Activity Tracker & PC Pain App Study

The fourth and final phase of the Activity Tracker and PC Pain App Study began December 28, 2016. The 12 PCers and their 12 matched normal controls wear a Withings Activite Pop tracker 24 hours a day and answer two questions every day about their PC Pain: 1) What was the highest plantar pain in the last 24 hours? 2) What was the average plantar pain in the last 24 hours? Each phase lasted at least four weeks during each season of the year. The study began in Feb 2016 and the final phase will finish during the end of Jan 2017. The purpose of this study is to develop validated baseline data for use in future clinical trials designed to reduce pain for PC patients by gathering a record of daily pain and activity levels for patients with Pachyonychia Congenita (PC) and normal controls matched by gender/age/location during the four different seasons of the year. A big thank you to our 24 participants and remember to keep tracking.

When There Is a Will There Is A Way: Time Management Tips

Paolo Cognetti, PC Advocate

Years ago, when I taught piano to kids and they were not ready for the lesson, I was often told “I did not have time to study!” My answer was inevitably, “This is not true, we all have 24 hours — you, me, and anybody else; it is all about how you choose to spend your time!” The statement is of course too sharp but it effectively emphasizes the decision-making importance and is a good antidote to that “self-absolution” attitude we are all inclined to.

If you would like to help and support PC project, but you are unsure that you can actually add one more thing to your busy life, you may find these short tips helpful.

Establish Priorities

As a PCer, it took me several years to go online and fill in the forms for the IPCRR. This happened for various reasons but one of them was most certainly that I had not put it in my priority list with a clear decision. Clarity of vision is essential for achieving your goals.

Manage time

Once you have your well-defined idea it is very important to ask yourself how you spend time. My suggestion is to create a table — all you need is paper and a pencil — with the days of the week and times and start writing down all the activities of your weekly routine. This is not a schedule you have to commit to, just a way to be aware of what exactly you do, how much time it takes and how much time is available. By doing so you can make a better use of your time. Focus on all those activities that can be downsized or removed and accomplished more effectively. For example, I basically thought about this article while driving from my house to the recording studio where I work. It’s just twenty minutes but twice a day for several days in a row makes it a bunch of time!

Set a realistic goal

Time management can not work wonders. It is not enough to have a clear goal and be aware of your weekly routine. You also need to set a goal that you can achieve and a reasonable deadline. Another factor comes into play: not all the parts of the day are the same. For example, after lunch — especially if it is not a light one — you are a little less productive because of the process of digestion. At night, after a long day at work, it might be almost impossible to accomplish tasks that require a lot of thinking.

A realistic goal and deadline should be fully compatible with your life.

Stay motivated

It is easy to begin with enthusiasm, but it is more difficult to maintain your initial commitment. If something goes wrong, try to identify your mistakes and to learn from them so that you will be less likely to make them again in the future. It is very important to keep your energy up and to develop patterns of positive thinking. Don’t panic or worry. Panic will cause loss of concentration, poor performance and will encourage negative thoughts. You will succeed.
GOALS
After the advocate training in Edinburgh, we received a lot of enthusiastic emails from patients with plans, ideas and goals. They were all awesome. We have asked a few of them to write an article.

PAOLO COGNETTI was the first. Wasn’t his article great and something we all need? Paolo had sent an email with a list of things he would like to do first. He said he was taking one step at a time. He is starting with the first one. He set a date to meet with his mom and help her complete the forms. As soon as he finishes step one, he will move on to step two and set another deadline.

1) Have my mother fill in the IPCRR forms.
   Do you have other family members that are not in the registry? Can you help them get enrolled?

2) Share my story
   We love having new stories on the PC Website to share. Email info@pachyonychia.org your story with photos. Also, you can share your story with your family, friends and colleagues.

3) Do and share with other Italians the procedures to get a percent disability and the disabled parking sticker.
   Have you had success getting disability parking or disability in your country or state? Write up how and what you did, send it to info@pachyonychia.org and we can add it to the PC Wiki.

