FROM A SCHOLARSHIP ESSAY

“I am so excited about the opportunity to attend my first PC Patient Support Meeting (PSM) and to get to meet other PC’ers and listen to their life experiences as I share mine with them.

I have to admit that I’m almost scared to meet others with PC. I’ve never before known anyone else with this same, painful condition -- I thought I was alone in this world until I accidentally discovered PC Project. It has changed my life.

Living with PC is so painful -- not only physically for us, but it hurts those who love us as well. They feel helpless. Having this PSM for individuals with PC and their families is an incredibly generous undertaking.

We not only want to meet with others who have PC. We also want to meet those who have offered so much of themselves to help ‘find a cure’ and to say THANK YOU. We never knew there were any such individuals out there.”

WHY ATTEND A PSM?

We asked PCers at the 2014 Edinburgh Patient Support Meeting (PSM) what advice they would give to someone who just found out they have PC and is considering attending a PSM. Here are some of their comments:

“I would say that the PC PSM is the best thing that could ever happen to a PCer. All people involved with the PSM are very caring, thoughtful, loving and comprehending. I would say not to think twice about attending because it is beyond incredible how life changing these PSM meetings are. New PC’ers will enjoy the idea of sharing with others, knowing that they are not alone.”

“When I first PSM meeting, for the first time I looked into the eyes of another person with the same disease, saw them move like me, saw them use the same tricks that I use to get through their day. This gave me an immense feeling of no longer being alone and it’s a wonderful feeling. I would say that I’ve never been as happy and confident before the PC PSM experience.”

“There’s nothing to be worried/nervous about, it’s amazing/comforting/helpful to be able to relate to other people with the same skin condition”

“We asked PCers at the 2014 Edinburgh Patient Support Meeting (PSM) what advice they would give to someone who just found out they have PC and is considering attending a PSM. Here are some of their comments:

“Please attend! It is very informative and the best thing for a new comer is meeting other people with the condition.”

“At my first PSM meeting, for the first time I looked into the eyes of another person with the same disease, saw them move like me, saw them use the same tricks that I use to get through their day. This gave me an immense feeling of no longer being alone and it’s a wonderful feeling. I would say that I’ve never been as happy and confident before the PC PSM experience.”

“There’s nothing to be worried/nervous about, it’s amazing/comforting/helpful to be able to relate to other people with the same skin condition”

“We asked PCers at the 2014 Edinburgh Patient Support Meeting (PSM) what advice they would give to someone who just found out they have PC and is considering attending a PSM. Here are some of their comments:

“Please attend! It is very informative and the best thing for a new comer is meeting other people with the condition.”

“At my first PSM meeting, for the first time I looked into the eyes of another person with the same disease, saw them move like me, saw them use the same tricks that I use to get through their day. This gave me an immense feeling of no longer being alone and it’s a wonderful feeling. I would say that I’ve never been as happy and confident before the PC PSM experience.”

“There’s nothing to be worried/nervous about, it’s amazing/comforting/helpful to be able to relate to other people with the same skin condition”

“We asked PCers at the 2014 Edinburgh Patient Support Meeting (PSM) what advice they would give to someone who just found out they have PC and is considering attending a PSM. Here are some of their comments:

“Please attend! It is very informative and the best thing for a new comer is meeting other people with the condition.”

“At my first PSM meeting, for the first time I looked into the eyes of another person with the same disease, saw them move like me, saw them use the same tricks that I use to get through their day. This gave me an immense feeling of no longer being alone and it’s a wonderful feeling. I would say that I’ve never been as happy and confident before the PC PSM experience.”

“There’s nothing to be worried/nervous about, it’s amazing/comforting/helpful to be able to relate to other people with the same skin condition”

“We asked PCers at the 2014 Edinburgh Patient Support Meeting (PSM) what advice they would give to someone who just found out they have PC and is considering attending a PSM. Here are some of their comments:

“Please attend! It is very informative and the best thing for a new comer is meeting other people with the condition.”

“At my first PSM meeting, for the first time I looked into the eyes of another person with the same disease, saw them move like me, saw them use the same tricks that I use to get through their day. This gave me an immense feeling of no longer being alone and it’s a wonderful feeling. I would say that I’ve never been as happy and confident before the PC PSM experience.”

“There’s nothing to be worried/nervous about, it’s amazing/comforting/helpful to be able to relate to other people with the same skin condition”

“We asked PCers at the 2014 Edinburgh Patient Support Meeting (PSM) what advice they would give to someone who just found out they have PC and is considering attending a PSM. Here are some of their comments:

“Please attend! It is very informative and the best thing for a new comer is meeting other people with the condition.”

“At my first PSM meeting, for the first time I looked into the eyes of another person with the same disease, saw them move like me, saw them use the same tricks that I use to get through their day. This gave me an immense feeling of no longer being alone and it’s a wonderful feeling. I would say that I’ve never been as happy and confident before the PC PSM experience.”

“There’s nothing to be worried/nervous about, it’s amazing/comforting/helpful to be able to relate to other people with the same skin condition”

“We asked PCers at the 2014 Edinburgh Patient Support Meeting (PSM) what advice they would give to someone who just found out they have PC and is considering attending a PSM. Here are some of their comments:

“Please attend! It is very informative and the best thing for a new comer is meeting other people with the condition.”

“At my first PSM meeting, for the first time I looked into the eyes of another person with the same disease, saw them move like me, saw them use the same tricks that I use to get through their day. This gave me an immense feeling of no longer being alone and it’s a wonderful feeling. I would say that I’ve never been as happy and confident before the PC PSM experience.”

“There’s nothing to be worried/nervous about, it’s amazing/comforting/helpful to be able to relate to other people with the same skin condition"
QUICK PC SURVEY
As PC research continues and we learn more about PC, new questions are often asked by researchers which are not included in the basic IPCRR registry questionnaire. We need your input and experience—PCers can provide the answers!

From time to time, we will be asking for your help with quick two-minute surveys. Please use the following link to respond https://www.surveymonkey.com/s/PCQuick1

January 2015 Informal PC Survey
1. Have you ever been referred to or treated by a pain specialist?
2. Do you have medical insurance?
3. Did you have problems sucking as an infant?
4. Are you bothered by a deep itch in or near your callus? How frequently does it occur?

NEW INSIGHT: PC & PAIN
Over the last ten years, we have repeatedly found that PAIN is the most debilitating feature for those with PC. Yet, we found there were more than 500 published articles on PC that did not mention PAIN at all. We have wondered about how this was possible. We have made good progress in including PAIN as a major feature for PCers. (See select publications listed below.) We still marveled at how it was possible that so few medical experts understood the pain that is such a major part of PC.

Over the years, we realized that PCers rarely go to doctors for pain. Many patients told us that doctors had told them “callus is not painful.” We learned that PCers usually only visit a doctor when they have an infection. We felt this was one reason PC pain was not mentioned in medical literature.

In an unexpected way, a very interesting new fact was recently learned which may be important in the area of ‘PC and pain.’

PC Staff were invited to participate in an American Academy of Dermatology Patient Advocate Task Force establishing measures for dermatologists under the new US Affordable Care Act (ACA).

In the call the topic of pain came up as a possible measure. The dermatologists on the call stated: “Dermatologists do not treat pain, especially chronic pain. They are not comfortable prescribing drugs that treat pain.”

We had a completely NEW INSIGHT about PC and pain! PC is a skin disorder. Those with PC are usually referred to dermatologists not to pain specialists. In fact, we think very few PCers have seen a pain specialist. Please help us by completing the survey this month and letting us know your own experience.

PUBLICATIONS
Search “pain” as a keyword at http://www.pachyonychia.org/research_articles.php to find publications about pain including: Pachyonychia Congenita Overview: The inherited nail dystrophy is associated with painful plantar keratoderma. The Dermatologist-April 2014.

CARE TIP CORNER
What are your best tips when you experience intense pain? How do you cope? Please email us at info@pachyonychia.org

“My feet have been unbelievably sore lately...super mushy and bleeding like crazy. I don’t know what’s going on. Right now I feel like all I’m doing with my life is getting through the pain each day. But I hope they get back to their normal pain.”

“I have been experiencing the most excruciating debilitating pain ever, way more than usual. It has been a burning blow torch feeling in my feet, heels and most specifically, sharp and shooting burning pain...it’s unbearable.”

WHO’S WHO
“The doctors are all the same...no one understands this bloody pain.
At 62 the phone it rings, it’s my younger sister and she is the best, She has been diagnosed—unlike the rest. Its K6b N172del!
At a PC Meeting only last year, I found lots of others —are all sincere.
Their feet hurt for all their years, some on crutches and in wheel chairs,
I can shuffle like all the rest but now have PC friends and those who care.”

WHO AM I?
Find this story and others at www.Pachyonychia.org. Add your experiences by emailing your story to info@pachyonychia.org.
PC SUPERHEROS
FUNDRAISING
For PC Awareness, Eveline Schloesser and her family sold PC keychains that Eveline designed. At Christmas they sent E-Christmas Cards and raised $50 more for a total $300 donation to PC. What great ideas to get us excited for PC Awareness in June.

Le Coeur Au Pied continues to help French PCers join the IPCRR patient registry, raise awareness and funds. This year they sent 10,000 Euros ($11550 US) to PC Project. We are grateful for all their work.

There are many of you who have joined the IPCRR registry, donated funds, raised awareness and supported PC in lots of ways. THANK YOU! We would like to feature what you have done or are doing. Please email information on to info@pachyonychia.org

PC PROJECT SERVICES
SUMMARY 2014
Patient Services (IPCRR)
• Served 161 new patients who contacted PC Project for the first time in 2014
• PC Project now serves over 1,500 patients who either have PC or a similar condition
• Provided 87 physician consultations for patients
• Sponsored genetic testing for more than 80 patients

• Over 600 letters sent to patients, local physicians, scientists, editors and others (and over 3000 emails)
• Monthly PC NewsBrief for patients, families and friends
• Maintained PC Social Media sites such as Facebook, Facebook PC Patient Chat, Twitter, Instagram, YouTube and Pinterest.
• Co-sponsored the 2014 Patient Support Meeting
• Held patient webmeetings
• Sent support letters for schools, work, insurance, etc. for numerous patients

Promoting PC Awareness
• Maintained PC website
• Contributed to 23 publications
• Sponsored special Grand Rounds training sessions

Science and Research (IPCC)
• Sponsored the 11th Annual IPCC Scientific Symposium for PC research
• Sponsored monthly IPCC Genetics Team webmeetings
• Organized quarterly physician webmeetings
• Edited Quarterly IPCC News for physicians and scientists interested in PC

EXPENDITURES 2013
Operating Services—$72,877
Misc Office $ 3,397
Fund Raising $ 22,708
Computers $  929
Rent $  5,400
Professional Services $40,443
Program Services—$238,790
Educational Outreach $ 10,771
Videos, Presentations $  500
Clinical Trials $ 78,761
IPCC Consortium $ 24,322
IPCRR Registry $ 98,166
Patient Support Mtg $ 18,009
Publications $  1,507
Scientific Meetings $ 4,258
Webinars $  478
Website $  2,018
Genetic Testing 2—$105,000

1 These numbers are estimated. The 2014 tax return will be filed in a few months and posted on our website.
2 Sponsor pays these funds direct to University of Dundee

RAISE RESEARCH MONEY
THE EASY WAY!
We have just learned that you can double the donations raised when you or friends shop online with iGive and Amazon Smile together. Here is what to do:
(1) Go to www.igive.com select PC Project as your charity.
(2) From iGive select the Amazon store and you will be sent to Smile.Amazon.com
(3) Choose Pachyonychia Congenita Fund.
(4) Go shopping and you’ll raise money for PC Amazon.Smile donates .5% and iGive donates .8% of your purchases so 1.3% of your total purchase will support PC Project. It costs you nothing extra to make this difference.
Thank you donors!

We received over $81,300 donations in 2014. Thank you! PC Project would also like to extend a special thanks to those sustaining members who provide regular donations to our organization. We are so grateful for your support!

Regular donations help provide evidence of support from the PC community which is essential in qualifying for grant funding. It enables us to achieve more advanced planning for research toward fighting for a cure, connecting & helping patients and empowering research. Regular donations of any amount are of great value in our overall program. Thank you.


We are so grateful for all who have supported Pachyonychia Congenita. Together we will make a difference!

See the above article “Becoming a Sustaining Member” to learn how to make your donations be automatically deducted either monthly, quarterly or annually.
"Most doctors looked at us puzzled without answers, and others simply shrugged and said they didn’t know what it is and that there is nothing that could be done. We had never known another person with this disease. My family and I felt completely isolated.

For the first time in my life, I could walk into a doctor’s office and tell them what it is called. After years of not knowing, finally learning my condition actually had a name was incredibly empowering!! It was also comforting to know that we are not alone.

Had Pachyonychia Congenita Project not existed, I doubt I ever would have been diagnosed and I firmly believe with all of my heart that without this organization’s tenacity, all of us who suffer from this condition would have gone on suffering, in isolation, in pain, without hope and even worse in acceptance of our fates. After being told for years that your condition is hopeless. I think many of us accepted being hopeless, I once did. Not anymore.” - A PC Friend.

We would love to hear your stories. Send your stories and photos to info@pachyonychia.org and your story might be shown next.

2015 PC AWARENESS
As June approaches, we are beginning to gather the 2015 PC Awareness plans. We are eager to know what you are planning. Please share your events with us by email to info@pachyonychia.org or through the PC Awareness Facebook page facebook.com/events/835912403138164

The links are also available on our website with an archive of past awareness events. There are many ways to find the Awareness information on our website: (1) Use the search box and type in Awareness. (2) Hover over the Donate/Help tab and choose How You Can Help. (3) Click the News & Events tab at the top of any page.

RARE DISEASE DAY
World Rare Disease Day, Saturday, February 28, 2015, is an annual observance held on the last day of February to raise awareness for rare diseases.

