active recruitment officially begins.

Thanks to all who have expressed interest in this historic clinical trial for PC. The feedback was extremely helpful. Furthermore, without the participation of PC patients who joined the International PC Research Registry (IPCRR), this trial would not be happening.
RECRUITMENT BEGINS FOR THE PHASE 2/3 CLINICAL TRIAL (VALO STUDY)

The time is finally here! Recruitment for the largest PC clinical trial yet has begun. Although only a portion of PC patients (those who live in the USA) will be able to participate in the trial, PC Project wishes to share this news with all PC patients and their loved ones everywhere.

Please note this part of the trial includes only three of the five types of PC. A parallel study is planned for the other PC types later this year.

Below is the official recruitment announcement:

SEEKING PARTICIPANTS FOR PACHYONYCHIA CONGENITA CLINICAL RESEARCH TRIAL

If you or someone you know has Pachyonychia Congenita (PC), you may be interested in participating in a Phase 2/3 clinical research trial for an experimental therapy that is applied to the soles of your feet.

Some of the requirements for participation include:

- Must live in the United States
- Must be at least 18 years of age or older
- Have a clinical diagnosis of PC, genetically confirmed to involve any of three keratin genes KRT6A, KRT6B, or KRT16
- If you have not been genetically tested, or if you do not have a copy of your genetic testing report, please contact PC Project at study@pachyonychia.org for more information about genetic testing.

NOTE: If you know your mutation because a family member was tested, you will need to do a confirming test through PC Project, so you will have your own report.

- Have not participated in a clinical research study in the past sixty days
- On a daily basis during the study, wear a wrist-based activity monitor and answer questions on a smartphone-based app.

Participants must be, in the opinion of the study doctor, able to understand the study, cooperate with the study procedures and willing to return to the clinic for all of the required visits.

You can find more information about the study and participating sites on www.valostudy.org.

If you are interested in participating, you can either:

- Complete the online form at surveygizmo.com/s3/4854812/Phase2-3
- Email PC Project at study@pachyonychia.org

BOSTON PATIENT SUPPORT MEETING
JUNE 20-22, 2019

IMPORTANT: If you are planning to attend this PSM, please book your hotel room BEFORE May 21, 2019. This is the deadline given by the hotel to receive the special rate. This is a great deal for a hotel in Boston. So please book soon! Meeting, hotel and registration details are available at: https://www.pachyonychia.org/2019psm/
This month is our Love Builders Giving Challenge. On 4.17 (or any day during this month), we’re asking our PC Project community to give it #All4PCProject. We need your help to reach our goal of 100 new monthly sustaining donors.

If we reach our goal, an anonymous donor will match every dollar. Your gift will have double the impact to help fight for a cure, connect and help patients and fund more PC research.

Become a monthly sustaining donor today and join the growing family of PC Love Builders at registry.pachyonychia.org/s3/LoveBuilders

Thank you to the following people who chose to celebrate their birthdays thinking of PC patients:

- Kali Bloedorn
- Mark Howell
- Kat Thangdora

Note: Kat set up a birthday fundraiser in honor of Cameron Baker, a PCer who passed away over a year ago. Kat wrote: “My best friend Cameron Baker had PC and despite the constant pain, he always maintained such a big, fiery heart. I wish I could do more to help. Thanks again!”

**Facebook Birthday Fundraisers**

Because PC Project is an accredited charity, there are zero fees when a person donates to PC Project through a Facebook fundraiser. Facebook fundraisers for PC are an excellent way to raise awareness and invite your family and friends to help support a cause that means so much to you.

**Walgreens Grant for PC Project**

Walgreens recently awarded PC Project a $15,000 grant to help support its programs and services for PC patients. This grant was made possible because one of PC Project’s Board of Trustees knew someone who worked at Walgreens. Thus, when PC Project applied for the grant, this contact was already aware of our organization. If you have connections at a company that offers grants or charitable sponsorships, please contact us and collaborate with us in applying for corporate funding.
PO Box 17850, Holladay, UT 84117 · www.pachyonychia.org · 801-987-8758 · info@pachyonychia.org

**Boston Patient Support Meeting**
**June 20-22, 2019**

IMPORTANT: If you are planning to attend this PSM, please book your hotel room by May 20, 2019 (that is today!) in order to receive the special discounted rate. This is a great deal for a hotel in Boston. So please book now! Reservations can be cancelled up to 72 hours in advance.

Meeting, hotel and registration details are available at: [https://www.pachyonychia.org/2019psm/](https://www.pachyonychia.org/2019psm/)

Currently, over 54 PCers, plus family members have registered to attend.

We can't wait to see you all there!

**Are You Ready to Talk About It?**

By Jack Padovano, Chair, PC Board of Trustees

June is Pachyonychia Congenita Awareness Month. It’s the one month every year where we join together as a community to raise awareness of PC and show the world how strongly we want effective treatments and a cure for this painful condition.

For this year, we’re asking a simple question: Are you ready to *Talk About It*?

For so many of us, living with PC, coming out to family, friends, strangers, about having PC and what it means for our daily life is very difficult. We worry about embarrassment, shame, people treating us differently, even concern that we may lose a relationship over something that we clearly had (and have) no control over. For me, coming out a gay man was actually easier than admitting to people I had PC. How silly is that? It’s only through the help of PC Project and people like YOU that I learned how to be brave and talk about my PC and how it impacts my life.

So, are you ready to talk about it? If you said YES, stay tuned for details on how you can participate in PC Awareness month and make a real difference in this fight against PC! If you answered no or you’re not sure, that’s ok too. There are plenty of ways to help PC Project move forward. Again, stay tuned!

**Pretty Nails for PC**

By Briannan Buchta, PC Patient

My name is Briannan and I have the K6a genetic mutation. This is something I would have never gotten clarity on if it wasn’t for discovering PC Project back in 2016. I’ve learned so much in such a short amount of time and I’m now able to connect to others around the world, thanks to our PC Patient Facebook chat. I have two small children that both also inherited the mutation and it has really motivated me to get more involved with contributing to finding treatments not only for them, but for all of us who suffer from PC.

June is PC awareness month and I really wanted to find a way to fundraise and take part in spreading awareness. I finally came up with an idea. I recently discovered Colorstreet Nail strips and for once I had a boost in confidence being able to make my nails pretty and feel good about showing them off. My whole life I would hide my nails and be jealous of how I couldn’t be part of the girls when they went to nail salons. So I signed up to be a Colorstreet Consultant and I am committing to contribute 100% of my personal commissions to the PC Project for charity. This is a way I am planning on fundraising and spreading awareness not only for June, but for as long as I continue to sell Colorstreet Nail strips.

Some PCers will be able to wear them just like I do, but I know everyone’s nail dystrophy is unique, so it may or may not work for everyone with PC. Regardless, Colorstreet can be for those with or without PC and every set sold will be contributing to PC. If you would like to learn more about these ColorStreet nail strips and how to order, I created a Facebook group called Pretty Nails for PC. Please join me and share with friends and family who would be interested. Thank you!

Link: [www.facebook.com/groups/Prettynailsforpc/](http://www.facebook.com/groups/Prettynailsforpc/)
PC Project hosted an impressive group of scientists, clinicians and drug developers from around the world who presented and collaborated on PC related research at the International PC Consortium (IPCC) Symposium in Chicago, May 7-8, 2019. With 18 speakers and even more interested researchers in attendance, PC Project is humbled to have this group on its team. As one key researcher said, “This is a special group and it’s a privilege to be part of it.”

After the IPCC Symposium, the PC Medical and Scientific Advisory Board, a smaller group that guides PC Project, met to discuss projects, goals and next steps for PC research. Following that meeting, PC Project had a table at the larger Society of Investigative Dermatology Conference in order to raise awareness and make new connections with interested scientists.
Reflections on the 2019 IPCC Meeting
By Jim Rittle, PC Board of Trustee

Last year, I had the honor of accepting the position of a PC Project Board of Trustee. (I’m still not sure what I’ve gotten myself into.) This year, I had the great privilege of attending my first International Pachyonychia Congenita Consortium Symposium or the IPCC Symposium. WOW, a mouthful and my head hurts. For those of you that don’t know, PC Project holds the IPCC Symposium just prior to SID’s annual meeting. SID is the Society of Investigative Dermatology and this was the 77th annual meeting. It is without a doubt a Who’s Who in dermatology from beginning research to end products.

