JERRY SEINFELD SHOW
For Immediate Release

Last night, January 14, was special for PC Project—an event that will benefit all PC patients around the world, as Jerry Seinfeld pledged the proceeds from his show in Salt Lake City to PC Project. These funds will be used for research and patient services.

Using the photos provided by PC patients with “Thank You, Jerry” signs (as well as signs with comments about having PC), we prepared a short thank you card video. You can view the video card at www.pachyonychia.org/video_presentation.php on the website.

With the video card, we included handmade scarfs with each family member’s name embroidered. Also, a special Superman Jerry Seinfeld ‘bobblehead’ (see page 3). We wanted to let him know that he is a superhero at PC Project.

Three 8’ banners with the image at the left were on display throughout the lobby. PC Project also had an information table in the lobby and a group of teen volunteers assisted in collecting donations. More details on this event and the significance to PC Project will be available in future newsletters. We are very grateful to a special PCer (an old school friend of Jerry Seinfeld), who made this amazing event possible.

The spectacularly beautiful Grand America Hotel in Salt Lake City donated rooms for Mr. Seinfeld and his staff to help PC Project gain the most benefit from this show. Special thanks to the Grand America Hotel for this major and generous contribution to help PC Project.
NEWS FROM YOUR CHIEF SCIENTIFIC OFFICER
Frances Smith, PhD

Some of you may be wondering what my new job entails.... the last two PC News Brief's have reported my travels around the USA to various meetings and to PC Project in Salt Lake City but what am I doing now back in Dundee?

How many of you when searching on the internet for 'PC, thick nails or keratoderma' have been directed to various medical/rare disease websites and found the information about PC is out of date? One of my tasks is to get these sites updated - a number have been done but still more to do. It’s amazing how many there are! If you come across any sites that you feel/think need updating please let us know and I'll contact the relevant person to get this done. PCer's - you are from all over the world and speak many different languages - there will be many sites that I will not find, or understand as my linguistic skills are bad! But you can help. Having up to date information easily available about the clinical features, the genes involved, genetic testing and caring for PC is so important and will help not only PCers and their families but also the doctors who diagnose and care for those with PC.

In line with this we have several drafts of potential publications that need completing and submitting to scientific/medical journals. One of these is a report of several PC patients from India who joined the IPCC and have had genetic testing. This paper has just been submitted to an Indian Dermatology Journal. It is important for us to publish these cases in a broad range of journals to reach as many doctors as possible, to educate them about PC.

I'm also still involved in the genetic testing of all new cases and discussing these monthly with the expert PC dermatologists at PC Project who perform your consultations so we can learn more about PC.

U.S.A. BUDGET: BIG BOOSTS FOR NIH & FDA
Recently, President Obama signed into law a year-end spending bill that funds the U.S. government through fiscal year 2016.

The National Institutes of Health (NIH) was a huge winner, receiving a boost of $2 billion and the Food and Drug Administration (FDA) was provided an increase of $133 million! The Orphan Product Development Grants Program at FDA will also receive a boost of $2.5 million in 2016. This is a significant achievement for the rare disease advocacy community, which has been fighting for increased funding for both agencies.

NOTE: The NIH has funded PC Project scientific meetings as well as PC research at TransDerm, Inc. Both of our Clinical Trials (siRNA in 2008 and Topical Rapa in 2014) received FDA clinical trial funding to cover a portion of the costs. The funding recently has been limited. This new legislation is important to our continue research efforts.

PC ADVOCATES TRAINING
The first six U.S.A. PC Advocates have held three training meetings. In the future, once we have developed the training materials and tested the PC Advocate approach, we hope to increase PC Advocates to other countries.

PC Advocates will serve to be the spokespersons for PC Project on behalf of all PC patients and on behalf of our scientific research community. As they move forward in training, they will develop the tools needed to carry the message of PC Project in both formal and informal settings.

PC Advocates will invite and enlist others (patients, physicians, researchers, donors) to join the PC community to aid in pursuing the goals of PC Project to benefit all PC patients. They will share the PC Project story, patient stories as well as current research efforts, clinical trial plans, fundraising needs and more. Their voices will be heard in many settings to increase awareness of PC.

PC PEER COACHES
We appreciate each of you who have volunteered to be a PC Coach to provide support to other patients who have questions or just need a friend. We are waiting on results of our grant application to fund the training program for PC Coaches to know when we will be able to begin this program. There are a number of details to complete before this training can begin — but it is definitely one of our goals for 2016. We will be in touch with you as soon as possible.
PC News Brief January 2016 Vol 11, No 1

Grunenthal Reception
On Wednesday, June 1, 2016, Grunenthal GmbH (a major worldwide pharmaceutical company that focuses on pain research), is hosting a special reception for patients with rare diseases.

The Reception will be at the Friar’s Club in Manhattan, NY beginning around 6:30 pm. Grunenthal will issue invitations to interested PC patients with more details.

We hope at least 10 PC patients will attend. If you would like to receive an invitation, or if you have any questions, please email Mary.Schwartz@pachyonychia.org.

On February 29, 2016 PC Project will join forces with rare disease patients and health care advocates in the U.S. and around the world for Rare Disease Day. Rare Disease Day is an annual awareness day dedicated to elevating public understanding of rare diseases and calling attention to the special challenges faced by patients and the community.

The theme for this year’s Rare Disease Day is ‘Patient Voice’. This theme recognizes the important role that patients play in expressing their needs and having a voice in their own care. This theme was selected to appeal to a wider audience including not only the patients, but patient organizations, caregivers, medical professionals and researchers that are also dealing with rare diseases each day.

We encourage our PC Project friends and family to get involved and use your ‘Patient Voice.’ Let those around you know what it’s like to have a rare disease, or care for someone you love who is impacted by a rare disease.

For information about Rare Disease Day in the U.S., go to www.rarediseaseday.us

For information about activities in other countries, go to www.rarediseaseday.org

Jerry Seinfeld Show—January 14, 2016—Abravanel Hall, Salt Lake City, Utah

Nearly 2,800 excited fans crowded into the theatre to enjoy a hugely successful stand-up-comedy show. At the end of the show, he thanked the audience for supporting the organization (PC Project) in the lobby.

We were able to give out thousands of small handouts about Pachyonychia Congenita, a number of people viewed our displays and talked with us about what PC is and how Jerry Seinfeld came to know about us.

We appreciate each of you PCers, members of the audience, and friends of PC who sent thank you tweets!
PC PAIN STUDIES
The 2011 Pain Study has now been published and is available on our website in the Published Research Articles. The 2014-2015 study will be published shortly.

We received this very valuable question in our email about PC pain studies — "Regarding the pain study, I assume we needed to 'prove' that there is pain associated with PC before we could consider treatments? I am simply curious."

Here is our response to this great question, which this patient encouraged us to share with all.

1. You and every other patient knows there is pain.
   QUESTION: Have you ever seen a pain specialist? Has there ever been a presentation or publication on PC at any pain association, conference, publication? Do you talk about your pain with physicians? If you say no to these 3 questions, you are quite a normal PCer -- and it is okay. But if we want to solve the PC pain problem, we have to find a way to focus and make others know there is significant pain.

2. In 2004 when we started PC Project, there were about 500 publications on PC cases. Not one mentioned pain. Some mentioned weird things (PC and deafness; PC and mental retardation; PC and...) but not one article mentioned pain.

3. Pain is a very complex area. To get people to focus on pain (not just how thick the callus is), we have to attract research interest from those who know pain. For the first time, in 2011, we had a pain specialist at the PC patient meeting. It was a start. In 2014, we had another respected pain specialist do a more complete study and we had a neurologist/pain specialist speak at the patient meeting. Patients may not have understood the reason, but those specialists got a lot from being there and have spoken to their peers about PC.

4. Apart from that, we now have the attention of one of the largest pharma companies, a firm focused on pain (Grunenthal). They are now looking at PC (a disease they had never heard of until April 2015).

5. So, short answer, YES. We have to demonstrate pain to get traction for research options to focus on pain. PC is a 'skin' disorder (even classified as a 'nail' disorder) and so it is relegated to dermatology -- and dermatologists are great. But dermatologists do not prescribe drugs for pain and do not conduct research on pain. We need some neurologists and other pain specialists to work with dermatologists on PC pain.

These are our first steps into the area of 'PC pain' so well known to PCers and PC pain that is so little known to the medical and research communities.

EMAIL FROM A PC MAMA BEAR
"Recently, my son had an infection in his foot. He gave me the heads up in advance and knowing he would be going out-of-state for a holiday, I wanted to be sure he was OK. I made an appointment with a paediatrician in an office that had late night openings. I printed out some info from the PC project website and took it with me.

Once the doctor came in I told him what the issue was and that it's part of David's Pachyonychia Congenita. I handed him the paperwork. He said, 'OK, let me see the spot.' David takes his sock off and the doctor says, 'Well, I am concerned about the excess calluses and the toenails.'

Deep breaths....the Mama Bear is about to come out... So I say, 'Yes, that is what this condition is, you know the paperwork I just gave you. The infection is right here, (pointing at my son's foot.) All I need you to do is prescribe the antibiotic for him and some antibiotic ointment.' He starts to hesitate and I said, 'The infection is right here, it hasn't come to a head yet and can't be drained, unless you hand me a scalpel and I will drain it right now.'

He said 'Seems you've done your homework...what kind of antibiotic did you say you wanted?' (LOL I should mention my son WOULD NOT let me near his foot with a scalpel!) I am so glad we have the PC Project to help not only with educational information but the connection we get to have with knowing we are not alone in this even when we think we are the only ones with it."

In Memoriam
Thank you for 160 £ donations received in honor of Edith Hunt
## 2015 BRIEF FINANCIAL SUMMARY

We are very grateful to the many who have donated funds, time and services to PC Project in 2015.

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<thead>
<tr>
<th>DONATED SERVICES</th>
<th>Total Hours</th>
<th>Total Value</th>
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<td>IPCRR patient consultations</td>
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<td>GeneDx (genetic testing)</td>
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<td>Fundraising Expertise</td>
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PC Project financial information is audit in January each year under GAAP standards by a qualified CPA firm.

### 2015 Income and Expense Report

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<th>Total 2015 Income</th>
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<td>Matching funds income</td>
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<td>Income from Awareness Events</td>
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<td>Fundraising Events Income*</td>
<td>$26,953</td>
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<td>Miscellaneous Income</td>
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**2015 Total Expenses** $373,856

- Program Expenses $279,268
- Operations Expenses $51,228
- Fundraising Expenses $43,360

**Expenses over/under income** $-41,943

*Includes $9,215 from 2015 Giving Tuesday. An additional $15,255 from 2015 Giving Tuesday was received January 2016.

### 2015 Donation Summary Report

- 213 total donors (the most in any year)
- 1 donor gave over $10,000 in 2015
- 5 donors gave $4,000 or more in 2015
- 15 Donors gave $1000 or more in 2015
- 14 monthly donors (our sustaining donors!)

Sponsor matching funds were $220,834

Funds paid for PC Research and Clinical Trials by an anonymous donor
- Stanford University $20,584
- University of Dundee $87,952
- PC Project Fellowship $75,000
- UC Davis $13,300
- **Total** $196,836

### Another Type of “Donation”

We continue to collect PC teeth. The National Institute of Health (NIH-USA) study on PC Teeth is on-going. Please let us know if you have a tooth or teeth to donate. We will send you the Consent Form and packaging. The teeth can be

- Baby teeth
- Molars that are pulled
- Teeth extracted for braces or any other teeth from children or adults with PC. Thanks.
PC Project Awarded Global Genes Grant

We are pleased to announce that PC Project was awarded a Global Genes Grant in the amount of $4,800 for costs in training Peer Coaches. The training materials will be completed and the training will begin shortly with travel to the in-person training session scheduled with the Peer Coaches.

For all who have volunteered to serve as a Peer Coach, we will be in touch shortly with further information. At present we are beginning the Peer Coach program in the USA and will extend it to other countries as the program is more fully developed.

Six PC Advocates Continue Training

Our first six PC Advocates are continuing training to be spokespersons and advocates for PC Project toward the goals to help all PC patients. We have started in the USA and will add additional advocates as the program is finalized.

Current PC Advocates are -
- Julie Bennett
- Christine Block
- Roseann McGrath
- Jack Padovano
- Janice Schwartz
- Stephen Wittmer

Three web training meetings have been held. Training will continue at a session the day before the upcoming IPCC scientific meeting in May 2016.

As our advisors reviewed individual PC stories, they advised that one-by-one, patient stories have a limited impact. These stories may be effective at Patient Meetings and in other specific venues, but to move us forward, PC Advocates must use their personal PC story to carry the more impactful message of the PC community including all PC patients and the difference PC Project is making with the hope that working together we will find a treatment for PC.

We need to raise millions of dollars to fund clinical trials. We need effective messengers. Our PC Advocates will lead the way in carrying the message of PC Project, educating on PC, and the need for support to a wider audience.

If we don’t tell the world how life is with PC and tell what PC Project means to PC patients, we cannot expect the world to support us.

We encourage everyone to develop a 2-1/2 minute “PC Story” that can be adapted to any audience (a) physicians (b) funders (c) patients (d) educators. And to use that personal story effectively to tell the PC Project story. You want to be able to easily and quickly explain -
- What is PC (many types)
- The impact of PC on your life
- The hardest thing about PC
- Managing/coping with PC
- The importance of PC Project and your hope for a cure.
JERRY SEINFELD DONATES $160,000 TO PC PROJECT

The website Look to the Stars has a short article about Jerry donating the proceeds from the Salt Lake show to PC Project. Also a link to our YouTube ‘Thank You’ video. [https://jerry-seinfeld-donates-show-proceeds-to-charity](https://jerry-seinfeld-donates-show-proceeds-to-charity)

Scholarship Award. PC Staff member, Holly Evans, received a scholarship grant to attend the Nonprofit Academy of Excellence at the University of Utah during February 2016.

Conference Pass. PC Project was awarded a free pass worth nearly $4,000 to the World Conference on Orphan Drugs to be held in Washington, DC on April 21-22, 2016. Roger Kaspar will represent PC Project at this conference.

Conference Passes. Through our membership in the Coalition of Skin Diseases, PC Project has received passes to the American Academy of Dermatology annual meeting which will be held in March in Washington, DC. The discounted cost to attend without a pass is $1,850. Roseann McGrath, PC Advocate, will attend with her husband Mike. Roger Kaspar will also represent PC Project at the AAD meeting. The CSD annual luncheon is held in conjunction with the AAD meeting.

BBB Wise Giving Alliance

“I’m happy to report that the Pachyonychia Congenita Project meets all 20 Standards for Charitable Accountability. Congratulations.” Email from BBB.

RARE DISEASE DAY FEBRUARY 29, 2016

To lead up to Rare Disease Day, NORD is holding a special countdown on social media. PC Project was lucky enough to be selected as one of 28 NORD member organizations to be highlighted. PC Project will be spotlighted on Wednesday, February 24, 2016 on the [NORD Facebook page](http://www.facebook.com/nordorg). We will also have a tweet posted in the Twitter Chat for rare diseases which will be on March 1st.

There are events around the world for Rare Disease Day. If you participate in your community, please let us know. And, please send a photo! And a little bit about what you do.

RESEARCH STUDY REQUIRES BIOPSY DONATION

Once again PCer, Ted Clark, donated a plantar biopsy for a special study at the University of Michigan. David Hansen, MD, one of our most dedicated specialists, donated his time, skill and clinic services to collecting the biopsy. You can see Ted is a PC Superhero! The numbing injection is always the most painful part. Also, for this biopsy, some special trimming was necessary, but Ted was a champion biopsy donor. We are grateful to all who are willing to participate and give samples when needed for research projects.

THE BURDEN OF RARE

In preparing for the International PC Consortium Annual Meeting, we often ask leading researchers to also invite young investigators who may be interested in a rare, genetic, skin disease. This thoughtful response was a powerful reminder of the challenges of a rare disease with no effective treatment.

“I don't know anybody around who is especially interested in this area. Because my country (Turkey) is not a ‘Research Country’, physicians usually don't want to engage with diseases (and families) whose treatment has yet been found. I haven't heard of anyone even from my dermatologist friends.” Volkan Okur MD Columbia University, NY

The statement is true for every country in the world including the USA (a country that is focused on research). If a rare disease has a treatment, physicians have something to offer patients and are glad...
to see them and prescribe for them. When a rare disease has a completely unmet medical need, the burden for these patients is far greater.

Ultra Rare Disease Burden —no effective treatment
- Misdiagnosis
- Misunderstanding
- Misinformation
- Isolation (feeling alone)
Pachyonychia Congenita
- Pain
- Appearance
- Time for care
- No treatment (unmet medical need)

MORE ON GENE THERAPY, EDITING OR CORRECTION
In the December 2015 NewsBrief (page 2) we wrote about this topic. It is important to understand that the gene therapy drugs we are pursuing for PC to stop the mutant gene from acting do not have any effect on offspring. There is a very big ethical debate at present about gene editing. An interesting talk was recently given by Sharon Terry, CEO of Genetic Alliance. Click here to view presentation.

THE RESEARCH PROCESS ......& why it takes so long to get results—Frances Smith, Chief Scientific Officer

A huge amount of data is collected every time someone registers with the IPCRR. This is continually added to as new PC patients register and as you update your information as your PC changes over time. This is all highly valuable and important to collect even if it is not used immediately. When physicians/scientists come up with a question we already have a good starting point of data.

Sometimes though when a study is being planned, we realise we are missing answers to a few questions that have been raised. This requires an addendum to be designed, sent out and answers collected. This all takes time. For all studies we want to collect data/information from as many of you as possible to make the results more meaningful. Thank you to all of you who have taken the time and completed these for various studies. All this information gets used and it can take some time for studies to be completed and reported back to you.

For some studies we may have an idea. For example, what type of pain do PCers experience? But to investigate and answer it in a way that will lead to PC pain being recognized and treatments being tested, we must validate the information.