4) I noticed in the location and demographic charts on PC’s website that there are 5 people in Italy genetically confirmed with PC. If possible, I would like to be put in touch with them. There are a few ways to get in touch with others in your country or state. One way is to post on the Facebook PC Patient Chat Group. Another way, after you have done genetic testing, you can email PC Project and give us written permission to release what contact information you’d like to release such as your name and email address. Then tell us who you want it released to e.g. those with PC in a certain country or state and you can even specify only those with a certain PC gene. You can include an introductory message you’d like us to send. We will then email your info and message to those you wanted. Hopefully, someone also wants to be in touch and will respond to your email.

5) Translate the IPCRR in Italian.
   I may need some help from a doctor.
   Let us know if there are certain parts of the website you would like to help translate.

We would love to hear about your goals.

QUEST FOR A TREATMENT
Calling All PCers…

With the help of 100 PCers….for 28 days, one time per day….we are poised to forever change the lives of current and future PCers…

PC Project is VERY excited to report that Mary Schwartz has been recruited to have an influential role in shaping the clinical trial endpoints for a novel treatment designed to help improve function, mobility, and activity for PCers. As part of this, Mary has recently learned from one of the world’s leading rare disease experts that the U.S. Food and Drug Administration (FDA) has strongly encouraged rare disease patient advocacy groups to collect specific information on how a disease impacts patients’ lives.

Here’s the exciting part:
PC Project and PCers have the opportunity to be the FIRST patient group in the world that collects this data in the manner that the FDA has recommended.

WE NEED YOUR HELP:
ENROLL NOW.

1. Click the link below and enter ONE (1) specific way you feel PC impacts your activity, mobility, and function THE MOST. You will track this same activity, mobility, or function over the next 28 days.

   You may choose one of the examples listed or list anything that is your personal choice in the ‘other’ field.

   While we know that overall pain is the problem for those with PC, the purpose of this survey is not to rate pain, but to identify the impact pain has on your life -- what you cannot do because of PC and would do if you did not have PC.

   registry.pachyonychia.org/s3/PROexamples
2. Once #1 has been completed, you will receive an email with a link and instructions to download an ‘app’ to your phone to rate daily how your chosen activity, mobility, or function affects you each day for 28 days.

3. Click once per day to report the impact on this activity, mobility, or function. We will send reminders to help encourage daily reporting.

ENROLL NOW. HELP CURRENT PCERS. HELP FUTURE PCERS. MAKE HISTORY WITH THE FDA....

AND ENCOURAGE 2 OTHER PCERS TO JOIN YOU IN THIS IMPORTANT EFFORT!

If you have any questions, please call 801-401-6300 or email Holly.Evans@pachyonychia.org

MEET THE NEW ADVOCATES
The second advocate training session was held Oct 2016 in Edinburgh, Scotland!

I am Tom Baker. I am a 38-year-old living in Swansea, Wales. I am an active person who loves the outdoors spending time mountain biking, surfing, and passing on this passion to my two boys, Timothy and Solomon. I spend my working day as a patent attorney advising in respect of intellectual property matters and helping protect inventions ranging from brain stimulation equipment to tent pegs. I also have K16 genotype which I have unfortunately passed onto Tim. At least I am able to relate to the difficulties he faces.

The biggest problem I face with suffering from PC is firstly that it hurts, and secondly that as hardly anyone else has ever heard of it, let alone has it, there is little understanding of the pain I put up with. Although I ‘look’ (subjective I know!) normal apart from some ‘hard skin’ on my feet, it is hard for anyone to appreciate what I am feeling in reality. This is sometimes frustrating, but nowhere near as much as not being able to walk or run as I would like to! I find I often get migraines at the weekend – this I attribute to the stress associated with the knowledge that I will need to be on my feet more and trying to plan my day where I will need to be on my feet accordingly. It isn’t helped by wanting to be more active with my two boys and being limited in the things we can do. I hope that someday there will be an effective treatment particularly for my oldest boy onto whom I have passed the condition.

---------------------------

Paolo Cognetti is an Italian composer and a pianist. He holds degrees in piano and composition from the Conservatorio di Musica “Luigi Cherubini” and he is a graduate from Berklee’s inaugural master program in scoring for film, television, and video games at its international campus in Valencia.

Winner of both Italian and international awards, he writes for visual media and theater pieces, and also performs as a solo piano artist.

---------------------------

I am Kieren Eyles and I am a Counselling Psychologist. Can you find his amazing story on the PC Website?

---------------------------

I am Phil Gard and I’m a 63 year old retired General Practitioner from England.