So, let me back up just a little. Some of you may know my father was a spontaneous PCer. I got it from him and passed it on to my oldest daughter. I want you to keep that in mind and I’ll come back to it. The story is too long as to how I found PC or what I’ve done over the years. A few patient meetings, a couple biopsies, FDA meeting panelist, and this or that. Now I find myself standing in a ballroom at the IPCC conference with some familiar faces. Some with name tags and I know I’ve heard their name somewhere, at some time, associated with PC Project. Full disclosure - I’m horrible at remembering names but I’m great with faces. Please remember that the next time you see me.

The 16th Annual IPCC Symposium started off with our Executive Director Janice Schwartz giving very personal opening remarks and thanks to all the attendees. After that the first speaker started. Hmmm, I said to myself - this is kind of basic. I understand all this because I’ve googled PC and have been to patient support meetings. That lasted maybe five minutes, and then I’m treading water in the intellectual deep end for maybe a few more minutes at best before I go down. After that, the afternoon was snacking and drinking coffee to keep my brain energized and engaged to try to follow along as best I could.

Day one consisted of presentations on the latest research that had familiar words like keratin and genetics. There were new terms to me like iRhom2 and ASOs. Pain mechanisms, transmission, and quantifying were a few more. Day two moved away from research and was directed more towards methods, production, application, development, FDA approval, and finally to large scale production.

Most, but not all the presentations included or were about PC. However, even the ones not about PC have applications to PC. What struck me as the most impressive was that all these research approaches were different. If nothing else, know that some very smart people are out there researching multiple possible solutions for PC.

In case you were about to ask, money came up. Who’s paying for each step and if they invest are they going to make money? Or, can you get a government grant for the step your working on in the process? Money, is an issue, the whole way, every step. We as patients don’t like it, but there it is, and this is the real world.

REGISTRY, REGISTRY, REGISTRY, the topic came up a lot. The PC Project registry is the game changer. For an ultra-rare disorder our side, to have a database on all PC Patients is priceless. Researchers LOVE data and this PC data will live for generations to come, enticing scientists and drug developers. Researchers and investors alike get very excited about the registry. If you know someone, or have someone in your family who hasn’t registered, I understand why they haven’t - because of years of no help, no treatment, and misunderstanding from doctors. Please believe me when I tell you that the sum of the registry is so much more than its parts – it’s us.

We are not a numbers game and personal connections matter. PC Project has fostered relationships with researchers for a very long time now and it showed at the IPCC. Long ago it started with our dear Mary Schwartz and without knowing, but maybe hoping, she passed on those connections and relationships to Jan who currently cultivates them so well. So much so that some researchers have even become monthly donors. Researchers donate both their time and their resources to PC Project and even some of their students have donated. These are college kids that don’t have a lot who have given to PC Project. It seemed to me that the attendees and the people they work with have a personal connection with PC Project. It’s not just what they do, they really care.

Lastly, I want to bring you back to my daughter. What drives many of us is helping others. We’ve learned to live and cope with PC. In a way we’ve made our peace with it. I’m extremely guilty of this and so everything I do at PC Project is for her. I have...
heard patients say many times at patient support meetings, to summarize, “We’re hoping for a cure for the children”. Children and treatment came up many times during presentations at the IPCC. I was sitting next to Jan when, during a question and answer session, children came up again and again. Jan made a very simple statement to me and I don’t think she realized how powerful it was. She leaned over and said, “EVERYBODY, wants to help the children”.

PC Project is run by PC patients, PC families, and PC supporters. There is no other agenda.

The PC Project website says it all: Fighting for a cure. Connecting and helping patients. Empowering Research.

I’ll add, if you are or know a PC patient, please help us, and future generations of children, and join the IPCRR at pachyonychia.org/patient-registry/.

**PC Patients in USA Still Needed for Phase 2/3 Clinical Trial (VALO study)**

PC patients in the USA are still needed for the clinical trial that involves evaluating the safety and effectiveness of PTX-022 (experimental topical rapamycin).

**NOTE: If you have already signed up for this study, you do NOT need to sign up again.**

PC Project supports this clinical trial. On the following page is a brochure with more trial details. This includes an overview of the study and the fact that travel assistance will be provided for the participating patient and one companion.

Below is the approved announcement for the trial recruitment.

**Seeking Participants for Pachyonychia Congenita Clinical Research Trial**

If you or someone you know has Pachyonychia Congenita (PC), you may be interested in participating in a Phase 2/3 clinical research trial for an experimental therapy that is applied to the soles of your feet.

Some of the requirements for participation include:

- Must live in the United States
- Must be at least 18 years of age or older
- Have a clinical diagnosis of PC, genetically confirmed to involve any of three keratin genes KRT6A, KRT6B, or KRT16
- If you have not been genetically tested, or if you do not have a copy of your genetic testing report, please contact PC Project at study@pachyonychia.org for more information about genetic testing.

**NOTE: If you know your mutation because a family member was tested, you will need to do a confirming test through PC Project, so you will have your own report.**

- Have not participated in a clinical research study in the past sixty days
- On a daily basis during the study, wear a wrist-based activity monitor and answer questions on a smartphone-based app.

Participants must be, in the opinion of the study doctor, able to understand the study, cooperate with the study procedures and willing to return to the clinic for all of the required visits.

You can find more information about the study and participating sites on www.valostudy.org.

If you are interested in participating, you can either:

- complete the online form at surveygizmo.com/s3/4854812/Phase2-3
- email PC Project at study@pachyonychia.org
WHAT IS PTX-022?
PTX-022 is a topical formulation of rapamycin, under development by Palvella Therapeutics to treat Pachyonychia Congenita (PC). Rapamycin has been shown to inhibit the production of a kinase called mTOR, which is involved in the production of certain keratin proteins. In disorders such as PC that are associated with overproduction of aberrant keratin proteins, the inhibition of mTOR may decrease the translation of mutant keratin-expressing genes and potentially block signaling pathways linked to skin fragility, keratoderma, pain and ambulatory impairment.

FOR MORE INFORMATION
For more information on VALO, PTX-022 or to inquire about participating in VALO:
- Contact PC Project (www.pachyonychia.org)
- Visit the VALO study website at (www.valostudy.org)
- Visit clinicaltrials.gov (NCT03920228)
- Visit www.palvellatx.com

PC PROJECT AND PALVELLA THERAPEUTICS PARTNERSHIP
The VALO study is Sponsored by Palvella Therapeutics in collaboration with the Pachyonychia Congenita Project. Since its founding in 2003, PC Project has connected PC patients, researchers and physicians throughout the world in a united effort to help those with PC. PC Project maintains the only PC patient IRB-approved registry, through which patients can access the most up to date advancements in research and receive free genetic testing.

VALO is a multi-center, Phase 2/3 study, evaluating the safety and effectiveness of PTX-022 in approximately 60 adults with moderate-to-severe Pachyonychia Congenita (PC) with either KRT6A, KRT6B or KRT16 mutations.