We must find experts in the right area who are interested and have time to do such a study. We are lucky to be working with a ‘pain team’ who were able to attend 3 patient support meetings in Paris, Edinburgh and Newark in 2013-2014 to carry out studies to help us understand pain and PC. For these studies they also need to collect data from non PCers to compare with that from PC patients. The findings from these studies are being analysed. We hope the data will be finalized soon and we will be able to report results back to you.

QUESTION: Would a vegan diet help to make PC better for my child.
ANSWER: Thank you for your recent email. While obviously a healthy diet is important to overall health, we have no data to support that a vegan diet would affect this disorder in any way. There is no direct connection between the keratin mutation and eating meat. However, a healthy diet that you find best for your overall health is, of course, a good choice.

QUESTION: Here is a link about CRISPR. Do you think we are far from the application of this technique?
ANSWER: CRISPR is only a new tool to aid gene sectioning. We have demonstrated the effectiveness of siRNA which is another tool with a similar ‘interference’ function. Whatever tool is used for creating the drug, delivery to the cells of the skin is still the main obstacle we have not yet solved.

QUESTION: My baby has a nail infection. What can I do? [A number of emails have asked about this problem.]
ANSWER: We’ve prepared a summary (next page) which include some tips from an experienced PC mom.
Swollen Nails in PC Babies—we call these infections, but most physicians say this is not an infection. Whatever the right term, many babies and children with PC have this problem at times. If you have a relationship with a pediatrician, physician or podiatrist who understands PC, that is always of great value. This is not intended to contradict medical advice in any way. The nail must be ready before the following steps can be taken.

ACTIONS MANY PCers TAKE WHEN THE NAIL IS “RIPE”
From one of our PC patients (a mom with PC who has two boys with PC) may be helpful. This is not medical advice, but experience advice.

“I looked at the picture and the nails look very familiar. Both my boys often got infected nails as little babies and they looked a lot like these nails. The nail on the left looks "ripe" right now. Both nails may be ready to have the pus released. I would be as gentle as possible and try to do one of two things (or both, depending on which thing works best):

1. Use clean (sterile), sharp, large nail clippers and see if I could make a hole in the nail. Probably start on the side, on the end near the tip. Do it when the baby is sleeping if possible. There will be pressure when the nail is first clipped and that can be very painful. So the softer the nail, the better and if baby is asleep, that's good too.

2. Use a sharp, clean (sterile) razor blade (I like a double-edged razor blade because it bends) and try to nick at the nail to make an opening, again from the side and end of nail nearest the finger tip. If nothing comes out, try near other areas. This may be a better way than the nail clippers.

- Whether a nail clipper or a razor blade is used, go slowly and gently and not too deep at first. The outer nail is "dead" but soon, inside the nail, there will be "live" nerve endings so be careful.

- Before trimming, if the nail doesn't seem soft, soaking it first by putting the baby's hand in warm water (not hot) for a few minutes will help. Also, after the nail is cut into and the pus comes out, soaking the nail again, or even running it under warm water will help.

The nail itself may fall off at some point once the nail starts to heal. Antibiotics may help the healing, but to heal quickly and to relieve the pain as soon as possible, getting the pus out is important and the pain relief will be almost immediate. Based on my experience, there will be pus in the nail, not clear liquid.

Also, baby pain reliever like baby ibuprofen will help as well. If I had to, I would give the baby some of that before trimming the nail. However, the greatest pain relief will come when that nail is gently cut into and the pus is released.”

NOTE: A topical antibiotic cream can be used after releasing fluid/pus (the same as for blister care.) Oral antibiotics may or may not be needed. If there are red streaks it is important to immediately see a physician for antibiotics. Other times nails can be treated and healed without antibiotics. You will learn to be able to know at what stage the problem is and when to see a doctor. If at anytime you are uncertain, see a doctor.
Rare disease day started as a European event 9 years ago that takes place on the 29th of February (ie. a ‘rare day’) or when it is not a leap year on the 28th of February to raise awareness of rare diseases. Now it is a world event with more than 80 countries participating.

**Rare Disease Day UK 2016**
To mark Rare Disease Day, Rare Disease UK hosted Parliamentary Receptions across the UK, at the House of Commons, London, the National Assembly for Wales, the Scottish Parliament and the Parliament Buildings, Belfast.

**Reception at the Scottish Parliament**
Frances Smith attended the reception which started with refreshments, a short welcome speech from Malcolm Chisholm MSP, followed by a series of short presentations. First, a patient with the rare disorder, progressive supranuclear palsy, and his wife discussed issues including getting a correct diagnosis, living with a rare disorder and the effect on family life. Alistair Kent OBE, the Director of Genetic Alliance UK then spoke about the role of Genetic Alliance - a national alliance of over 180 patient organisations working to improve the lives of patients and families affected by all types of genetic conditions. This was followed by Jamie Hepburn MSP, the Minister for Sport, Health Improvement and Mental Health who announced the funding of £6m investment by the Scottish Government for the Scottish Genomes Partnership, a medical research collaboration between Scottish universities and the National Health Service. Through collaboration with Genomics England this large sequencing project hopes to offer more rapid diagnosis to rare disease patients or a diagnosis to those with an unknown disorder. Genetic diseases will become better understood and new ways to test, manage and treat these diseases will be developed. The final presentation was by Professor Zosia Miedzybrodzka, a clinical geneticist at the University of Aberdeen. She spoke of her hope of a diagnosis, through the Scottish Genomes Partnership, for some of her patients with unknown rare disorders.

**Facts about rare diseases**
- A disease or disorder is defined as rare in the USA when it affects fewer than 200,000 people
- A disease or disorder is defined as rare in Europe when it affects fewer than 1 in 2,000

There are 239 genetically confirmed PC patients in Europe (countries in the EU). This means that PC affects fewer than 1 in 3.1 million.

PC is ultra rare and so very rare that to achieve our goals, we need to ensure that every patient and every family member joins the IPCRR and is counted.

- There are between 6,000 - 8,000 recognised rare diseases
- Of these, it is estimated 2,000 are rare skin diseases. PC is one of the rarest of the rare skin diseases.
- 80% of rare diseases have genetic origins. Other rare diseases result from infections (bacterial or viral), allergies and environmental causes or are degenerative and proliferative
- 75% of rare diseases affect children
- Rare diseases are disablign: the quality of life of patients is often compromised

**Fact:** Collectively rare diseases are not rare.

- Approximately 30 million people in the USA are living with a rare disease
- Approximately 30 million in Europe are living with a rare disease
2016 PC AWARENESS
As can be seen from the information Frances Smith shared about rare diseases (page 1), PC is one of the rarest of the many rare diseases. We are small in numbers, but together we can make a difference for PC.

Although June is PC Awareness Month, PC Awareness efforts can happen anytime throughout the year. Pick your date and what you want to do.

Each family can hold an event (large or small) in their own community.

Some examples from past years include:
⇒ Printing and sending brochures to friends and coworkers
⇒ Posting social media messages on Facebook, Twitter, Instagram
⇒ Hosting a dinner for family and friends
⇒ Holding a yard sale, bake sale, bike-a-thon, carnival
⇒ Setting up collection boxes
⇒ or just chatting about PC with a few friends

Here are some tips for planning your fundraising or awareness action for 2016:

1. Choose an activity that you like. Your activity can be large or small but it will be most successful if you choose something that interests you. Remember that you are the ‘heart’ of the PC action in your own community.
2. Include your friends and family in your project or activity. The more people the better. It is really good for your family and friends to be involved in spreading the word about PC. The more they learn and become involved the more effective we can be at spreading the word about PC.
3. You can register your activity with PC Project by sending us an email, photos and other information.
4. Publicity! If you LIKE our PC Project Organization Page, you can then post on that page. You also can click onto the attached PC Event page and post your event there. This link will take you directly to the PC Project Events page for 2016 https://www.facebook.com/events/503550539833327
5. Don’t forget to have fun while you are planning/holding PC Awareness efforts.

2016 PC EVENTS PLANNED!
Talk with your family and friends and plan what you will do in 2016. Several awareness actions/events have already taken place and many are planned. Please help us achieve our goal of at least 100 PC events in 2016!

FIREWALKING EVENT 2016
From Julie Peconi:
Both my husband, Tom and son Timmy 8, have PC (K16) and as Timmy gets older his feet are causing him more and more pain. As a result, to show him our support and to help raise valuable funds and awareness for PC Project, we wanted to do a big event this year! When a good friend suggested...
firewalking as a symbolic way of raising money for a condition where it hurts to walk, we thought it was perfect! For those of you who have never heard of it, a fire walk is basically walking barefoot unharmed over hot burning embers and coals. Although the fire is very hot, with a temperature as high as 1200 degrees Fahrenheit, walkers are trained beforehand to walk safely across.

I have hired a company called Planit Firewalking who have been brilliant and have helped me plan the event at each stage. The Firewalk will be held on Saturday April 23 up at our local Cricket Club in Mumbles, Swansea, Wales.

We hope to have 40 people signed up to walk and will make it a family event, with a flaming bbq serving hot burgers and hot dogs! We have asked each walker to raise a minimum sponsorship of £100 so if we can achieve our target of 40 walkers, even deducting the cost of Planit Firewalking we should be able to raise a good sum of money for PC Project, while raising awareness of PC and the pain it causes.

UNICORNS 4 PC SET 2K OBSTACLE RACE IN THE UK
From Debbie Gregory:
My daughter, Ellie (age 11) and her friends are doing a 2k obstacle race, it's similar to an army assault course. They have come up with a team name 'Unicorns 4 PC' and they have a logo.

Her dad Neil, is going to run the adult race the day before with a few of his friends who will all get sponsorship too.

PC Awareness 2016 Has Begun!

2016 PC AWARENESS
Seven events held or planned...

1. Seinfeld Show in Salt Lake City
2. AAD/CSD meetings / USA
3. Rare Disease Day / Scotland
4. Firewalking / Wales
5. Unicorns 4 PC / UK
6. Brochures to physicians / USA
7. Golf Tournament / USA
8. Plan now to have a PC Awareness Event in your community and at least 100 PC Awareness efforts around the world in 2016.
9.
10.
11.
12.
13.
14.
15.
...
GOAL: 100 Events in 2016

What’s the deal with Jerry Seinfeld still doing stand-up? Not that there's anything wrong with that. Find out for yourself when the man who helped invent the modern sitcom comes to town. A portion of the proceeds to benefit the Pachyonychia Congenita Project. Abravanel Hall, Jan. 14, 7 pm., artix.org or 801-355-ARTS

Thanks to Chris Misiano, the first 2016 Pachyonychia Congenita Awareness Event was the Jerry Seinfeld Show.
WANTED

This month, (in addition to images from your PC Awareness efforts), we are especially asking for images of PC fingers and toes after nail removal. This isn’t a treatment many PCers have experienced and the results are mixed. We expect the article on PC Nail Removal (first begun in 2012) will be published soon. Recognizing that we need to better understand why some nail removal procedures are successful and some not, we will immediately begin to gather new data using an updated IRB-approved questionnaire. More images from those who have had nails removed will be helpful.

Two of the four who stopped at the booth were PC Project friends: Roger Kaspar, CEO/TransDerm and Manor Argarwala (New Delhi, India). Both are members of the International PC Consortium. The Coalition of Skin Diseases (CSD) also holds a luncheon in conjunction with each annual AAD meeting and Roseann and Mike also attended and participated at the luncheon representing PC Project very effectively.

PC Project will send application information to families who are participating with us in the IPCRR (PC registry.)

PC Advocate Roseann McGrath and her husband, Michael, attended the 74th Annual Meeting of the American Academy of Dermatologists held March 4-8, 2016 in Washington, DC. Roseann wrote “There must be 10,000 attendees” and she is correct. There are a large number of dermatologists. However, there are only a few interested in rare keratin disorders.

Roseann and Mike manned the booth for the Coalition of Skin Diseases (CSD) and she writes “The booth was in a good location, a lot of foot traffic, but unfortunately not a lot, if any (well 4) came by the booth.”

Organization of Rare Disorders website is rarediseases.org. There are over 7000 rare disorders/diseases many of which are genetic. Also, PC is not always 'inherited' - at least 40% of cases are spontaneous (not inherited).

POST: Happy National Rare Diseases Day! I use the word "happy" because it is a joyful word of celebration. We not only celebrate this day and the awareness it can bring (thank you PC Project and NORD) but also those whose lives have been affected by a rare disease and their abilities to still live life with great joy. This is my Alex (K16) one of the happiest and most active little boys you will ever meet. His daycare has nicknamed him "The Galloping Giggler" as PC has not slowed him down nor affected his spirit.

RECENT POSTS ON OUR FACEBOOK PAGE

POST: I would like to point out PC is not a disease as its not contracted it's an inherited condition due to gene mutation

REPLY: Thanks for your comment. PC is a disease and a disorder. The definition of disease or disorder is difficult, but something “that is not simply a direct result of physical injury”. The National Organization of Rare Disorders website is rarediseases.org. There are over 7000 rare disorders/diseases many of which are genetic. Also, PC is not always 'inherited' - at least 40% of cases are spontaneous (not inherited).

Tooth Donor Awards This Month

🌟 Kiera Chapman
🌟 Kaden Chapman
🌟 Lily Ziebell

The NIH research on PC teeth is continuing and initial reports will be presented soon.
Why is it so important for PCers to initiate awareness events? Well, we are small. And because we are small we have to have big voices. Large organizations succeed with lots and lots of small voices. Because we do not have the numbers that other organizations have we must have loud voices. Every single PC voice is extremely important to raising awareness and raising funds for PC.

The most important place to let your voice be heard is in your own community. No one else in your community will sound the trumpet for PC. Many simple projects that do not generate a lot of money will generate a lot of publicity and awareness. Your network will also reach people who are not responding to traditional methods of fundraising for PC. Your project is about reach. Through your events you will spread the word and encourage others to participate with you. Be vocal about PC and PC Project. Tell your friends, family, neighbors, co-workers, doctors.

Whatever you do, make your awareness event your own. Choose something that will be fun for you and your family. Fundraising idea websites include


Find a project you like and that will be fun to do!
If you haven’t yet joined the registry, please do that right away. It takes about 30 minutes and your data has the power to bring more research for PC.

INTERNATIONAL PC RESEARCH REGISTRY (IPCRR)
Holly Evans, Program Director
The IPCRR is powerful because of you. Participation in the IPCRR leads to more research for PC. This is why we ask all those affected with PC to join the registry and participate with us even if a relative has previously joined. Each person adds to the data and expands the knowledge about PC.

During March 2016 there were 15 new IPCRR participants from Australia, Bulgaria, Israel, United Kingdom and United States (AL, CA, FL, ID, IN, MT, NJ)
0 updated IPCRR forms
7 Genetic testing kits sent out
9 Genetic Testing Reports sent out

After you complete the forms online and have your consultation and genetic testing, here are a few ways you can continue to make your Patient Voice heard in the IPCRR:
1. Respond to emails, surveys & addendums sent only to IPCRR participants
2. Invite and encourage all affected relatives to join the registry at http://registry.pachyonychia.org/s3/IPCRR
3. Regularly update your/your child’s data and add new photos at https://registry.pachyonychia.org/s3/UPDATE
4. Share photos with us of your “normal” PC. We want to also see the bad times like a bad blister, infection or other problem. These help teach about PC.

INCORRECT INFORMATION
There is a massive amount of incorrect information about Pachyonychia Congenita online and in printed materials. Our PC website (www.pachyonychia.org) is a reliable source of correct and up-to-date information on PC.

However, on the PC website, we include all articles published about PC and related disorders. The best articles are highlighted on the ‘Select Lists’ on the right panel. Some keys to help you evaluate information and articles.
1. Is genetic testing information included? If not, the report may not really be about PC! An example is an article on ‘recessive’ PC. All of the information on our website states that PC is a dominant disorder. This is important. After nearly 8 years, genetic testing showed that the patients said to have ‘recessive PC’ actually have a completely different disorder.
2. Does the information contradict information listed on ‘What Is PC?’ on the PC website? Many single case (or single family) articles incorrectly connect things found in that family with PC. For example, although testing is conducted to establish PC, the article says that hair loss or deafness or some other characteristic is associated with PC. You can rely on the PC website information. Or, feel free to send us an email for an answer to PC questions you may have.
3. Another key is to look at the dates of the publications that are cited. Remember, the genes that cause PC were discovered by Smith/McLean about 1995. Before that date, there were many assumptions and guesses as each case was examined. Most of the best publications on PC have come after we had more than 200 patients in the IPCRR (about 2008 or after).

Social media posts may often have inaccurate information. We hope our responses to several recent posts on our PC Facebook page have been helpful and provided correct information.
(a) PC causes loss of teeth. No. This is not shown in the data available for those with genetically confirmed PC. Other similar disorders do cause loss of teeth.
(b) PC is not a disease because it is inherited. We explained that PC can be described as either a disease or a disorder.

Importance of Genetic Testing
Frances J.D. Smith. Chief Scientific Officer, PC Project
There are many rare skin disorders - some, rarer than PC but they may have some similar features. Some-
times determining which clinical features are linked to a disorder and which are co- incidental can be difficult and confusing. For a doctor it is important to gather a detailed clinical history from a patient to guide appropriate genetic testing, to confirm the clinical diagnosis and identify the exact mutation. Why is this important?

1. Inheritance pattern of a disorder - autosomal dominant (one copy of the mutant gene, like PC) or recessive inheritance (two copies of the mutant gene, like desmoplakin, below). The mode of inheritance determines the risk of passing a disorder on to your children.

2. Care of your disorder - can share information/tips with others with the same disorder.

3. For some disorders, monitoring of symptoms that aren't visible is important. For example, in patients with mutations in the desmoplakin gene, skin blistering occurs soon after birth and then nail dystrophy and palmoplantar keratoderma develop that can look similar to PC. However, hair is sparse and tightly curled. Confirming the disorder by genetic testing is extremely important. Desmoplakin is found in desmosomes - a type of junction that forms tight links between adjacent cells in skin and also in cardiac muscle. Therefore, in addition to skin fragility & sparse tightly curled hair, the heart can also be affected. Regular monitoring of these patients is necessary to detect any cardiac abnormality early.