How will you celebrate rare disease day in your area?

This is an opportunity to join a global effort and show support for improved access to treatments and medical representation for individuals and families affected with rare and genetic diseases. On this day, various activities take place in the United States, Europe, Russia, Japan and around the world to raise awareness and donations for the foundations that support and serve these communities.

Many individuals meet with state and local representatives to discuss their concerns regarding their disease, or plan special events, large and small, to support the cause and show they care.

This is a great opportunity to spread the word about Pachyonychia Congenita on Rare Disease Day and begin the effort toward 2015 PC Awareness Month in June.

2015 NEWARK NEW JERSEY PC PATIENT SUPPORT MEETING (PSM)
Register as soon as possible at www.pachyonychia.org/news_events. Need help registering see last page or contact us.

“Definitely attend a meeting! You will find it very interesting and realize you are not alone living with such a rare condition. You will make new friends too.” - A PC Friend
IPCRR Participation
Your participation makes a huge impact on all aspects of PC research. Each PCer who takes the thirty minutes to complete or update the IPCRR forms adds to the PC data. More data lead to more publications in quality scientific journals. Publications lead to more doctors and scientists becoming interested in research for PC. The larger group of researchers leads to new collaborations, innovative ideas, questions and other pathways forward. The results are returned to the individual PCer in clinical studies and trials for new treatments — and sometimes in new questions for PCers.

PC Project uses all of the information provided by patients to make a difference. We could not do this without each individual participant in the IPCRR registry. THANK YOU.

GRDR
The Global Rare Disease Registry (GRDR) is a project from US National Institute of Health (NIH) National Center Advancing Translational Sciences (NCATS). NIH/NCATS is the newest center at NIH and was created by Congress to speed the delivery of treatments to patients. The goal of the GRDR is to create a single database of information for patients with rare diseases and with this database, to accelerate the development, dissemination and use of new knowledge to improve the health and quality of life for millions of people. PC Project and the IPCRR has been selected as one of six registries participating in this pilot program. This will open more opportunities for sharing PC data with researchers who become interested in PC.

Genetic Testing
In 2009, realizing that PC is a dominant disorder, PC Project reached out to patients mentioned in an article about so-called ‘recessive PC.’ Based on data in the registry, we felt these patients did not have PC, but some other disorder.

It has taken the researchers six years and the cooperation of family members providing additional samples — but these patients now know their specific gene and mutation. This answer sometimes comes quickly to those of you with PC — but for this family, the ongoing effort by PC Project and those working with us on the Genetics Team has been gratefully received.

We are pleased that now a correction will be published clarifying that PC is a dominant disorder and that there are no ‘recessive’ cases. The ‘unsolved cases’ are reviewed each month during the IPCC Genetics Team Web Meeting. Knowing of preparation of a paper for a new disorder caused by mutations in the CAST gene, the team noticed many similarities between the cases — and the mutation was confirmed for this family. The findings for this new disorder have recently been published in the American Journal of Human Genetics in a paper entitled Loss-of-Function Mutations in CAST Cause Peeling Skin, Leukonychia, Acral Punctate Keratoses, Cheilitis, and Knuckle Pads by Lin et al. (including PC Project).

This is great example of the knowledge gained from the IPCRR data — and learning what is and what is not PC.

PC Clinical Trial
Many of you, including all of us at PC Project, are anxiously awaiting the topical sirolimus clinical trial results. After the completion of the trial, the data cannot be released until those overseeing the trial review and lock the data. That was completed the first week in February and we expect the data reports to be released shortly.

We found some interesting information about clinical trials in a publication by the Pseudoxanthoma Elasticum (PXE) patient sup-
port group and wanted to mention a few points we thought would be of interest to you:

- Clinical trials are structured research experiments used to advance medical knowledge and patient care.
- Clinical trials are the way that we gather evidence to determine whether or not something works and how well it works.
- The participants in clinical trials are sometimes called Heros because they are testing something that is unknown.
- The first consideration is to answer what formulation of the treatment is right for this condition?
- The next consideration is what dose is optimal and what can someone tolerate.
- Ultimately the researcher will be working what FDA and/or EMA to apply to have the treatment relabeled. This means that the treatment will be named as appropriate for the new condition.

Quoted from www.pxe.org/about-pxe/faq/clinical-trials-faq

**SURVEY RESULTS**

There were 61 PCers who responded to the January 2015 News Brief Quick PC Survey. Thank you! Here is a review of the results. These responses will be very valuable in a number of projects PC Project is currently pursing.

Do you have medical insurance? 88.5% said YES

Have you ever been referred to or treated by a pain specialist? 14.8% said YES

Did you have problems sucking as an infant? 15.8% said YES

Are you bothered by a deep itch in or near your calluses? 61.7% said YES

We will continue to have additional Quick PC Surveys as we have projects that may lead to new research discoveries and collaborations for PC.

**NIH Keratin Study**

Researchers at the NIH Human Genome Project recently completed a study on tooth enamel and discovered that certain keratins, including those involved in PC, are active in tooth enamel. The NIH researchers contacted PC Project and asked that we collect some PC teeth for examination in their continuing research.

We are in the processes of organizing this study and getting Institutional Review Board (IRB) approval. Once we have approval, we will send a direct email survey and request for teeth to those in the IPCRR.

**PC SUPERHEROS FUNDRAISING**

The January News Brief listing 2014 donors to PC Project did not include any donors to PC UK because PC Project did not receive any donations from PC UK in 2014. PC Project does not receive a report of donors to PC UK and has no control over the PC UK funds.

PC UK funds were used to pay costs of the PC Patient Support Meeting (PSM) held in Edinburgh in October 2014. And, PC Project has applied for reimbursement of some of the scholarship award funds given for the 2014 PSM.

At the PSM meeting it was exciting to see the laboratory equipment purchased by PC UK for the McLean/Smith laboratory at the University of Dundee.

W. H. Irwin McLean and Frances Smith at the University of Dundee are leaders in our genetic testing and in all PC Project efforts. Irwin and Frances started PC UK so that those in the UK can add Gift Aid to their donation. This gives an extra 25pence for every £1 donated. We wouldn’t be where we are without these two PC Superheroes and the Dundee team, but we don’t control or report donations contributed to PC UK.

SAVE THOSE TEETH: If your child loses a baby tooth, if you have a wisdom or other tooth removed, please save it! Even a small piece of a broken tooth can be studied. Place the tooth in a plastic bag, put in the refrigerator and save it until this study can be launched.
CARE TIP CORNER
What socks work best with PC?
Please email us at info@pachyonychia.org

“I’m addicted to wicking socks. Wicking socks are made with synthetic fibers that wick away the moisture from feet and claim to be anti-blistering...My feet aren’t soggy and clammy like they used to be when I’ve worn socks and shoes. This has significantly reduced the terrible itching that usually comes when I take off my shoes, and most of all, I can wear socks and shoes longer before the pain of simply wearing them starts setting it. Now, wicking socks aren’t a cure, and I still hurt when I walk, but they have helped me considerably enough that I am hooked for life.”

There are many different types of wicking socks. You can find wicking socks at most stores and online stores. (Remember to use iGive.com when shopping online.)

“I was at the beach and saw a guy selling a product called sand socks. I saw a lot of people on the beach wearing them because the sand was hot. I bought a pair that I will wear next time to the beach. But this looks like a much better option to the swim shoes I have been wearing. This product is a thin sock that provides breath-ability and has a sole on the bottom to provide support. Protect feet from: Hot & Cold Sand Dirt & Mud Sun Blisters & Rashes “
http://www.sandsocks.org/

Visit the PC Wiki to see other tips and tools at www.pachyonychia.org/wiki
I don’t know why he did or why he didn’t run home and simply ask our mom to come and get me in the car. Without embarrassment or fanfare, my brother simply carried me home many times during those years.

There’s no doubt I’ve been blessed by a family who has carried me. I am a spontaneous mutation of PC and when I was young there was no PC Project for information or support. My parents tried their best. My dad trimmed my feet until I was old enough to do it myself. I remember times when my feet were particularly bad or I was simply discouraged about doing the activities I wished I could. I didn’t get sympathy, but a simple, sincere, “I’m sorry you’re hurting” from my parents carried me, letting me know I was loved, not alone and I would be okay.

Today I’m still “carried” by family and other good people in my life. I have a husband and children who carry me in too many ways to even write here. I have extended family who carry me, including a mother-in-law who started PC Project because of her love and genuine desire to make a difference for me, my children and for every single person with PC in this world.

I’m carried by those who work and volunteer at PC Project. Knowing they’re working for a treatment and cure carries me, especially on hard days, which sometimes can be often. The dedication of those involved is unparalleled. It’s apparent every day that this is not just a job for those at PC Project. They are deeply invested, heart and soul. The same can be said about the doctors and scientists on our team. I’m amazed and humbled by their commitment to us.

I’m carried by other PC patients around the world who’ve let me know I’m not alone, who understand what I’m going through. Some I’ve met, others I’ve connected with through email and social media. Even if I’m not as active myself on those sites, I still “see” them and knowing they’re there carries me.

I desire to live a life where I, too, can carry others. I hope I can lighten loads and relieve pain, maybe not physically like my good brother carried me, but in other ways. We all have the capacity to give of ourselves. I’m so grateful for those who continue to carry me each day. Life is so much better when we all carry each other in the best ways we can!”

We would love to hear from you. Please share your PC stories and comments by emailing them to info@pachyonychia.org.
NEW PC VIDEO
We have been working with Spy Hop Phase 2 Productions creating a basic video about Pachyonychia Congenita specifically for sponsors. The video turned out great and we now need your help to share the video. We have many different formats available to you for distribution, including: DVDs, flash drives, web links and downloadable versions. Please email info@pachyonychia.org and let us know what version you need and how many copies.

We invite you to share this video with your friends, family, neighbors, and potential donor/sponsors. With spreading the word comes funding for more research to help each PCer. We are so grateful for every donation whether it be $1, $10, or $10,000. Every penny counts.

2015 PC AWARENESS
June is the month designated for PC Awareness. We would like to start featuring your events. Please let us know what you are planning to do. It could be sending brochures to friends and coworkers, posting social media messages, having a dinner, yard sale, bake sale, bike-a-thon, carnival, collection boxes, or chatting about PC with a few friends please email info@pachyonychia.org your plan.

We hope that each and every individual family will participate and hold some event (large or small) in your own community. You can even share the new PC video with local businesses. Individually we are small, but together we can make a difference.

PC Awareness can be done anytime throughout the year. Don’t forget to have fun while you are doing it.

Here is one example of having fun, reaching goals and raising funds for PC: Andreas Berroth, PC Fellow post-doctoral scientist at Stanford University, wanted something to help him regain motivation towards accomplishing his New Year’s resolution weight loss goals. So, he challenged his co-worker at TransDerm, Roger Kaspar.

If Andreas reaches his goal by a set date, Andreas will donate $100 and Roger will donate $300 to PC Project. However, if Andreas does not reach his goal he has agreed to donate $300 to PC Project and Roger kicks in $100. (In all cases PC Project is the big winner!) What a fun challenge. Thank you Andreas and Roger!

By the way, the folks at Trans-Derm are open to additional challenges, either as individuals or as a team (as we all know, they prefer to work together as a team). It may be fun for us to challenge Trans-Derm to become leaner (hopefully not meaner) with the expectation that this would translate into even more productivity in reaching our goals of effective PC therapeutics!

For example, maybe we could challenge TransDerm and make pledges for each 5 lbs (up to a fixed limit – we don’t want this to get unhealthy) that they collectively lose in 2015 ending with a weigh-in at the Patient Support meeting in June? If TransDerm rises to the challenge, our pledges would turn into donations to PC Project? Thoughts, interest, challenges? Email your ideas etc. to info@pachyonychia.org

Andreas Berroth
RARE DISEASE DAY
PC Project attended the first annual Utah Rare Disease Symposium at the University of Utah last month. The symposium was held to inspire, motivate, raise awareness and spark collaboration among the many rare disease stakeholders — patients, families, legislators, medical students, health care providers, researchers, administrators, industry representatives, and more. The topics focused on diagnosis, treatment and support for rare diseases.

Roger Kaspar, CEO TransDerm, Inc. attended the first annual Rare Disease One Day Conference at University of California, Davis. This was the first symposium of its kind to take a look at rare disease research, the obstacles and new approaches that are shaping the future of research and how to treat difficult diseases. Experts shared their perspective, the challenges and their paths to success in driving innovation towards finding cures for people living with rare disease. Speakers and panels from academia, industry and patient advocacy groups shared perspectives on driving innovation towards finding cures for people living with rare disease. Dr. Kaspar was on the panel and shared his efforts including those for PC.

Next year the world wide rare disease day will fall on the most rare day, February 29, 2016.

PC RESEARCH
Topical Rapamycin—The Phase 1b Clinical Trial results are still being analyzed. A follow-up questionnaire for those who participated is being developed and will take a few weeks to get IRB approval.

Nail Study—A new study has been proposed by Robert Rice to do a proteomic analysis of PC nails vs non-PC nails. The study will be to analyze four different samples from each PC Gene and compare them with samples from non-pc samples. If you would be willing to participate in this study by providing a clipping of a nail, please email us info@pachyonychia.org

PC QUICK SURVEY
Please take this month’s informal PC survey about your health care professional as well as blisters at https://www.surveymonkey.com/s/PCQuick2

HIGH SCHOOL SENIORS
Oh the places you’ll go.
Today is your day!
Your mountain is waiting.
So … get on your way!
--Dr. Seuss

Congratulations, you’re almost done! Graduation is a time to celebrate your accomplishments as well as plan for your future. Each of you have a story to tell and have overcome so much to get to where you are. We want to feature all of the graduating seniors in our May PC News Brief.

REQUEST: Please send us your stories, your achievements, goals, plans for the future and anything else you would like to add. Also, please send us a photo or two of the special things you do. Email info@pachyonychia.org before April 30th so you can be included. We can’t wait to hear from you.