VALO STUDY
Overview of Study Visits*

1. STUDY PERIOD 1: SCREENING

4 WEEKS

- 1 clinic visit
- Informed consent
- Eligibility check
- Lab samples
- Start daily diary and wearing activity monitor
- Continue normal routine (foot care and activity level)
- Optional video interview

2. STUDY PERIOD 2: OPEN LABEL

12 WEEKS

- 3 clinic visits
- 2 home nursing visits
- Eligibility check
- Daily dosing with PTX-022
- Continue daily diary and activity monitor
- Continue normal routine
- Lab samples
- Optional video interview

3. STUDY PERIOD 3: RANDOMIZED, DOUBLE-BLIND

12 WEEKS

- 3 clinic visits
- 2 home nursing visits
- Eligibility check
- Twice a day dosing with either PTX-022 and/or placebo
- Continue daily diary and activity monitor
- Continue normal routine
- Lab samples
- Optional video interview

4. STUDY PERIOD 4: FOLLOW-UP

8 WEEKS

- 2 telephone calls
- No longer taking PTX-022 or placebo
- Phone call from study doctor to see how you are doing
- Continue daily diary and activity monitor, return at end of study
- Continue normal routine
- Optional video interview

SUB STUDY
If you are not eligible to enter the Open Label study period, you may be invited to participate in a 28-day sub-study that will look at the amount of PTX-022 in the blood.

28 DAYS
- 2 clinic and 4 home nursing visits

TRAVEL ASSISTANCE
Travel assistance will be provided for participants in VALO and up to one caretaker to accompany them. Transportation (airfare, trains, car services) and housing (hotels) will be booked for you through a travel service that specializes in helping participants in clinical trials.
TOGETHER, WE ARE STRONGER THAN PC

For PC Awareness Month this year, we are focusing on the fact that while we may feel isolated and sometimes discouraged while living with this rare, painful disease, together, as a PC community, we are stronger than PC.

Some of the questions we’ve been asked to consider this month are:
- How can we be, and stay, strong in the face of PC?
- Why should I share my PC story?
- Am I ready to talk about it?
- Why does my involvement matter?
- Why should I give?
- How are we stronger than PC?

We have already received feedback from many of you from this campaign. We’ll published some of your experiences in next month’s newsletter.

And to all of you who have already held events or made efforts to increase PC awareness this month—thank you! Because of you, we ARE stronger than PC!

ARE YOU A PC WARRIOR?

By Jim Rittle, PC Patient & Board of Trustees

While PC Project has existed, there have been more than a few people who have quietly gone about their lives, business, and work. In the circle of life they have hovered over and around PC Project and Pachyonychia Congenita in many ways. Always keeping PC in mind, helping when they can, forwarding the cause, and bringing awareness to PC whenever possible. Some of these people existed in their roles even before PC Project was formed.

I’d like to introduce you to a new term I’ve recently been introduced to - a PC Warrior. A PC Warrior is defined as a person who gives of themselves in the battle against Pachyonychia Congenita in any and every way they can. They do not cover one simple title, but many. They are strong in spirit, body, and mind, knowing this race is not a sprint but a marathon. At each opportunity they give something of themselves to chip away at PC. Everyone knows that many personal battles have been lost to PC. But that does not in any way mean we give up. This fight takes many forms but follows the same pattern. We get knocked down and we rest..., we treat our wounds and we heal...., and then, oh yes then...,WE STAND UP AND FIGHT AGAIN.

Patient or family member, have you completed the registry, do you advocate, have you donated or raised funds? Have you done everything you can do to support this cause?

Life always throws us curves. But, do you circle around and come back to PC, or do you only think about it when it’s convenient for you? Pachyonychia Congenita is a genetic, sometimes generational, and relentless problem that has proven itself a worthy adversary and will not go down without a fight.

What more can you do to become a PC Warrior?
June is
Pachyonychia Congenita
Awareness Month

Pachyonychia Congenita (PC) is an ultra-rare genetic skin disorder.

Chronic Pain is one of the distinct characteristics of PC. Painful calluses and blisters form on the feet and the hands.

Currently there is no specific treatment or cure for this condition.

More awareness and more research is needed to help these patients. For more information, see the excellent work the PC Project group is doing via their website.

www.pachyonychia.org

#PachyonychiaCongenita
#StrongerThanPC

know chronic pain
**Birthday Fundraisers for PC**

Happy Birthday and a huge thank you to those who celebrated their birthdays by creating a Facebook fundraiser for PC:

- Addy Gaston
- Debbie Gregory
- Linda Harvey
- Amanda Lawrence
- Kimberly Noemi

We've noticed that those who make the fundraisers personal get a lot of support from their followers.

Here's an example of Debbie Gregory's Facebook fundraiser description:

“For my birthday this year, I'm asking for donations to Pachyonychia Congenita Project. I've chosen this charity because this is the condition that my beautiful Ellie lives with every day. She is the bravest person I know, despite living most days in pain she does not let that stop her and lives her life to the full.

“The PC project have been fantastic for Ellie, supporting her (and us), connecting us with other families as well as continually looking for a cure and I hope that you'll consider contributing as a way of celebrating with me.”

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**PC Patients in USA Are Still Needed for the Phase 2/3 Clinical Trial (VALO Study)**

Study centers across the USA are still accepting PC patients for the clinical trial that involves evaluating the safety and effectiveness of PTX-022 (an experimental topical rapamycin). Anyone over 18 who lives in the continental US and who has a known mutation in K6a, K6b and K16 should fill out the interest form.

If you are interested in participating, you can either:

- complete the online form at [surveygizmo.com/s3/4854812/Phase2-3](surveygizmo.com/s3/4854812/Phase2-3)
- email PC Project at [study@pachyonychia.org](mailto:study@pachyonychia.org)

**NOTE: If you have already signed up for this study, you do NOT need to sign up again.**

PC Project supports this clinical trial. You can find more information about the study at [www.valostudy.org](http://www.valostudy.org). Details will include an overview of the study, clinical trial sites, travel assistance for the participating patient and one travel companion and compensation for participation.

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**Supporting PC Via AmazonSmile**

PC Project continues to receive a small percentage of every purchase you make through AmazonSmile when you designate Pachyonychia Congenita Fund as the charity of your choice. AmazonSmile is available in the USA and also in UK. Happy shopping!
The 18th Annual Pachyonychia Congenita Patient Support Meeting (PSM) was held June 20-22, 2019 in Boston, MA. Sixty-three PC patients, along with their loved ones, clinicians, scientists and drug developers spent a day and a half learning from one another about how to best live life with PC.

A big thank you to our PC advocates and other volunteers who helped with registration, gave presentations, and led group discussions, including teen and children’s discussions.

Some program highlights included having patients group according to their affected gene. Patients who wished to could show visiting clinicians their feet and other PC symptoms. This educated physicians about PC in a profound and easy way.

Other program discussions and topics included PC research, clinical trials, PC pain, the social and emotional impacts of PC, being a loved one of a PCer, taking control of our health, how to help children and teenagers navigate life with PC, and being stronger than PC.

Special Report: Boston Patient Support Meeting
By Jennifer Hart

My doctor once told me I was his worst and best patient all rolled into one. What a compliment he was giving me! Let me elucidate. He said I was the worst because I wouldn’t sit down but the best because I’d never let PC beat me. How right he was!

Having struggled with pain from my PC over my entire lifetime, I had become resigned to just dealing with this as best I could. I had fallen off the radar of any specific medical care for the condition, but was satisfied that the podiatry treatment which I receive was the best support which I could expect. Then my daughter introduced me to the PC Project! My daughter Rebecca, who does not suffer from PC, works in Clinical Research so is very pro-active about all things new in human medicine. However, having been shown some of the information about the group by her, I still felt a fair amount of trepidation about joining as I did not want to get embroiled in a group just feeling sorry for themselves. This is, of course, as far from the case as is possible from the attitude of my fellow PC’ers who I met at the recent patient meeting in Boston. We are indeed warriors and determination is in our DNA!

I decided to book to attend the meeting, very much persuaded by my daughter that I would never know if I didn’t try. I’m not sure when my daughter started quoting me, I’ve used that phrase so many times in her childhood! Although having received nothing but warmth and welcome from both Jan and Holly in advance within various emails, I must admit to still feeling very nervous as I arrived at the venue. I shouldn’t have worried. The PC ambassador group at the desk was wonderful, and I immediately felt empowered by the possibility of what I was to hear and learn.

Over the next two days we enjoyed a variety of sessions, some informative and tutorial, some Q&A and others patient discussion groups. The mix was excellent. I found myself laughing out loud on the first evening when I realized that we were ALL limping. Now I felt “normal” and not the odd one out. This was in fact the first time in my life I had actually met a fellow PC’er.