4. For a specific disorder knowing the genes involved (their function and where they are found), the exact mutations (the type - missense, deletion etc and the number of different mutations), and the inheritance pattern, together with the numbers of patients determines and drives forward specific research for development of future treatments. This is why, for PC, it is important for PC Project to gather this information and for as many affected family members as possible to enrol in the IPCRR.

**EXAMPLES from the IPCRR**

**Do these two unrelated patients have PC or a different disorder that mimics PC?**

Patient 1 has thickened nails (nail dystrophy) and mild callus (keratoderma) on their feet. On closer examination eyelashes were noted to be thin and sparse and eyebrows were sparse.

Patient 2 has nail dystrophy, mild keratoderma and some hair loss since childhood.

- Neither patient has plantar pain, oral lesions, cysts or follicular hyper keratoses.
- Do they have mild PC? Are the sparse eyelashes, eyebrows (patient 1) or hair loss (patient 2) related to the nail dystrophy and keratoderma disorder? Yes! Do they have PC? No!

Screening PC keratin genes revealed no mutations but a mutation was identified for both patients in the gap junction protein, connexin 30. Mutations in connexin 30 cause Clouston syndrome, an autosomal dominant disorder, with nail and skin involvement plus hair changes. Hair loss distinguishes it from PC but this can range from very mild (and easily missed) to total alopecia (baldness.) Treatments for this will likely not be the same as treatments for PC.

**PC ADVOCATE**

**REPRESENTS PC PROJECT**

PC Advocate Roseann McGrath and her husband, Mike, attended the American Academy of Dermatology (AAD) meeting held March 4-8, 2016 in Washington, DC. Roseann and Mike helped to staff the Coalition of Skin Diseases (CSD) booth and attended the CSD luncheon to represent PC Project. Roseann provided a detailed report and materials from the meeting. Thank you both!

**OBSERVATIONAL STUDY: PAIN APP AND ACTIVITY TRACKER**

24 individuals (12 PCers with their 12 ‘normal controls’ who match age/gender/location) have completed the first four weeks (Section 1) of this study. The participants are consistently providing their data. The information being gathered is fantastic. We believe this will be a landmark study in establishing the impact of pain for PC patients. The next section of the study will begin in June. If you’d like to join the study send email to info@pachyonychia.org
**PARTICIPATION is the way to ensure your Patient Voice makes a difference.**
The chart below lists 10 opportunities PC Project has offered to those in the IPCR and the scores for 25 PC patients.

**What is your participation score?**
2. Become a donor. Even a small amount makes a difference and a small monthly donation is very valuable.
3. Provide a biopsy if asked.
4. Be active on Facebook PC Patient Chat — and help guide others to PC Project services. LIKE and follow Pachyonychia Congenita Project on social media sites.
5. Host a PC Awareness Event (large or small these matter!) See page 1 of this NewsBrief for more on this opportunity and plan your 2016 event now.
6. The PROMIS survey is closed, but may be collected again. When we have a survey be sure you respond. You are a major part of the PC patient population.
7. Addendums and surveys are sent out as we seek to know more about PC (such as nail removal, use of retinoids, and other special interest research).
8. The 2011 and 2014-2015 pain studies are complete. But other studies will be offered in the future. Will you join in?
9. Activity Tracker and Pain App study is on-going now. If you have plantar pain and want to join this study, send an email to info@pachyonychia.org
10. UPDATE your IPCRR data.
   - Adults – every year
   - 6-18 – twice a year
   - Under 6 – 4 times a year
   - Under 1 – every month

Two additional ideas to increase your participation with PC Project:
- Visit [www.pachyonychia.org](http://www.pachyonychia.org) website often to read and learn. Please send us your feedback.
- Share your story with us either for the website or just anonymously. Explain how PC affects your daily life? What are your best care techniques?

Remember we are here for you! Please contact us if you have a question, problem or need any service from PC Project.

Together we can make a difference for all those with Pachyonychia Congenita.
We are excited to report on some of the PC Awareness events for 2016. We encourage everyone to do something large or small to raise awareness, raise funds—and have fun!

Firewalking Event Summary
By Julie Peconi (Baker)

On April 23 at the Mumbles Cricket Club in Swansea, Wales, 40 brave people accepted the Firewalk for PC challenge, walking barefoot over hot coals measured at 1200 degrees!! The event was organised by Julie and Tom Baker to raise money for research into PC. Both Tom and their son, Timmy, age 8, have PC-K16.

The team from Planit Firewalking were in charge of all things fire related and provided an opportunity for 40 people to walk across the coals. While Planit Firewalk owner, Errol, trained the Firewalkers out of sight in the marquee, his sons, Jack and Joel, stoked the fire getting it nice and hot!! Halfway through the training, the Firewalkers were able to have a look at the fire and as you can see by the pictures, the fire was very real! After all 40 firewalkers were feeling ‘strong, powerful and magnificent’ Errol brought them back out where one by one, cheered on by the crowd, all walked across the burning embers! The turnout for the event was amazing and several spectators from out of town said that they could really feel the community spirit.
As part of the event, Tom and Julie also organised a barbeque selling burgers and sausages stacked with hot peppers and fresh produce donated by a local farm shop. Friends also made fire themed cakes which sold out. Overall the event has raised in excess of £4000 for the PC Project while also raising publicity for PC!

TIPS ON HOSTING AN EVENT
We asked Julie to share her six tips for success.
(1) Choose an event:
We researched companies who put on fire walk events in the UK. After speaking to a couple, we went with Planit Firewalk. The cost for them to host the event was £1200 for a maximum of 40 walkers, with each additional walker charged at £40. We gave each walker a goal to raise £100, which covers the cost of the walk and raises a reasonable amount per person. We didn’t want the total to be too difficult for each walker to raise.

(2) Choose a venue for the event:
We approached 3 different venues (a) a local pub (b) a café next to the beach and (c) the local cricket club. All were keen to host the event without charge, both because it is a charity event and also the potential sale of drinks!

(3) Organise your event:
Once the date was chosen and booked with the venue and Planit Firewalk, it became real that we needed people to actually walk! This was the daunting bit as 40 people seemed like a large number. The response was terrific however – it really captured the imagination, and the first 30 walkers were signed up with relative ease! Filling the remaining 10 places took a bit of work persuading certain individuals, but in the end 40 places were successfully filled.

(4) What else can be done to supplement the event:
As part of the event, we also organised a barbeque selling burgers and sausages stacked with hot peppers and fresh produce donated by a local farm shop. The barbecue was a success. We brought in 150 burgers and 150 sausages. It was an epic 3 hours, with 5 of us manning the barbecue, taking money, unpacking cheese and cutting rolls. We added trimmings of lettuce and tomatoes and hot peppers and hot sauce which was a success. All the burgers were sold and only a few uncooked sausages remained. The barbecue alone raised over £500! There was also a cake stall with homemade donated cakes, pre-packaged sweets in bags selling for 50p, and also a stall selling homemade sangria. Each was a great success.

(5) Promote your event:
As the date approached I took to Facebook relatively often to keep the date of the event in people’s minds. We probably had over 300

In fact many people commented on how it was nice to be raising money for a smaller charity. One person said afterwards that ‘I had never hear of PC, to be honest, but when it was explained, I’d walk on anything to help anyone that has to live with this’.

The event was so successful that the Bakers hope to make it an annual event!
people attend, with many just coming for the barbecue and to watch. It was surprising the number of people watching who said they would like to do it if we organised a similar event again.

(6) Rate the success (what made it successful):
Planit Firewalk was excellent. The walkers were taken off prior to walking for ‘training’ which was hilarious! It will be remembered by all who were there for years to come. During the walk itself the spectators made it a real party atmosphere, cheering each walker as they walked the 4 or so steps across the coals.

**SIMPLE WAYS TO RAISE FUNDS WITHOUT AN EVENT:**
- Collect coins - have a jar in the house/office & put in those small coins that fill up your purse & pockets.
- Go out for dinner with friends - skip starters or dessert or drinks & donate the cost.
- Recycle scrap metal - find out about your local scrap metal merchants and what they buy. Then check your sheds/yards for old bits of scrap metal/house hold appliances, car batteries etc. Ask your friends/neighbors - many will find things they’ve been meaning to get rid of for ages.
- Recycle phones or ink cartridges. There are companies that pay for these and supply a box to put at your work or other businesses to collect these.
- Other ideas—every effort large or small really does help.

**EVERY AWARENESS EFFORT MAKES A DIFFERENCE**
The van Der Laan family from The Netherlands continues to raise funds in many creative ways. For example, their local market sells ‘banana boxes’ for a 1€ donation to PC Project. This year they sent 60€ from this project! And the market likes to post the certificate PC Project sends to them! Almost everyone has a market near them. What a great idea!

**GLOBAL GENES**
hosted an interesting webinar *Understanding Gene Therapy*. The meeting recording and slides are available at [https://globalgenes.org/understanding-gene-therapy/](https://globalgenes.org/understanding-gene-therapy/)

**WORLD CONGRESS-ORPHAN DRUG DEVELOPMENT**
Matt Morgan, PC Project, and Roger Kaspar, TransDerm, attended this conference held April 20-22 in Washington, DC to gain insights into ways PC Project can continue to press for treatment for Pachyonychia Congenita, an orphan disease.

**BETTER BUSINESS BUREAU BBB**
has given PC Project a top rating confirming PC Project meets all 20 standards for excellence.

**PACHYONYCHIA CONGENITA PROJECT-EUROPE** has been officially recognized as a charity in the UK. More next issue.
We need you! This month we have lots of requests for ways you can participate directly in the research process and help with various research projects.

Please let us hear from you. If you have any questions, please call us at 801-401-6300 or 877-628-7300.

**Biopsies.** We need to collect 2 or 3 biopsies from genetically confirmed PC patients (KRT6, KRT16 or KRT17) for a project to develop PC stem cells for possible future use in gene correction techniques. These biopsies are much easier than the ones we often need from the affected areas on the soles of the feet. These can be from hip, thigh or any part and can be done by your local physician. There has to be special packaging that we will provide. Also, we will pay any costs not covered by insurance. If you are interested, please contact us at info@pachyonychia.org.

**PC Wiki.** We constantly add your tips and suggestions to our PC Wiki. Let us know your ideas and what you find most helpful in caring for your PC.

**Blood Samples.** As most of you know, we use saliva samples for DNA testing. And, we have collected biopsies many times for specific research studies. A new study from researchers in Germany indicates they can conduct some additional research using blood samples. If you have PC and are interested, please email us at info@pachyonychia.org. We will provide the collection kits and pay any lab costs.

**Help From Parents.** Do you have any good ideas for choices of shoes and socks for children with PC? From a young age when they are just starting to walk? What are your experiences? Please send your suggestions and comments to info@pachyonychia.org.

**A Tribute to Our Friend**
As some of you may know, our dear friend and fellow PCer, Harry Stergar is on hospice. Although his long hair is gone, his smile and spirit and determination are as strong as ever. Harry decided that since he is on a lot of pain killers, he should give samples at this time — and so on April 21, he arranged with his physician for a visit for biopsies. Roger Kaspar kindly travelled from TransDerm to help with the collection, but due to a travel mix-up, Harry and Debbie actually had to go to the appointment twice! Harry has now also arranged to have teeth pulled for research! He has often filled our email with messages of encouragement, with laughter and with great ideas. We love you, Harry!
Pachyonychia Congenita Awareness

Organized by: Pachyonychia Congenita Awareness

We appreciate each of you who have ordered PC tee-shirts! We invite you to send the link above to your family and friends and invite them to order as well. We know shipping is expensive outside the USA and will try to do other campaigns in connection with Patient Support Meetings so everyone can have an opportunity to participate. We encourage each of you to host something in your own community for PC Awareness month. In this News Brief we highlight a number of amazing projects by individuals. This is wonderful. Our goal is ‘less patient and more empowered’ and as you hold an event you are empowered to speak about PC and spread awareness around you.

Tee-Shirts for PC Awareness

We invite all friends of PC Project to raise awareness of Pachyonychia Congenita and raise funds for research, patient support and clinical trials. Please join us with Pachyonychia.

Buy a shirt today to help create awareness for Pachyonychia Congenita

All funds raised will go directly to PACHYONYCHIA CONGENITA FUND

43 items  $730 raised

50 goal

Select Item

Next Level Jersey T-shirt
$ 25  Unisex - Heather Grey

Buy / Donate

Orders are delivered about 2 weeks after the campaign closes.

Ends June 15th
NEW LOGO FOR PC PROJECT
At the recent IPCC scientific meeting, Dr. Ofir Artzi from Tel Aviv, Israel, used this ‘heart in feet’ logo in his presentation. We liked it so much that with his permission we’ve adopted it for our PC Awareness 2016 campaign. Thank you Dr. Artzi (who by the way, is the leading physician for one of the clinical trials now being developed for PC).

ENGLAND—Darling Ellie Gregory, age 11, and her friends formed the Unicorn Team and ran an obstacle course to raise funds for PC. Her Dad also formed a team and entered the race to raise funds. Way to go!

We invite every PC patient/family to do something big or small to become empowered on behalf of your PC condition.

Have Fun! Raise Awareness! Raise Funds for PC!

FRANCE—Mother’s Day Donations for PC Project
In March, Marie Jose Billeau (at left in photo) spoke about PC Project to Julie Hutchison (center in photo). Julie and Marie Jose read together the monthly PC News Brief and read about doing a good deed. But, Marie Jose said, "What can I do?" Julie had a good idea. She sells organic beauty products from London and she donated a lovely little bag of beauty products as a fundraiser for PC. Marie Jose writes “Well, in France, we have Mother's Day on May 29th, so my husband and my children gave "euros" for PC Project and surprised me with this bag of wonderful products. We are very happy that Julie contributed a little to help PC Project and now I will go to the post to send you the check. JE T'EMBRASSE TRES FORT!”
We did an ice cream social with our bike club for awareness on Tuesday night. It was great and we raised $500. We are doing a second night next Thursday with the mountain bike club. (Additional donations have also been received at PC Project.)

Maryland, USA—Kislal Family Letter and Cupcake Sale for PC

Dear Friends,

Ela Kislal was born with a rare condition known as Pachyonychia Congenita (PC). The disease is caused by a chromosome mutation which affects the production of keratin. PC patients experience thick nails, painful calluses and blisters, cysts, follicular hyperkeratosis, and a white film on the tongue. PC patients experience almost constant pain.

June is PC Awareness month. All money raised will be donated to the Pachyonychia Congenita Project. PC Project was invaluable to our family when Ela first encountered difficulties related to her condition. They provided free genetic testing to diagnose her disorder and they paid for our attendance at a patient support meeting which provided a chance to meet others with the disease, and to share information about how to best manage her condition. In addition, PC Project hosts an annual research symposium to bring together scientists and physicians for developing and delivering effective treatments for PC, and has sponsored more than 18 clinical studies.

Please consider buying a cupcake. All proceeds will go to PC Project. Also, feel free to take a look at the pamphlets and help yourself to the small paper books. Your help is much appreciated!

FRANCE

Friends and Family of Beatrice Wannamacher 1973-2014 Yard Sale for PC Awareness

We were especially touched to see this event held in remembrance of our dear friend, Beatrice Wannamacher who died unexpectedly at the young age of 41. Beatrice provided many services to PC Project and to LeCouer au Pied (the PC group in France). This is a wonderful example of never forgetting our loved ones and doing something special to raise funds to help others who suffer as Beatrice did.

Thank you! Merci!
**NEWS FROM PC PROJECT**

Since the last PC News Brief in May 2016, lots of important things have been accomplished at PC Project and we’ll highlight a few in this Newsletter.

1. IPCRR continues to grow with an average of two new patients registering every week and now over 1660 registered patients. Please remember to update your information—we can provide a link that lets you skip a portion to make the data input even quicker. And if you haven’t ever filled out the forms—please do that. It is important and it is making a difference.

2. PC Advocates were selected and 8 participated in training prior to the IPCC scientific meeting (see comments in this newsletter.) We will add additional PC Advocates both in the USA and in other countries. In the last News Brief we included a ‘check your score’ article. We urge you to participate in every way with PC Project— we want you to be ‘less patient and more empowered!’ The PC Advocates all scored very high (some 100% in participating in every opportunity they have been offered!) They have attended Patient Support Meetings, updated their IPCRR, answered each survey, etc. All PC genes are represented in this group of highly accomplished individuals. We know they will be able to represent all PC patients with a united voice and help us expand our efforts.

3. The 13th Annual International PC Consortium (IPCC) Annual scientific meeting was held in Scottsdale, AZ.

4. Clinical Trials. It takes an enormous team effort to develop proper protocols that will result in effective outcomes and gain approval of the FDA (USA) and other regulatory agencies in each country. Each trial will be focused on specific groups of patients and in specific localities with physicians who are able to conduct the trials. Informal patient experiments will not move us forward to approved treatments with costs covered by medical systems and/or insurers. We understand how eager physicians and patients are to try these things. We hope we can work together in a coordinated manner to be most effective and gain real results. Many things that have been tried have not been found effective enough to take forward. Also, the details of the trials are not always obvious. For example, in topical rapa some of the individual patient trials have used a formulation that delivers no active drug and are, therefore, not resulting in any successful outcome for these patients.

5. The following trials are currently in design at PC Project. We don’t know if any of these trials will be approved or successful in helping those with PC but these are the best options at this time:
   A. BOTOX INJECTIONS
   B. TOPICAL RAPAMYCIN
   C. NEW RETINOID DRUG
   D. TARGETED siRNA

6. Publications. We continue to publish articles about PC. We have many articles now in the leading dermatology journals and now our focus is to also have articles in pain, podiatry and pediatric journals. It sometimes takes 2 or 3 years for a publication to be completed.