I have always had weird, sore feet, my father and grandfather had it and my son’s got it.

As a child and then a medical student I was told that I had a variant of Epidermolysis Bullosa Simplex.

I was diagnosed with a specific mutation in the K6C keratin gene confirming Pachyonychia Congenita by PC Project when
I was 59. K6c is one of the milder variants of PC. For me, PC has meant and means having rather unpredictably sore feet which are much worse in summer, being restricted in what I can do and wear and knowing that all days are going to be uncomfortable to some degree and some are going to be painful and pretty unpleasant.

I need to ration my walking and standing on a daily basis, knowing that if I overdo it today, I will pay for it tomorrow.

Other PCers have a much harder time, getting about indoors on hands and knees, needing to use wheelchairs, regularly using opiates to control the pain and allow some degree of mobility etc.

PC isn’t an easy condition to explain. What are those hard skin lumps on your feet? Why do they hurt? Everyone knows that hard skin doesn’t hurt. Why do your nails look like that? You ought to get some anti fungal pills from your doctor.

PC is ultra rare, so normally someone with PC has never met anyone else other than a family member.

Most doctors (including this one) have never heard of it, and most dermatologists have never seen it. This makes it a very isolating condition.

Having a diagnosis is psychologically important and makes it much easier to explain the limitations of life with PC. It also allows you to meet other people with PC and to talk to dermatologists that understand what PC is like.

My name is Melanie Hettler, I’m 19 years old and I live near Frankfurt, Gemany. I'm in teacher training with the subjects English and Spanish at the University of Wuerzburg. I am a very musical person, as two of my favourite hobbies are singing and playing the guitar. Also, I'm really interested in languages. I can speak German (of course), English, Spanish, and a little bit of French.

I found PC Project last year and got diagnosed with PC a couple of months ago. My brother Manuel who is 17 years old, and my mother also have PC. I’m happy to finally put a name on something I've been struggling with my whole life.

Hi, I’m Pamela Ibanez Triguero from Spain. I am 34 years old and was born with PC. It was a very hard blow for my parents — there wasn't information like there is now. We only knew it was an ultra-rare disease. Since I was very small, I had to listen to comments from everyone. People would ask if I have fungi or burns. They told me to try this cream, or do this thing. There was daily pain, and having to explain on top of that was difficult.

I cannot wear heels and today I cannot walk. I hate the summer and the heat because my feet really hurt. It is the hardest thing I have had to bear, but over time, I’ve learned to live with it. I am a strong and courageous person and few things can stop me. My baby Hugo has been a great challenge the last 6 years, but the effort has paid off. He is healthy and happy. Many people have worked very hard to reach that goal — geneticists, gynecologists, and PC project — because without their genetic test results, we could not have succeeded.

Thanks!

Thank you also to my dermatologist who knows very well how to treat me. Few things that they have prescribed me have gone wrong. I have seen a better quality of life and my pain and blisters have disappeared considerably! I know I'm very lucky, and for this reason, I want to help fellow PCers. I am already thinking about my next challenge — a cure for everyone!
I am Katri-Anna Lehto. I live in the countryside of SW Finland with my husband, our two children, a dog and two cats. I work as a medical secretary and enjoy swimming and cycling. I am a K16 as are my mother and my 9-year-old daughter. My life changed when I discovered PC Project and got a diagnosis 12 years ago. No more shame or feeling like I’m alone in the world with this - instead, I’ve received a lot of help from PC Project and other patients, and - finally - got access to benefits like a parking badge and a wheelchair.

My name is Soe Mattijssen. I’m 22 years old and live in the Netherlands. I’m still living with my parents but have my own apartment in their home. I study Nutrition and Health at the University nearby. I have a different study program than the other students, because of the pain and low concentration level that I have. This is caused by Pachyonychia Congenita (type K16). The pain it causes, also makes that I’m sitting in a wheelchair, to get the pressure off my feet. Like the soles of my feet, the palms of my hands also contain blisters and calluses which are painful, especially when the calluses crack. My nails are not only growing forward but also growing up, so when I don’t file them, they become very thick. They are very sensitive and get infected quite easily, because they are porous. Next to my study, I like to do sports and hang out with friends.