CARE TIP CORNER
We received the following PC tips from past Care Tip Corners. Send tips to info@pachyonychia.org.

Re: Socks
“I have found Experia Powered by Thor-Lo Pads socks to be the most comfortable. They have padding!!! I buy them at Dicks Sporting Goods.”

Re: Intense Pain
“I was having such constant pain, especially at night, and even if I hadn’t been on my feet a lot. One of the PC doctors suggested taking pain medication morning and night for a while to break the pain cycle. And it worked! Even though it is mid summer here, I was OK. He also suggested I not put my feet into cool water to ease the pain which I usually did, as it expands the outer layer (stratum corneum) and could make it more sensitive. He said using cool gel packs would be better. His advice about various pain medications was terrific and about pre-dosing if I knew I was going to be on my feet a lot that day.”

Re: Shoes
“I find Sketchers really help with comfort, not 100% but more bearable in daily life.”
PC SUPERHEROS

Julie Peconi has been volunteering for PC Project for the past few years, helping contact patients and assisting with our Facebook pages. She is married with two kids—Timothy and Solomon. Her husband Tom Baker and son Timothy both have PC. Their family has participated in many events raising awareness for PC along with donating funds for the cause. Thank you to Julie!

We can say these same things about many of you. Thank you for all you do.

Have you seen a “PC Superhero” in action? Please email us info@pachyonychia.org. We’d love to hear your stories, see photos and be able to say thanks.

IPCRR GRAPHS

At the top of any page on the www.pachyonychia.org website, the last menu item is PC Data. This is where the graphs and charts from the International Pachyonychia Congenita Research Registry (IPCRR) data are posted. They are updated monthly and the information is very interesting.

There has been a lot of discussion on the PC patient chat group on Facebook about where PCers are. The PC location chart (right) is posted on the IPCRR Graphs tab and lists each country, US state and the number of PC patients who live in that area. There is also an interactive map of PCers.

You are the pieces to the PC puzzle. Thank you for participating.
Collect used ink cartridges, old cell phones, etc. and raise money for PC. Follow this how-to to get free shipping and print the label to send your collection of items to Plant Green. Currently only for USA and Puerto Rico.

**Recycle for PC through Planet Green**

1. On the How You Can Help page scroll to the bottom and click the Planet Green link.

2. On that page click the Sign In button on the top right hand corner.

3. On the enviropartner section log in and click come on in and skip to step 7. If you don’t have an account choose, “Don’t have an account? Sign up.”

4. In the middle on the right side you will see a Let’s Get Started option. Fill out, click Let’s Go.

5. Create your password and proceed by choosing “Come On In.”

6. Click the Print Label photo to take you to the screen to complete.
PC Project is evolving. There are so many good things that are under way which we absolutely believe will bring about the long sought goals of effective treatments to help with PC pain—our first target.

If you compare PC Project with other organizations, you will quickly understand that we need to engage and involve many more people—both paid staff and volunteers—in order to continue to accomplish all that is needed.

We have spent time evaluating and have a plan of how to move from only a full time volunteer (Mary) and one paid employee (Holly) to the place we want to be. The proposed plan is outlined in this Special Edition NewsBrief. We wanted you to be the first to know!

ADDITIONAL BOARD MEMBERS. Several additional members will be added to the Board of Trustees to provide the support needed with the additional staff.

STAFF POSITIONS. These are paid positions. Salaries will vary based on position, skills and experience. A benefit package is available for full time staff.

NEW—Executive Director. (see organizational chart attached)

NEW—Program Director. The Board has named Holly Evans as Program Director. She will be based in Salt Lake City, UT and will oversee the Int’l PC Research Registry (IPCRR).
- Part/time clerk
- NEW Part/time Nurse or Genetic Counselor will be PI on the IPCRR and provide medical assistance and coordinate the Peer-To-Peer Coaches (see below).

NEW—Office Manager based in Salt Lake City. This will be either a full/time or part/time position. If part/time, then a bookkeeper or other assistant may be hired.

NEW—Development Director. May work from any location; prefer full time, but may be part time. Some travel will be required to visit donors, organize fundraising events and assist the PC community in hosting local events.

NEW—Scientific Outreach Director. Mary Schwartz will fill this position temporarily and coordinate with the Int’l PC Consortium (IPCC). May work from any location. Some travel will be required to attend meetings, visit universities and corporations to encourage PC research and publications.

VOLUNTEER POSITIONS—this is an opportunity for patients, family, friends to be involved in all that is happening at PC Project. The time needed will vary according to each individual’s availability so that the burden never falls on any one family. This will be gradually developed and continue to grow over time.

(1) Peer-to-Peer Coaches. Will serve from home. We will initiate training so that PCers and/or family members can help other PCers access the best information and medical care. The number of hours required will depend on the individual volunteer’s schedule and the needs of the PC community; however, apart from training sessions, we expect this will be less than 10 hours/month.

(2) PC Patient Advocates. Will serve from home. Advocates will assist both the Development Director and the Scientific Outreach Director. Advocates will be able to select when/where they are able to travel to meetings, government offices, educational outreach efforts and fundraising functions. We will initiate training and provide tools to empower active advocates for a consistent message about Pachyonychia Congenita and PC Project.

If you or someone you know have qualifications and skills for these positions, please send resume to info@pachyonychia.org.

2386 East Heritage Way, Ste B, Salt Lake City, UT 84109 · www.pachyonychia.org · Phone 877-628-7300 · Email: info@pachyonychia.org
A NOTE FROM MARY: On behalf of the Board of Trustees, I am delighted to send this special PC News-Brief with this absolutely thrilling message. I am eager to work with the Board to find and fill these essential positions — both paid and volunteer. I will continue to play a role at PC Project as needed.

We are at a pivotal point at PC Project where we have to decide whether or not to become a fully-staffed and funded organization — or simply a ‘hobby’ organization. This plan will move us forward, involve many more patients/family members in meaningful roles using their passion and talents — and take us ever closer to an effective treatment for PC. We need to find the funds to properly staff PC Project. Mary Schwartz

A look at the future of PC Project...

NEW—Executive Director, PC Project
The Board of Trustees will provide oversight to the Office Manager, Program Director, Development Director and Scientific Outreach Director, until an Executive Director is hired. These positions need to be filled and functioning as the search for an Executive Director is conducted. The Executive Director may be filled by one of these individuals or by a new hire in addition to these staff members.
2015 Patient Support Meeting—Newark, NJ
The meeting was held June 14-16, 2015 at the Marriott Renaissance Airport Hotel. At the meeting, there were 60 PCers plus family members, physicians and scientists for a total of 115 attendees. The meeting theme was working together to make a difference. No single bee can make honey. It takes a hive! This meeting used a hive of PCers participating to make a wonderful meeting.

It all began at registration with a welcome from the McGrath and Filoso families. Then, a ‘welcome to New Jersey’ by Nicole Smith. The Smith family prepared a special New Jersey gift basket which the Jennings family won!

Presentations
- PC Around the World (Schwartz)
- PC Genetics (Smith)
- Pain (Pappagallo)
- Characteristics of PC (Hansen)
- Patient Adaptations (Bravo)
- IPCC Meeting Report (Wittmer)
- Cell and Mouse Models (Chen)
- 3D imaging of PC skin (Rittié)
- Research/Clinical Trial (Kaspar)
- PC Living With Me (Padavano)

NOTE: Many of these presentations will soon be available to view at www.pachyonychia.org.

Panels & Discussions
- Pain, Stigma and Support Panel
  - Alexis Baker, Jason Cluff, Roland Holliday, Peter Niketeas, Amanda Soderstrom
- Employment Choices Panel
  - Diane Butler, Jack Butler, Robert Lindfors, Roseann McGrath, Christopher Misiano
- Parenting PC Kids
  - Julie Bennett, Antonietta Giofu, Gwen Tate
- Teen & Young Adult Discussion
  - Stephen Wittmer
- PC-K6a Group Discussion
  - Jason Cluff, David Lock
- PC-K6c Group Discussion
  - Angela Haynes and Frances Smith
- PC-K16 Group Discussion
  - Kerry O’Keefe, Jessica White
- PC-K17 Group Discussion
  - James Andrade, Harry Stergar and David Hansen
- Q&A Closing session

Kids Program—led by Sally Despain, Chris and Jake Kaspar. There were many Kids Program activities and games including extracting DNA, a Mad Scientist who made slime-to-go, a PC kids discussion (led by Jim Rittle and Kylie Wittmer), and even a marshmallow attack on Roger Kaspar.

Pain Study—In addition to the presentations, panel discussions and breakout sessions, the final section of the 2014-2015 Pain Study and Dermatology Exams was completed at this meeting.

Special Thanks to our Medical & Scientific Experts
Jiang Chen, MD, PhD, Stony Brook School of Medicine, New York, NY
C. David Hansen, MD, University of Utah, Salt Lake City, UT
Alain Hovnanian, MD, PhD, Imagine Institute, Paris, France
Roger L. Kaspar, PhD, TransDerm, Inc., Santa Cruz, CA
Laure Rittié, PhD, University of Michigan, Ann Arbor, MI
Frances J.D. Smith, PhD, University of Dundee, Scotland UK
Eli Sprecher, MD, PhD, Sourasky Medical Center, Tel Aviv, Israel

Always working, collaborating and making plans, even during meals.
Above: Pain, Stigma and Support Panel
Below: Employment Panel

Question and Answer panel with the physicians and scientists
2015 PATIENT SUPPORT MEETING EVALUATION

OVERALL RATING 4.4 on a scale of 5

| Better than other meetings I attended | 6/19 |
| Same as other meetings I attended     | 11/19 |
| Not as good as other meetings I attended | 2/19 |

SCHEDULE

| Excellent | 27/35 |
| Okay      | 8/35  |

PRESENTATIONS

| Helpful | 34/35 |
| Not helpful | 1/35 |

PATIENT DISCUSSION

| Helpful | 30/35 |
| Not Helpful | 5/35 |

PSM COSTS & FREQUENCY

| Continue As Now * | 19/35 |
| Costs paid fully by meeting fees | 1/35 |
| Use Funds Only for research | 2/35 |
| More Often (Every Year in US) | 8/35 |
| Less Often (Every 3-4 years in US) | 5/35 |

WHAT LIKE BEST

| Connect with other patients | 17/32 |
| Connect with staff, researchers, doctors | 15/32 |

*Held every other year in USA; costs are $45,000+

Typical Positive Comments

- Breakout sessions are a very useful part of the meeting. You learn a lot and can relate to many things discussed.
- Breaks, meals and time with other patients were terrific.
- Employment session was excellent. It was impressive to hear meeting others with PC was the best part.
- It’s important to balance PSMs with research so holding PSMs every few years with web meetings in between might be best.
- Since the PSMs are costly it may be prudent to have attendees pay the full cost so money is used for research, etc.
- Jack’s letter was the best. (See this on the PC website front page.)
- Open discussion and meeting other PCers was my favorite part of the conference.
- Presentations gave me a better understanding of scientific aspects of PC.
- Schedule & meals presented lots of time to mingle and meet.
- Scientific presentations were in fairly short bursts that didn’t stifle the mind.
- There is tremendous value in meeting in person but it does not need to be paid for from PC funds. A much higher percentage of cost could be shared by attendees in a financial position to pay their own way with PC funds helping pay for first time.

Suggestions to improve

- No teen activities this year.
- Some of it was too scientific and over my head.
- Wanted a group activity; maybe to NYC.

Here are comments from PCers who attended their first PSM in 2015 to encourage you to attend next time.

No one puts pressure on you to show your pc or to talk.

You should definitely come. Everyone was nice and supportive. Everyone is like a big family there.

The atmosphere is supportive and the meeting is educational.

Just do it! You will be welcome and accepted. These people understand like no one else can. A very informative and positive experience.

Please join us. We can get one step closer to finding a cure.

Just Do It!!!!
WHAT’S NEXT?
At the end of the recent Patient Support Meeting this question was asked. It surprised us because we are not at a ‘stopping’ point. So, in our mind, the question really is ‘What is on-going?’

In the opening address for the meeting, Mary Schwartz presented some of the on-going and future goals and projects. Here is a summary of those points:

CONTINUING GOALS FOR PC IPCRR—We need to enroll every PCer; numbers do matter!
- Questionnaire—to gather accurate data from those with PC
- Genetic Testing—provided free so every one with PC can know their own specific gene/mutation and this data can drive research forward
- Publications—to introduce PC (an ultra rare disorder) and educate physicians and scientists with accurate information

IPCC—annual scientific meetings to attract the best and brightest and most cutting-edge researchers

CLINICAL STUDIES—we have completed two Phase 1b Clinical Trials through the FDA:
(1) siRNA (TD101). This holds the most promise, but is the furthest from being available to patients. We continue to work towards solving the delivery issues for siRNA.
(2) Topical Sirolimus-Rapamycin (TD201). The results of this clinical trial were presented at the IPCC meeting in May and again presented at the PSM in Newark. Only 2 of the 14 patients in the trial showed clear/good results. That is usually not sufficient for a trial to move forward. However, those patients who benefitted want this taken forward. And, the physician and scientist involved would like to further develop this treatment. Will funding be available to do that? No one knows for sure at this time.

We have many small studies on-going at all times involving a few patients in each study. Every study teaches us and leads us to additional studies. Since many of these are ‘off label’ studies, the results are not published, but the information is used for each step forward.
(3) Statins. 4 patients with mixed results.
(4) Botox. 12 patients treated so far. Mixed results.
(5) Topical Pain formulation. Studies in 3 or 4 centers will begin shortly.
(6) PC tooth study. Please send any teeth that are pulled or baby teeth that fall out.
(7) Imaging, biopsy and tape stripping studies are all on-going.

We all would like to have a cure for PC — something dramatic that absolutely ends the disorder. We’d like a gene change! In 2004, that was our goal — a cure! However, that type of treatment is not yet a part of the real world for even the most common genetic diseases.