The following articles are about to be published:
   a. The 2014 Pain and Clinical Exam study.
   b. The results of the nail removal survey from 2011.
   c. The results of the biopsy project in August 2014 and the follow-up exams in AZ and NY. This will be a major article in PAIN (the leading journal for pain.)
   d. A second article in a podiatric journal.

6. We also arrange individual responses to patient questions and needs. Recently a family in Spain had questions. Dr. Ramon Grimlatt kindly translated for us to help this family. Each family and patient is important to us at PC Project.

We feel we need to do a better job of letting you know that we are working hard and clearly focused on our mission goals:

**Fighting for a cure**

**Connecting and helping patients**

**Empowering research**

**WANTED**

Thank you for the amazing, quick responses for blood and biopsy donors. We are arranging matches for various projects (PC-type to the proper study) and will be in contact with each on you in the next months. Thank you again!
IPCC SCOTTSDALE, AZ
MAY 10-11, 2016
Frances Smith
Chief Scientific Officer

Nearly 50 physicians and scientists from around the world gathered on May 10-11, 2016 in Scottsdale, Arizona for the 13th Annual Research Symposium of the International Pachyonychia Congenita Consortium (IPCC). The first day focused on current research including the genetics of PC and other rare skin disorders, studies on different aspects of keratin biology in relation to PC, developments in EB research and therapy and sweating and ways to measure it. Another topic was pain and understanding pain including a presentation reporting the initial results of the pain study carried out at 3 patient support meetings in 2014/15. Histology of the nail and nail removal as a PC treatment were also discussed. On the second day presentations and discussions concentrated on proposed clinical trials and the importance of measuring clinical trial endpoints to evaluate these studies.

PC ADVOCATE TRAINING
A training session was held on May 9, prior to the IPCC meeting with 7 specialists and the 8 PC Advocates who then joined the IPCC meetings the following two days. Here are comments from a few of the PC Advocates.

Christine Block, PC Advocate
I was honored to attend the advocate training and research symposium in Arizona May 9th-11th. As advocates we met for further training to understand all forms of PC and be able to effectively communicate with patients, physicians, and others. This was also a time for us as advocates to come together and form a team to continue working toward a cure for PC. We also attended the research symposium where approximately 25 researchers and physicians from all over the world presented their current studies on PC treatment. The collaboration of these researchers and physicians was impressive. They all are putting in significant time and effort to find a cure for PC.

As a parent of a child born with a spontaneous mutation, K6a, this was also my first opportunity to meet other PC patients and learn from them. It was an emotional time to listen to the pain everyone with PC lives with on a daily basis but it was also empowering. We came together and became friends and a PC family. The advocates are strong, driven and amazing people.

After spending 3 days in these meetings, I left inspired to continue doing my part to help PC project find a cure. My part is to con-
We, the PC Advocates are blessed, honored and so pleased to advocate for the patients and to educate the physicians, researchers and scientists (to put a face to their “cells”).

One issue, however, that befuddles me is if these doctors, physicians, scientists from around the globe can come together to collaborate and find treatment for our ultra rare orphan disease, why can’t the patients who make up the IPCRR, our patient consortium, find the time to register and be genetically tested (and update the information each year.) This is the way to thank them and do our part to build towards success.

Jack Padovano, PC Advocate

PC Advocate training lesson learned: Being an advocate is about using your voice to make a difference. The PC Advocate training taught me the power of using my voice to self-advocate on behalf of myself and other people with PC. I learned how to speak in factual terms about PC, how to get and evaluate information, find out who will support me in my journey, know my rights and responsibilities, problem solve, listen and learn, reaching out to others when I need help and friendship, and learn about self-determination.

The PC Advocate Program was based on the premise that I have a powerful story to tell about my life with PC and can use it in a way to educate multiple audiences including other PC patients, scientists and doctors, and people who have never heard of PC. I am delighted and honored to be a PC Project Advocate!

IPCC lesson learned: It truly takes a village to find a treatment and eventual cure for PC. The village includes PC Advocates, PC patients, the hard-working PC Project team, and a collection of the world’s smartest (and nicest) scientists and doctors across the world. The IPCC is a special club of doctors and scientists working in clinical interested in collaborating efforts to develop and deliver an effective treatment for PC. These folks are truly committed to helping and unlike most organizations like IPCC, these folks leave their egos at the door. They have one goal: to treat and cure PC.

As I sat and listened to each doctor present different aspects of their work with PC and related disorders, I was blown away by the level of intelligence, commitment, and downright hootspa of each person. In between sessions, I was honored to be able to ask questions since most of what they presented went way over my head! No matter, they took the time to connect and made sure I walked away a bit more educated.

Stephen Wittmer, PC Advocate

I want to give my heart-felt appreciation to PC Project for giving me the opportunity to attend the 2016 IPCC in Phoenix. The meeting felt like a family reunion with scientist and physicians happy to see each other again and welcome new attendees with open arms. Even a high ranking member of the National Institutes of Health was in attendance. What a pleasure it is to be able to spend my free time at breakfast, lunch and dinner discussing my rare disease with them. These are
individuals who whole-heartily care about finding a treatment or cure for Pachyonychia Congenita. The best part about it is that PC Project has done an exceptional job of finding the best scientists and physicians in the world to join their team. The advocate training that we received on the first day was a delight. Getting to know the other advocates was wonderful, and I am looking forward to helping other PCers as they struggle with the same problems that I have lived with all of these years. I can only imagine the fear that a mother would have after having a baby with a spontaneous case of PC.

What a comfort it would be to be able to speak with a patient advocate who could tell them that everything is going to be alright and this is not the end of the world. In fact, you will find that your child will be a special blessing that you could never replace.

PC Project is not just about finding a cure or treatment for us, but it is also a valuable tool for helping patients who struggle with this rare disease on a daily basis. Nothing is better than knowing that we are not alone, and thanks to PC Project that is not the case anymore.

FROM THE PC PROJECT

SPAIN-Claudia Avella
(mother of a toddler with spontaneous PC-K6a)
Hello, We'd like to share how we care for our toddler’s nails. Our son is a light sleeper, so it’s not possible for us to cut his nails while he is asleep. During the first year, Dad cut his nails while Mum breastfed the baby who used to concentrate only on the breastfeeding, making nail care easier to do. At around the first year of age it became increasingly difficult to do this because baby was no longer distracted by feeding. We changed our strategy and made nail-cutting more frequent so that baby would get more used to it (and we have also gotten more used to it). Since baby has a bath most nights, we integrated cutting or filing one or two nails each night as part of his bedtime routine after his bath. This is also a two-person job. Dad cuts nails while Mum shows baby a short age-appropriate video on her smartphone (2-4 minutes) and controls the other hand/feet, etc. Baby is now more used to nail care than he was before and cooperates much more. I hope this can help others.

Andrew Gaskill
Response to request for information on shoes for children.
Regarding shoes and socks -- when our son first started walking we had him in Toms shoes. We found them to be more breathable and flexible than the typical sneaker scaled down to child size. He’s 5 now and we have him in Plae shoes, for the same reason. If I had known about Plae shoes earlier we would probably have had them earlier, but I don’t know how small their sizes go, or if the smaller sizes are as good as the larger sizes. For socks we just always find nice thick socks to cushion and prevent rubbing. We also let him go barefoot as much as he wants, and he likes being barefoot. On one hand I worry it might make the callouses on his heels thicker, but I think there’s also a natural exfoliating effect as well. He has always had the thickest callouses on the sides of his big toe, and the most painful blisters between his toes, and going barefoot reduces that irritation, and it keeps feet dry too.

Response from an unidentified PCer to a recent online post suggesting that those with PC should not have children. PC Project does not give direction in regard to this topic and respects each person’s decision on this topic. We felt this response was worth sharing:

I'm sorry but I find your comment (suggesting those with PC be sterilized) to be extremely offensive and short sighted. One of the hallmarks of this disease is the feeling of isolation and the inability to feel normal, yet finding someone who loves you and to create a family with that person is one of the best things anyone could do and gives purpose to an otherwise empty life. Yes, there is a possibility of passing on PC but passing on the joy of life outweighs any unknowable variable of life with PC. And to simply “stop breeding” which would somehow eradicate genetic conditions is laughably dumb. My mother had a spontaneous PC mutation, should she have been put down? Should she be doomed to life the rest of her life alone? These things will happen no matter what. And with medical advancement happening faster than ever, what’s to say by the time me or any other young person with PC starts a family that there won't be a way fight it better than ever.

NOTE: Currently over 40% of the confirmed PC cases in the IPCRR registry are ‘spontaneous’ and are not inherited.
Grunenthal Patient Day  
June 1, 2016

Grunenthal pharmaceuticals is a company based in Germany and dedicated to pain research. They have recently started a focus on rare disorders involving pain. Those attending the Patient Day were top level executives from Germany and from Grunenthal USA as well as staff members involved in day-to-day research and development at the company.

Four rare disorders were asked to present their story at this all-day meeting held at the Omni Hotel in New York City. The groups were:
- Complex regional pain syndrome
- Duschenes Muscular Dystrophy
- Pachonychia Congenital
- Parkinsons

We were very grateful to be invited. I hope every PC patient will at sometime be able to see the presentation given by Janice Schwartz on behalf of all PCers. In 20 minutes she changed lives! At least that is what people told her afterwards. She explained how PC affects her life, the hardest things about PC and how PC patients manage their PC. She did it with humor, short/clear illustrations that demonstrated her pain (without bemoaning her pain!) For example, she had a photo of when she was a camp counsellor and because it was too painful to walk around the craft tables, she just climbed up in the middle of the table and sat that way to help the kids. Her choice of pictures was effective. As she said “I’m telling you my deep, dark secrets and so now we have to be best friends!”

And she explained how those with PC look completely normal and often mask/hide their pain because it is difficult to explain to others. At the end of the day’s sessions (which were from 7am to 5pm), the Grunenthal CSO, Klaus-Dieter Langler, asked that we form small groups for each of the 4 disorders and come up with things that Grunenthal will do to help us.

Here is our list:
1. Publicize Pachonychia Congenita in the ‘pain world’ so that people know that pain is a major part for those with PC.
2. Develop some type of training to help those with PC be more familiar with pain, how to talk about it, etc.
3. Provide suggestions for our website to add more pain information.
4. Provide suggestions to revise our IPCRR questionnaire to add better pain questions.

In addition to the above, they will have representatives attend our Patient Support Meeting in Edinburgh and help us gather Patient Reported Outcome Measures (what patients want from a clinical trial.)

The PC team at Grunenthal has already been helping us with statistical analysis which we have never had access to previously. The data from the PROMIS questionnaire which many of you completed has now been analyzed. The data from the Pain App and Activity Tracker is now being evaluated. And, their team is helping us with comments and review of our clinical trial protocols.

We are very glad to have this help from this company. Those working with us are among the best people we have ever met. They are careful not to promise to develop or deliver a ‘miracle’ drug for PC. They currently have no product to sell for PC. I believe the will do what they have agreed to do—and this is an enormous help!

Grunenthal Pain Networking Reception  
June 1, 2016

Friar’s Club, New York City

Following the day long meeting, a ‘Pain Networking Reception’ was held where physicians and scientists specializing in pain gathered along with the Grunenthal staff, the four groups who presented in the day meeting and others. Six PC patients from NY joined Janice the day meeting and others. Six PC patients from NY joined Janice at the reception. Here are comments from several:

Peter Niketees—I had the privilege to attend the US Pain Networking event sponsored by Grunenthal Pharmaceuticals. To be in the presence of so many doctors and scientists solely dedicated in trying to relieve pain gave me and the other PCers hope that a solution will be found. I had the opportunity to speak to Marco Pappagallo (MD) and Mark Field

About Pain—there is excellent information on the Grunenthal website for patients regarding pain—what it is, how to talk about it, etc.
grunenthal.com/grt-web/Grunenthal_Group/Patients/Understanding/en_EN/265300325.jsp
(PhD) who have spent the last 25 years working on pain relief. I asked if PC pain was different from other diseases. He said yes. His explanation is beyond me to relate his answer. All the PCers present at the reception, and those not present, are grateful to Grunenthal USA and Grunenthal GmbH for their devoted work on our behalf.

Fran Sargianis—Attending the Pain Reception offered an enlightening experience. We were able to discuss our PC traits with a medical professional who was interested in learning anything and everything about PC which will help them to create a solution for our pain. Typically visits with medical professionals result in negative feelings as we are told there is no cure or that we can experiment with different meds. This visit provided a positive feeling as we answered their questions of scientific curiosity. We were not known as a specific number on a piece of paper. Hopefully with the face to face approach the scientists were able to gather important information that could help to change our (pain) future.

Mary Howard—The Pain Reception on Wednesday night was a first for us, so I had no idea what to expect. There were many people milling around talking. We got to talk to various doctors and scientists. I was very impressed at how interested everyone seemed to be in PC. They listened to us and asked questions. I left with a feeling that they truly will work hard to try to figure out how to help us with all our problems.


Best Western Edinburgh Capital Hotel
Friday, October 28th - welcome dinner starts at 5:30 pm
Saturday, October 29 - all day presentations, discussions and meals
Sunday, October 30 - half day meeting with the closing luncheon at 12.30 pm

REGISTER FOR MEETING at surveymonkey.com/r/2016PSM Please register as soon as possible There is no cost to register. Fees are paid separately.

MEETING FEES Before September 1, 2016: £50 per person 15 years or over After September 1, 2016, the fee increases to £60 per person 15 years or over Fees paid on arrival at the meeting increase to £75 per person Meeting fees are waived for PC patient and one family member attending a PC Patient Support Meeting for the first time and are also waived for all those under 15 years of age. PC Project and our sponsors pay 70% of the meeting costs which include the meeting room and equipment rental fees and food including Friday dinner, Saturday breakfast, lunch and dinner, and Sunday breakfast and lunch as well as meeting break snacks.

REGISTER FOR HOTEL at reservations@edinburghcapitalhotel.co.uk or by phone 0131 5359988. You are not charged until you check in at the hotel. There is no cancellation fee if you cancel 24 hours ahead of arrival. Be sure to register direct with the conference hotel. £50 for two persons per night (double or twin beds) £70 single person per night Those under 15 are charged only £5; those 15 or over are full rate. Please note that you are attending the PC Project/University of Dundee Meeting. If you are not a hotel guest, you will be charged a ‘delegate’ fee to attend the meeting. Do not use online or other booking services to book your hotel room for this meeting.

APPLY FOR SCHOLARSHIP FUNDING at surveymonkey.com/r/PSMScholarship After you have registered for the meeting and booked your hotel reservations, you can apply for a scholarship as needed to cover meeting fees, travel, hotel costs. Application Deadline is July 1, 2016 and awards will be announced August 1, 2016.

If you need assistance in completing the forms or if you have any questions, please contact info@pachyonychia.org

PACHYONYCHIA CONGENITA—EUROPE

We are very pleased that after nearly six months of preparation and effort, the new Pachyonychia Congenita Project Europe has been recognized as a charity. The entity was founded in Scotland (as a SCIO) and is a part of PC Project. All funds and expense will be a united effort with PC Project.

Officers include Mary Schwartz, Frances Smith (both PC Project staff members) and Phillip Gard (PC Project MSAB and Steering Committee member.) As many of you know, Dr. Gard is a retired physician, a PCer and one who helps us often with the physician consultation calls.

Those who live in countries with Gift Aid (where the government adds a percentage to all donations to recognized charities) can now donate direct to PC Project Europe and apply the gift aid request. We will soon add a page to the PC website to provide direct links and additional information.
NEW FEATURE. Starting this month, each issue will feature an article by one or more of the PC Advocates on a topic of interest/importance to those with PC. Readers are invited to add their comments via email or posting on our Patient Chat Facebook Page.¹

¹ Note: The Facebook Patient Chat is for those in the IPCRR. If you haven’t taken time to join the patient registry please do that today — it is the way we are able to move forward.

Back to School
By Christine Block,
PC Advocate
mother of 7 year old spontaneous PCer

Summer is flying by and soon we will be starting a new school year. The school supplies are starting to show up in the stores and the kids will find out which teacher they have next year.

There are always worries when kids start a new school year such as will they like their teacher. But when you have a child with PC there are a few more concerns and anxieties.

I have found one of the best ways to deal with my daughter’s PC is to make sure her teacher is aware of her PC right away at the beginning of the year. I started meeting with her teachers when she was in preschool and 4K.

Last year as she started Kindergarten, I contacted her teacher during the first week of school and set up a meeting with her and the gym teacher. We sat down and I explained what PC is and how it affects my daughter. I also took along one of the brochures provided by PC project that explains PC.

The school nurse also has a copy of the PC brochure in her records.

I have found that explaining things early and being proactive can help avoid problems down the road.

I tell the teachers that I don’t want them to limit my daughter or stop her from doing things, but she may have foot pain and need to sit down at times. I ask her teachers to listen to her and believe her if she is complaining of foot pain and explain that my daughter will do anything she can to participate. If she says something is too much she is really in pain. I explained that occasionally she has had nail infections and those are very painful for a few days.

I also encourage her teachers to contact me with any questions or problems so we can trouble shoot together. My daughter has learned that she can trust and talk to her teachers when she is having problems with her PC.

I hope everyone has a great 2016-2017 school year!

Share your comments on this topic. Tell us what things you have found that are effective for back to school.

At what age are kids able to talk to teachers on their own? Age 10? Age 12? Age 15? Or?

Do you have a special experience in helping a child with a difficult experience at school?
PC Awareness 2016 Events
The Buchta family held a local candy drive at their school to help spread some awareness and knowledge about PC.
Great idea!

We appreciate everyone who did something to help others learn about PC and raise funds for support and research.
Thank You!