I am Juli Peconi. I am a Canadian who has married an Englishman (Tom Baker) and ended up in Swansea, Wales. I love spending time crafting, even on occasion with my 2 boys, Timothy and Solomon. I work part time in the field of clinical research in Swansea University and for Skin Care Cymru, a charity giving a voice to those with skin condition in Wales. I volunteer with PC Project.

I am Sylvie Potier and I am 46 years old. I worked for 21 years in insurance and for the last 5 years I no longer work – I’m on disability. I got PC by spontaneous mutation.

The most difficult period for me was a teenager. I have a lot physically and mentally suffered from PC during this period. But I have parents and a brother who understand me and who were still there!

I have lived alone for 2 years with my son François (18), who also has PC. We support each other and we help each other every day. My goal is to succeed as a mom with experience with PC and allow François to flourish despite the pain and the difficulties created by PC. I think I succeeded. François lives well with the PC, never complains, and is a warrior. He was 4 times the champion of France archery in schools. He is my pride and my energy.

I created the French association Le Coeur Au Pied in 2006 to allow all PCers to know the PC Project, give them hope for a treatment, and not be isolated.

If you would like to ask any questions of our now 18 advocates, please address your email to advocates@pachyonychia.org
THE PC MAILBOX
“I wanted a way to support PC Project in a small but meaningful way. So I started "Cans for a Cause". I placed a large box outside the entrance to the kitchen at my job and would collect cans/bottles. Once or twice a month I would cash them in and put the money in a pot for PC Project. After almost 2 months I collected 1080 cans and $54.00. Enclosed is a copy of the flyer I made and the money we raised. Happy Holidays! Jessica White”

What a wonderful idea!

Email Question:
Can pain occur even without the blisters?

Answers:
PC-K6a “I don’t know. A few days ago, my feet were hurting immensely when I stood on them. A terrible, burning pain. I actually reached down to look at my calluses to pinpoint the source of the pain because the pain was on my soles and the sides of my feet. My heels are typically a mess - mushy, bloody, with the blood vessel/nerve ending combo that hurts. And yet, as I looked more carefully at my feet where they were hurting so much, the pain was in areas with calluses but with no blisters. I also find the pain can radiate all over my feet. I just had this experience so it’s fresh in my mind. I am only one patient, but to me the pain and it’s source is still sometimes a mystery. When there are calluses or blisters to be popped, the pain is easy to pinpoint. But sometimes my feet hurt and I really don’t know why.”

PC-K16 “This is a difficult question. I rarely have any visible blisters, but I am certainly in pain every day. In my experience, the areas that are free of callouses are usually not sore. On the other hand, if there is a blister or a callous forming underneath, there is certainly pain before anything is visible.”

PC-K16 “We aren’t entirely certain but my husband would say no. This is because if he walks on the arches of his feet where there are no blisters he doesn’t experience pain.”

PC-K6c “I would have thought it would depend on the presence or absence of calluses.”

Have an answer? E-mail us at info@pachyonychia.org

BE SURE TO FOLLOW US ON OUR SOCIAL MEDIA!

facebook.com/pachyonychia
@pachyonychia
pinterest.com/pachyonychia

Thank you for your donation.
PC Project has a new leader!

We are delighted to announce to all our PC patients, PC family members and other supporters, that PC Project has recruited a new leader who will be familiar to many of you. In February 2017, Professor Irwin McLean, the scientist who discovered the first gene mutations that cause PC in the 1990s, was appointed by PC Project’s Board of Trustees as Chief Executive Officer (CEO) and Chief Scientific Officer (CSO). Dr. McLean’s appointment is highly supported by PC Project’s founder – the truly amazing Mary Schwartz, who has been tremendously generous and is much loved by the PC community across the globe. The appointment followed the January departure of the previous CEO, Cindy Atha, who moves on to other pursuits. We thank Cindy for her hard work and efforts while she was CEO and wish her health, happiness and every success in all that the future brings.