PC is many different things—for most PCers (whether K6a, K6b, K6c, K16 or K17), the plantar pain is the thing they would most like changed. We know that the cysts are also a huge problem for many. We have attracted researchers to help us learn more and try options in these areas. We have studies on-going in many countries, many universities and some companies. We have also gathered information on thickened fingernails and toenails, nail removal, the ‘first bite’ pain, shoes/socks/insoles and other topics of information to PCers. All of the information is shared on the PC website.

PC PROJECT NEW GOALS
QUALITY MEDICAL CARE FOR EVERY PC PATIENT—with over 2000 rare skin diseases, we cannot expect medical experts to know about PC which is one of the rarest of the rare skin disorders. We must begin an aggressive Educational Outreach program including
- Continuing Medical Education (CME credit) for physicians and other medical staff to teach about PC. The costs for this range from $50,000 to $300,000 and up.
- Grand Rounds with multiple PC patients teaching doctors. The costs vary with each institution where these are held.
- PC Natural History which we will begin to compile. By updating your information in the registry regularly, you will help us create a PC Natural History – without the $million dollars$ otherwise needed.

Please help us build a PC Natural History by updating your IPCRR information. So far 42/690 have updated — that’s a start!
⇒ Adults—once every year
⇒ Age 6-18—twice every year
⇒ Under 6—four times a year
⇒ Under 1—every month

Please help us with this project.

PC PROJECT STAFF
Another is to add Staff Members to PC Project to achieve all our goals!
**PC AWARENESS**

June is PC Awareness month and we are beginning to hear about events we would like to share with you. We hope you will take these ideas or come up with others and raise awareness in your local area in June or anytime you can. Please let us know what you do.

Kate Fairbrother’s work has a charity day where staff pays for dressing down, then they have a bake sale, bingo and sweepstake. The PCer had them feature Pachyonychia Congenita as their charity this quarter and £1050 were raised for Pachyonychia Congenita. Read more at [www.compassltd.co.uk/over-1k-raised-for-ultra-rare-genetic-skin-disorder-charity/](http://www.compassltd.co.uk/over-1k-raised-for-ultra-rare-genetic-skin-disorder-charity/)

The van der Laan family asked a local vegetable shop, The Pumpkin to raise funds and awareness. The store sells banana boxes for 1 euro and donates funds raised to Pachyonychia Congenita.

The Cruz family sent an email to their friends and family about PC June Awareness. There have been five donations already from that one email.

Victoria Barbier put donation boxes at local newsagent stands.

**Annamarie Baulmer** is selling special PC Nail wraps.

The Jennings Family is having an online facebook Jamberry nail wrap party all month long. You can view her fundraiser at [www.facebook.com/events/1651877531714380/](http://www.facebook.com/events/1651877531714380/)

Richard Stedman completed a 100 mile endurance horse race. You can read his wife’s blog about it at [enduranceintrospection.com/an-old-wifes-tale-od-100/](http://enduranceintrospection.com/an-old-wifes-tale-od-100/)

The March 2015 PC NewsBrief, reported a challenge at TransDerm between co-workers Andreas Berroth and Roger Kaspar. Unfortunately, Andreas didn’t reach his goal and lost. However, the challenge brought in a $400 donation ($300 from Andreas and $100 from Roger) to PC Project. Roger and all the scientists at TransDerm said they are open to any challenges either individual or as a team. Contact PC Project with your challenge to TransDerm.

At the patient support meeting last week, “Give Me Some Skin” sweatshirts and PC t-shirts were auctioned off and other attendees bid to raise $1,210.

Also, during the meeting, Roger Kaspar, challenged attendees to a swimming competition. Roger said he would donate $100 if he lost. Theo Misiano won the competition and Roger made the $100 donation to PC Project.

Whether you plan a big or small event, hold a dinner, send a letter, challenge TransDerm or post about PC on social media, it all counts and makes a difference. Let us know your plans. Post on the PC Awareness Facebook event page at [www.facebook.com/events/835912403138164/](http://www.facebook.com/events/835912403138164/) or send us an email to info@pachyonychia.org. We would also love any photos from your event. We are excited to hear about your activity. We are grateful for all your efforts and being engaged in the PC family ‘hive.’

“I guess this side won. At least my wallet is lighter!”

Andreas Berroth, PhD. PC Fellow at Stanford University
**PC News Brief**

**July 2015**

**Vol 10, No 8**

**PC STUDENT STORIES**

We will feature *PC Student Stories* each month. School days can be difficult at times for those with PC and your story may be just what another PC family needs to read to encourage them and their students. We welcome more PC Student Stories and would like to include yours!

**Theo Misiano** won a vocabulary competition based on completing analogies. The competitive meets are held 3 times a year and students learn 75 new words for each meet. The tests have 20 questions each time. Theo was the only 5th grader out of 23,000 participants to get a perfect score at every meet for a total of 60/60! Way to go, Theo! *(Theo is the son of Chris and Hilary Misiano.)*

**Evan Delanty** graduated from 5th Grade this spring and will be entering his first year of middle school in the fall at Three Rivers Charter School in West Linn, Oregon. Of particular significance, Evan embraced his IP Independent Project, A seasonal academic challenge of each student’s choice. Evan chose to study Medical X-Rays and did so with focus and depth for over 2 ½ months. He embraced his IP experience fully and invested considerable energy into each of his projects. We are very proud of Evan’s willingness to jump right into a new challenge and come out with a smile on his face and a positive attitude. Evan received an Exemplary grade for Academic Achievement and an Admirable rating for his Efforts in his IP Project of Medical X-Rays. In addition, Evan also received Exemplary grades in Math, Study Skills and Art. Evan just completed his fifth year of private piano lessons! Great work, Evan!

**Ava Delanty** successfully completed her first year of high school and will begin 10th grade in the fall at West Linn High School in West Linn, Oregon. A serious student, Ava had a full college prep course load including Japanese and AP Human Geography. Her hard work placed her as a member of the “Lions Den” which honors WLHS students who have earned a weighted GPA of 3.5 or higher for both first and second semesters. Ava became a member of the WLHS Debate team and has just completed her sixth year of private piano lessons.

An active Senior Girl Scout, she participated in TAMBU, an outdoor skills competition for scouts in the 6th-12th grades where without parental help girls compete in skills such as knots/lashing, Dutch oven cooking, orienteering, backpacking/camping skills, fire building and more. During the summer 2015, Ava served as a Program Aid for a Girl Scout day camp where she is counselor for a group of 4th grade girls.

Ava received a certificate of Standards of Mathematical Practice/Repeated Reasoning in Geometry. We agree with her teacher who wrote, “Thank you for always looking for patterns and generalizations that allow for shorter methods of solving problems without compromising the integrity of the problem.” Great accomplishment!
On Spring Break 2015, the family visited Japan where they took a cooking class and an air tram over Mt. Fuji. (Evan and Ave are the children of Sarah and Eddie Delanty.)

Kylie Wittmer, 19 years old, graduated in May from Greenville Christian School in Greenville, TX. She received the Jonathon Award for being the best friend to all students. In August, she will be attending the University of Mary Hardin Baylor in Belton, TX. She will be majoring in film studies.

Ashley Wittmer, 20 years old, has just been awarded a $3,000 scholarship for making the President’s List (for full-time students with a 4.0 semester average) two semesters in a row at the University of Texas-San Antonio. She is on track to graduate next spring (one year early) with a BA in Public Health concentrating in Epidemiology. She will then continue with one more year of classes to prepare her for Medical School. (Ashley and Kylie are the daughters of Stephen and Ali Wittmer.)

Henry Martin Bjelkerud-Altbark a toe-tapping star! “Of course! I would like to BRAG about Henry who had his first ever show at kindergarten - taking center stage and continuing to dance no matter what letter we were singing about.” (Henry is the son of Mikael and Martina Bjelkerud.)

Nathan Schwartz was Valedictorian of his class at Cottonwood High School in Salt Lake City. He received numerous awards and scholarships and was cheered by his many friends and classmates. Nate is leaving to serve a two year mission for his church and will return to Brigham Young University in 2017.

Sam Schwartz is an honors student at Utah State University. This summer he received a grant from the university’s College of Science for a project which overviews a variety of mathematical models behind Latin American voting systems. As Sam says “although a little nerdy, the project is starting to uncover some really interesting results.” Sam hopes the project will, if nothing else, lead to a fascinating capstone experience for his undergraduate career as he pursues his double major in math and computer science. (Sam and Nate are the sons of Janice and David Schwartz.)

Allison Block is 5 years old and in kindergarten. While riding to her 4K class, Allison looked at her fingernails and told her mother that she has lucky fingernails. When asked what she meant, she said her nails are lucky because they don't break or snag like her sister’s or Mother’s or Daddy's. She said sometimes she pinches one and the nail will hurt and eventually fall off, but her nails don't break so she is lucky. This surprised her mother because just a couple weeks ago she pinched a finger and had a really swollen sore finger for about a week. A week later her nail fell off. Her mother says “From my perspective it was much worse for her than anything I have with my nails but Allison doesn't know any different. I thought it was an interesting perspective that she came up with out of the blue. She is a tough kiddo.” Great attitude, Allison!

Please brag a little and share your stories or stories of students in your family (from pre-school to graduate school). What school are they attending? What do they like most? Have they received a good grade? Or an award? Help us tell these great PC stories!
THE PC E-MAIL BOX

Email from a young adult about the June 2015 PC News Brief: “I LOVED this news brief!! This email just made us smile all day. Thanks!

Email from a mom: I see that I can buy PediPaws on Amazon Smile, where PC project gets a tiny percent--this is how I do all of my shopping! I hope you’re seeing the proceeds. YES! YES! YES! We receive a percent from all of you who shop at Amazon Smile. And if you sign in to Amazon Smile through iGive.com, we get a little more!

SPECIAL until July 31, if you sign up for iGive.com, we receive $5.00 — and it costs you nothing. Please sign up and shop for PC.

Email from a mom: I have one of his teeth that has fallen out (he thinks the tooth fairy has taken them.) Would you like it for your study? We will also update his registry information. YES! We do want to have any teeth that are available. And, thanks for remembering to update the IPCRR data.

PC SOCIAL MEDIA

Facebook post June 2, 2015: “One question PCers, where are you? City, state, country? “ Did you know there is an interactive map of all PCers on the PC website? Here’s a link—enjoy! http://www.maptive.com/ver3/PCers

DEVELOPMENT DIRECTOR

I feel very blessed to start a brand new position with PC Project. I am a PC patient along with many others in my extended family. My activities will be focused on raising funds to meet the objectives of PC Project which are “Fighting for a cure. Connecting & Helping patients. Empowering Research.” My first priority will be to activate the PC community regarding fund raising efforts. I plan to “reach out” to those who have helped in the past and to ask them again to help with the burden of supporting our organization. Also, I will be calling on some to take on the larger task of helping other patients getting started with PC awareness and fundraising. For years, PC Project has carried the majority of the financial burden for many projects such as genetic testing, PC registry maintenance, and patient studies. In order to have long term success, we must all work together and do whatever amount, small or large, to help contribute to the cause. I look forward to getting to know you all as I begin my endeavors. Please contact me anytime. Stephen Wittmer Stephe witten @pachyonychia.org.

REPORT ON PC AWARENESS

We are grateful to many in the PC community who participated in PC Awareness Month—June 2015.

JULIE PECONI writes “although I haven’t been doing much creating lately I have been doing PC Awareness throughout the year through our craft business, selling online and at school fetes. So far we’ve raised £130.” This reminds us that PC is every day and all year long. Our efforts for PC Awareness need to be on-going with special events held whenever most convenient in your local areas.

AUTHOR DONATING PROFITS FROM BOOK SALES

Armel Abundis is donating in honor of his daughter, Catana. We’ve received $100—thank you!

ANNUAL BAKE SALE

Once again the Block family of WI raised funds with their bake sale at Nine Mile County Forest during the WORS mountain bike races.

ADORABLE ONE YEAR OLD

Naomi Merriman’s family raised $460 for PC Project with a flyer to friends and family. A sponsor will match this $2-for-$1. The flyer is attached—it is a great example!
You can now enjoy a new story on the PC website as Jack Padovano shares his ‘PC Living With Me’ experiences.

We invite you to share your story on the website. If you have questions, just send us an email and a photo.

Jack has also joined the PC Project Board of Trustees. To see information on all of the Board of Trustees and Medical and Scientific Board Members, click on the About PC Project tab.

You can learn a lot about PC by using the various charts and graphs available on the PC Data tab. This is where the interactive map showing where each genetically confirmed PC patient is located. The IPCRR Summary Data combines your answers to the questionnaire with all the others in the registry — so you can see where you fit in the PC syndrome. If you’d like to have a copy with your own data answers marked, please let us know. We prepare this for all of the Educational Outreach sessions where PC patients work to help physicians learn more about the ultra rare skin disorder, Pachyonychia Congenita.
I am working on my second month as Development Director, and I have had many good talks with some of you. I am learning how PC affects each one of us differently. My personal opinions of what PC consists of are being changed. Before working here, I only knew the PC in my extended family. We have an unusual K6a mutation which is located at a ‘splice site’ on the gene. This causes variations in PC across family members. Now, I am learning how others deal with PC in a whole different way. For instance, for those with K17, who suffer from painful cysts, I learned that the pain aches throughout your entire body and that even a couple of cysts can keep you awake at night. Due to my thinner finger nails, my PC can be hidden with the exception of limping when I have walked too much. Yet, others with PC must constantly endure looks, glances and questions about their thick nails. I understand now, more than ever, that PC affects us both socially, emotionally and physically.

I want to thank all of you who have donated your time, money and items to raise funds for PC Project. One of you graciously donated a one week beach vacation to be auctioned off at a golf tournament. Others have been inspired to go outside of their comfort zone and ask a friend or acquaintance to donate. Our kind board chair, Lex Udy, set up an appointment with a longtime friend who owns a real estate development firm. He showed him our wonderful fund raising DVD, and then, gave him a plea that included the fact that helping PC Project also helps other diseases as well. His friend became emotionally connected and he gave from his heart. He pledged $2,000! The key to me is that Lex took the initiative to set up a specific time to meet with this friend.