So, my experience attending my first PC Project Patient Support Meeting has been positively life changing for me. I definitely have made new friends, but have also realized that I’m not alone in continuing to remain the best and the worst patient!

The last two pages of the NewsBrief contain a collage of photos from the Patient Support Meeting. Enjoy!
STRONGER THAN PC
AWARENESS MONTH

Thanks to all who bravely shared PC stories, raised funds or made donations during PC Awareness Month. The Block family hosted their annual ice cream awareness and fundraising social. Briannan Buchta sold Color Street nail products to raise awareness and funds all month long in her clever “Pretty Nails for PC” campaign.

Steve Hall is doing a “Run, Jog, Walk 100 Miles” in August to raise funds for PC in honor of his daughter, Sara, who suffers from PC. Read more on his JustGiving Fundraising page at https://www.justgiving.com/fundraising/Steve-Hall35

Many PCers also shared their stories on social media. Letting others know they have PC is not an easy thing to do. The interesting thing is not one person had a negative experience. In fact, many PCers were pleasantly surprised at the positive responses they received.

Michael McCullough shared his story on Facebook, along with some photos of his feet and reported, “My Facebook responses have been awesome. Some of my friends have never seen my feet before and have been blown away by the photos. Some of them have shared my story. One woman in particular shared the story with both her daughters who are in the medical field.”

In his shared story, Michael first gave an explanation about PC. Then he told about life with PC. Here is a portion of what he shared:

I want to discuss a typical day in the life of someone with Pachyonychia Congenita. Each day can be very different. We have good days and bad days. Good days are those when some of us can walk and stand with mild discomfort. There is never a wonderful day. We are limited to the amount of time we can stand or walk. Running is out of the question. I played baseball when I was a kid. For me, hitting a double was torture. I could make it to first base with some speed but the resulting effort brought on a burning sensation in my feet. To have to run to second base wasn’t possible, the burning was too intense. To put it in real terms, if my life depended on it, I could probably run a block.

When I attended grade school, getting new shoes was an awful time. In the early 1960s, the dress shoes were terribly hard unlike the materials they use today. You literally had to break them in over a number of weeks. That was a bad experience. I cannot be thankful enough for the shoe manufacturers of today’s footwear. What a difference they make.

From day to day, we never know what kind of day it’s going to be. Sometimes, in bed, the weight of a simple top sheet or comforter over your feet is unbearable. I cannot tell you the number of times I’ve hung my feet out of the bed to sleep in peace. High humidity, rain, and snow are usually a bad day. In my case, the skin surrounding the calluses gets very taught. Each step stretches the taut skin making it feel as if it’s about to tear. In some cases, this actually happens. Sometimes a hot shower will soften the taut skin enough to get you some temporary relief.

I always walk with my head facing down to the surface I am walking on. Concrete, steel, and wood flooring are the safest for me. Natural grass or gravel is brutal to walk on because you cannot judge what’s underneath you. I look down because I need to make sure I am not treading on a stone. I need to see what’s in front of me; otherwise, I can trip or fall...
because one callus took the brunt of landing on a stone. For me it’s all about balance, in other words, spread the pain across the whole foot rather than isolating it on one or two calluses.

I mentioned earlier that my feet require maintenance. Even though I consider the callus dead skin it actually grows and thickens if not trimmed after so many days. I usually trim them every 7 to 10 days using single-edge razor blades. Some of the patients use Dremel tools or power grinders to maintain their feet.

Although my dad was well aware of our issues, he never told me not to play baseball or football. My mom also never let me feel like I couldn’t play like the other kids. So I did, and over time I built a tolerance for pain that I could handle. That changes as you age. It becomes much harder to do what you did in the past.

PC Project has been making a real difference in our lives. They have brought on many scientists and physicians from all over the world in an attempt to give us some relief and possibly a cure. If you have any interest in getting more information please check out the following addresses:
info@pachyonychia.org or www.pachyonychia.org

Another story was sent to PC Project to be added to the website by the parent of a PCer named Olivia. You can read Olivia’s story in English and in Spanish on the website at
https://www.pachyonychia.org/patient-stories/olivia-castro-lay/

Not only did Olivia’s family send photos of Olivia smiling and doing things she loves, but they also sent a collage of what affects her the most at her current age—the nail infections. Thank you to Olivia’s family for sharing.
PHOTOS FROM THE 2019 BOSTON PC PATIENT SUPPORT MEETING
VALO PHASE 2/3 CLINICAL STUDY... 
PCERS AND PC COMMUNITY 
CONTINUE TO STEP UP!

By Emily Cook and Wes Kaupinen

While studies of experimental treatments for rare diseases almost universally struggle to identify enough patients willing to enroll in a particular clinical study, individuals with PC and the collective PC community have distinguished themselves through a very impressive level of participation in the VALO Phase 2/3 study of Palvella Therapeutics’ investigational PTX-022 (QTORINTM rapamycin) for pachyonychia congenita (more information on the study here: [clinicaltrials.gov/ct2/show/NCT03920228](https://clinicaltrials.gov/ct2/show/NCT03920228)).

As of mid-August, the VALO study has enrolled more than 30 PC patients in only a few weeks since the study officially commenced. To put this into perspective, many studies of experimental treatments for rare diseases take months, and in some cases years, to achieve this level of clinical study participation.

The ability to achieve this level of participation is a testament to each and every PC patient enrolled in the study (who, along with their families, are in many cases traveling by plane to clinical site visits), the many dedicated clinician investigators who have prioritized PC patients and this study in their busy clinical and research practices, and the study coordinators and managers at each of the clinical sites for their tireless efforts to manage every detail of the study. PC Project also deserves special recognition for all that it’s contributed, ranging from its efforts to help finalize the design of the study to its ongoing efforts in communicating with individuals across the country who have expressed interest in enrolling in the study.

Importantly, the VALO study is continuing to enroll up to 60 patients across the U.S. If you or a family member are interested in learning more about participating in the VALO study, please visit the following link: [pachyonychia.org/valo](https://pachyonychia.org/valo)

In summary, thank you to all of those individuals who have made valiant efforts, and in many cases significant sacrifices, to participate in this study.

**IMPORTANT NOTES ABOUT THE TRIAL**

- If you are curious about participating in the trial, NOW is the time to complete the interest survey at [surveygizmo.com/s3/4854812/Phase2-3](https://surveygizmo.com/s3/4854812/Phase2-3). This is not a commitment to participate, merely an opportunity to discuss with a trial coordinator more about the trial, including the fact that trial expenses are covered.

- If you have already completed the survey and haven’t yet been contacted by a trial site, don’t worry. You will be contacted soon and do not need to resubmit the survey.

- For those who didn’t progress further than the first screening in the trial, please know your participation was and is still important to the trial as a whole.

- For those at any stage in the study, please remember this is a double-blinded study. All information about your situation needs to be kept confidential and not shared with others in the PC community in order to keep each person’s experience as unbiased as possible.

- To those unable to participate for any reason including age, location, language barriers, etc., we appreciate that you would be in the study if you could. We know your heart is with everyone participating on behalf of those who cannot.
More patients are still needed. Below is the official IRB approved recruitment announcement as a reminder of who can currently participate in the VALO trial:

**Seeking Participants for Pachyonychia Congenita Clinical Research Trial**

If you or someone you know has Pachyonychia Congenita (PC), you may be interested in participating in a Phase 2/3 clinical research trial for an experimental therapy that is applied to the soles of your feet.

Some of the requirements for participation include:

- Must live in the United States
- Must be at least 18 years of age or older
- Have not participated in a clinical research study in the past sixty days
- On a daily basis during the study, wear a wrist-based activity monitor and answer questions on a smartphone-based app.
- Have a clinical diagnosis of PC, genetically confirmed to involve any of three keratin genes KRT6A, KRT6B, or KRT16

If you have not been genetically tested, or if you do not have a copy of your genetic testing report, please contact PC Project at study@pachyonychia.org

NOTE: If you know your mutation because a family member was tested, you will need to do a confirming test through PC Project, so you will have your own report.