Irwin McLean PhD DSc FRS FRSE FMedSci MAE
Professor of Genetic Medicine, University of Dundee
Principal Investigator, Division of Biological Chemistry and Drug Discovery, School of Life Sciences
Scientific Director, Centre for Dermatology and Genetic Medicine
Honorary Consultant Clinical Scientist in Dermatology and Genetics, UK National Health Service
CEO/CSO, PC Project
**Biography of the New CEO/CSO**

Although he has a very Scottish name and works in Dundee, Scotland, Irwin McLean is originally from Northern Ireland and trained at The Queen's University of Belfast where he graduated with an honours degree in Microbiology in 1985. He gained his PhD in Human Genetics from Queen's in 1988, based on his studies of protein abnormalities involved in muscular dystrophies. Following a period of postdoctoral work 1988-1991 in the Department of Medical Genetics, also at Queen's in Belfast, he learned a range of DNA technologies and started to apply them to identify the causes of inherited skin diseases, which were completely unknown at that time. He moved to the University of Dundee in 1992 and there, working in the laboratory of Professor Birgit Lane, he made a number of key discoveries in the field of hereditary skin conditions. This pioneering work included the breakthrough in 1994 of the first mutations in the genes encoding keratins K16 and K17 as the cause of PC. This discovery also predicted that mutations in K6 would likely also cause PC. Indeed, this was proved when the first mutations in K6a were discovered by Dr Paul Bowden at Cardiff University in Wales in 1995. Later, Irwin and his long-time co-worker and friend, Dr Frances Smith, and their research group, discovered that mutations in K6b and K6c also cause PC, thus completing the genetic story of the disorder that we are all too familiar with today.

In 1996, Irwin set up an independent research laboratory at Thomas Jefferson University, Philadelphia. Based on his success there, he relocated his group to the University of Dundee, in 1998 where he was based at its Medical School for ten years. From 2008, his laboratories have been based in the School of Life Sciences, which is the top-rated university research unit for biomedical sciences in the UK.

Following his early work on PC and many other rare genetic skin diseases, in 2005/6 Irwin’s group made a significant and pivotal breakthrough in the understanding of the most common human skin disease – eczema – with the discovery of mutations in the skin barrier gene filaggrin. This work showed that eczema, allergic asthma and many types of allergies, including hay fever, are all linked by the same mechanism, which is the failure of the skin’s barrier function. People with filaggrin mutations (up to 10% of the population) have “leaky” skin that lets foreign material that is invisible to the naked eye, such as allergens and chemicals, into the skin, so firing up the immune system and leading to eczema and other allergies. This discovery completely changed the eczema/asthma/allergy field and so for this work, and also his work on PC and similar skin disorders, Irwin and his group have won many awards in the field of dermatology, genetics and general medical sciences. For example, in 2013, Irwin was elected as a Fellow of the Royal Society (FRS), which is the UK’s national academy of science and the world’s oldest organisation of this type. In 2015, he was also awarded the Royal Society’s Buchanan Medal “for his major contribution to our understanding of the genetic basis of heritable skin diseases” which is only awarded to a single individual once every two years. Irwin has published more than 300 articles in the scientific and medical literature to date, including many in the top genetics and dermatology journals, reporting his lab’s many pioneering discoveries in the field of inherited skin diseases. He is also an elected Fellow of the Academy of Medical Sciences (FMedSci), the Royal Society of Edinburgh (FRSE) and in 2016, he was elected as Member of Academia Europaea (MAE), which is a Europe-wide academy of science.

Irwin has been very heavily involved in PC Project from its outset, and has been on the Medical and Scientific Advisory Board (MSAB) from its inception in 2004, as well as the Steering Group that guides the organization’s research priorities. Because he himself discovered the genes for PC, Irwin has a very strong personal commitment to supporting PC research and PC families. In particular, he is committed to the development of therapies for PC, and has been involved in this undertaking for well over a decade.

Prior to the existence of PC Project, Irwin’s laboratory was one of the few diagnosing the disease using advanced genetics methods. This effort was led by Dr Frances Smith and since the launch of PC project in 2004 she has continued to do all the genetic diagnosis worldwide for PC as well as any other patients who present to PC Project with closely related conditions. This work is critically important for the International PC Research Registry (IPCR), which we need in order to study the natural history of PC and to allow future clinical trials of new medicines to help treat or even cure PC.
Since 2004, Irwin and his group have been running the European PC Patient Support Meetings (PSMs) in Scotland, France and other locations in Europe and he has also attended a number of the North American PSMs and one in Asia. This extensive involvement in the meetings has led to especially close patient contact. In addition, Irwin runs patient meetings for the skin blistering disease epidermolysis bullosa (EBS; caused by mutations in K5 or K14) as well as other skin diseases that are closely related to PC. He is always in search of ways to increase awareness of rare, neglected skin diseases like PC and others across the medical and scientific communities. Considering these disorders as a group helps him to leverage funding for keratin diseases such as PC, EBS and many other rare diseases involving mutations in any of the 54 human keratin genes.