Some of you have been so kind to include PC Project in your Estate Planning. I want to encourage others to do the same if you have the means. It is a great way to donate to PC Project and at the same time pay fewer taxes. The best way to get started today, would be to contact your estate attorney. In the upcoming months, I will provide greater detail on including PC Project in your Estate Planning.

I would like to encourage everyone to use our website as a valuable source of information. I am still going there to look at the video archives from past meetings, to view charts of where we are located, to easily donate, to view past issues of the PC NewsBrief, to update my information on the Registry, to look at past fundraising ideas, to read peer-reviewed articles in scientific journals and more. Remember to share the website link with physicians, educators, and possible donors so they can visit the website for information.

Behind the Scenes

One of the many ‘behind the scenes’ tasks being taken care at PC Project are the monthly meetings with the IPCC Genetics Team. New case studies for those being genetically tested are prepared for review by the team. This monthly meeting allows the International Pachyonychia Congenita Research Registry (IPCRR) to run efficiently with a very high rate of success in identifying the specific gene and mutation for hundreds of patients. Having a well maintained registry sets us apart from many other rare disease organizations, but it requires a lot of resources and hard work behind the scenes by PC Project staff.
PC-K17 PARENTS MEETING
PC Project recently sponsored a web meeting for parents of PC-K17 children. Several PC patients participated in answering questions and sharing experiences. Here are some of the questions/answers which we think will be important to all PC families:

QUESTION: Our daughter is now 2 1/2 years old. At what age do parents recommend telling one's daughter that she has PC and how have you done so within your families -- particularly those where the child is the only one in the family with PC?

ANSWER: PC-K6a Parent
I don't recall a special way of telling my PC children. They just knew they were different from others in the household who didn't have PC and as the differences were noticed, they were talked about as needed.

ANSWER: PC-K17 Patient
Well seeing I didn't know I had PC until I was 16, I lived with it long before I knew what PC was.

ANSWER: PC-K6a Parent
My daughter is 5 years old. She knows she has callouses and her feet and nails are different. I never really sat her down and told her she has PC. I dealt with it one question, issue at a time. When she was about 3 1/2 she asked why her feet were different than her sister’s. Our family and friends all know about her PC. We try to be very open with anyone who asks. Her older sister knows that her sister has PC. Again we never sat down and had a discussion but have answered questions as they come up.

ANSWER: PC-K17 Patient
I was just diagnosed (as an adult) and my parents or myself never knew what it was. I am just learning of the disease now. I am very excited to have someone (doctors, others with PC) looking closer to find something that helps.

QUESTION: What strategies have you employed / taught your children to employ to deal with teasing about their nails, calluses, etc?

ANSWER: PC-K6a Parent
Role plays, talking about it before it might ever happen about how to respond. Teach them how to simply answer questions if asked. Talk with teachers beforehand as needed to let them know about your child's needs. Each child is different. Some may not want any extra attention. Some may need it.

ANSWER: PC-K17 Patient
I also am a spontaneous case so no one in my family could help me. I learned to soak and trim calluses when needed. Seeing my feet did not bother me till school age.

ANSWER: PC-K6a Parent
My child is better at answering other kids questions than I am. She will simply say that it is just the way her nails/feet are made. At 5 that is all she has had to do and the other kids have always just accepted it. We also taught her from a young age that God makes everyone different and special.

ANSWER: PC-K17 Patient
Never let anyone push you around or make fun of you. Bully's will go away.

ANSWER: PC-K17 Patient
I was just diagnosed (as an adult) and my parents or myself never knew what it was. I am just learning of the disease now. I am very excited to have someone (doctors, others with PC) looking closer to find something that helps.

QUESTION: What advice can you offer to parents to best support our child - both emotionally and physically?

ANSWER: PC-K6a Parent
Love your child, raise them in a happy, stable family. Focus on their strengths, what they can do. Still give them chores and responsibilities within their abilities, spend time with them. When things are hard and they hurt, you can simply say, "I'm sorry this is hard." Show empathy but not pity. But let them know you're still glad they're part of your family. Let them know they have much to contribute in life and to other people. Teach them to serve others and to think of others, especially by example or having them serve with you.

ANSWER: PC-K17 Patient
Of course think it's much tougher being a female with PC. (As a boy) I was talked about as a child and seemed to grow out of it quickly.

ANSWER: PC-K6a Parent
Stay open to her questions. Help others (teachers, daycare, friends) understand PC. Let your daughter choose what she does and doesn't do. It's very tough but you must teach them to employ to deal with teasing about their nails, calluses, etc. Help them when needed. Teach them to serve others and to think of others, especially by example or having them serve with you.

ANSWER: PC-K17 Patient
But let them know you're still glad they're part of your family. Let them know they have much to contribute in life and to other people. Teach them to serve others and to think of others, especially by example or having them serve with you.

ANSWER: PC-K6a Parent
Still give them chores and responsibilities within their abilities, spend time with them. When things are hard and they hurt, you can simply say, "I'm sorry this is hard." Show empathy but not pity. But let them know you're still glad they're part of your family. Let them know they have much to contribute in life and to other people. Teach them to serve others and to think of others, especially by example or having them serve with you.

ANSWER: PC-K17 Patient
Stay open to her questions. Help others (teachers, daycare, friends) understand PC. Let your daughter choose what she does and doesn't do. It's very tough but you must teach them to employ to deal with teasing about their nails, calluses, etc. Help them when needed. Teach them to serve others and to think of others, especially by example or having them serve with you.

ANSWER: PC-K6a Parent
Stay open to her questions. Help others (teachers, daycare, friends) understand PC. Let your daughter choose what she does and doesn't do. It's very tough but you must teach them to employ to deal with teasing about their nails, calluses, etc. Help them when needed. Teach them to serve others and to think of others, especially by example or having them serve with you.
**QUESTION:** When does the pain start? **NOTE:** This varies with each individual and each type of PC is different on when callus begins; some with PC have little or no callus.

**ANSWER:** PC-K6a Parent
The worst pain and walking came in elementary school. Not sure there was anything to be done to delay the inevitable. As a middle aged adult, the pain is almost 24/7, but in younger years, I was able to attend school, including college, work, etc. Not sure if I would have been up to that with the sharp pain levels I have now. I think everyone is different though.

**ANSWER:** PC-K17 Patient
Again I don't remember cysts as a child. I do remember the small "pimples" around my waist and under my armpits. Cysts are now my biggest PC problem. I go to a surgeon 2 or 3 times a year. Most of them I lance myself after hours of soaking and using topical lotions to soften it up. I'm not saying to do your own surgery but I do what I need to do :-)

**ANSWER:** PC-K6a Parent
The calluses started shortly after she started walking. Her first blisters were around 8-9 months when she was standing more. At first they would heal and fall off, now she has constant calluses at age 5. Using lotion regularly helps some. We try to make sure she has good shoes. I find if her shoes start to get tight or small her calluses are worse.

**ANSWER:** PC-K17 Patient
At the time of puberty is when most flare-ups with cysts start or get worse; also worsens with menstrual cycles. **DO NOT TRY TO POP A CYST LIKE A PIMPLE!** My parents used to hold me down when I was 5 and try to open it with a needle, then proceed to squeezing. This doesn't work and causes more aggravation to the area. Hot compresses or baths and time for it to head is all you can do unless the doctor lances it (or injects it).

**QUESTION:** Is there anything you know now in dealing with PC that you wish you had known years ago and/or that you'd tell someone with PC -- whether in terms of coping strategies and/or particular items (clothing/products?) that were helpful/harmful to you?

**ANSWER:** PC-K6a Parent
Do all you can while you can. Remember life is good and find the good, even in the small things, in each day. Enjoy the journey of life. Read the PC website for care ideas and then try out what works best for you. Remember not all people with PC are the same so you have to find what works best for you. Don't compare yourself to others but know there are some great tips out there that just may work for you. Again, search the website for the best ideas..

**ANSWER:** PC-K17 Patient
The biggest thing in my life was meeting other PC people just to relate and know I'm not alone.

**ANSWER:** PC-K6a Parent
Shoes that breath and are the right size. We buy new shoes more often. [Remember—SOFT SHOES]

**ANSWER:** PC-K17 Patient
I cannot wear any tight clothing or underware because of the cyst.

During the webmeeting, we also shared PC Data from the website showing where patients live, the various mutations and more.

**CARE TIP CORNER**

**NOTE:** We have just learned that a portion of our PC Wiki with information on tools is not visible on our site. We are working to fix this as quickly as possible.

All tips we receive are available on the PC Wiki which we update every few weeks. Please send us any/all care tips and we’ll share in every way we can.

“June 2015: While I was at the patient meeting, I heard someone talking about having to wear safety shoes or boots at work and how uncomfortable they were. I wanted to mention an alternative called OSHATOES. OSHATOES slip over your regular shoes and have composite safety toes that meet the exact same OSHA standards as steel toed boots. Just another option.”
MORE AMAZING PC KIDS

Timmy Baker, of Wales, UK has recently completed Year 3 and is looking forward to the summer! Although he has several calluses on his feet this has not stopped him yet from playing his favorite sport this year: football. He had a successful season with Mumbles Rangers and was even chosen Player of the Season! He has also earned his green belt in karate.

Timmy enjoys many activities which don't require him to be on his feet- one of his favorite things to do is mountain biking. He recently came third in a cross country race! He also plays the cello and was a runner up in his school talent contest for playing. He loves math and was selected to take part in a small group in school which was given more challenging math problems to work on.

He also received two head teacher awards this year: one for excellence in problem solving and one for good manners and being polite. We are really proud of Timmy! NOTE: Timmy and his dad are featured on the PC website front page. We'd love to have photos and stories from you/your family.

5TH ANNUAL PC GOLF TOURNAMENT

For the last five years, the PC Project staff has organized a golf tournament as our local fundraising program. We wanted to tell you about two amazing employees who work in the same building where PC Project offices are located. We decided to share a flyer with the employees and ask them to help us gather raffle prizes. Here is how two employees responded to our invitation—

1. One man, a gifted potter, gave us one of his beautiful hand thrown vases (value $125)
2. Another employee, a very shy lady, asked Home Depot for a prize and they gave her a Shop Vac! She said she is usually too shy, but she told the manager she knew the people and knew the money went to research. She has two other places that have told her 'yes' but she hasn't yet picked up the items. AMAZING! What an example she is of someone who just tries -- and look what happened!

We are sharing this because there are many in your community who will be glad to help you with your projects to raise funds for PC research. We are doing our best to continue our $2-for-$1 match on all of your efforts.

THE PC E-MAIL BOX

We had a great question from a PC high school student who is beginning to think about applying for college admissions. This student is a good student and accomplished in student government, etc. Her question is “Should I include information on PC and how it has helped me become more compassionate, etc. in my college admissions essay? Have any others included information on PC in their essays? What about for scholarships?”

We had several really good answers—all saying 'yes' tell how PC has made you a better person!

Here’s an answer from one mom: “I think there was one essay in particular where my son wrote about swimming and how it was something he could excel in even with PC. I don't remember him writing in a way that evoked pity, but more something that became a strength. Frankly, the world of applications and scholarships is so competitive, I absolutely think anything a student could do to distinguish him or herself from the many others is a good thing. I don't mean it to be something to make people feel sorry for you, but anything to give you an edge, make you different, make them think you are different and can add to their program/school/etc. is a good thing. And yes, that may mean play the PC card - meaning, the pachyonychia congenita card - in a positive way, of course. For many of us with PC, we don't like to be different, but in this very competitive world of applications and scholarships, I think being different is good. Does that sound just awful? If two people have exactly the same basic qualifications, you must find something that puts you just above the rest. Appealing to the human heart is not a bad thing and people do respect others who have overcome hard things - probably because everyone must overcome hard things. So again, not to ask for pity - no one gets an award because someone feels sorry for but to share a human story of triumph. That's okay, I think.

It's true, if the core qualifications are not there, nothing can help that. Still, it's back to if two people are just the same and they are competing for the same scholarship, admission slot, etc., the trick is to get them to take YOU. You need to show you are not a cookie cutter student, but someone unique and special who would add to their school or program.”
**Topical Rapa (Sirolimus)**
Since the trial data is now locked, and the post-trial data collected, we are pleased to be able to publicly report some findings from the study.

**Post-trial Questionnaire**
Thinking about your PC while in the study, which PC feature was your MOST bothersome feature? PAIN 11/11

<table>
<thead>
<tr>
<th>Q27: Did TD201 help?</th>
<th>One foot seem to be better than the other, less pain and the calluses didn't grow as fast</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Softer, less initial and long term pain, reduced callous, easier to abrade</td>
</tr>
<tr>
<td></td>
<td>I would actually put a maybe. I had some really good days I didn't expect, but since the calluses never went away and I still hurt overall, it's hard to be sure.</td>
</tr>
<tr>
<td></td>
<td>I felt a decrease in callus thickness, and I noticed a Better texture to the calluses- more dry with less blood vessels.</td>
</tr>
<tr>
<td></td>
<td>It helped one foot for awhile than screwed the other one up and both feet were compromised.</td>
</tr>
<tr>
<td></td>
<td>Softening the callous and reducing the burn.</td>
</tr>
<tr>
<td></td>
<td>Odor control and temperature control, feet were cooler and not as sweaty.</td>
</tr>
<tr>
<td></td>
<td>It seemed to slow the growth of the callous</td>
</tr>
<tr>
<td></td>
<td>Although it did not stop the growth of callus, the growth was slowed down</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Q29: Changes after trial?</th>
<th>Didn't noticed much until after a couple of weeks and the pain is stronger and continued to the next day or longer</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Feet went back to 'normal' eg thicker and harder calluses. more red spots.</td>
</tr>
<tr>
<td></td>
<td>More blood vessels and thicker callus</td>
</tr>
<tr>
<td></td>
<td>After stopping the drug I noticed more flare ups in the form of blisters which made me realize that I had fewer flare ups during the clinical trial overall. My symptoms seemed to even out during parts of the trial.</td>
</tr>
<tr>
<td></td>
<td>My feet are dry and cracking again and they burn on a regular basis.</td>
</tr>
<tr>
<td></td>
<td>Odor increased. feet were warmer and became more uncomfortable quicker.</td>
</tr>
<tr>
<td></td>
<td>Callouses thickened back to what I consider &quot;normal&quot; more quickly.</td>
</tr>
</tbody>
</table>
NATURAL HISTORY STUDY FOR PC
A major effort is underway at PC Project to create, for the first time, a ‘natural history study’ for Pachyonychia Congenita. While such a study usually costs $300,000 up to $1,000,000, our plan is to create this valuable PC Natural History dataset with little or no cost by having individuals regularly update their information in the Int’l PC Research Registry (IPCRR).