PC T-Shirt Campaign
PCers, friends and family model their PC T-shirts!
Thanks to all who joined in this PC Awareness effort.
We raised $1,149.41 which will be matched $2-for-$1. This will provide meeting scholarships or genetic tests for patients or additional research funds.
Thank you!
PC Awareness 2016
PC AND PAIN

Although pain is the most basic and widespread concern for patients with PC, over the years PC has been classified as a 'nail disorder' and a 'skin disorder' with the focus mostly on appearance. Since 2011, PC Project has worked to shift the focus to alleviating the pain patients experience.

We have learned some valuable lessons and will share them with you in the hope that these are helpful points.

1. Those who specialize in pain may be anesthesiologists, neurologists, internists, etc. and may or may not work in a pain clinic.
2. The pain specialist is a somewhat new speciality and there is a wide variety of skill and practice across the pain field.
3. Since the type of pain a PC patient has may be different from other types of pain, you may need to find a specialist willing to learn more about PC. It is different than 'low back pain' or other types of chronic pain and the source of PC pain is not fully understood.
4. If you want to have help specifically with your pain, the country and community where you live will have a major impact on the services available and who provides those services. For example:
   - In the USA dermatologists do not treat for pain or prescribe for pain. Therefore, patients who are referred to dermatologists may not have an opportunity to receive help with pain from their dermatologist and may need to request a referral to a pain specialist.
   - Non-US dermatologists do prescribe pain medications and treat patients for pain in hospitals and out-patient cases. So, outside the USA, the dermatologist may be the best help for your pain.

Learning to talk about your pain can be valuable. We found a lot of helpful information on pain at the Grunenthal website. There is a link to help you talk with your physician about your pain.

Check it out at: see if it’s helpful: http://www.grunenthal.com/grt-web/Grunenthal_Group/Patients/Understanding/en_EN/265300325.jsp

PC PROJECT MAIL

Where PC is found? — Everywhere! PC is not found more in one country or among one ethnic population than any other. It is found everywhere and is rare everywhere.

- Do you find PC is more prevalent in Scottish or those of Scandinavian or Eastern European descent?
  No. It is the same everywhere and ethnicity is not a factor.

- I was referred to a consultant dermatologist in xxxxxx as they said I was a rare case as this condition is mainly seen in people of Afro-Caribbean descent and I am Caucasian.
  No. It is the same everywhere and ethnicity is not a factor. The information (even from specialists!) is simply not correct.

- ...it is proven that gluten makes it worse for people with Dermatitis herpetiformis, Eczema, Psoriasis. So I guess it would also make it bad for people with PC don't you think?

Response from Frances Smith,
Chief Scientific Officer PC Project
“Thank you for sending us these links. As Mary mentioned, psoriasis and eczema are very different disorders from PC. As stated in the links you sent a gluten free diet may help some people with psoriasis/eczema by reducing skin inflammation but probably only in cases where gluten is a problem food and if they have some of the known symptoms of gluten allergy such as digestive issues. It's unlikely to help with PC unless you have a gluten allergy as well and then it might help with any inflammation but not with callus formation or pain.” NOTE: The links imply it is proven helpful, but the actual research articles do not say that.

EXECUTIVE DIRECTOR

PC PROJECT

Mary Schwartz will be retiring and is working with the PC Board on an active search to find the new Executive Director who will lead PC Project forward.

Here is a link to the job posting which includes information on how to apply. http://jobs.nonprofitjobmarket.org/jobseeker/job/29395705/

Please share with anyone interested. Relocation to Salt Lake City is optional if the applicant has the necessary skills to work remotely.
Hi! My name is Roseann McGrath. I’m 50 years old and reside in a suburb outside of Philadelphia, PA with my husband, 2 beautiful step daughters, our 7-year old golden retriever and our newest family member our grandson who is 6 weeks old.

Pachyonychia Congenita (PC) is a condition I have. Sure, it’s part of my life but I won’t let it define me. So when choosing a career path, since People are my Passion, the field of Human Resources was an obvious choice. HR has been my profession for 30 years, the last 21+ of which I’ve served as the Director of HR for the 9th largest municipality in our state. With PC you know that becoming a professional athlete is out of the question, but not much else is! It has been my personal decision to never let PC get in the way of my desire to make a difference and effectuate change.

It’s all about perspective! Instead of focusing on what I couldn’t do, it was my mission to focus on what I could! Since I couldn’t play sports in high school, I worked from the age of 16 a minimum of 20 hours per week. Those 5 years of solid work experience helped me secure my first job in the spring of my senior year of college. It was hard work, a positive attitude, and determination that led to opportunities and promotions over the 8 years. Each step forward prepared and qualified me for a remarkable accomplishment of achieving my current senior level director’s position at age 29. I say this not to praise myself, but rather to inspire you to not allow PC to interfere or limit you, but rather for PC to motivate you!

My position as a HR Director is a highly responsible senior/executive leadership position where I am accountable for 700+ employees and oversee my own HR team. We are committed to partnering with all employees to ensure a safe, secure, comfortable work environment of excellence, quality performance and legal compliance. I handle all the hiring, firing, union negotiations, employee benefits, training, payroll/compensation, pensions, all policies & procedures and key programs of Workplace Violence, Substance Abuse, Harassment/Discrimination, Equal Opportunity, Safety, Wellness, American with Disabilities & Family & Medical leave Acts. It is my job to inform all employees of their rights, expectations and obligations in accordance with Federal and State laws.

PC has never hindered me from what I want to do, who I want to be or what I want to accomplish professionally or personally. With that said I might have had to channel my energies in more sedentary directions, but my career is extremely challenging and one I would highly recommend. In fact, who better to be the head of HR but a woman with a disability? And who herself occasionally needs time off through the FMLA. My PC challenges have made me a leader who understands pain and suffering so I can feel compassion for our workforce, and because I’m a fighter I can be the tough cookie I need to be at times. As I prepared these remarks, I read what was written about me on our Township website and literature. One line stood out. It said, “Perhaps Roseann is best known for her caring and compassionate leadership of the Township’s most valued resource; its people.” It occurred to me that my philosophy of life is what pervaded my career. If God gives you lemons, make lemonade!

In closing, yes PC is something we have to deal with, but don’t let it limit you - let it guide you to a fulfilling career path, and to pursue your dreams. I know I’m living mine. My job led me to meet my wonderful husband who is also the Superintendent of Police. Now that’s a job with lifelong benefits!
I want to spend on my feet? This is very important to consider. What are you willing to do in order to have a career/job that allows you to be off your feet more?

PC Project wants to help in these important areas. We think you can help one another and we ask for your comments and feedback — whether you have the exact career that you love or wish to make some big changes. What tips and suggestions do you have about work/jobs/careers and PC? Please let us hear your story!

10 Questions To Ask Yourself When You Don’t Know What’s Next In Your Career

1. What do others see as three of my strengths? Try this right now – go to your Facebook, or hop on email, and ask your friends and family to describe your top three strengths. Once you’ve gathered this information, look and see if there is a trend or pattern in the strengths people see in you. Once you weed out the qualities that people jokingly, does a majority say you are compassionate? Talkative? Organized? Finding out how others see you and what they see as your strong points can help show you what path to follow.

2. What do I enjoy doing for other people? After you’ve looked outward for information, it’s time to look inward. What is it that you willingly like to do for others? Is it plan parties? Is it organize their office for them? Is it cooking them a meal when they are sick? Figuring out how you instinctively help others is a reflection of what you truly enjoy doing.

3. What am I drawn to reading? What magazines do you subscribe to? What non-fiction books draw you in? What blogs do you enjoy reading? Is there an overall theme to these? Do you realize you are subscribing and reading blogs about interior design? Or are you fascinated by books that talk about traveling the world?

4. Who do I look up to and why? Maybe you don’t have a personal hero, but chances are you have someone you admire because of what they do and how they do it. What does that person do, and what specifically draws you to their field? Is it that they’re diligent and you admire their work ethic? Or do you like that they’re building something from scratch? Look for career characteristics you want to emulate.

5. What tasks make me feel the happiest? Is there something you enjoy doing on the weekends? Something that makes you wake up with a smile when you realize you get to do it? Is there something that gets your blood pumping and makes your eyes light up? Even if it’s something like video games, that’s legit. It just has to make you feel alive.

6. What do I know I do NOT want to do? As much as you are trying to figure out what you DO love to do, you also have to realize what it is you know you DON’T want to do. I personally hate math, I am not too fond of science, I know I have a black thumb, I don’t particularly like being outside for long periods of time, and I don’t like camping and hiking and climbing, etc. etc. Make a list of things you know you don’t ever want to do to ensure you know what direction you definitely should avoid.

7. What do you want to be remembered for? I know it’s kind of morbid to think about how you want to be remembered at the end of your life, but it is important to consider. If you could fast-forward to that point – what is it that you want people to say? What do you want people to remember you for doing? If your passion doesn’t feel like it’s applicable to this one, that’s fine. Many people would prefer to be remembered for things other than their career and there’s nothing wrong with that.

But this question will help you realize what your core values are. And that is something that you should know and learn from, no matter what field you are in.

8. When you were 10, what was your dream job? You may have to ask relatives or childhood friends for this one if you can’t remember. This method isn’t definite on where you might want to be now – but it can be indicative of where your interests are.

9. If money wasn’t a consideration, what would you want to do? This one is a kind of tongue in cheek, since we all know that money is a consideration and we can’t afford to pretend otherwise. We all have bills to pay, and many of us have a hefty student loan debt. But for just a few moments, it’s worth it to stop and wonder what our goals would be if money wasn’t a primary factor.

10. What do I see as the most important career objective? What’s the first thing that comes into your head? Is it money? Recognition? Is it passion? Or how your job will help/effect others? There is no right answer here. The goal is simply to figure out what’s going to motivate you to go to work everyday, and help you find more of a purpose in your professional life.

From: The Color Coded Life by Amanda Oliver, Aug 2015
held by those with PC. Here are just a few jobs we know PCers have:

- Artist
- Attorney
- Auto Mechanic
- Beautician
- Business Owner
- Computer Programmer
- Construction Worker
- Investment Broker
- IT Specialist
- Laborer
- Marketing and Advertising
- Minister
- Nurse or Nurse Practitioner
- Non-profit Administrator
- Physician
- Podiatrist
- School teacher
- Scientist
- Truck Driver
- University Professor
- Website Developer

Please complete a quick survey at www.surveymonkey.com/r/PCWork and tell us about the work you do. Please also answer these questions or include your comments about PC and employment:

- How does your work affect your PC?
- Would you recommend your career for others with PC?
- If you could, would you change your work to something else?
- What suggestions do you have for our PC kids (under age 18) about their future job choice?
- Would a letter from PC Project to your employer be helpful?
- Would job counseling from PC Project be helpful?

PC COMMUNITY—
ALL ABOUT SHOES

Shoes are the most important clothing choice for girls and women with PC. Recently we gathered comments from several PCers from different countries about shoes they wear.

(1) A MOTHER OF AN 8 YEAR OLD DAUGHTER WITH PC

This one is hard for me to answer because I don't have PC. I encourage my daughter to wear tennis shoes and cushioned shoes as much as possible. She does like to wear sandals and other shoes that I don't think are supportive. I have decided I have to let her decide. They are her feet and if she chooses to wear shoes that hurt she might not choose those shoes again. (I look at it as a life lesson for her.)

We have gotten some sandals and dress shoe hand-me-downs from cousins that I would never choose for her, but she likes some of the styles. She will wear them to church at times but I try to convince her not to wear them to school all day. If she insists on wearing shoes that I think will bother her feet I try to send along another pair of shoes to change into if needed.

She had a pair of Ugg type boots during the winter that she loved. I thought they would be too hot and not supportive enough but they were her favorite shoes. She wore them out. I think she liked the sheepskin cushioning inside and she liked the style.

Anyway, I've found I can't always predict what feels best for her. I just try to provide good shoes. We just bought her a pair of Sketchers tennis shoes with the memory foam inside and she really likes them and says they are comfortable.

(2) FROM A YOUNG ADULT PCer

I have attached pictures of the shoes I wear regularly (next page). I find skater-style trainers the most comfortable for day to day wear (here are two pairs of 'Vans' brand, 'DCs' also have worked well for me.) If I want to dress up a little more, I find a pair of boots or brogues work well for me.

No matter what shoes I wear, I have to be able to fit a pair of 'shock blockers' insoles in them. These are the most comfortable insoles that I have come across, and once they fit well, they make even cheap quality shoes quite comfortable! They're available at oregonaero.com if anyone is inter-
ested in them. Each pair lasts me years with the limited amount of walking I'm able to do.

For me, the most important part of any pair of shoes (once they fit well) is the support or cushioning under the soles of my feet. The next biggest obstacle is trying to keep my feet cool, which is a hurdle I have not yet crossed! I'm not personally comfortable wearing shoes that show my bare feet, even though I'm sure they would help with that problem for me. The picture of wedge-sandals that you sent looks like they would keep the feet nicely cool, which is great, though I know every PC-er has a very different list of what helps and hinders.

I've never had trouble with shoes not giving enough arch support, because the worst pain for me comes from the pressure points around my heels and the balls of my feet.

I know, as a teen especially, it's important to feel comfortable and confident and stylish, so my advice to any young PC-ers is to wear what makes you comfortable, in either physical or a mental way, depending on what way you feel on the day. Some days I'm most comfortable in my worn-out old shoes that fit like a glove, and other days I'm most comfortable when I wear a cute pair of shoes that match my outfit, even if they make my feet sore. Just be sure that any your shoes aren't doing damage, like rubbing at sore spots that could create or worsen calluses, or cause blisters to form. It's no good wearing shoes that look nice if they make your PC worse long term, and the only person that can be sure of that is the person wearing the shoes themselves. Sometimes the shoes you least expect can be the most comfortable, and vice-versa. Try anything out, and you'll learn pretty quickly what's good for you, and what's not worth the trouble.

(3) A PC BUSINESS WOMAN

My "dressy" shoes for work are flats:

My everyday shoes (to and from work, casual days, etc.):

My "dress" shoes (weddings, parties):

My fall/winter shoes for work:

I wear Sketchers "Go Walk" a lot and wear a lot of pants these days to work since heels or wedges are not comfortable for me. I'm K6a, and do not find anything but flats comfortable. I cannot walk barefoot, wear heels, wedges or anything with arches.

(4) A PC MOM WITH PC KIDS

The kind I like the most are good quality, soft leather loafer type shoes. I have them in brown and black. They are breathable, not too heavy, have good support and have a soft padding to walk on. I usually need to wear socks with them. This brand is Naturalizer.

The next photo is a pair of Sketchers. They are canvas shoes. They have a very soft padding on the bottom but also have a lot of support.

Because the sides are breathable, they don't get too hot and they are very light weight.
My boys with PC wear canvas shoes more than any other type of shoes. They like brands like Vans because they are well made and comfortable. I don't know if girls like Vans or not. But the Sketchers shoes are kind of the same idea.

All three of us with PC like shoes that are not hard - not hard leather or hard plastic. The shoes must be able to bend and breathe. I sometimes wear tennis shoes that are running shoes because they are more breathable, but even those make my feet too hot after less than an hour.

UNDERSTANDING PC PROJECT, Frances Smith, PhD, Chief Scientific Officer

What do you know about the 2 major parts of PC Project?
Do you know where to look to find out more information?

1. The International Pachyonychia Congenita Research Registry (IPCRR)
The IPCRR is central to our success in developing therapies for PC. Patients who enrol in the IPCRR are helping physicians and scientists discover more about PC. By joining the registry you are also connecting, building and strengthening the community to help those now with PC and for future generations. Data from the IPCRR patient registry is available on the 'PC Data' tab on the website. This information is changing all the time as new patients register and is regularly updated. Check it out & look for
(i) how many patients are there with confirmed PC mutations?
(ii) how many PC patients are there in your country/state?
(iii) how many have mutations in the same keratin gene - K6a, K6b, K6c, K16 or K17?
(iv) how many different mutations have been found in your gene?
(v) is there anyone else in the IPCRR with the same mutation as yourself?

2. The International Pachyonychia Congenita Consortium (IPCC)
Formed in 2004, the IPCC is now a group of more than 200 scientists and physicians from around the world, who are interested in providing clinical or research services related to PC. Twenty-one members of the IPCC form the Medical and Scientific Advisory board and eight of these are on the IPCC Steering Committee for PC Project. The annual IPCC symposium is a time when at least 50 of the IPCC members meet in person for two days to discuss and present recent discoveries and the progress in developing and delivering effective treatments for PC and related disorders. The Steering Committee meet in monthly webmeetings to discuss specific projects to move PC research forward.

Can you name one or more members of the IPCC Steering Committee?
Check out the ‘about PC Project’ tab on the website to learn more about PC Project.

ADVOCATE TRAINING FOR EUROPEAN PC ADVOCATES
Training for PC Advocates from Europe will be held on Friday, October 28, beginning at 12 noon. Those participating as PC Advocates are:

- Tom Baker-Wales
- Paolo Davide Cognetti-Italy
- Kieren Eyles-England
- Philip David Gard-England
- Christoph Hemmesmann-Germany
- Melanie Hettler-Germany
- Katri-Anna Lehto-Finland
- Sylvie Potier-France
- Srinivasan Ramamurthy-India
- Siddharth Srinivasan-India
- Pamela Ibáñez Triguero-Spain

If you live in a European country and you’d like to apply to serve as a PC Advocate in your country, please send an email to Mary.Schwartz@pachyonychia.org.

There are no specific duties for PC Advocates. They serve as volunteers who have some training to be able to represent PC Project at meetings, with other patients and in other capacities.

Our USA PC Advocates continue to train and serve in various ways to help PC Project. Two PC Advocates serve on the PC Board.