Participants must be, in the opinion of the study doctor, able to understand the study, cooperate with the study procedures and willing to return to the clinic for all of the required visits.

If you are interested in participating, complete the interest survey at surveygizmo.com/s3/4854812/Phase2-3

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**PC and Career Choices**

Many of our PC friends are young adults, many who are attending trade schools and colleges, and who are thinking about their future jobs. Because of their unique skin condition, PCers often plan ahead, and planning ahead about career choices certainly is wise. There are a wide variety of careers which PCers choose. Sometimes it takes a little thinking outside the box to find a career choice that is right for a person with PC.

Forbes.com had an article about planning for careers. (https://www.forbes.com/sites/ashleystahl/2018/08/29/3-steps-to-develop-your-career-plan/#65aa01d34910). The article suggested the following:

1. **Self-reflection.**
   Besides reflecting on your career path, you should also reflect on yourself and your values, skills and passions. When you begin to understand yourself and what you want, you’ll be able to more easily create a plan that suits your goals and your lifestyle. (For a PCer, this may mean thinking realistically about the pain you currently experience and whether or not your PC will stay the same or get worse.)

2. **Goal setting.**
   Setting goals is the key to a successful career plan. SMART goals are specific, measurable, attainable, relevant and timebound goals that all set you up for success in your goal setting and achievement. Goals should be written down and shared with a friend or coworker.

3. **Develop a plan.**
   After assessing your values, skills, and passions, and after setting SMART goals, now it’s time to really dig into developing a plan to get there. It’s time to make career decisions. Here are some ways you can make those difficult decisions:
   - Make a pros and cons list.
   - Evaluate how each path aligns with your values.
   - Think about the future consequences of each path (including how it fits with the way PC affects you).
One advantage of being in the PC community is the opportunity to discuss career options with other PCers. Some are able to stand for substantial amounts of time and walk fairly long distances. Others who are not able to stand or walk have chosen more “sit down” jobs, whether they be desk jobs or car/truck jobs. Others are able to get accommodations for their work. For example, a teacher may teach using a stool to sit on, or an officer worker may find a foot rest under a desk to make their feet more comfortable. Others work from the comfort of their homes where they can keep their shoes and socks off and rest their feet while they work. For others, PC is so debilitating they are on government disability. There really is no comparing when it comes to what each person with PC can and cannot do.

How has PC affected your career or your plans for a career? Are there adaptations that can be made for you to have a career you didn’t think possible? We want to hear your career experiences, especially if you have advice for the younger generation in planning for careers as a person with PC. Please share your stories by email info@pachyonychia.org.

**Career Success: from the PC Mailbox**

Steve Clark, a PCer, has chosen a career in which he can excel in and stay mostly off of his feet. Steve’s mom, Linda Clark, shared with PC Project Steve’s recent career successes:

“Just wanted to share a huge event that our son, Steve achieved in. He is a truck driver, pulls triples, and was competing in the Utah State Truck Driving Competition. He won first place in his division, triples, and then won first place in the entire competition! Here are the two belt buckles presented to him for winning first place and Grand Champion:

Steve is now representing Utah at Nationals, being held in August in Pennsylvania. He has tried for 8 years, always winning in his division, but this is the first time winning the whole shot! He never gave up, a lesson for us all.”

**Congratulations Steve for excelling in your chosen career! And good luck at Nationals!**

**RARE Champion of Hope**

PC Project was nominated with 200 other organizations and individuals as a Rare Champions of Hope. Only nine were selected and one of those was PC friend and advocate, James Valentine, attorney at Hyman, Phelps & McNamara, PC.

James’ efforts have been influential in helping PC Project progress on the pathway to effective treatments. We first met James in 2017 at the Salt Lake City Patient Support Meeting where he led a meaningful patient-focused discussion to begin guiding efforts to learn what life is truly like for a PC patient and what a meaningful treatment would look like from a patient’s perspective.

James later served as our guide and moderator at the critical 2018 EL-PFDD meeting, where even more patients shared their voices with key FDA officials. He’s since helped with PC focus groups and patient surveys. He attended and presented at both the 2018 London and the 2019 Boston Patient Support Meetings, learning from patients and in turn, helping our patient community understand the basics and the importance of clinical trials.

James continues to advocate for PC Project and PC patients, referencing our organization on panels, social media and in other forums, all the while helping to guide the process of the current Phase 2/3 VALO Study.

Congratulations, James, for fighting for PC patients! You truly are a RARE Champion of Hope for us!
Opportunity to help PC Research

Thank you to all PC patients who are part of the International PC Research Registry (IPCRR) and who have already participated in the recent registry addendum questionnaire. Over the weekend after first sharing it with you, we received more than 130 responses and completed surveys continue to pour in. This kind of support is the reason PC research is moving forward.

If you have PC and have NOT yet participated in the short survey, please do so now at: registry.pachyonychia.org/s3/8HS-PC

This is a special opportunity to help the scientists and physicians explore additional aspects of PC that are not on the original questionnaire. Some of the survey questions may not apply to you. Simply answer each question the best you can, all the while knowing you are furthering the cause of PC.

Again, thank you for your participation. Your answers are extremely valuable.

VALO Phase 2/3 Clinical Trial Update

The trial continues to progress with many patients at different stages. The trial is close to full recruitment. Only a few more participants are needed before the study is completely full.

Remember all costs of travel are included for each patient and one companion, plus a small compensation for participation. If you have K6a, K6b, or K16, live in the USA, are fluent in English and are over age 18, please fill out the interest survey here: surveygizmo.com/s3/4854812/Phase2-3

Once you complete the survey, you will be contacted by a site coordinator to discuss the trial details.

PC Project and Le Coeur Au Pied Represented 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 (MSAB). In addition, they received the contact information of new researchers who are interested in being connected to PC Project.

Marie and Alice are perfect examples of volunteers in the PC community helping and making a difference in their own area of the world.

Note: Le Coeur Au Pied is a group for French PCers which supports PC Project. The president is Sylvie Cierpucha. This group is exceptionally effective in patient recruitment for the IPCRR patient registry, information sharing and fundraising for PC Project.
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

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.”

Read the full interview here:
globalgenes.org/2019/09/12/rare-leader-janice-schwartz-executive-director-pachyonychia-congenita-project/

Facebook Birthday Fundraisers

Happy birthday to Briannan Buchta, a PC patient and to Morgan Eads, sister to Ryle Defebaugh, who has PC. Both women set up a birthday Facebook Fundraiser for PC Project.

This is a simple and easy way to raise funds for PC. Just go to the Pachyonychia Congenita Project Facebook page: facebook.com/pachyonychia/ then click “Fundraisers” and you’ll find a link that takes you step-by-step in creating your own fundraiser. When a fundraiser is created for PC Project, an official charity, there are zero donation fees. And, when a person writes specifically why they create the fundraiser, their friends and loved ones respond.

In Briannan’s case, she wrote: “It’s officially my birthday month. I know so many of you donated last year and also supported my fundraiser. It is so appreciated. Well it’s that time again for my birthday fundraiser. The PC project is something that hits close to home for me and my family who suffer from PC. Any little donation helps. Thank you all so much!”

Month after month, our hearts are filled with gratitude for the members of our PC Love Builders group. These are donors who give an ongoing gift each month for PC. Thanks to each one of you for being part of the solution to help others with PC and to advance treatments. There is no way this work could go on without you!

To set up a monthly donation to PC Project, go to the donation page on the website, pachyonychia.org/donate-help/ select a donation platform and then choose to be a monthly donor.
FROM THE PRIVATE PC PATIENT
CHAT GROUP ON FACEBOOK:
HOW TO DEAL WITH BULLIES

Because only 500 members of the PC community are part of the private PC Patient Chat group on Facebook, we will occasionally share posts in the PC News Briefs.