You may have some questions, such as:
• What is this?
• Why is so important?
• How will this really help me or my children/grandchildren

We are sharing slides from a presentation by Dr. Anne R. Pariser of the FDA which we feel clearly explain how important this PC Natural History Study is — if we hope to ever have any drug approved for PC (in the USA or elsewhere in the world.) Here is what we are asking you to do
• Adults – every year
• 6-18 – twice a year
• Under 6 – 4 times a year
• Under 1 – every month

UPDATE YOUR IPCRR DATA NOW
To make updating even quicker, we have implemented a suggestion from Carl Gogol. It should take each person less than 30 minutes to update their IPCRR data. Create a million dollar asset for PC research just by updating your IPCRR information! Here is the link to update https://registry.pachyonychia.org/s3/UPDATE

IMPORTANCE OF NATURAL HISTORY (NH) STUDIES IN RARE DISEASES
Anne R. Pariser, M.D.
Associate Director for Rare Diseases Office of New Drugs
Center for Drug Evaluation and Research

Natural History Studies
• Purpose: To inform drug development
  – Marketing approvals require design and conduct of adequate and well-controlled studies
  – Designing A & WC studies requires a scientific foundation upon which to build
    • Knowledge of disease NH is an essential element in the scientific foundation of any clinical development program
  – Rare diseases, in general, are poorly understood
    • Important and essential role for NH studies in rare disease drug development (IND phase) to facilitate efficient clinical development

Rare Diseases and Orphan Drugs
• What is different about rare diseases and Orphan drugs?
  – Diseases are usually poorly or incompletely understood
    • Generally, the lower the prevalence, the less well we tend to understand them
  – Small populations
    • Limited opportunity for study and replication
  – Highly heterogeneous group of disorders
    • 7,000 different diseases
    • Often high phenotypic diversity within individual disorders
  – Usually little precedent for drug development within individual disorders
  – Often requires more (and more careful) planning than non-Orphan
    • Need a solid scientific base upon which to build an overall program
Importance of Natural History Studies in Rare Diseases
by Anne R. Pariser, M.D. (continued). If you would like a copy of Dr.
Pariser’s full presentation, please request at info@pachyonychia.org

Key Points

#1 NH data contribute to scientific foundation upon which
 drug development programs can be built
  - Rational, scientifically-based drug development requires an
    understanding of the disease
  - NH describes the disease - independent of individual
    investigational agents
  - Most informative when NH study data are available early in
    development
    • Ideally before design of efficacy trials

#2 Patient and caregiver involvement is important
  - Engage all stakeholders early and on an ongoing basis

Key Point #3

- Monolith\(^{10}\) (mon • nth • lith)
  - an obelisk, column, large
    statue, etc., formed of a single
    block of stone
  - Something having a uniform,
    massive, redoubtable, or
    inflexible quality or character

Rare diseases are a highly diverse collection of disorders
  - Design and conduct of clinical development programs are
    highly individualized
  - Dependent on disease and population under study,
    understanding of the intervention and its expected impact on
    the disease

Key Points #4

Drug development as a continuum
Efficiency ≠ corner-cutting

THE PC E-MAIL BOX

Email Treasures from a fantastic teen!
We met for the first time at the PC conference this past June. I'm the baseball player!
I apologize for how long it took me to send this thank you, but I can not describe how thankful I was to attend the conference.

After the first night, I went back to my room, and I was literally in tears. Seeing someone with PC, who I could relate to, was the most humbling feeling ever. It was so refreshing to actually talk to someone that truly experiences the same things that I do. I'm struggling to convey in words my emotions from the meeting, but I don't think words can justify how I feel. I apologize, but I can assure you it was life changing. So thank you! You are truly the best!!

...and from a PC ‘senior’
Thank you for your Aug 2015 PC newsletter. This was an exceptional newsletter - very interesting and informative - one of your best. I am a 66-yr-old PC'er (K6a spontaneous mutation), and reading how parents are teaching their children with PC about this condition made me think back (many years) about how my parents handled this same situation and how they never held me back from doing anything I wanted to try.

I'm sitting here with very sore, painful feet today, wishing I could do more, like I was able to do when I was younger. The only things I can do to help with PC research at this point is (1) leave PC Project a donation in my will

2386 East Heritage Way, Ste B, Salt Lake City, UT 84109 · www.pachyonychia.org · Phone 877-628-7300 · Email: info@pachyonychia.org
(which I have done), and (2) continue to volunteer to participate in any future PC research studies.

**DEVELOPMENT**

**Funding PC Project**

Our Board of Trustees has a committee headed by Jack Padavano working to create a Development Plan to help us focus on how to raise the funds needed to carry forward the work at PC Project.

We want to continue the research, registry, genetic testing, publications, clinical studies, patient meetings, website updates and other program services that have been the core of PC Project since 2004. To do this, we must fill the staff positions and we have to find ways to increase the funds available to PC Project. The Development Plan will be a key in this.

Stephen Wittmer recently helped us in making patient contacts and other projects in the months he served as Development Director. We appreciate his service and wish him well in his new opportunities. Stephen will continue as our first PC Advocate and will represent PC at the American Academy of Dermatology Legislative Conference in Washington, DC this month.

If you order be sure to let him know you support PC Project. If you order be sure to let him know you support PC Project.

**Book Sales**

Armel Abundis, father of a PCer, has sent $100 donation from book sales. He has offered 1/2 of the profits from his most recent book. If you order be sure to let him know you support PC Project.

<table>
<thead>
<tr>
<th>BOOK</th>
<th>COVERS</th>
<th>DESCRIPTION</th>
</tr>
</thead>
<tbody>
<tr>
<td>Two Asturians</td>
<td>[Image]</td>
<td>Armel Abundis, father of a PCer, has sent $100 donation from book sales. He has offered 1/2 of the profits from his most recent book.</td>
</tr>
</tbody>
</table>

**Thank you for shopping with PC Project in mind. The money raised this quarter was enough to cover the cost of 4 saliva kits for genetic testing for four PC patients. We can receive much, much more if every one will remember to use iGive.com when you search or shop. Do you like to shop on Amazon? If you access Amazon.com through iGive your purchase will raise .8% through iGive and .5% through Amazon Smile = 1.3%.

If you go direct to Amazon.com, we receive nothing. If you go direct to Amazon.Smile, you miss out on earing the extra .8%!

Going through iGive, your shopping is exactly like Amazon (or any of the other hundreds of online shops.) It costs you nothing — and could help PC Project. Let’s remember to use iGive as our entry to shopping online and select Pachyonychia Congenita Project.

**DEVELOPMENT**

Funding PC Project

Our Board of Trustees has a committee headed by Jack Padavano working to create a Development Plan to help us focus on how to raise the funds needed to carry forward the work at PC Project.

We want to continue the research, registry, genetic testing, publications, clinical studies, patient meetings, website updates and other program services that have been the core of PC Project since 2004. To do this, we must fill the staff positions and we have to find ways to increase the funds available to PC Project. The Development Plan will be a key in this.

Stephen Wittmer recently helped us in making patient contacts and other projects in the months he served as Development Director. We appreciate his service and wish him well in his new opportunities. Stephen will continue as our first PC Advocate and will represent PC at the American Academy of Dermatology Legislative Conference in Washington, DC this month.

**Book Sales**

Armel Abundis, father of a PCer, has sent $100 donation from book sales. He has offered 1/2 of the profits from his most recent book.

If you order be sure to let him know you support PC Project.

**iGive.com**

Change online shopping for good.

<table>
<thead>
<tr>
<th>website</th>
<th>affiliate</th>
<th>percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>iGive</td>
<td>AmazonSmile</td>
<td>.8% through iGive and .5% through Amazon Smile = 1.3%</td>
</tr>
</tbody>
</table>

If you go direct to Amazon.com, we receive nothing. If you go direct to Amazon.Smile, you miss out on earing the extra .8%!

Going through iGive, your shopping is exactly like Amazon (or any of the other hundreds of online shops.) It costs you nothing — and could help PC Project. Let’s remember to use iGive as our entry to shopping online and select Pachyonychia Congenita Project.

**MASTER THE ART OF PHILANTHROPY**

**4 WAYS TO MAKE YOUR GIFTS GO FURTHER**

We found the following ideas really excellent in a brochure from a local hospital. PC Project (aka PC Fund) is a 501(c)(3) public charity in the USA. As each state and each country have different laws, please consult with your bank, attorney or financial advisor as you consider these options for furthering the work for Pachyonychia Congenita.

- **Pocket the tax deductions.** When you make a donation to PC Project now, we both can benefit. Your generosity enables us to continue working diligently toward our goals and you receive an income tax charitable deduction (USA) when you itemize.

- **Keep your accounts intact.** An easy way to make a generous gift is to make a bank account payable on death (POD) to Pachyonychia Congenita Project. The savings are there for you if you need them during your lifetime, after which we receive them to expand and continue services for PC patients.

- **Eliminate the tax bite on retirement plan assets.** Taxes can consume a substantial portion of your retirement plan assets after your lifetime. For this reason, many people use their IRAs to make gifts to charity, such as PC Project, eliminating the taxation of these assets.

- **Live now, give later.** Many people consider gifts through their estates the perfect way to provide support because they’re able to help a cause that has meant a lot to them or a loved one without immediately giving up assets.
**PC AWARENESS**

Although June is PC Awareness Month, a fundraising effort can be held anytime.

**YARD SALE**—A $175.00 donation was received from Jessica White and her son, David Mortier. This seems to be an annual event for Jessica and David. We appreciate their efforts.

**GOLF TOURNAMENT**—Our local awareness event was held on August 22, 2015 at Crater Springs Golf Course in Midway, UT. We collected over $4000 in raffle and auction prizes. Start by contacting local businesses online for donations. One idea you can use for your local PC Awareness events is to contact universities in your area/state. We received sports baskets from four different Universities.

A week stay in Myrtle Beach, SC was donated by PCer Jamie Callahan-Simpson sold at auction and with the matching funds will bring $3000 to PC research.

This is our 5th year to host this local PC awareness event. We had 138 golfers including Utah Attorney General Sean Reyes and former All American football tight end Chad Lewis. Each member of PC Board of Trustees participated by golfing or sponsoring golfers.

We hope the total profits will be over $10,000 this year — and that those attending will become donors or supports for PC Project and be aware of this ultra rare disease.

**PLAN NOW TO ATTEND**

Starts Friday
October 28

Ends Sunday
October 30

Edinburgh
Capital Hotel

Registration will begin in October 2015

The Question and Answer feature was so popular last month, we want to make it a regular monthly feature. Please send your PC Questions to info@pachyonychia.org.

We’ll print questions and answers monthly as those are available. This month’s question is “Where would you like us to hold the 2017 PC PSM?” Here is the link to the survey so you can vote for your favorite USA city for the 2017 Patient Support Meeting.

https://www.surveymonkey.com/r/PCQuick9

**NEW OFFICE MANAGER**

We are delighted to introduce and welcome our newest staff member, Jessica Martin. Jessica joined PC Project on September 1 as our first ever PC Office Manager. She brings experience, skills and a delightful attitude.

Jessica has been married for 18 years to Nate Martin. She is the mother of two boys, ages 16 and 11. She was raised in Edmond, Oklahoma, moved to Utah while in high school and immediately fell in love with its mountains and beauty.

Jessica has worked in the telecommunication, automotive racing and medical industries in various office roles for over 20 years. She is excited about her new position with PC Project and helping make a difference for those living with PC.

When she is not working, Jessica enjoys camping, skiing, reading, photography, gardening and spending precious time with her family.

The Martin Family
RESEARCH STUDIES

PC patients continue to help with a variety of studies including biopsy samples, ‘proof of concept’ studies and other efforts to find an effective treatment for PC.

A major goal is to develop clear endpoints. With PC pain, which varies so much day-to-day, it is difficult to answer did it work or not? If pain is ‘better’ is that good enough? These are challenges in drug development. A lack of PC natural history data is hindering progress in some cases (see pgs 1-2).

In mid-September, the Steering Committee of the Int’l PC Consortium will hold a two-day meeting with PC Project staff to prioritize the more than 40 studies, select the most promising and review the funding costs of taking those studies forward.

CUTEST KIDS EVER—
FROM ALL OVER THE WORLD

Welcome to our newest little ones!
Clockwise from top:
USA-California
Japan
Czech Republic
USA-Georgia
Canada

WE LOVE ❤️ PC KIDS

Each month we will feature pictures of our PC Kids—Share your kid photos with us!

PEER-TO-PEER COACHES AND PC ADVOCATES

We haven’t forgotten those who have volunteered in these areas. We hope to have training materials by the end of the year. In the meantime, we continue to connect families.