Since he arrived back in Dundee in 1998, Irwin has generated research funding well in excess of $50 million. The ability to fund-raise is particularly important for PC Project’s long-term future and is especially important as we move into clinical trials of new medicines for PC that are in development. Irwin has always been extraordinarily generous with his time, and therefore, it will come as no surprise that he is taking no salary for his post as CEO/CSO of PC Project. He has also agreed not to accept any research funding from PC Project at any time in the future, so that he has no conflict of interest. Because of Irwin’s international reputation in the dermatology field, the University of Dundee has very generously allowed Irwin to work for PC Project from within his academic post and also to allow PC Project’s headquarters to be run from Irwin’s offices at no additional cost.

Irwin McLean’s research group is now pushing hard to develop therapy for PC and related skin conditions. In 2013, Dr Robyn Hickerson moved from TransDerm Inc to Dundee, where she now has her own independent research position and is spearheading the efforts of the McLean/Hickerson labs to get new medicines into patients with PC and closely related conditions. Robyn and Irwin have recently been awarded almost $3 million in new research funding to move their work rapidly towards the clinic. We look forward to sharing more information about this exciting research in future newsletters.

Irwin is already settling into his new post of CEO/CSO of PC Project and starting to make some changes to the organization to strengthen it and ensure its success. Holly Evans will take on the role of Patient Support Officer (for the Americas) and will remain in Salt Lake City. Holly has 10 years of experience in the day-to-day running of PC Project and should be familiar to all the readers of this News Brief. The PC Project office will have a new address and telephone numbers from March 1st 2017 and Holly will remain as the main point of contact for PC Project. The headquarters of the organization will be based at Dundee and Irwin will be supported by his Personal Assistant, Margaret Barton, who is also Patient Support Officer (for Europe). Dr Frances Smith has taken on the role of Chief Geneticist and will be running the genetic diagnostic program for PC Project, which is critically important to allow us to continue building our patient registry and to enroll patients in clinical trials. In addition, Irwin and Margaret have already raised considerable new funding to underpin PC Project’s activities around the world.

We will keep you posted of any further exciting developments as they emerge. Having an internationally renowned scientist leading the organization who has a close personal interest in PC and in helping patients is an incredibly positive move for PC Project. Irwin is in the process of shutting down several other research programs in his laboratory and is giving up other directorships so that he can focus his time on PC Project and therapy for keratin diseases.

Watch this space – exciting things are happening!
PC AWARENESS MONTH
June 2017

June is PC Awareness month and now is the time to plan what you will do this year to raise PC awareness. No matter what you may choose to do, please join with us to celebrate and support those with PC, to spread awareness of PC, and to join forces with other PCers. It doesn't matter what you decide to do, but we hope you say “yes” I will do something to join with every other PCer in some way in June (or another date this year) and make a worldwide “PC connection.” Once you have decided what you are going to do, please let us know about your plans at info@pachyonychia.org. We would also love to share photos and the stories of your events. Below is an announcement of a wonderful awareness event.

PC AWARENESS—WIGGLE SPRING SADDLE SPORTIVE
Laura Harris, PCer

I was inspired to fundraise for the PC Project on my way home from my first patient support meeting last year. Initially I was apprehensive and didn’t know what to expect from the meeting – wishing I could stay in my hotel room instead of going downstairs to register! I am glad I made it downstairs, as it turned out to be a great experience and learning opportunity. I got to meet lots of lovely people and despite being quite shy and not very vocal, I still view it as an important experience and a turning point in my life. I saw the fundraising and good work others had done and I thought that I needed to do something too. We are the masters of our own future – we are the ones who need to be actively spreading the word about PC and doing what we can to help, after all we are only helping ourselves!