We plan to add Canadian, Central and South American and Asian PC Advocates in the future as this program evolves.
2016 Edinburgh Scotland—PC Patient Support Meeting
October 28-30, 2016
Best Western Edinburgh Capital Hotel
Friday, October 28th - welcome dinner starts at 5:30 pm
Saturday, October 29th - all day presentations, discussions and meals
Sunday, October 30th - half day meeting with the closing luncheon at 12:30 pm

SCHOLARSHIPS FOR TRAVEL AND HOTEL—DEADLINE EXTENDED TO AUG 15
Apply for scholarship at www.surveymonkey.com/s/PSMScholarship
Be sure to register for the meeting and book your hotel reservations first. Apply for scholarships as needed to cover meeting fees, travel, hotel costs.

REGISTER FOR MEETING at www.surveymonkey.com/r/2016PSM
Please register as soon as possible. There is no cost to register. Fees are paid separately and reduced fees apply until September 1, 2016. If you plan to attend the meeting, please register now to help us plan the event.

MEETING FEES
Before September 1, 2016: £50 per person 15 years or over
After September 1, 2016, the fee increases to £60 per person 15 years or over
Fees paid on arrival at the meeting increase to £75 per person
Meeting fees can now be paid through www.justgiving.com/pcproject-europe—Please note for PSM.
Meeting fees are waived for PC patient and one family member attending a PC Patient Support Meeting for the first time and are also waived for all those under 15 years of age. PC Project and our sponsors pay 70% of the meeting costs which include the meeting room and equipment rental fees and food including Friday dinner, Saturday breakfast, lunch and dinner, and Sunday breakfast and lunch as well as meeting break snacks.

REGISTER FOR HOTEL by email
at reservations@edinburghcapitalhotel.co.uk
or by phone 0131 5359988.
Please book now. You are not charged until you check in at the hotel.
There is no cancellation fee if you cancel 24 hours ahead of arrival.
Be sure to register direct with the conference hotel.
£80 for two persons per night (double or twin beds)
£70 single person per night
Those under 15 are charged only £5; those 15 or over are full rate.
Be sure to confirm these rates with the hotel and note that you are attending the PC Project/University of Dundee Meeting. The hotel has generously given us the same conference facility and rates as in 2012 and 2014. If you are not a hotel guest, you will be charged a 'delegate' fee to attend the meeting. Do not use online or other booking services.

There are currently 24 PCers plus 15 family members registered for the meeting.

If you need assistance in completing the forms or if you have any questions, please email
PC Project at info@pachyonychia.org.
PC PROJECT APPOINTS NEW EXECUTIVE DIRECTOR

Staff members at PC Project (Mary Schwartz, Frances Smith and Holly Evans) are thrilled to introduce the new Executive Director for PC Project to our IPCC members.

Mary says “I am certain the goal we have all shared over the last years — to develop and deliver effective treatments for PC patients — will move forward and be achieved under Cindy’s leadership.”

Cindy Byers Atha is a Healthcare Executive with a 26-year history of commercial experience in a series of increasingly responsible sales and marketing roles at Atossa Genetics, Depomed, Inc., and Amylin Pharmaceuticals, innovative biotech/biopharmaceutical companies, as well as AstraZeneca, one of the world’s leading pharmaceutical companies. She most recently was the Vice President of Sales and Marketing at Atossa Genetics where she led commercialization efforts for pharmacogenomic testing and medical devices for Breast Health. Previously, she was Vice President of Managed Markets and Trade with Depomed overseeing Managed Care Sales and Marketing, Pricing, Contracting, Trade, and Distribution.

While at Depomed, her team was recognized by Healthcare Distribution Management Association (HDMA) as 2014 DIANA Winner for Best Manufacturer and while performing a similar role at Amylin, her team was twice recognized for “Best Marketing Programs and People” for a small manufacturer and achieved the 2012 DIANA Award for best new product introduction.

Ms. Atha began her pharmaceutical career in field sales with Merck & Co. and then moved to AstraZeneca where she advanced into Senior Sales Leadership. She was the recipient of more than 16 sales awards for exceptional individual and group performance and AstraZeneca’s highly prestigious “Leadership Excellence Award” and “Global Challenge Award.” She helped bring over 16 new pharmaceutical products to market. Early in her career, she was recognized by several key healthcare clients as the best account representative in the industry. She gained formative experience as an Account Executive with Scientific Technologies, promoting the sale of research and medical equipment to leading biotechnology companies and medical centers.

Ms. Atha also serves on the Board of Directors for Operation of Hope, a non-profit organization that provides facial reconstructive surgeries to poor children around the world. She is a member of the Academy of Managed Care Pharmacy and a volunteer mentor for BoomStartup.

She received her Bachelor of Science in Zoology from North Carolina State University in Raleigh, North Carolina. She and her husband William reside in Salt Lake City, UT.

WELCOME CINDY!
BE A PC ADVOCATE
We need every PCer and every PC family member to be advocates for PC and PC Project. An advocate is one who supports a cause to help others.

To further help patients and family members be effective in this role, we have developed a training program of several webmeetings and one in-person training session.

Seven individuals in the USA have completed the training and are eager to serve in whatever ways their circumstances permit. Another ten individuals in Europe are now joining the training and will participate at the upcoming Patient Support Meeting and we’ll introduce them in a future NewsBrief.

Our PC Advocate training will continue in the months ahead as we hope to expand awareness, understanding, research and funding for Pachyonychia Congenita through PC Project. The next training will be for PC Advocates in Canada and USA.

We want to be sure that each individual in our PC community thinks “I Am PC Project” in my own home, on my own street, in my own city, or in my own country. We want to engage and empower everyone with PC to be able to speak up and speak out about PC. We want to help them have the information and tools that will enable them to be effective in this role.

To achieve the goals we have, to find an effective treatment for everyone with PC — we need everyone to do what they can where they are.

Those who are able to have the extra training and be designated “PC Advocates” are here to help as others work to spread PC awareness.

INTRODUCING PC ADVOCATES AND OUR NEW “CONTACT AN ADVOCATE PROGRAM”
If you’d like to ask a patient a question or be in touch with a PC Advocate in the USA, please send an email to Advocates@pachyonychia.org

You can address your message to a particular advocate or we will share your email with one or more advocates for you. The advocates will respond directly to you. Please understand, they are patients or family (not medical professionals), they have very busy lives and want to help and support PC Project and all affected by PC. As we introduce this option to connect you, we hope you will understand this is a new effort and help us succeed in this effort and give us feedback.

In the future, we’ll add advocates who are fluent in languages other than English. The Advocates are writing articles for the NewsBrief and we’ll include bio information with those articles.

NOTE: One K17 advocate has completed a portion of the training.
UNDERSTANDING
PACHYONYCHIA CONGENITA
Frances Smith, Chief Scientific Officer, PC Project

Genotype and Phenotype - very similar sounding words but they mean different things:

Genotype - the genetic makeup of an organism, i.e. the information within your genes. The PC genotype is determined by mutations in any one of 5 keratin genes, KRT6A, KRT6B, KRT6C, KRT16 or KRT17.

Phenotype - is what you see - the physical characteristics of an organism determined by the genetic makeup and environmental influences. In PC, the phenotype - what you see - is thick nails, calluses, blisters, oral leukokeratosis, cysts and follicular hyperkeratosis.

The collection of detailed clinical information together with the genetic testing results from those enrolled in the IPCRR allows us to look for genotype/phenotype correlations within PC. See PC website 'PC Data tab' for summaries of genotype/phenotype observations. There are differences in the PC phenotype depending on the PC genotype. For example, what you see in PC-K6a is different from PC-K17. Oral leukokeratosis is often present in infants with PC-K6a whereas it is rarely present in PC-K17. As the number of individuals within the IPCRR continues to grow, we can narrow things down further and look for correlations between different mutations within the same gene. In PC-K16 we now know there can be a major difference in the nail findings based on the specific mutation with some having few nails affected.

TOPICAL SIROLIMUS UPDATE

We are pleased and excited to officially announce that topical sirolimus (TD201) for PC patients will be advancing towards its next human clinical study. TransDerm has recently partnered with Palvella Therapeutics, a Philadelphia-based company focused on relentlessly and selflessly serving individuals suffering from rare diseases (Palvella, in Finnish, means “to serve”), to rapidly advance the program. Palvella's core competencies involve developing and commercializing novel rare disease therapies in the US, Europe, and other geographies. Many of you will have the opportunity to hear more about Palvella and meet their leadership team in the months to come.

Consistent with the goals of any initial study of an experimental therapy in a rare disease, the Phase Ib study of topical sirolimus elucidated several key learning points about the potential for this therapy in PC. The study met its primary objective of demonstrating the safety of topical administration of sirolimus cream for the treatment of painful plantar keratoderma in PC. Topical sirolimus demonstrated an excellent safety profile: there were few local and systemic adverse events, no serious adverse events, and overall the topical sirolimus cream was well tolerated.

The path forward for topical sirolimus in PC will now involve the Palvella and TransDerm teams working closely together to build upon the novel formulation developed at TransDerm and explore the potential to optimize that formulation for the benefit of PC patients. Once that analysis is complete and the FDA has been properly engaged on the status and plans of the program, topical sirolimus will be poised to enter its next clinical study, which will more thoroughly evaluate therapeutic efficacy.

Thanks to all of you for your engagement over many years to advance this promising therapy for PC patients. Success going forward will again necessitate the same extraordinary level of collaboration, and we will again aim to harness the collective wisdom and expertise to design and enroll the next clinical study.

Wesley Kaupinen
Palvella Therapeutics

Roger Kaspar,
TransDerm
SHOES AND PC
A. Bravo. DPM
Bio: Dr. Bravo (PC-K6b) graduated from Temple University School of Podiatric Medicine. He began private practice in 1986. He is a PC Advocate (see page 2).

Finding the right shoe gear for a PC patient can be a frustrating and challenging endeavor. Due to the foot pain inherent to PC patients, finding the right shoe gear can make a dramatic difference in one’s comfort level. There are many factors to consider in selecting shoes.

The first and foremost factor is to get a proper fit in the shoe. Though quite basic and seemingly obvious, this at times can be overlooked. A shoe that is too tight will place excessive pressure on the foot and lead to early blistering and subsequent pain. However, a shoe that is too loose allows for excessive movement and subsequent friction within the shoe can also prove to be quite uncomfortable. This at times can be difficult to discern, as the tendency seems to be for people to choose shoes with a little “extra” room, however this can be counterproductive. A general rule of thumb is to allow just under a fingers breadth of room between the ends of the toes and the end of the shoe. An easy way to check this is to have someone trace the outline of the foot on a piece of paper when full weight is being placed on the foot, and pull the insole of the shoe out of the shoe and lay this on the tracing of your foot. The outline of your foot should fit the shape of the insole of the shoe.

If the toes need to be squeezed together to fit, this will likely cause problems.

The breathability of a shoe can have a profound impact on comfort level. Many shoes are now available with a mesh upper which dramatically improves breathability. Most running shoes and walking shoes can be found with a breathable mesh. Open sandals can provide obvious ventilation benefits, and there are sandals available which have adequate support and cushioning, and which cover the toes, if one would like to have the thickened nails of PC not be visible. Manufacturers such as Keen, Birkenstock, Clarks, etc., and others, have sandals meet this criteria. Certain shoes, such as Clarks, have a ventilation system built into the sole of the shoe which some people find helpful.

Oftentimes PC patients will understandably try to maximize the cushioning within a shoe. However, this needs to be done judiciously. If the entire structure of the shoe is too soft and cushioning, this will lead to excessive movement of the foot within the shoe, subsequent friction, and increased blistering. An appropriate accommodative insole or orthotic placed within a shoe which offers good support and structure, can be quite helpful. Shoes which have a stiff, rocker-bottom sole on the outside, and an appropriate insole on the inside, can be quite helpful in redistributing weightbearing pressure and peak mechanical forces from the symptomatic lesion sites on the foot. As an example, a good quality hiking boot such as Asolo can be quite helpful in this regard.

A walking shoe/sneaker called the Swiss MBT shoe can offer similar results. Shoes that are quite flimsy, and twist easily in one’s hand rather than have some structure to them, usually are less than optimal choices for patients with PC, due to the friction will develop within the shoes.

Many people with PC find that certain shoes make the feet too hot, significantly increasing their symptoms. This needs to be judged on individual basis, because certain shoes that may be excellent in other regards, may have an insole to which one is sensitive, making them difficult to wear. As an example, I have had Rockport shoes which can be quite helpful, however the foam lining of the shoe causes a great deal of heat buildup, sensitivity, and discomfort. I remove this and substitute my custom foot orthotics and the problem is solved.

Integral with choosing the proper shoe is selecting the proper socks to wear. All cotton or wool socks are generally best avoided, as they tend to accumulate a great deal of moisture and heat within the shoe. Moisture wicking socks, of which there are many brands (Wigwam, Nike, Darn Tough, ThorLo, etc.), generally can be helpful. Wearing nylon stockings within the shoe likely will exacerbate heat buildup and perspiration, and is often best avoided. Barefoot walking can be helpful in shoes which will absorb perspiration and keep the moisture away from the skin. However can be problematic in other shoes. For people who live in cold climates,
winter boots with thick felt liners, such as Sorel, can be helpful as the felt liner will absorb a considerable amount of moisture. If a prescription for Bilateral Foot Orthotics is needed, it can be used in conjunction with the felt liner.

Once a well fitted shoe is selected, attention can then turn to an appropriate insole. Though individual responses may vary, a properly fitted insole or orthotic can often provide dramatic improvement. There are literally hundreds of types of materials available for insoles and orthotics, and the proper material needs to be tailored to the individual patient. This can be a frustrating search, as initial poor results with an insole or orthotic tend to lead the patient to abandon further endeavors in this regard. Consultation with a professional skilled in the selection of insole and orthotic materials, design, and fabrication can be quite beneficial.

Alternating shoes daily and changing shoes halfway through the day, or even several times a day, can be helpful by reducing repetitive pressure to the same spots on the foot.

There are many shoes which fit the above-mentioned criteria, and some of these are listed below:

Hoka: Running shoe which has a good rocker-bottom insole

Pedor, Drew, Dr. Comfort: Have shoes with added depth to accommodate insoles and orthotics

Asolo, Oboz: Have hiking boots with good support and good rocker sole.

Abeo: Have attractive comfort dress shoes which can accommodate an orthotic.

Finn Comfort: Very expensive, however very well made, proportioned to the foot, cushioned, supportive.

Wolky, Birkenstock, Keen, Clarks: Have sandals and shoes with good breathability and support.

One needs to remember that there can be a great deal of variation in how different PC patients respond to the same footwear. However if one keeps in mind basic general guidelines such as adequate fit, ventilation, support, and consideration of various over-the-counter and custom insoles, hopefully some improvement in comfort level can be achieved.

NOTE: We will add these shoe names to the PC Wiki on the website along with other hints from PCers.

FROM THE PC EMAIL BOX

“I had back surgery and they suggest walking several times a day for 20 minute intervals. My feet cannot handle that. They don’t believe me. They say: Feet cannot hurt that bad.” The physical therapist keeps saying the best thing is a lot of walking which I just can’t do. I told the surgeon about PC and asked him to call PC Project. He seemed interested, but never called PC Project. They gave me a prescription for bilateral orthotics. Is there anything PC Project can do to help insurance pay for the cost of the orthotics.”

IN A PERFECT WORLD

Our heart broke when we read this email. We sent a letter of support, but trying to be of help after a surgery is not effective. Here is what we wish had happened and a much better result. Our wish email: “Thank you so much for helping me with my back surgery. Having the dermatologist and pain specialist connect with my surgeon prior to the surgery made all the difference. Also, please tell the physical therapist ‘thank you!’ for the information she shared and for advising a recovery program that worked for someone with PC! Also, I’m glad to report because the medical team understood how important the bilateral orthotics were in this situation, they were able to get the insurance company to agree to partial payment which has really helped.”

QUESTION OF THE MONTH

Have you ever had a prescription for bilateral orthotics?

Yes/No

If yes, were they helpful?

Please explain

Send to: info@pachyonychia.org

MANY THANKS!

Attached is a marvelous collection of responses from the survey in August NewsBrief from 15 PC patients who took time and shared their experiences. We think it is very valuable information! Enjoy!
PERIPHERAL NEUROPATHIC CHANGES IN PACHYONYCHIA CONGENITA

This extremely important article will be published shortly in PAIN which is the number one ranked journal for pain research. It will make a very important difference in being able to have PC recognized not simply as a ‘nail disorder’ (where it has been classified) to a disease with debilitating pain.

This article and the research was made possible through a collaboration of many individuals:

1. The scientist, Michael Polydefkis, PhD (Johns Hopkins), outlined the research needed.
2. Holly Evans, PC Project staff, coordinated travel and other details needed to bring 15 patients (from eight states and two countries) to Salt Lake City for the biopsy collection.
3. The physician, C. David Hansen, MD, donated his time to collect the biopsies.
4. Another scientist, Roger Kaspar, PhD (TransDerm, Inc.), assisted in the collection and prepared the biopsy material in special formulation for shipment to the research lab.
5. Michael Polydefkis and his team at Johns Hopkins conducted the analysis of the biopsy material over many months. They collected ‘normal control’ samples to compare with the PC samples.
6. After nearly two years of effort, the research was prepared for publication. After submission, additional examinations of the patients were needed and Michael Polydefkis travelled to AZ and to NYC to meet with many of the patients who had donated biopsies. With this additional data, notice was received that the article will be published.
7. Without the patients and each of these partners this amazing achievement would not have been possible. THANKS TO ALL!

PC AND ITCH

Another example of patient participation leading to research insights relates to PC and Itch. A few months ago, Thomas Magin (Leipzig University) described a possible mechanism by which itch could be triggered and was interested in analyzing serum samples from PC patients. In response to a request in the April 2016 News-Brief, a number of patients offered to provide samples.

The first PC sample was sent and the researchers found the same mechanism in the PC sample and reported “this research is highly promising.” Dr. Magin requested additional samples and three PC patients were recruited to donate blood samples. Once again, Dr. David Hansen and his staff donated their time and clinic to collect and prepare the blood samples.