This last month several have helped as coaches for other families with a new diagnosis of PC and sucking issues for some PC babies (usually PC-K6a). This is a great help. Having a new baby is a challenge — and then to have a diagnosis of PC can seem completely overwhelming. It is great to hear from someone who understands and knows ‘it will be okay.” We try to match coaches by genetic types as well as ages of the babies/children/teens. PC is a syndrome and each type is different—so it is important that ‘coaches’ are matched with families they help.
Clinical Trial Planning

In September 2015, members of the Int’l PC Consortium (IPCC) Steering Committee donated their time for a two-day meeting in London, England to plan the next clinical trials to take forward for Pachyonychia Congenita.

The first day was focused on clinical trial selection. Quality presentations were given on nearly 40 possible treatments. The presenters included Eli Sprecher, Chair, Roger L. Kaspar, W. H. Irwin McLean. Edel O’Toole, Mary Schwartz, Frances J.D. Smith and Maurice A.M. van Steensel. Phil Gard and David Hansen have also provided input as members of the steering committee.

Each option was researched by the presenters prior to the meeting so that full data on each was available. To avoid bias, the various options were randomly assigned to the presenters. Possible studies were from completed preliminary trials, from both patient-reported and physician-reported treatments gathered by PC Project during the past 12 years.

After extensive discussion, three drugs were unanimously selected as the best options. These are:

1. Botox Injections
2. Retinoid (new drug)
3. Topical Sirolimus Phase 2

The second day focused on clinical trial planning. This includes points such as (a) what center/physician to use (b) how many patients to enroll (c) what are enrollment criteria such as age, sex, mutation, etc. (d) what to measure (e) how to measure. All of these points may make the difference between a trial being approved or not by regulatory authorities and funders and a trial providing necessary evidence of safety and efficacy. This step is so essential.

Following the meeting, work immediately began on drafting clinical trial protocols. The IPCC Steering Committee (including Phil Gard, David Hansen, Edel O’Toole and PC Project staff) have been active in discussions on measurement devices. Also, initial contacts are being made with drug manufacturers to seek funding support for the clinical trials.

We Need Patient Input

We have no validated baseline pain or quality of life data that can be effectively used in a clinical trial. Before any clinical trial starts, we need to collect baseline information from at least 60 PC patients. We’d like to have 100 or more responses. Please help.

Step 1 — Answer this survey to help us know if you already use a smart phone, a pain app or an activity monitor. Only 3 questions.

LINK: https://goo.gl/TvTrv2

Step 2 — Take the PROMIS 29 Survey. It takes less than 5 minutes and give us this quality of life baseline data.

LINK: https://goo.gl/Oy8Qy5

Step 3 LATER — We will be selecting a pain app and an activity monitor for clinical trials. More on this next month.

Important News — Effective October 15, 2015, Frances J. D. Smith, PhD will serve as the PC Project Scientific Director. Dr. Smith was one of the researchers who discovered the PC genes in the mid-1990s. She has been the director of genetic testing for PC Project at the University of Dundee and has often given presentations at Patient Support Meetings and numerous professional conferences.

This is a major and important step forward for PC Project and we are delighted to work closely with Dr. Smith. She will continue to supervise the genetic testing for PC, coordinating efforts at the University of Dundee and elsewhere. She will oversee PC Project’s scientific efforts, including clinical trials and publications and will be Principal Investigator for the IPCRR. We enthusiastically welcome Dr. Smith.
AMERICAN ACADEMY OF DERMATOLOGY
The American Academy of Dermatology Association (AADA) is the legislative action group of the AAD. Annually, the AADA sponsors a lobbying session in Washington, DC. The AADA sponsors patient advocates to attend the session and asks them to assist in lobbying congress on topics of interest to dermatology and the AAD. PC Project has participated in this event a number of times to raise awareness.

In late September 2015, PC Patient Advocate, Stephen Wittmer and his son, Kyle, attended the AADA Conference. During this conference they had the opportunity to have a long session with their own congressmen and to visit other offices. Kyle and Stephen both spoke of their struggles with PC and asked them to continue fighting for more funding for research.

In connection with the AADA annual legislative session, the Coalition of Skin Diseases hold their Annual Meeting. This is an organization that unites groups such as PC Project that are focused on therapies for skin disorders. At the CSD final dinner, Kyle gave a speech about living with PC which ended in a standing ovation. Both Stephen and Kyle had a great experience.

NOTE: If you are interested in attending the 2016 AADA Legislative (lobbying) session as a Patient Advocate to represent Pachyonychia Congenita please email us at info@pachyonychia.org

GLOBAL RARE DISEASE REGISTRY
Since 2010, the Int’l Pachyonychia Congenita Research Registry (IPCRR) has been selected to participate in the NIH/NCATS GRDR® Program.

Now in Phase 3, the GRDR provides an important opportunity to extend the power of data provided by PC patients. The GRDR statement says “Our IPCRR de-identified data integration into GRDR will allow query by investigators to accelerate research across all rare diseases and to develop novel diagnostics and therapeutics for patient benefit.”

Stephen Wittmer wrote “This Congressman Knows PC” Congressmen John Ratcliffe with PCer Kyle Wittmer September 2015

Creating our PC Natural History—IPCRR Data Updates—Please Update Your Data Today

Our PC Stars! Thank you.
☆ Total Updates = 86
☆☆ Updated More Than Once = 9
HAPPY BIRTHDAY AT CHUCK E CHEESE
An adorable six-year old PC Princess in Maryland—we fell in love with you at the 2015 Patient Support Meeting!

Amazing five-year-old, PCer from New Zealand travelled with his family for a holiday in Thailand. He and his family had an awesome experience and he even got to pet a sleeping tiger cub (above).

WE LOVE 💖 PC KIDS
We love and enjoy hearing from you and learning about the wonderful things PC kids are accomplishing. Please continue to send us stories and photos. From newborn babies to college grads we’d like to feature their photos and stories. Please send photos and information to info@pachyonychia.org. We are eager to share more ‘PC Kids’ stuff and we’ll include this special page each month when we have new information.

HELP PC RESEARCH—RAISE MONEY AS YOU SHOP
Make this Holiday Season a real win for PC Project and PC research. We can receive much more if you and all your friends will remember to use iGive.com when you search or shop online.

EXAMPLE: Do you like to shop on Amazon? If you access Amazon.com through iGive your purchase will raise .8% through iGive and .5% though Amazon Smile = 1.3%. If you go direct to Amazon.com, we receive nothing. If you shop direct at Amazon.Smile, you miss out on earning the extra .8%!

Please remember to use iGive as your entry to shopping online and select Pachyonychia Congenita Project as your charity to benefit. Going through iGive to shop at Amazon and hundreds of other online shops, your online shopping experience does not change. It costs you nothing extra and helps PC Project raise funds.
OBAMA SIGNS RARE CLINICAL TRIAL COMPENSATION LEGISLATION INTO LAW

President Barack Obama (USA) signed a bill into law that will compensate patients for participating in clinical studies of rare diseases. An update to a 2009 law, the Ensuring Access to Clinical Trials Act of 2015 will allow patients with rare diseases to collect up to $2,000 per year without having the compensation count as income that could jeopardize eligibility for Supplemental Security Income and Medicaid. The bill S. 139 — also repeals a five-year sunset clause in the 2009 law.

The National Organization for Rare Disorders (NORD) praised the bill’s passage, saying it will remove income-related barriers to trial participation. According to NORD, only a few hundred of the roughly 7,000 rare diseases in the have approved treatments.

Tip For Infection Site—
"A simple, but handy thing, I learned in a recent hospital visit — doctors draw a line at the edges of the red part of an infection site, to see if the infection worsens. I drew a line on myself and see the infection has subsided a little and it is really good to see that rather than guessing." Kim Goode on Facebook.

Tip for Shoes—
many patients have recently written that they find Sketchers brand shoes very helpful with their PC.

EU and UK PLAN NOW TO ATTEND

Starts Friday October 28
Ends Sunday October 30
Edinburgh Capital Hotel
Registration will begin in November 2015

RAISE YOUR VOICE: RARE DISEASE DAY 2016

Date: February 29, 2016 Global Theme: Patient Voice
Global Slogan: Join us in making the voice of rare diseases heard
By definition, each rare disease affects a small segment of the population, yet combined the 7,000 known rare diseases affect 30 million people - that is 1 in 10 of us. Many do not realize how common rare diseases are and that our community has tremendous unmet medical needs.

This is a worldwide effort, and there should be details online for your specific state or country. For more information from NORD (USA) see http://rarediseases.org/for-patients-and-families/connect-others/rare-disease-day/

Collaboration— ‘become more powerful by bringing like-minded organizations together.’ To enable collaboration PC Project is a member of UNA, NORD, Genetic Alliance, Coalition of Skin Diseases, Global Rare Disease Registry and participates in collaborations whenever possible.

Fundraising— ‘never met a donor who was tired of donating to something they care about.’ Donors are friends who know and care about someone with PC. To sustain our work at PC Project we have to effectively tell others our PC stories. Friends want to become donors; we have to show them how. Use the PC dvd on YouTube.
The spotlight this month is on our amazing, wonderful PC Patient Community. No group in the world is more motivated to make a difference and help us move research forward to find an effective treatment for PC! Here are a few of the ways you have made a difference this month.

**PROMIS 29 SURVEY**—You did it!!!
We received 88 responses within 24 hours and now have a total of 114. Thank you so much.

With over 100 responses, we have enough data to send for analysis to see whether this is a tool that may be useful in our clinical trial design protocol to measure effectiveness of a treatment. Each step leads us to our goals.

**SMART PHONE SURVEY**—130 responded to this quick survey. 65/115 use iPhone 42/115 use Android 4/115 use other 15/130 do not use a smart phone

As a result of your great response, we were able to develop a pain app for iPhone and Android. We will share this with anyone who’d like to track their pain. And, we will have a small baseline survey soon.

**PC & TEETH**
Please send PC baby teeth, molars or teeth pulled for braces etc. to PC Project.

**Tooth Donor Awards to**
★ Tim Baker
★ David Mortier

The IPCRR main registry data shows that PC does not have a negative effect on dental health. PC does not cause cavities. The white plaque in the mouth called leukokeratosis is excess keratin that is white when it is wet. Currently a researcher from the National Institutes of Health (NIH) is doing a study on the role of K16 in teeth. We are working with the NIH to provide sample teeth for analysis, but it is not connected with decay or cavities. They have begun analyzing donated teeth.

Thanks to the willingness of a PCer who reached out with a short email to his friend, Jerry Seinfeld, all PCers will benefit. This is the biggest opportunity for fundraising we’ve ever had. We are grateful beyond words. This will make a difference in our research funding, scholarships, and program services. [PS—the show is sold out!!!]
**Publications on PC**

We often mention the importance of publications about Pachyonychia Congenita. It is one of our major ‘outreach’ efforts and serves to raise awareness of PC and attract researchers. We have posted 16 new articles since 2014 on our website which now has more than 650 PC-related research publications.

A new publication is available on the NIH National Library of Medicine website. It is dated October 2014, but we just became aware of this posting. Unfortunately, there is a misprint in the first paragraph where it says that with PC you get calluses and “cysts on the bottom of your feet”! Later in the article it is clear that for those with PC-K17 they get cysts all over their body. Maybe this mistake will just be a reminder to be grateful that cysts do not usually form on the bottom of the feet. They are working to correct this. Here is the link to this pamphlet—http://www.niams.nih.gov/health_info/pachyonychia_congenita/default.asp

**Important Meetings for PC Project**—Over the last three weeks, Frances Smith, our Chief Scientific Officer, has attended three important meetings in addition to working with PC Project staff for a week in Salt Lake City to plan a series of articles, grants, and programs for PC Project. She has now returned home to Dundee and continues her work with PC Project.

**Nord Rare Diseases and Orphan Products Breakthrough Summit**

**Oct 20-21, 2015 Arlington VA**

Discussion included how to promote rare disorders. Although some rare disorders are taught in medical school, in practice physicians see them rarely. We need to supplement their knowledge. Physician education should be ‘clinician’ education - to include general practice physicians, nurses, podiatrists, pain specialists, as well as dermatologists — all that may be involved in care.

Key points from sessions-

1. Need engagement with patients to conclude what is meaningful to them.
2. The importance of the 'patient voice' - patients/patient groups covering discussions about:
   - Patients are experts in living with their condition
   - The patients’ main complaint may not be what clinicians think is most important
   - Patients need to identify what is important regarding treatment benefit - a small improvement may greatly improve quality of life
   - Patients need to be as active as possible in working to develop and evaluate new treatments

We’ll feature notes from the FASTER CURES meeting and the 2015 PeDRA meeting in next month’s PC NewsBrief.

---

**PC Project planning together!**

Left to Right: Holly Evans (Program Director), Frances Smith (Chief Scientific Officer), Mary Schwartz (Acting Executive Director), Janice Schwartz (Patient Advocate), Jessica Martin (Office Manager).
Photos! Photos! Photos! We are often asked for photos of PC ‘people’ that show the person — and their PC. The following three photos were recently posted for the GRDR (Global Rare Disease Registry) website. We definitely need more photos like this—cute, fun “people” who also show signs of PC. Please help us!

We have used this great photo for more than 10 years in publications and posters showing PC thickened nails and this adorable kid (who is now a grown up teenager.)

An important researcher in the USA asked us to provide a photo so that he could make a poster for his lab and we sent this photo. He wanted those working in the lab to know that they were not just studying ‘cells’ and that there were real people relying on them. We need many, many more pictures of PC feet — somehow connected to fantastic PC people!

We had to really search, but we were delighted to find this new treasure—definitely a princess!! This photo helps to show the precious baby and for those who are aware or who are learning about PC, it shows the sign of leukokeratosis with PC.

We also WANT more new PC STORIES for the PC website and for the website home page. When you share your PC story you help others know they are not alone. If you need help with this, please email to info@pachyonychia.org

One PC patient updated their IPCRR information last month. We don’t yet have enough data to begin to compile PC Natural History so important for clinical trial preparation. Please continue to update your information.


PC Project has applied for a Global Genes Grant to fund preparation of training for the Peer Coaches program. We begin training early in 2016.