The following question was recently asked of the group: “Any ideas of things you have said to bullies or would recommend PC kids say to bullies when teased about their PC?”

Here are some answers to consider, especially now that many PC children have headed back to school:

♥ I’ve been thinking about this since first reading the question and dealing with truly malicious bullies is one thing, where kids probably shouldn’t engage and should just laugh, turn around and walk away. But most kids aren’t malicious, just ignorant. They might hold their nose and say “Eew! Gross!” when they see PC feet. If this happens to you, you can just say “It’s not gross. It’s just different. It’s called PC and sometimes it really hurts. If you want I can show you more and tell you all about it.” Try not to believe anyone who says it’s yucky, disgusting or gross (or that you should just take care of your feet more). Remember that’s it’s not your fault that you have this and feel sorry for anyone who feels so insecure that they feel like they have to belittle you for your condition.

♥ We have different color eyes, different kinds of styles, different types of nails. What’s the difference?

♥ As a child I never really handled bullies in a way I would condone others repeating. However, as a parent of a PC child I take a different approach. Every year I have a meeting with his new teacher first and explain it to her. Then I ask her when would be a good time to come in and talk to the class about it. I go up and introduce myself and tell them we’re going to learn some super advanced biology today. Stuff that’s usually for big kids to learn but that they’re special so they get to learn it now. I tell them about PC, how it happens and etc. Just the basics in terms that are kid friendly. Then I tell them my son has it just like I do and explain a little about how it looks/feels and what it’s like to have it and live with it. When I’m done I give them the chance to ask me questions and share how they feel about it. The only year he was ever teased was kindergarten because that was the only year I didn’t do it. He’s going into 4th grade now and we’ve never had another problem with bullying.

♥ Probably nothing I should repeat.

♥ That’s a great way to approach it. Unfortunately, I think my daughter would die of embarrassment if I did. She hates it being mentioned. I just print the brochures off the PC website and give to any new teachers. My son is still young enough that he’s not worried about being embarrassed just yet but the time is coming I’m sure!

♥ I’m beautifully different and beautifully me. I’m so unique that there’s only 792 people that we know of in the world like me! I can’t control the fact that I was born different, but you can control the way you treat me!
I got this a lot so I feel like it should be addressed... “No, I’m not contagious. Yes, you can play with me.”

For me, I’ve noticed the issue is that it’s not often solely bullies, but other kids who are trying to make sense of things and end up saying something flippant or ignorant which embarrasses the PCer. There are also those who are insecure and put other people down and choose the more painful words to get the most impact. Things I’ve heard are that I have “claws”, “oh those are like dog’s nails” and “your nails are dirty”, in which case, I’ve never come up with a good retort as a kid. In essence, I never tried to educate or correct many because to me, someone who was so direct with assumptions wasn’t someone I wanted to be around. As an adult I’ve practiced; “If you ever want to be my friend, we can talk about why you’re wrong about my nails but otherwise, back off.” Some kids also trick you to showing your nails by playing games where they compare hands or feet. Kids can practice learning how to NOT engage in things they feel uncomfortable doing. I think it helps to rehearse phrases so it becomes easier because usually we are all taken by surprise when people make fun of PC. You can practice 3 very different responses that go from kind or dismissive or educational depending on what the child wants out of the interaction.

As a mum of a son who had PC, a wife to a husband who has PC and of other family members I find it quite difficult when my son is upset by what another child had said about his “nails being dirty” and “why are they like that” I used to get annoyed at children who had previously asked these questions several times but still felt the need to draw attention to different areas. My husband is still very sensitive about this at times as he had bad experiences as a child at school. It may be easy for me to say as a non-PCer, but I want my son to be proud of who he is. I say everyone is different in their own way. We are all very inquisitive, especially children, and usually give him an example of when he himself has asked question about someone he has seen who looked different for whatever reason. I try to play it down without ignoring the situation and find a brief explanation to close friends is often enough. My son has a close group of friends who he feels happy to take socks off in front of when at swimming. As he says, they are my mates and look out for me. Definitely explaining the situation to teachers is a help and I believe encouraging the child to be confident to explain a little themselves too. My son will soon be moving to high school so am worrying what he may come across when needing to be more active whilst enduring huge blister and cracks in skin that seem so deep and the worst “festered fingers” as we call infected nail beds. Meeting new kids, older kids can be more ignorant and less understanding of differences fingers crossed all will be well and we have done enough to make him feel confident in himself.

I come from a family with 7 kids. 6 of us have PC we were all called nasty nails growing up. As a kid I would walk right up to other kids calling me nasty nails and tell them to go ahead and make fun of me for something I was born with and can’t change. Most kids wouldn’t know what to say or do and just awkwardly laughed and walked away.

I had a VERY deep and raspy voice until I was about 12. So of course, I was called man voice along with nasty nails. Speaking up and explaining that my voice was odd because of a medical disorder and that they can feel free to make rude jokes had a tendency to stop kids.

We would love to learn about your experiences and thoughts about how to handle bullies as a person with PC to share on the website. You can email them to info@pachyonychia.org.

If you want to be in the Facebook Pachyonychia Congenita Patient Chat private group, you must join the Registry or be a close family member to a PC patient. Because many PC patients hide their PC, this is a safe place for PCers to gather and share tips and how they live life with PC.
PC News Brief

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Connecting Around the PC World

PCers Jennifer Hart and Natalie Kendrick met through the Pachyonychia Congenita Patient Chat, a private Facebook group where those with PC share thoughts and tips about living with PC. [https://www.facebook.com/groups/pachyonychia/](https://www.facebook.com/groups/pachyonychia/)

Jennifer and Natalie discovered they only live five miles away from one another in the UK and arranged to meet in person. As they talked, they discussed ways to increase the profile of PC Project in the UK and the importance of everyone with PC joining the Registry.

These women demonstrate the power of PC Project growing globally. If you want to be in touch with PCers who live near you, please contact PC Project and request we share your contact information with PC patients in your part of the world.

PC Project at the AADA Annual Legislative Conference

PC Parents Barbara Feinstein and Aaron Klein (their daughter Audrey is K-17) represented PC Project at the American Academy of Dermatology’s Annual Legislative Conference, held September 8-10 in Washington, DC. The meeting brought together more than 200 dermatologists and representatives of patient advocacy organizations to lobby Congress on issues affecting dermatology and patients. In addition to raising awareness of pachyonychia congenita among Members of Congress, doctors and patient groups, Barbara and Aaron also pressed legislators, including Representative Jamie Raskin (D-MD) [See photo below] to prioritize funding for rare diseases.

Shopping For PC

As the holiday shopping season approaches, don’t forget that when you use AmazonSmile to make your purchases, you can choose to support Pachyonychia Congenita Fund and Amazon will give PC Project a small percentage. Every bit counts. Thank you for supporting Pachyonychia Congenita Fund by shopping [smile.amazon.com](https://smile.amazon.com) or [smile.amazon.co.uk](https://smile.amazon.co.uk)

Another Way to “Donate”

We continue to collect PC teeth. The National Institute of Health has an on-going study on PC teeth. If your baby teeth come out or you are having any teeth or molars extracted, please save them and send them to PC Project. This month’s tooth donor award goes to Caidan Martinez, a 7 year old PCer, who donated two of his baby teeth this month. Thank you Caidan!
PC Project Board of Trustees Welcomes New Board Member Kay Dee Holmes

Kay Dee was reared in Idaho Falls, Idaho and received an Associate’s degree from BYU-Idaho. But, her schooling took her to the other two “corners” of the state of Idaho. She received a Bachelor’s Degree in history from Boise State University and a law degree from University of Idaho. She is licensed to practice law in Idaho.

Currently, Kay Dee works for the University of Idaho negotiating contracts and ensuring compliance with state and federal law. But, she is especially passionate about drafting contracts in plain English and eliminating legalese. In her free time she enjoys being outdoors, road trips, reading, studying history, and binge watching a good TV show or watching movies (when she can find one she enjoys!).