MOVING RESEARCH FORWARD

The PC patient community has a direct impact on the research goals and direction of PC Project in developing treatments and cures for PC. Also, the patient community is making great contributions to help researchers gain a better understanding of this rare disease. The above are just two of the exciting and important devel-
Developments that are a result of patient participation. Additional research on PC samples is ongoing in the laboratories of Pierre Coulombe (Johns Hopkins) and Roger Kaspar (TransDerm, Inc.) and other research facilities.

GLOBAL GENES RARE PATIENT ADVOCACY SUMMIT
Cindy Atha recently represented PC Project at the Global Genes 5th Annual Rare Patient Advocacy Summit September 22-24 at the Hyatt Regency Resort in Huntington Beach, CA. More than 800 patients, advocacy leaders, industry professionals, and healthcare philanthropists came together to unite in efforts to eliminate rare disease challenges. Global Genes awarded over 100 scholarships for meeting participation and PC Project was one of the organizations to be awarded a scholarship for participation in the meeting, which carried the theme “Learn. Connect. Inspire.”

The keynote address for the meeting focused on Rare Disease in the 21st Century and the importance of everyone working together around the globe. Many themes reiterated throughout the meeting focused on all stakeholders working together which included industry, patients, academia, and government. This includes partnering, collaborating, leveraging technology and speaking with one voice.

Cindy participated in several workshops that can help progress the goals of PC Project. She attended a social media workshop to gain insight as to how to better use social media to communicate our message. Although PC Project is on social media, we can improve our presence and impact. PC Project will have to decide its best platform for its audience and then pick a purpose. For example, will PC Project use social media to educate or serve as a call to action? Then, we will need to choose partners within social media such as rare disease organizations, manufacturers, and institutions. This expansion of social media reach will evolve over time but will be a key forum for PC Project communications.

Cindy also gained value from other workshops such as grant writing, fund raising, and generating interest in research and development. Many of the concepts from these workshops PC Project is already doing but Cindy also picked up some best practices that we can incorporate into our future plans.

The most impactful part of the meeting for Cindy was the accomplishments of the patient advocates. Many of these advocates told very impactful stories about how their rare diseases had impacted their lives and inspired them to take actions. These patients were truly empowered and were responsible for driving much of the awareness, education, and research pertaining to their respective rare diseases.
AMERICAN ACADEMY OF DERMATOLOGY ADVOCATES (AADA) LEGISLATIVE MEETING
NOTE: Each year the AADA hosts the Coalition of Skin Disease (CSD) members and others from the dermatology community in Washington, DC for training sessions on how to be an effective lobbyist. The AADA also arranges meetings for attendees with members of the US Congress. This year PC Advocate Roseann McGrath and her husband, Michael McGrath represented PC Project. The following is her report of the AADA 2016:

"Thank you for the opportunity to attend and represent us at the CSD and AAD conferences as well as lobby on Capitol Hill Sunday, Monday and Tuesday. Wow! What an experience! The 3+ days were exhilarating and exhausting. I'm embarrassed to say I didn't know how our own government operates. But I certainly received a quick education!

The intimate experience and relationships built with the CSD are beyond measurable. The AADA training was overwhelming, but once into the setting, I believe we did great. The training on Sunday and Monday was intense. We went from 8:30 straight through to 5:30 each day. The key "asks" and "leaves" were healthcare access and treatment, most especially what is happening regarding prescription drugs, the 21st Century Cures Act and maintaining funding for medical research at NIH, NCI, NIAMS & CDC.

At the first two sessions with the Pennsylvania Senators there were eight of us: two dermatologist, two dermatology residents, Mike, myself and two other advocacy patients.

The two sessions with Pennsylvania Representatives were much more intimate with Mike, me and one other patient representative. Mike actually knows and has collaborated with them. I was blessed to be able to tell my story at all four sessions as well as my own unique perspective as I administer a 12 million dollar Health Care budget with 2 - 3 million dollars spent on self-funded prescription drugs. As a Human Resources Director and as a patient I was able to bring life to my own story while discussing in depth the Health Care situation. Although Mike already had a connection with a couple congressmen, we were able to strengthen his connections and build others by being public servants. Again, thank you for the opportunity to attend, represent us and tell our story!"

2nd from left Roseann McGrath, PC Advocate. Right Mike McGrath, Roseann’s husband.
TOP TEN WORDS EVERYONE SHOULD KNOW ABOUT PC

Jack Padovano, PC Advocate

We all have a story. And we know that PC is a part of that story -- the burden of living with a rare disease, the isolation it causes, the pain, appearance, and time it takes for care.

But how many of us really understand what PC is? And why should it matter? The answer is quite simple -- knowledge is power. And who doesn’t want to feel more powerful?

Most fellow PCers and the incredible caregivers we surround ourselves with can become more powerful by understanding the finer points of PC. Why? So they can explain it to a doctor, teacher, friend, son, daughter, sibling, or even a stranger. Sharing knowledge leads to understanding which leads to empathy, and eventually a treatment and cure.

So let’s dig in.

Most of us know the formal definition of PC, but let’s review it again:

Pachyonychia Congenita (PC) is an ultra-rare genetic autosomal dominant skin disorder. PC is caused by a mutation in one of five keratin genes KRT6A, KRT6B, KRT6C, KRT16 or KRT17. Keratin genes are responsible for production of keratins, which are tough, fibrous proteins that form filaments to support skin cells and give them shape and strength. Keratin filaments help cells handle pressure and stretching. With PC, the filaments do not form properly, causing extreme cell fragility. The specific gene and specific mutation involved affects the disease condition for the PC patient. These different types of PC are properly classified as PC-K6a, PC-K6b, PC-K6c, PC-K16 and PC-K17 (related to the affected keratin gene where the mutation is located).

WOW! I know, quite the brain twister. So, let’s break out and define ten words highlighted above that are contained in the definition.

PACHYONYCHIA CONGENITA: pronounced "Pack-e-o- neek-e-uh" "kuhn-jen-i-ta"-- pachy means thick; onychia refers to nails; congenita means something present at birth.

ULTRA-RARE: estimated to impact 2,000 to 10,000 people worldwide.

AUTOSOMAL DOMINANT: one copy of the gene causes the condition (the opposite is recessive, meaning a person must have two copies of a gene to have the condition.)

MUTATION: a permanent change in the DNA sequence (code) of a gene -- a mutation may be disease causing or harmless.

KERATIN: tough, fibrous proteins that form the main structure of hair and nails.

GENES: a part of the DNA that contains “instructions” telling the body how to produce specific proteins -- human beings have about 25,000 genes.

PROTEINS: chains of chemical building blocks called amino acids -- proteins form the basis for most of what the body does, such as digestion, making energy and growing.

CELLS: the basic building block of all living things -- the nucleus of a cell contains DNA.

PC-K6a, PC-K6b, PC-K6c, PC-K16 and PC-K17: Type of PC -- if you don’t know your type, find out by joining the registry! Register right now. Registry link: www.pachyonychia.org

DNA: (deoxyribonucleic acid) substance within a cell that carries a person’s genetic information -- DNA is made up of four similar chemicals (called bases) that are repeated over and over in pairs.

If you’ve read this far I’m proud of you! And if you wish to dig deeper, go to the PC website where you can find an amazing glossy of additional words and definitions related to PC. Glossary link: www.pachyonychia.org

Now, as you share your story, you can add the detail to better educate your listener, transforming both of you into more powerful allies in the fight to find a treatment and eventual cure for PC.

FIRST PC MEETING IN SPAIN

OCTOBER 21-22, 2016

MADRID, SPAIN

We appreciate CIEMAT providing meeting space and the assistance of Fernando Larcher, PhD of CIEMAT in helping with meeting arrangements.

In addition to Dr. Larcher, Roger Kaspar, PhD (TransDerm) Alain Hovnanian (Imagine Institute Paris) and PC Project staff members (Cindy Atha, Holly Evans and...
Frances Smith) will attend the meeting with the patients.

Coffee breaks for the meeting are courtesy of Ciberer and we are grateful for their support.

FOCUS ON PC AND PAIN
PC Project staff continue to encourage research and discussion on understanding and treating PC pain.

The following list of analgesics that have been used in pain and itch was provided by a dermatologist who is a member of the International PC Consortium. It is a relatively complete list with the first few being the most promising in published studies.

1. **4% Amitriptyline and 2% Ketamine in a cream base.**

2. **Doxepin - available as Zonalon cream.** Used mostly for itch. Reference: I don't know of any good studies for pain.

3. **Lidocaine** used as a patch, cream, ointment or gel at 2-5% strength. Has been only marginally helpful in most patients.

Next are interesting potential agents that have not been extensively studied:

4. **Topical Botox** - it does produce decreased pain when injected but used mostly for sweating along with cosmetic uses.

Reference: I don't know of any studies showing percutaneous penetration to the level of the dermis.

5. **Naltrexone 1% cream** - this is often used systemically for opioid reversal but works well systemically for itch, not sure about pain. No information on topical usage.

6. **Butorphanol** - cousin of Naltrexone but little topical information.

7. **Capsaicin** - available as topical cream, very painful with first applications but occasionally used for chronic pain. It blocks the TRPV1 receptor releasing substance P from nerves.

8. **Cannabinoids** - this is topical marijuana and has a lot of anecdotal reports but few studies. Apparently blocks receptors found on cutaneous nerve fibers

9. **Gabapentin** - frequently used orally to modify chronic neuropathic pain. Little information on topical use. I think where the medication is tolerated it might be useful as an oral agent but many patients can't tolerate the adverse effects.

Next are agents available for itching but most seem to be not very effective for itch or pain but are on the market:

10. **Tacrolimus** - like a topical steroid but less effective

11. **Calcipotriol** - Vit D analog used for itching

12. **Pramoxine** - combined with steroids for itch, usually mildly helpful

13. **Tryptase** - found it on a list but I have little information (said to inhibit the protease-activated receptor 2)

14. **Acupuncture** has been used for itch and pain

GIFT AID AVAILABLE FOR DONATIONS TO PC PROJECT EUROPE
For those employed in the UK, your donation can be increased by clicking on the purple JustGiving icon on the donation page on the PC website.

On the JustGiving site, remember to click the “Gift Aid” request in Step 3.

This is so important in helping to raise funds for PC Project. Be sure to claim this extra 25%.
**PC AWARENESS ANYTIME**

With weather being much too hot in June, the Jennings family decided to hold their PC Awareness Event in September.

They had a very successful event sharing information about PC and raising funds.

While June is the month we designate as “PC Awareness Month” it is great to remember that events can be held anytime.

And, sharing information about PC is something that we all need to do as often as we can in our families and communities where we live.

**PATIENT SUPPORT MEETING**

**EDINBURGH, SCOTLAND**

**OCTOBER 28-30, 2016**

This will be the 15th Patient Support Meeting for Pachyonychia Congenita. Attendees will include:

- 34 PCers
- 28 family members
- 15 specialists
- Total 77 attendees

There are 10 PCers who will be attending their first PSM. This is such a special event and we’ll report details in the next PC News-Brief.

**PC ADVOCATES**

Joining the USA PC Advocates, a group of patients from many countries in Europe have participated in four web meeting training sessions. They will join in a final in-person training session just prior to the Edinburgh PSM and will help to lead many of the meeting sessions.

- Tom Baker—Wales
- Paolo Davide Cognetti—Italy*
- Kieren Eyles—England
- Philip David Gard—England
- Melanie Hettler—Germany
- Pamela Ibáñez Triguero—Spain
- Katri-Anna Lehto—Finland
- Soe Mattijssen—The Netherlands
- Julie Peconi—Wales
- Sylvie Potier—France

* a professional musician and composer, Paolo will present a concert for the PSM. DVDs of his music will be available for sale with all proceeds funding PC research.

**PC SURVEY—5 QUESTIONS**

This survey for those in the IPCRR will help as we build knowledge about PC and how it affects patients. We hope all 800 with PC will respond and help in this effort.

https://www.surveymonkey.com/r/PC2016-10

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*A great turnout for the Yard Sale to benefit PC Project held in September 2016. Plan and hold your own PC event when it is most convenient and the weather just right.*
**Spain PSM Report**

Claudia Avella, PC Parent

A mini Patient Support Meeting was held in Madrid on the 21st and 22nd of October. It was the first time that a PSM was organized in Spain. We were met on the first day with a lovely welcome by PC Project staff and experts. We had dinner and afterwards we were up past midnight talking and getting to know each other.

We found out about PC Project’s latest initiatives such as updating the web page and translating materials to different languages. We learned that the IPCRR registry is our best tool for research into possible future treatments and that this is one of the reasons that it’s important to update the registry regularly, so that changes along PCers’ lifespan can be document-

ded. We heard how using social media to spread the PC message is another thing that we can do and that we should do it proudly. In addition, we were able to ask a panel of experts our questions and learn about the latest research advances related to PC.

One PC patient shared her life story with us, starting with her diagnosis during early infancy, and she did it from the heart. I am very grateful to her for doing such a good job in bringing together and mentioning so many of the things that I have felt, thought or wondered about in my (as yet) limited experience with PC. One of her conclusions was that it’s better to try and see things with a little humour and not dwell on the difficulties. I will do my best to carry this out and teach my toddler the same way.

The kids who attended the meeting also enjoyed some great activities during the sessions. I think this was an amazing solution for the families that participated and that both parents and children were more comfortable this way. The children dressed up as scientists and had a good time making toys and customizing their own caps amongst other fun things.
For me, personally, as a mother of a toddler with PC, it was great to get to know others in the same shoes. It was a valuable opportunity to be able to talk to other parents with children with PC and to meet PCers who did their best to describe their symptoms and what things help them so that I can try to understand and manage my own child’s symptoms better.

One of the best things that has happened as a result of this meeting is the creation of a WhatsApp group including the participants at the meeting. This has allowed us to keep in touch with each other since the meeting and share relevant information. Being able to help each other is essential because what makes PC project successful is that we all work together. PC patients, family members and scientists can all contribute and work as a team, supporting each other to achieve our common goals and not less importantly, by “just being there” for each other.

EDINBURGH PSM REPORT

Julie Peconi, PC Parent Advocate

We have just returned from a fantastic 3 days in Edinburgh in Scotland attending the 15th Annual Pachyonychia Congenita Patient Support Meeting (PSM). The conference was fittingly called PC: The Power of You!

Before the official meeting started, on Friday afternoon there was a training session for new PC advocates. These 10 individuals, from all over Europe and the UK, have volunteered to take a more recognised role in representing the PC Project and in teaching others in their communities about PC. (Dec NewsBrief will introduce these new advocates.) In this training, the PC Advocates were taught how to best explain what PC is, the clinical symptoms of PC and the importance of telling their own PC stories. Many clinicians and physicians were also on hand to help with this training. (We will be starting another group of US Advocates. If you are interested in training and being a PC Advocate, please email info@pachyonychia.org.)

Friday evening was the drinks reception and dinner. For many of us it was the first time that we got to meet Cindy Atha, the new Executive Director of PC Project. And, it was a pleasure! Cindy’s enthusiasm and passion are a perfect fit for PC Project and it was nice to meet her in person. Mary Schwartz was deeply missed and everyone is grateful that she will remain a resource to PC Project Staff. Friday’s dinner was also a fantastic opportunity to see other PCers we hadn’t seen since the last European PSM and to meet many who had come to a PSM for the first time.

On Saturday, there was a full day of presentations. These ranged from understanding PC and the genetic causes of PC to a discussion of how PCers manage and deal with their pain. We also learned of some of the exciting research that is going on as scientists work towards effective treatments to PC. This included research into the role of keratin and also microneedle delivery systems and sweat tests. In the afternoon, Dr. Phil Gard gave a brief overview of Patient Reported Outcome Measures (PROMs). These are typically questionnaires which can help measure how well a treatment is working for a specific patient group. We then split into our mutation groups to discuss and identify possible outcome measures for PC. This is an extremely important task as these outcome measures will be needed to assess the effectiveness of any experimental treatments for PC.
Following this, PCers shared their care tips and there was a panel discussion question and answer session.

Saturday night was a really special evening. After the raffle (which raised an impressive £195!), we were treated to a piano concert by Paolo David Cognetti. His performance was simply amazing and so heartfelt.

On Sunday, there was another great discussion on Patient Reported Outcome Measures as the scientists and clinicians in the room sought feedback from the delegates. We also learned of the next stages in the proposed research. There are so many exciting things going on! This was followed by a presentation on the importance of fundraising for the PC Project and information on how to effectively tell one’s PC story. These are both important topics as they present a chance to tell people of the burden of PC and the burden that a rare disease can bring. The meeting closed with the presentation of the first ever PC Spirit Award which was awarded to Sylvie Potier for her great work in recruiting PC patients to join the International PC Patient Registry. Well done Sylvie!

The PC children and their siblings were treated throughout the meeting to a fabulous kid’s programme. They did all sorts of activities including building with marshmallows, science experiments and volleyball! Holly and Angie, thank you- you did an amazing job organising it!

Throughout the whole meeting the amazing drive, commitment and dedication to finding effective treatments to PC was everywhere! We learned so much from not only the wonderful scientists and physicians that were there, but also fellow PC patients and their friends and family members. As one newly diagnosed delegate said: “This was my first meeting, and although it seemed daunting at first, I’m so...
glad I went. I met some amazing people, learnt a lot, and it has inspired me to share my story and fundraise for the project. It’s a very scary prospect to let everyone in on my secret, but I feel it needs to be done to get knowledge of PC out there. Definitely attending the next meeting!

The overall message of our time in Edinburgh was one of unity and empowerment - each of us has the power to make a difference and with so many of us working toward the same goal, this power is strong! Thank you so much to the PC Project for bringing us all together!

Hope to see you at the next PSM!
RESEARCH THROUGH BIOPSIES

New PC Publication
In 2014, 16 PC patients and 3 'normal control' subjects (husbands of PCers) donated biopsies. Ten of these were sent to Johns Hopkins University (Michael Polydefkis/Mike Caterina) for a Neuroanatomy Study searching for understanding of PC pain. In 2016, Michael Polydefkis conducted follow-up exams on PC Patients to further this research. The findings have now been published in the leading pain journal and are available on the Pachyonychia.org research articles section.