PC Patient Advocates schedule first webinar. Topic: What is a PC Patient Advocate?

The disease spotlight on the NORD website the first week of December will be Pachyonychia Congenita. Beginning Nov 30, view at http://rarediseases.org/

Tuesday, December 1, 2015 is Giving Tuesday a major international day dedicated to giving. Watch for our upcoming emails to help you and others participate in this great event.

Some types of Pachyonychia Congenita cause oral leukokeratosis.
HELPING TO FIND A CURE

I’m actually happy because... I have a husband with a heart in the right place. People with PC in their family, may have grown up in nice and wonderful home that gave support. But my mom was the first and it was horrible what she needed to put up with. She tried to do better but her parents were still around, so we got the same really awful dose of shame.

I have thought about it. A cure! Not for me so much, but I’m thinking of my daughter, and the children. My daughter is now 14 and even if she doesn’t want to speak about it, I know that she thinks about it....How am I ever going to get a boyfriend/husband? All of the thoughts are so hard because it’s hard to be a teenager anyway. She asked me what a boy would say (about her feet). She wouldn’t show her new girlfriends.

It really hurts in my heart because I do understand. I also know that this will pass. She is gorgeous. Friends won’t care if they are good people and she is going to get a good husband.

What is horrible about this PC is the pain, if there only where something against the pain.

We all very much want an effective treatment to have a pain free life for those with PC! Let us know your questions! Respond and participate in surveys. Your input is really important to our success.

FINDING AND BUILDING A RELATIONSHIP WITH A DOCTOR INTERESTED IN YOU—one thing we are often asked is for a referral to a “physician who knows about PC.” We want to introduce another way to help every PC patient have a physician who cares.

We have identified key points from patients who have an established a relationship and have an effective medical care giver. It makes such a difference. Unfortunately, many PC patients only try to get an appointment when they have an urgent need and too many PC patients expect doctors to know about PC. It is okay that a physician will most often have to learn about PC from you. What you want is someone willing to take time to learn.

FACT: Travelling or trying to see a “physician who knows about PC” will usually bring disappointment because no matter how much a physician knows about PC, there is no effective treatment. Your relationship with the provider is important and the provider can learn about PC.

FACT: The relationship between you and you doctor is the most important part of your care. Building this relationship takes time.

STEPS: (1) Before you have an urgent need, you should “shop” for a qualified professional; find someone you like in your local area. Set aside your hope/expectation that this doctor will already know about PC; that is very unlikely and is not the important thing. This may be a dermatologist (skin specialist), a general practice physician, a physician assistant, a podiatrist or other professional. Just as every shoe does not fit, you must shop until you find a fit for your personality.

(2) Then begin your part to educate this caring doctor about PC. This will happen over a period of time. It will not happen with one appointment. It will not happen when you have an urgent need. You will need to teach about PC a little at a time. Doctors are busy, but if you develop a relationship, it will happen. That is your responsibility as a patient with a rare disease. A quote from a member of the audience at the NORD meeting explains: “I am a nurse and a patient, but I had never heard of my rare disease until my diagnosis.”

FACT: It is okay (maybe good) when a doctor tells you he has never seen PC or doesn’t know anything about it. Don’t get mad. This really gives you an opportunity to offer to help the doctor learn. Accept the fact that all rare disease patients have this burden of teaching their care givers about their disorder. PC Project will help you in every way. Contact us.

FACT: Your personal genetic testing report is extremely valuable in working with medical experts. Few are trained in genetics, but your report is so important to establish your indisputable diagnosis. It will help you avoid a situation that happened to this patient: “The physician laughed and said have you diagnosed yourself or what?”

For additional ideas visit www.pachyonychia.org and watch the 2011 Patient Meeting presentation by Dr. David Hansen: Becoming an In-
“Just a quick email to say how awesome and inspiring I found Giving Tuesday to be!! It was amazing to watch the comments and the number of people sharing your posts! It really felt like everyone was pulling together! Thank you! I hope too that this was reflected in the donations!!”

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

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

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

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

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

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

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

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

FASTER CURES MEETING NOV 1-3, 2015 NEW YORK

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

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

PEDRA (PEDIATRIC DERMATOLOGY RESEARCH ALLIANCE)

NOV 6-7, 2015 IRVING, TX

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

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

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

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

PC Project is already working with a number of PeDRA members and hopes to have a formal PeDRA project focused on PC during 2016.
(PC Project News & Notes)

- **PC Natural History** is so important for clinical trial preparation. Please continue to update your information online using the IPCRR registry forms and including updated photos of PC.
- PC patients are invited to attend the Patient Day reception in Manhattan, NY in June 2016. This will be sponsored by Grunenthal Ltd. (a pharma company focused on pain.) If interested in attending and helping us spread awareness of PC to important researchers and executives, please contact PC Project.
- **FDA Awards NORD $250K.** To further the study of rare diseases, the U.S. Food and Drug Administration (FDA) has awarded a $250,000 grant to the National Organization for Rare Disorders. NORD will use the grant to develop 20 natural history studies for 20 rare diseases based on a lottery system. PC Project will apply and hope we win this ‘natural history’ lottery.
- **2016 PC Patient Support Meeting.** Edinburgh, Scotland. October 28-30, 2016. A flyer will be sent to all in Europe and Asia so you can register soon.
- **2017 PC Patient Support Meeting.** Salt Lake City, Utah in June 2017.
- **PC Patient Advocates** held their first webinar and a second session will be in mid-December.
- The **disease spotlight** on the NORD website this week is Pachyonychia Congenita.

---

**PC News Brief December 2015 Vol 10, No 12**

Dec. 7, 2015

www.rarediseases.org/Pachyonychia-congenita-project/

**Pachyonychia Congenita Project**

Posted by Christina Jensen

**PC Natural History**

1. What does it mean to you personally to be a patient organization serving the rare community? It is at love about. With a disease as rare as Pachyonychia Congenita, patients feel alone and often scared. Others around them do not understand the pain that they are dealing with while trying to live a normal life. We are glad that PC Project gives our patients an organization to turn to for support, education and a way to connect with others who have the same disease and really understand their daily struggles. We have seen rare having a rare disease affects each individual and are passionate about providing a community of patients, researchers and physicians that is actively fighting for a cure, connecting & helping patients, and empowering research. Together we can make a difference.

2. What do you find your patient community values most from your organization serving the rare community?

- PC Website pachyonychia.org - a website specifically designed to provide education and support for Pachyonychia Congenita patients, family members and for clinical and research professionals.
- PC NewsBrief - a monthly newsletter for patients providing news and tips
- PC Patient Support Meetings - annual meetings for patients
- PC Clinical Studies - more than 40 clinical studies have been completed
- International Pachyonychia Congenita Consortium - over 150 professionals dedicated to collaborating on PC research

Those affected with Pachyonychia Congenita and their family members best express what these PC Project activities mean to them.

"My daughter found information about this project and for the first time, we had hope."

"Today is the first day I discovered the name of the condition and am elated to find this site! I am literally brought to tears at the thought of not being alone in this. Thank you so much for starting this project. God bless you all.""I have PC since I was two-three years old. But I didn’t know the name of the disease until I was 52 years old. I don’t know anyone else who has PC. I was happy and died when I read about the PC organization and all the people with the same pain and difficulties and all the people who want to help to find a cure."

"It wasn’t until recently that my brother found this site and I never fully appreciated how much a bit of information can change one’s life. Suddenly we don’t have to try and explain what he has ... and put up with people who belittle what he has by suggesting he simply cut them off. You have essentially given us a voice. Thank you."

"After the first night at the PSM, I went back to my room, and I was literally in tears. Seeing someone with PC, who I could relate to, was the most humbling feeling ever. It was so refreshing to actually talk to someone that truly experiences the same things that I do. I’m struggling to convey in words my emotions from the meeting, but I don’t think words can justify how I feel. I apologize, but I can assure you it was life changing. So thank you! You are truly the best!"

3. What are some of the challenges your organization has faced?

The greatest challenge is finding any effective treatment for Pachyonychia Congenita (PC). Other challenges include:

- Although PC patients experience excruciating, constant pain from the time they begin to walk and throughout their life, PC does not cause death and, therefore, the serious nature of this disorder is often minimized.
- Even with few patients in the world (and only about 300 in the USA) we still must interest physicians and scientists, support clinical trials, provide educational outreach and other services to make a difference for patients – the same as organizations with many, many patients.
- With a rare disease misdiagnosis and misinformation is common so a challenge is to publish correct information based on a larger patient population.
- Focusing attention on pain as the main disabling feature of PC (rather than thickness of nails)

4. What’s been your most successful awareness campaign and/or fundraising event?

PC Awareness Around the World each June encourages patients and friends to conduct events in their local areas. Because we have patients in over 50 countries, there is no center for PC fundraising. Empowering individuals has been important to expand our reach, increase awareness and bring some additional funding. Patients have successfully held a variety of events including garage sales, car wash events, dinners, donation boxes, bake sales, golf tournament, spin-a-thon, horseback ride, bicycle rides and more. One very creative idea is a grocery market that sells empty boxes for $1 and donates that money to PC research. Our biggest fundraiser was sponsored by TransDerm, a pharma partner, who sponsored a Dualathon and raised nearly $30,000 for PC research and services.
We appreciate so much the wonderful photos you are sending. Please continue to send us photos of your adorable PC kids — and of wonderful PC adults, too. These photos send a message that no words could convey properly. The photos are so important in all of our work to help others learn about PC and how it affects lives. We want others to know the patients and not just the disease. Your photos are wonderful. Please continue to send photos showing fingers, toes, callus, etc. on beautiful pictures of you and your children showing them and their PC—with or without a sign. We want to find an effective treatment—a cure—for these adorable children.

Pachyonychia Congenita causes my nails to grow very thick.

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

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

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

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

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

<table>
<thead>
<tr>
<th>PC-K6a</th>
<th>PC-K6b</th>
<th>PC-K6c</th>
<th>PC-K16</th>
<th>PC-K17</th>
</tr>
</thead>
<tbody>
<tr>
<td># responded = 6</td>
<td># responded = 1</td>
<td># responded = 1</td>
<td># responded = 6</td>
<td># responded = 3</td>
</tr>
<tr>
<td># of responses = 26</td>
<td># of responses = 1</td>
<td># of responses = 4</td>
<td># of responses = 22</td>
<td># of responses = 18</td>
</tr>
<tr>
<td>Highest pain = 10</td>
<td>Highest pain = 3.7</td>
<td>Highest pain = 2.7</td>
<td>Highest pain = 10</td>
<td>Highest pain = 10</td>
</tr>
<tr>
<td># responses at 10 = 8/26</td>
<td># responses at 3.7 = 1/1</td>
<td># responses at 2.7 = 1/4</td>
<td># responses at 10 = 1/22</td>
<td># responses at 10 = 6/18</td>
</tr>
<tr>
<td>Lowest pain = 1</td>
<td>Lowest pain = 3.7</td>
<td>Lowest pain = 0.8</td>
<td>Lowest pain = 0.6</td>
<td>Lowest pain = 3</td>
</tr>
<tr>
<td># responses at 1 = 1/26</td>
<td># responses at 3.7 = 1</td>
<td># responses at 0.8 = 1</td>
<td># responses at 0.6 = 1</td>
<td># responses at 3 = 2/18</td>
</tr>
<tr>
<td>Average pain = 7.2</td>
<td>Average pain = 3.7</td>
<td>Average pain = 1.6</td>
<td>Average pain = 6.9</td>
<td>Average pain = 8</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Patient</th>
<th>Patient</th>
<th>Patient</th>
<th>Patient</th>
<th>Patient</th>
</tr>
</thead>
<tbody>
<tr>
<td>1 = 5</td>
<td>1 = 3.7</td>
<td>1 = 2.7</td>
<td>1 = 4.4</td>
<td>1 = 3</td>
</tr>
<tr>
<td>2 = 6</td>
<td></td>
<td>2 = 4.1</td>
<td>2 = 6.1</td>
<td></td>
</tr>
<tr>
<td>3 = 6.5</td>
<td></td>
<td>3 = 10</td>
<td>3 = 10</td>
<td></td>
</tr>
<tr>
<td>4 = 7.5</td>
<td></td>
<td>4 = 8</td>
<td></td>
<td></td>
</tr>
<tr>
<td>5 = 10</td>
<td></td>
<td>5 = 8.4</td>
<td></td>
<td></td>
</tr>
<tr>
<td>6 = 9.1</td>
<td></td>
<td>6 = 8.7</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Patient</th>
<th>Patient</th>
<th>Patient</th>
<th>Patient</th>
<th>Patient</th>
</tr>
</thead>
<tbody>
<tr>
<td>1 (one entry)</td>
<td>1 (one entry)</td>
<td>0.8 to 2.7</td>
<td>1 (one entry)</td>
<td>1 (one entry)</td>
</tr>
<tr>
<td>2 (one entry)</td>
<td></td>
<td>2 (one entry)</td>
<td></td>
<td>2 (one entry)</td>
</tr>
<tr>
<td>3 (one entry)</td>
<td></td>
<td>3 (one entry)</td>
<td></td>
<td>3 (one entry)</td>
</tr>
<tr>
<td>4 = 3 to 7.5</td>
<td></td>
<td>4 = 0.6 to 8</td>
<td></td>
<td>4 = 0.6 to 8</td>
</tr>
<tr>
<td>5 = 8 to 10</td>
<td></td>
<td>5 = 5.9 to 8.4</td>
<td></td>
<td>5 = 5.9 to 8.4</td>
</tr>
<tr>
<td>6 = 1 to 9.1</td>
<td></td>
<td>6 = (one entry)</td>
<td></td>
<td>6 = (one entry)</td>
</tr>
</tbody>
</table>

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

INSTALL THE PC PAIN SCALE to Android or iPhone/iPad

Android – add to home screen


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

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

iPhone/iPad—add to home screen

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

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

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