PC Awareness

Supporting PC Project comes in many forms. PC Patient, Candida Torres DeJesus taking PC awareness to a whole new level (see photo).
Phase 2/3 VALO Study: Frequently Asked Questions

“Do I get paid for being in the study?”
Yes, you are paid for your time and effort. The amount is up to $1,200 if all visits and assessments are completed.

“How can I participate if I can’t take time off of work to travel to clinic visits?”
At the end of September 2019, a new site opened that can see VALO patients on the weekends. If you would like to participate at this site, you can contact PC Project.

“Does the study provide financial reimbursement for babysitting?”
Yes, the study provides reimbursement for respite care (care for children, spouse, seniors, animals, etc.). You will have to provide receipts or documentation.

“The nearest site is 3 hours away, too short to fly. What if I can’t drive, don’t want to drive and/or don’t have someone to drive me?”
The study provides transportation to and from the clinical site for you and a companion. In this case, a car and driver would be provided to you to assist with the round trip. The study also provides financial support for transportation (train, bus, gas mileage, tolls, airfare), lodging and other costs incurred as a result of study participation.

“How can I find out if I might be taking a medication that I can’t take during the study?”
First, don’t stop any medications that have previously been approved by your doctor. If you are interested in participating, please contact PC Project so they can connect you with a clinical trial clinician that can answer your questions to see if you are a candidate for the study.

“When will study enrollment close?”
Study enrollment may close as early as the end of 2019….but we need more patients! Please complete the short form at https://www.surveygizmo.com/s3/4854812/Phase2-3, if you are interested in participating to get started.

“What if the study drug works for me during the study, can I still take it after I finish the study?”
An extension study will be opening at the beginning of 2020, which will allow any patient that received study drug in the VALO study, and the patient and clinician believed helped the patient, to continue to use experimental PTX-022 (QTORIN™ topical rapamycin). The study drug will be provided at no cost to the patient.

For more info:
valostudy.org
Clinicaltrials.gov
NCT03920228

Version 1.0 / 22Oct2019
Do you know someone who has PC? A family member or friend?

Are they 18 years or older?

Are they interested in helping to test a potential first treatment for PC?

We need your help to inform your friends and family about the opportunity to take part in our VALO clinical trial and help us to treat PC in the future. Participation is voluntary.

Please invite your family member or friend to sign up for the study here: https://www.surveygizmo.com/s3/4854812/Phase2-3

Invite your family member or friend to contact PC Project at 801-987-8758 or info@pachyonychia.org with questions or to learn more.
Seeking Participants for Pachyonychia Congenita Clinical Research Trial

If you or someone you know has Pachyonychia Congenita (PC), you may be interested in participating in a Phase 2/3 clinical research trial for an experimental therapy that is applied to the soles of your feet.

Some of the requirements for participation include:

- Must be able to travel to sites in the United States. Travel may include driving and flying.
- Must be at least 18 years of age or older
- Have a clinical diagnosis of PC, genetically confirmed to involve any of three keratin genes KRT6A, KRT6B, or KRT16
  - If you have not been genetically tested, or if you do not have a copy of your genetic testing report, please contact PC Project at study@pachyonychia.org for more information about genetic testing.
  - **NOTE:** If you know your mutation because a family member was tested, you will need to do a confirming test through PC Project, so you will have your own report.
- Have not participated in a clinical research study in the past sixty days
- On a daily basis during the study, wear an activity monitor and answer questions on a smartphone-based app.

Participants must be, in the opinion of the study doctor, able to understand the study, cooperate with the study procedures and willing to return to the clinic for all of the required visits. Your participation is voluntary.

If you qualify for study participation, you will be compensated for your time and your travel expenses to and from the clinic for you and a companion will be covered.

You can find more information about the study and participating sites on www.clinicaltrials.gov and www.valostudy.org.

If you are interested in participating, you can either:

- call a participating site directly, or
- email PC Project at study@pachyonychia.org
Extraordinary Match Opportunity for Giving Tuesday 2019

On Giving Tuesday, December 3, 2019, PC Project will join in a global day of charitable giving fueled by the power of social media. For the past few years, PC Project has successfully raised money on Giving Tuesday to help fund the mission of finding a cure for PC.

This year, all donations to PC Project will be matched two-to-one by a generous, anonymous donor. That means for every dollar donated, another two dollars will be given!

This is an extraordinary opportunity for monetary gifts to go even farther for PC research and patient services. This match will be honored for all Giving Tuesday donations starting now: pachyonychia.org/givingtuesday/

Watch in the coming days for more information about participating in Giving Tuesday activities and ways to be part of the PC Painkillers, PC Project’s official Giving Tuesday Team.

2. A robust patient registry filled with data from genetically diagnosed patients for research and clinical trials.

As the only organization in the world to advocate for PC, PC Project actively works to achieve both, all the while supporting patients who are navigating life with this chronically painful disease.

The PC community is strong and thriving. But these critical programs can only continue with your help. Please make your dollars go farther than ever before during this end of year campaign by donating to this worthy cause.

Then, on Giving Tuesday, share memes and messages with your contacts on social media and through email and include WHY you support PC and PC Project, along with the link to donate: pachyonychia.org/givingtuesday/

When you tell your friends and family that you support and trust a charity and why, they are more likely to support it as well. Show the world that together we are #StrongerThanPC.

Why Give To PC Project On Giving Tuesday or Anytime of Year

In order to have effective treatments for PC, two key things are needed:

1. Collaborative research that will translate into meaningful therapeutics.
**Be Part of the Solution for a Cure**

In addition to helping on Giving Tuesday, there are two important things a PC patient can do:

1. **Join the International PC Research Registry.** Every single person with PC needs to be documented as having PC. Researchers and drug developers are literally looking at the numbers in the registry and making decisions about whether or not to help PC patients. Encourage family members with PC to join as well. Be “counted for a cure”.
   
   registry.pachyonychia.org/s3/IPCRR

2. **If you live in the US or Canada, are over 18, and have K6a, K6b or K16, please consider participating in the current clinical trial.** All expenses are paid and this trial is paving the way for all future studies. If PCers don’t participate in the trial, who will? Trial information available at pachyonychia.org/valo/

**SAVE THE DATE!**

**2020 PC Patient Support Meeting**

The Annual Patient Support Meeting will be held in Roissy, France (near Paris), June 4-6, 2020. Thank you to PC advocate, Sylvie Potier, and her associates at Le Cœur Au Pied, for scouting hotels and helping with planning. Registration and meeting information will be available next month.

**A PCer Making a Difference:**

**Tom Baker Educates Dermatologists**

PC Advocate, Tom Baker, recently attended a Royal Society of Medicine Meeting of dermatologists. Tom courageously educated approximately 60 dermatologists about PC by showing his PC symptoms and how PC affects him personally. If you are invited to a meeting or grand rounds, please contact PC Project info@pachyonychia.org and we can try and arrange for other patients to join you and provide educational brochures for you to share.

This is Tom’s experience in his own words: “After not seeing a dermatologist outside of the patient support meetings with PC Project, I decided to see a dermatologist at our local hospital. During the appointment I explained that I wasn’t necessarily looking for a cure for PC (although that would be nice!) but I wanted to raise awareness and understanding of the condition.

From my own experience of meeting others with the condition I am always surprised how the symptoms vary so much. Further, some symptoms appear visually similar to the symptoms associated with completely different conditions. In my case, as I don’t have thick nails, the name ‘PC’ doesn’t really describe my symptoms of foot calluses either. I therefore don’t envy a dermatologist trying to make a diagnosis.

As a result of this, my dermatologist suggested she present my case at the Royal Society of Medicine meeting of dermatologists that happens a few times a year, where cases that are particularly rare or of interest are presented and the patient can attend to actually show the symptoms and talk to the attendees. I recently attended such a meeting and was pleased to be able to show around 60 attendee dermatologists my feet. In particular, I was able to explain the difference between regular ‘hard
Skin that builds up on top of regular skin for many people, and the hard skin PC patients have that is protecting underlying raw flesh and why therefore you can’t simply cut all the hard skin off. This seemed to help explain why it hurts so much! The attendees were interested, and the experience was positive – hopefully this will help lead to PC becoming more and more widely known and leading to more people being correctly diagnosed, particularly if they don’t have the more obvious PC symptom of thickened nails.”