I have recently developed an interest in cycling (relatively low impact to the feet), so I thought what better way to raise money than to take part in a sponsored bike ride. On 26th March myself and a few friends will be taking part in a bike sportive – some are cycling 40 miles and some 70 miles. I used the fundraising as my opportunity to be open with everyone in my life and tell them about PC and how it affects me. Opening up via Facebook was one of the hardest things I’ve ever done – my heart was pounding! But the relief I felt afterwards was immense and the support I’ve received has been amazing. Once I’d signed up to the sportive, I started a JustGiving page and linked it to PC Project’s page. I set my initial target as £500 which has been hit and surpassed and I’m hopeful this will increase more before the big day. Training for me started over Christmas. My legs felt like lead and my bike weighed a ton, burning off those festive chocolates was not easy! I’m hoping the day itself will be fun and that we’ll be successful in raising awareness for the PC Project. Please wish my team and I luck!

I would also encourage anyone thinking of fundraising to just do it – what do you have to lose? Setting up my JustGiving page was very simple and it was easy to link it with PC Project’s page. When you first start your page up, it will ask you who you are fundraising for. Just search for Pachyonychia Congenita and it will come up immediately. I am also very happy to help anyone who wants to start fundraising – please just ask me!

If you wish to sponsor me, please find the link to my JustGiving page below and thank you so much in advance for anything donated, large or small.

https://www.justgiving.com/fundraising/Laura-HarrisPCProject

We hope to see your event and ideas here next month!
16th Annual Pachyonychia Congenita Patient Support Meeting, Salt Lake City, Utah USA June 15-17, 2017

SLC International Airport is the closest airport.

1. **REGISTER**: Please register for the meeting at [https://www.surveymonkey.com/r/2017PSM](https://www.surveymonkey.com/r/2017PSM)

2. **BOOK YOUR HOTEL ROOM** at R&L Hotel Downtown online at [https://goo.gl/8LTYe9](https://goo.gl/8LTYe9) or by calling 801-521-7373 and state you are with group code PAC0615G. The special discounted rate is $99/night. Please book hotel rooms at the conference hotel.


   The meeting fee covers Dinner on Thursday, Breakfast, Lunch and Dinner and breaks on Friday, Breakfast, Lunch and break on Saturday, all meeting handouts and programs. This fee is heavily discounted because PC Project is covering more than 90% of the actual cost. The meeting fee is paid by PC Project for first time attendees. Others may apply for scholarship funding as needed.

   - Please pay meeting fees either by credit card at [https://www.givedirect.org/donate/?cid=12278](https://www.givedirect.org/donate/?cid=12278) and note for PSM or by check payable to PC Project.

   - If you wish to arrive at the hotel early (before Thursday) or stay longer (checkout Saturday), please call the hotel and depending upon space the hotel will try to honor the discounted $99/room/night rate.

4. **SCHOLARSHIPS**: If you need assistance for travel, hotel accommodations or meeting fees, please complete the Scholarship Application available at [https://www.surveymonkey.com/r/PSMScholarship](https://www.surveymonkey.com/r/PSMScholarship) Scholarship Applications are due May 1, 2017.

If you cannot complete the meeting registration or scholarship application online, or if you have any questions, please contact PC Project.

### MEETING SCHEDULE

**Thursday, June 15**
- 4-6:00 pm Registration
- 6:30 pm Dinner

**Friday, June 16**
- 8:00 am Breakfast
- 9:00 am Session
- 12:00 pm Lunch
- 1:30 pm Session
- 6:30 pm Dinner

**Saturday, June 17**
- 8:00 am Breakfast
- 9:00 am Session
- 12:00 pm Lunch
- Meeting ends
The last News Brief announced PC Project's new CEO and CSO, Professor Irwin McLean and PC Project's office move. Since then, this summer has been a climb, with ups and downs but always moving forward with lots of amazing things coming about.

One of those amazing discoveries is that because of the International PC Patient Registry (IPCCR), established since 2004, we have found that 20% of patients who were previously diagnosed with PC actually have other keratoderma disorders that can resemble PC. Keratoderma means thickening of the skin and PC is just one of many rare palmoplantar keratoderma (PPK) disorders where skin on the hands and feet is thickened. So, what can we learn from the differences and similarities between PC and other keratoderma disorders? And how should we help these individuals? We know that these individuals have symptoms similar to PC and yet have no support, no help and no one to turn to for advice. During the 2017 Patient Support Meeting held in Salt Lake City, Irwin asked the attending PCers what we should do to help those with similar keratoderma disorders. The PCers agreed that these individuals should be included. We wholeheartedly agreed and want to be an inclusive group for all keratoderma patients. Dr. Edel O'Toole created this slide to test some of the specialists. Can you tell which ones are Pachyonychia Congenita and which are other keratoderma disorders? (Answers on next page)
Which ones are PC? #3, 6, 8, 10, 11, 15 & 17