B Pam, K Byrnes, M Schwartz, CD Hansen, CM Campbell, M Krupiczkojc, MJ Caterina, M Polydefkis.
Peripheral neuropathic changes in pachyonychia congenita.

RESEARCH THROUGH BLOOD

PC and Itch Continued
Two more blood samples were donated by PCers at Dr. C. David Hansen’s clinic. The samples have shown promising results. Now, the researchers are planning the steps forward that will require additional samples and information. We may be contacting you for some blood in the future.

COPING WITH PC WHILE WORKING EVERYDAY

Stephen Wittmer, PC Advocate
At first thought, I was going to write an article of instruction for PC patients on how to cope with our rare disease while working every day. Then it occurred to me that all of you already experience the same pain that makes it difficult (to say the least) to get moving after a couple days of work. I am going to tell about my life these past couple of months in the hope that it will encourage others who are in the same boat as I am in. For some reason, if we know that others are out there experiencing the same, it helps us to deal with it better. It being Pachyonychia Congenita, of course.

Back in August, I decided to make a big change and try a new adventure. With several issues pressing me, I wanted to leave for a while. So I decided to try a new profession. As a child, I loved shows like “B.J. and the Bear” and movies like “Convoy” and “Smokey and the Bandit”. Back in those days, even my Grandpa’s Cadillac had a CB radio. So against the wishes of my wife, I signed up for trucking school and nine months of “on the road” experience. I knew, however, that I could pull the rip cord and bail at any time. And, oh how I wanted to do that after the first couple of days! And, oh how I wanted to do that after the first couple of weeks. I thought this would be a good job for my feet because of all of the sitting while you drive, but what I found out was that there are long hours without changing your socks, and miles of walking at truck stops and customer yards. At the Walmart distribution centers, for example, I would have to walk a half mile from my loading dock to the office for paperwork.

A far cry from the pampered world I had created back in Texas, a place where I never had to walk more than a few steps from my car to wherever. Even working on a ranch wasn’t bad with an all-terrain vehicle toting me around. But now I had done it. I had to dig down deep and find another gear. Was I too old for this? Could my feet handle this at my age? I knew that I had pushed my feet much farther in the past. I have always had a difficult time finding the sweet spot of not being on my feet enough (which caused them to be too tender for me) and being on my feet too much. This time, I was going to find a healthy median, but it required some honesty with my co-driver. I swallowed my pride, and I told him what I was dealing with. I said it wasn’t pretty, but since we are going to live 24/7 with each other, he was going to see it. I would have to let my feet air out for a while everyday. I would have to trim my feet in the truck, but I would clean it up. I was born with this problem, and it is not contagious. Three of my five children have it...This was all it took. Being honest with him instead of trying to hide my PC. My whole life, I have had the tendency to hide my disorder. Now, I am being more like my children. They don’t mind wearing flip flops in front of strangers. They don’t cover up their feet when a house guest walks into the room. And everyone loves them!

After three months of the trucking life, I have crisscrossed the country a dozen times, and I have found that there is still more life left in me. I probably won’t be doing this a year from now, but it has
opened new doors and new opportunities for me. I am currently spending a week at home after being gone for the past six weeks, and not only do my feet feel good, but my overall health is better. I have lost weight, and I am full of energy. I am thrilled to know that I still have a lot left in me, and I am looking forward to the years ahead, for me and for PC Project.

**RAISE FUNDS BY SHOPPING**

*Online shopping USA & UK*

Pachyonychia Congenita Project Europe is now listed on Give as you live. A UK site to raise a portion of your online shopping to be donated to PC Project Europe. [www.giveasyoulive.com/join/pcproject-eu](http://www.giveasyoulive.com/join/pcproject-eu)

**iGive.com** is the USA site to raise funds while you shop or search online. [www.iGive.com/PCProject](http://www.iGive.com/PCProject)

Please remember to use these sites when you shop online. It costs you nothing extra and helps PC Project raise funds.

For those not in the USA or UK, if you know of an online shopping site for your country, that we should join, please let us know.

**2017 Utah PSM**

The 2017 Patient Support meeting will be held June 15-17, 2017 in Salt Lake City, Utah. Save the date! Watch for registration and more information next month.

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**QUICK PC SURVEY RESULTS**—We received responses from 72 PCers from October’s NewsBrief survey.

**When was the last time you visited a physician specifically to treat your PC?**

- Never: 7 PCers (10%)
- Within a month: 14 PCers (19%)
- Within a year: 16 PCers (22%)
- 2-5 years: 15 PCers (21%)
- Over 6 years (many were 20+ years): 20 PCers (28%)

**How many times have you seen this physician regarding your PC?**

- N/A - I do not see a physician for my PC, 19 PCers (26.4%)
- 1 time: 20 PCers (27.8%)
- 2 times: 5 PCers (6.9%)
- 3 times: 3 PCers (4.2%)
- 5 or more times: 25 PCers (34.7%)
- 20 PCers (27.8%)

**What specialty was this physician that you visited to help manage your PC?**

- Dermatologist, 38 PCers, 57.6%
- Podiatrist/Chiropodist, 23 PCers, 34.8%
- Primary Care Physician/GP, 16 PCers, 19.7%
- Pain Specialist, 3 PCers, 4.5%
- Nurse Practitioner, 1 PCer, 1.5%
- Emergency Care Physician, 0 PCers, 0%

**Has this physician prescribed any medications to help with your PC pain?**

- Yes: 25 PCers (36.8%)
- No: 43 PCers (63.2%)
**NEW PC STORY**

We love new stories. If you’d like to submit your story please email it to info@pachyonychia.org. An English version of this story will be available on the PC website. Can you find it?

Mi nombre es Diego Avila, tengo 7 años y vivo en Venezuela en una ciudad llamada Valera, con mis padres y mi hermano, Fernando de 10 años. Fui diagnosticado clínicamente por un dermatólogo con Paquioniquia a los 3 meses de edad, nací con dos dientitos y con varias uñitas de mis manos y pies de color oscuro, al mes de nacido comenzaron a cambiar de color y hacia los dos meses a engrosarse, mis dientes se cayeron y el espacio de los dientes permanentes que vendrían a los 5 años fue preservado con prótesis.

En mi país no realizan los exámenes genéticos necesarios para confirmar el diagnóstico de esta condición. Cuando ya había cumplido los 2 años, un día mis papás buscando en internet información supieron de Paquioniquia Project, los contactaron y les enviaron mis fotos y mi historia médica, al recibirla, la organización decidió darnos su apoyo para poder realizarme las pruebas sin ningún costo, mis papás enviaron las muestras solicitadas y mi condición fue confirmada.

Hasta ahora solo presento hiperqueratosis folicular en los codos y algunas infecciones en las uñas de las manos que me producen inflamación y un poco de dolor, mi mamá sabe curar estas infecciones, yo colaboro cuando debe tratármelas y mejoran en pocos días.

Cuando inicio cada año escolar o una actividad extra escolar, mis padres les hablan a todos los instructores o maestros sobre mi condición para que puedan explicar a mis compañeros y sus padres. Me han enseñado a explicar a otros el nombre de mi condición y sus síntomas, aunque me molesta un poco cuando me preguntan muchas veces por ella, he ido aprendiendo que se debe especialmente a la curiosidad por ser una condición muy rara de la que antes no sabían nada, así que respiró y trato de tener paciencia para responder con calma. En algunas ocasiones he sido molestado por otros niños por mi condición, pero mis instructores o mis papás siempre me apoyan y con calma explican a esos niños lo que deben saber para comprender mejor lo que me ocurre.

Soy un niño muy inteligente y muy activo, tengo muy buenas notas en mi colegio y practico varios deportes, ahora mismo hago tenis, natación y fútbol, me gusta ir a la piscina y al cine, me encanta invitar a mis amigos a jugar a mi casa con mis legos, mis juguetes y el play de mi hermano. Para mis amigos, mis uñitas no hacen ninguna diferencia entre nosotros, para mi familia tampoco, en mi casa, mis papás con las consideraciones por mi edad me exigen y tratan igual que a mi hermano mayor.

A veces tengo preguntas sobre lo que me pasa y se las hago a mis papás, quienes siempre me aclaran mis dudas, así repita las preguntas cada tanto tiempo. Cuando mis papás me mostraron esta página, valoré saber que habían otros niños con mi condición porque en mi país aún no he conocido a otras personas que la tengan, cuando me preguntaron si quería que pusiéramos mi historia en PC Project, les dije que sí y les pedí que colocaran que me sentía un niño amado y respetado a pesar de mi condición. Algún día iré visitarlos en uno de sus encuentros anuales para conocer sus historias y darles a conocer más de la mía.
**The PC Mailbox**

**Question:** I have the excess skin on my tongue (called leukokeratosis) but normally it's just on the sides and just a little on top. Sometimes it's even un-noticeable. But every so often it flares up randomly, becomes thicker, almost with hard lumps and it's really tender, sore and hard to eat. Has any research been done on this? Is there something I should avoid? I can't see a pattern for myself, so wondered if others had any insight.

**Answer:** The International PC Research Registry shows that 53% (401/754) of all genetically confirmed PCers have leukokeratosis (K6a has the greatest percentage of PCers who say they have leukokeratosis at 89%, K6b 27%, K6c 18%, K16 36%, K17 25%) and only 16% (66/401) report the leukokeratosis as somewhat painful. The majority of PCers do not have pain with leukokeratosis. PC Project asked a few PCers (see responses below); however, if you have any comments please email info@pachyonychia.org.

**PC-K16** “I do have leukokeratosis but usually it doesn’t cause pain. When it does it’s because of aphas-thas that I develop in moments of my life when I’m really stressed out or debilitated.”

**PC-K6a** “Mine hasn’t been that thick since I was pre-teen. It does get painful when that thick. I find that L-lysine tablets help but need to be taken long term. You can get them at most health food or vitamin stores.”

**PC-K6a** “I have white on my tongue & insides of my mouth, too, but not to this extent and it’s never painful. In my 67 years, it’s always been there & I’ve never done anything to it.”

**PC-K6a** “My leukokeratosis fluctuates, too, but it is rarely painful, and I’ve never been able to identify anything specific that aggravates it. I will say the ‘cleaner’ I eat and the more water I drink (vs soda, etc), the better it seems to be. I also think an antiseptic mouthwash helps. Health professionals have suggested scraping it with a tongue scraper but I’ve never tried it! I’m pretty sure my friend could tell a difference after quitting smoking - for the better. I pray this is some help!!”

**PC-K6a** “Yes, I suffer as explained and my tongue is exactly the same. When it’s extra bad I rinse with luke warm water and smidgen of salt. I notice my tongue is worse when I talk a lot which I think is from my tongue rubbing on my teeth. I absolutely cannot eat anything spicy. It’s literally agony so I stay away from spices even pepper. If I have a cold or am rundown my tongue also flares. I also suffer on my gums the same as my tongue. I use glyoxide. Hope this helps”

**Email:** I just had to share a couple pictures of my amazing Allison. Last weekend we had a birthday party for Erin our oldest. Erin chose a climbing party on the rockwall at the YMCA. Allison made it to the top and had a great time climbing. She doesn’t let PC stop her.

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**Best Gift This Holiday Season**

*SPECIAL ADVERTISMENT*

Roseann McGrath, PC Advocate

Happy Holidays, but not so happy for our poor piggies (feet) and hands. Decorating, baking, entertaining, gift, food and other shopping is enjoyable for non-PCers, but with being so busy it is sheer utter agony for us. Thankfully
modern technology and the internet has hopefully been a saving grace for us PCers. As Janice noted in a recent post, we hate “real stores,” so shopping on-line is the best gift for PCers. And not only does it help our feet and hands but if you shop through an organization that rewards a charity of choice, like Amazon Smile, it’s what I call a “win-win.”

Although we live our lives always trying to hide our hands and feet while fighting the pain, at this hectic season we must be good to ourselves and realize it’s more than okay to use walking aids and wheelchairs. In fact, it is a must! If we don’t limit and help ourselves then we become the bah-humbugs. I don’t know about you, but I get short, snappy and miserable with those I love the most! We want to enjoy the holidays, not make ourselves miserable from overdoing it and using all our energy to fight the pain when we should be holly and jolly. So the best gift ever is to be good to yourself/take care of yourself, which ultimately is being good to those who love you and you love the most. At this beautiful holiday season, I’m sending you the warmest wishes with a heart full of love for a very happy and healthy New Year with a Cup of Cheer to us fabulous PCers.

Let’s put our vanity aside and admit, yes we have a disability, but there are far worse things in life. We have to be grateful for our many blessings, enjoy our family and friends. The best gift we can give ourselves, and all those who love and care about us, is to take care of ourselves. We are not putting ourselves first, but those we love first. Although the pain is constant and chronic, do all you can to not make it crippling.

So the best gift ever is to be good to yourself/take care of yourself, which ultimately is being good to those who love you and you love the most. At this beautiful holiday season, I’m sending you the warmest wishes with a heart full of love for a very happy and healthy New Year with a Cup of Cheer to us fabulous PCers.

**INTERNATIONAL CONFERENCE ON RARE DISEASES & ORPHAN DRUGS (ICORD) 2016**
*Cape Town, South Africa*

Pachyonychia Congenita was represented at the ICORD meeting by Kim Ansley, MD. Dr. Ansley referred a patient for genetic testing through the International PC Research Registry (IPCRR). She presented a poster on Pachyonychia Congenita during the conference. We are so grateful to her and to each of you who join the registry and raise awareness for PC.

**21ST CENTURY CURES ACT**

Earlier this week, President Obama signed into law the 21st Century Cures Act, a game-changing bill for medical innovation. The bill extends the Rare Pediatric Disease Priority Review Voucher Program, which incentivizes the development of new therapies to help more than 15 million children with rare diseases. It also aims to improve the development process of orphan therapies for rare diseases, speeding up the time it takes new medicines to get to patients. The 21st Century Cures Act provides greater focus on patients being more involved in the FDA review process and allows clinical trials to be designed with fewer patients, less money, and easier to achieve goals incorporating more “real world” evidence. The bill earmarks over $4.8 billion over the next 10 years for the National Institutes of Health (NHI) to fund the BRAIN Initiative, the Precision Medicine Initiative, and the Cancer Moonshot. There is also a $1 billion allotment to fight the opioid epidemic and $500 million for the FDA and includes provisions to improve mental healthcare in the US.
TELEMEDICINE
2016 NORD presentation
During the recent Rare Diseases & Orphan Products Breakthrough Summit in Arlington, VA, Dr. David Flannery delivered a very promising presentation on telemedicine and the role that it can play in the diagnosis and treatment of rare diseases. As Medical Director of the American College of Medical Genetics and Genomics, Dr. Flannery has utilized telemedicine to exchange key patient information within the state of Georgia but sees this technology as a promising resource for delivering quality healthcare.

What is telemedicine? Telemedicine is the use of electronic communications to provide clinical health care from a distance. It helps eliminate distance barriers and can improve access to medical services that would often not be consistently available in more remote communities.

Telemedicine can offer several benefits to patients, especially to those that suffer from rare diseases. It can allow access to locally unavailable medical services that may only be available at select medical facilities or institutions. It can also mean less travel for the patients, especially if traveling is difficult or expensive. And, telemedicine can reduce time off work or away from school.

For the physicians, telemedicine can allow multiple physicians and specialists to collaborate pertaining to patient cases. It also allows the physician to maintain the patient in his or her “medical home,” allowing the physician to observe the patient while staying within the home under normal circumstances. It can also ensure that the primary care physician remains involved in the patient care as other specialists are included in the care, thus providing additional training to the primary care physician. The Primary Care Physician will have multi-specialty encounters with physicians that may be better versed and have more experience with a specific disease.

There is also the potential for telemedicine to be incorporated into schools as telehealth clinics. Some of the school systems within the United States are implementing these programs. In these school-based clinics, the child is escorted from class by a school nurse, etc. and then connected to the physician(s) via telemedicine. Then, if the physician orders pharmaceutical prescriptions or locally provided services, he or she can send those for the child via a local provider of such products or services.

Telemedicine is still in its infancy and faces several challenges. Some insurance companies reimburse it dollar for dollar as if the patient is in the room with the physician, but this reimbursement depends on the state. Some states have parity laws and some do not but this reimbursement information is usually available on the internet. Also, within the United States, doctors have to obtain medical licenses in the state where they are treating the patients because medical license boards are responsible for the quality of care delivered in their respective states. So, for telemedicine to be of great value to rare disease patients, the governments will need to enact medical license reforms.

As telemedicine becomes more available, rare disease patients will need to talk with their physicians to see if telemedicine is appropriate for them based on their medical needs. Hopefully, within the near future, geographic factors may not limit access for those suffering from rare diseases.

REMEMBER TO RAISE FUNDS FOR PC BY SHOPPING
UK - Give as you live is a UK site to raise a portion of your online shopping to be donated to PC Project Europe. www.giveasyoulive.com/join/pcproject-eu
USA - iGive.com is the USA site to raise funds while you shop or search online. www.iGive.com/PCProject

2017 Utah PSM
June 15-17, 2017 in Salt Lake City, Utah at the Hotel RL Salt Lake City. Registration information will be posted later this month. We hope you will plan to come!

“It may be daunting to go for the first time, but it’s one of the best experiences you’ll ever have. In no time, you will have made many friends that will last a lifetime and that will possibly understand you better than anyone else can. Everyone has so much to learn about PC, and I feel that it’s when we come together that we can teach each other. When we separate, we no longer feel alone in our pain, no matter where in the world we are. You have to experience it to believe the difference it makes to your life.”

“This is the friendliest, most welcoming group of people who will help you understand and manage your condition. They have it too - you can talk to them and they will listen. They know what it’s like.” - A PC patient