**Shop for PC**

Remember, as you do your holiday shopping, please use the links below. Choose to support “Pachyonychia Congenita Fund” and a portion of your purchase will be donated to PC Project. Every bit counts. USA/Canada: [smile.amazon.com](http://smile.amazon.com) or [iGive.com](http://igive.com) Europe: [smile.amazon.co.uk](http://smile.amazon.co.uk) or [Give As You Live](http://www.giveasyoulive.com)

**IPCC Symposium Report**

A critical part of PC Project’s mission is to empower research. In May 2019, PC Project hosted the International PC Consortium (IPCC) Symposium in order for scientists to present and collaborate regarding PC-related research.

A report of the Symposium was written by key members of PC Project’s IPCC. Although this report was written for IPCC members, and portions will soon be published in the British Journal of Dermatology, if interested, [click here to read the Report of the 16th Annual International Pachyonychia Congenita Consortium Symposium.](http://www.pachyonychia.org)

**PC Project Represented at 2019 PeDRA Conference**

PC Project participated at the PeDRA (Pediatric Dermatology Research Alliance) Annual Conference in Chicago, November 14-16, 2019. PC advocate and patient, Jim Rittle, attended the patient track, along with his daughter, Kaelyn. Janice Schwartz, from PC Project, spoke on how the Externally-led Patient Focused Drug Development Meeting with FDA has benefitted PC Project. This 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 other patient advocate leaders.

PC Project presented a poster and had a PC 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.
2020 France Patient Support Meeting

The annual Patient Support Meeting will be held in Roissy, France (near Paris), June 4-6, 2020. Thanks to PC advocate, Sylvie Potier, and her associates at Le Coeur Au Pied, for scouting hotels and helping with planning. Registration and meeting information are available at https://www.pachyonychia.org/2020psm/

Vallo Study Update

Ten more PC patients are needed to enroll in the VALO clinical trial. The study is on track to have enrollment close by the beginning of February 2020. If you are interested in participating in the study, please indicate your interest as soon as possible at surveygizmo.com/s3/4854812/Phase2-3 or Pachyonychia.org/valo/

Palvella’s team reported, “Some PC patients have now entered Phase 3 of the study. Interest from the PC community continues to remain strong with more than 75 patients having entered the study as of mid-Dec and nine clinical study sites actively enrolling. We are very thankful and appreciative for the intense engagement and collaboration from all clinical study investigators, PC Project and of course PC patients.”

Giving Tuesday 2019 Success!

Because of you, and with the help of over 380 donors, Giving Tuesday was truly a day of giving for the PC community. Please accept our heartfelt thanks for your support now and always. You are the reason PC Project is thriving, research is in progress for PC, and PC patients have hope for a less painful life.

Each contribution made for Giving Tuesday and through December 31, 2019, will be matched 2:1.

Donate at pachyonychia.org/donate-help/

From the bottoms of our feet, we thank you!

The following meme was created by Jack Padovano, PC patient and chair of the PC Project Board of Trustees as an entry in the meme-making contest:

Living with Pachyonychia Congenita is a curse. Generous supporters like you are a blessing.
Giving Tuesday Highlights

Many PC supporters joined the PC Painkillers Team, sharing their PC stories, creating personal Facebook Fundraisers and/or sending emails, explaining how PC affects them. Personal connections to PC are powerful motivators for people to give. People give to people! Below are just a few examples:

Barbara Feinstein and Aaron Kline, parents of a PC child, sent heartfelt letters to all their contacts, telling about PC, PC Project and how PC affects their child. Significant donations were received through their effort.

Without being asked, PCer Michael McCollough, posted thank you messages to donors who gave through PC Project’s Facebook fundraiser. Mike’s expressions of gratitude for donors demonstrated his team-focused attitude.

Patients and loved ones filmed themselves answering the question: “Why I Give to PC Project?” Some were filmed in different languages and all were shared during Giving Tuesday. Click here to view these videos on the PC YouTube channel. And, please “subscribe” to the channel, too. We need 1,000 subscribers to be able to live stream from a cell phone.

PC memes were shared by patients and their loved ones on social media. Winner of the meme-making contest, Kerry Briggs-Evans and her son with PC, Tony, entered 11 memes. This one was a favorite:

A member of the International PC Consortium of scientists and clinicians, Dr. Mark Field, owner of Analgesic Innovation, saw the social media posts and created a video just for PC. Please click here to enjoy or share the video.

A hardware shop in Kerry Briggs-Evans’ town kindly offered to put a donation box on their counter. An announcement was made on the town’s Facebook page. Here is a picture of the donation box featuring Tony:

Celebrating and Helping PC

Lynn and Larry Block, grandparents of Allison Block, celebrated their 50th Wedding anniversary and chose to help PC at the same time. Here is a letter from Lynn to the PC family:

"On Oct 26, 2019 my husband and I and our children and grandchildren celebrated our 50th Wedding Anniversary and the 97th birthday of Lynn’s mom, Margaret.

Instead of accepting "things" we asked that friends and relatives wishing to "give" us a gift, to please consider a donation to the PC Project in honor of our granddaughter, Allison Block or to the Cancer Center where I and Allison’s mother have been treated.

Everyone was so generous, we could not believe what happened. We were given enough in
donations to give $1500 for PC, as well as $500 to our Cancer Center to assist patients. We are so grateful for your Giving Tuesdays and donation matches!

I’ve included a photo of my husband and I dancing (we have ballroom danced for 30 years) and a photo of Allison dancing for our photographer. Although she was limping an hour into the party, she loves to dance and wouldn’t give up. Our prayers are with all people with PC and with the researchers and doctors trying for a cure and/or pain relief.”

I thought about my life and all its ups and downs. I thought about my PC and how it has impacted me personally. Intellectually I understand PC won’t kill me like pancreatic cancer or a heart attack might, but I wondered if it could cause death. And I think the answer may be yes. Take my father for example. He drank too much and eventually drank himself to death. The official coroner’s report said heart failure. But I know better. He suffered from severe depression caused in part by his constant PC pain. So, he numbed the pain with vodka. Lots of it. The day we cleared out his apartment we found empty one-gallon vodka bottles everywhere. Did PC cause his death? Not directly, but certainly indirectly.

I also think about a story I heard from a father who shared his son’s struggle with drugs as a result of his PC. His son died tragically as a result of those drugs. Did PC cause his death? Not directly, but certainly indirectly.

Looking back at my father’s life with PC, my own, and now my daughters, it probably explains why I’m so driven to help PC Project achieve its mission of fighting for a cure, connecting and helping patients, and empowering research. And it's why I’m writing this article today. I don’t want any of us to die indirectly from PC. Not one of us!

If I told you that you or a loved one who has PC is going to die because of PC, what would you do differently? What would you change? Would you become more involved to move our mission forward?

My wish (hope) today is that we take our pain and turn into a force for good. A force that includes:

• Sharing your PC story so more people learn about PC
• Becoming a PC Warrior (volunteer).
• Raising money for our cause
• Donating
• Joining the PC registry

Don’t let another person die from PC. Get involved and help PC Project achieve its mission.

**In Loving Memory**

**Robert Hansen**, the great uncle of Allison Block, recently passed away. Allison’s grandparents, Robert and Marilyn Bramer (Marilyn is Robert Hansen’s sister) are monthly contributors to PC Project. Because Mr. Hansen also supported PC Project, donations were made in his memory and in honor of Allison.

PC Project expresses sincerest condolences and thanks to the Hansen, Bramer and Block families at this tender time.

**What if PC caused death?**

*By Jim Rittle*

A close friend of mine has been given 5 months to live. Once the initial shock began to wane, I began to reflect on my own life, my own mortality, and what kind of end may be in store for me. Pretty heavy stuff I know.