What are the other ones? 1=DEPPK (diffuse epidermolytic PPK Keratin 9); 2=Tylosis; 4=FAH (focal acral hyperkeratosis); 5=Punctate PPK (AAGAB mutation); 7=DNEPPK (non-epidermolytic); 9=Naxos; 12=DLE (discoid lupus erythematos); 13=EI (Epidermolytic ichthyosis Keratin 1); 14=Limescale remover; 16=Mal de Maleda

2017 INTERNATIONAL PC CONSORTIUM SYMPOSIUM APRIL 25-26

The theme for the annual 2017 International PC Consortium (IPCC) meeting held in Portland, Oregon, was "New Horizons in Keratoderma Research." In the audience, there were many faces of our dear physicians and scientists who have continuously worked on PC over many years. In addition, we welcomed new physicians and scientists who focus on different keratoderma research to join the IPCC meeting to help expand our horizon and reach new heights of research and discovery. The meeting was wonderful and there was a buzz of excitement for the way forward for PC.

2017 PATIENT SUPPORT MEETING EXPERIENCE JUNE 16-18

BY ERIN WEIR, PCER

Despite living with PC for my entire life (although not knowing what it was for most of that), I had never met another person with the condition. That all changed when I stepped into a hotel lobby in Salt Lake City on June 15, 2017, and my life changed forever as I discovered the amazing community of patients, caregivers, physicians, scientists, and others who make up PC Project.

The 2017 Pachyonychia Congenita Patient Support Meeting was held in Salt Lake City, Utah, from Thursday, June 15 to Saturday, June 17, and was attended by numerous patients, caregivers, scientists, physicians, and others involved with PC Project. The presentations and panel discussions covered a vast array of topics relevant to those living with Pachyonychia Congenita, from very exciting news about ongoing research into treatment for PC, and PC pain, to care tips and day-to-day life with PC. The theme of the meeting was PC: The Power of You, and it was an idea that was returned to again and again throughout the three days of presentations, panels, and group discussions, not to mention the informal conversations that took place throughout the meeting.
One of the highlights of the meeting, for myself as well as others I have talked to, was the focus group held on Saturday morning. This intense and often emotional discussion gave PCers the chance to share in great detail the ways that PC impacts their lives. The information gathered and stories shared will help to gain recognition of PC by the FDA, and to lead to future clinical studies. As well, on a personal level, the focus group helped PCers to really realize that they are not alone, and that they are part of this amazing community. Attending the Patient Support Meeting was an incredible experience, and I very much look forward to the next one. I strongly encourage anyone who is thinking about attending to go ahead and do it, you will not regret it!

**BY JEFF CHRISTENSEN, PCER**

My son Oliver (age 2) and I (age 35) both have a K17 mutation. I have had K17 symptoms since I was born, and am a spontaneous mutation. I recently found this out through the genetic testing with PC Project in 2016 and am so glad we did. Knowing what I have, and now what my son has, is a relief in that we know there is support, research, and hopefully someday a cure.

The conference this year was a first for myself and family with our new information about PC. My wife, my son and I were all able to attend. Salt Lake City was a great location, as it was not too far to travel from our home in Boise, Idaho. The time of year was definitely ideal for traveling, for vacation time from work, and eventually working around school schedules. The individual sessions were very informative and provided insight on many different aspects of PC.

The overall atmosphere of the conference was very welcoming and inviting. It was laid back and very well organized. The PC staff did an amazing job pulling it all together. We were impressed with everything from meal services to childcare. I know my wife was a little nervous at first on how the childcare would work, with this being our first conference. And you never know how a toddler will react to a new setting and new people. It gave us both peace of mind knowing our son was close in proximity and having a good time with caring people.

I found that the report on research being done by Irwin and Robyn was very promising and real. It seems that their hearts and minds are 100% dedicated to
finding a cure. It is certainly reassuring to see progress and such an open line of communication from them and everyone involved.

I learned a lot at the conference about the science behind PC. The sessions were very informative and presented in a way that broke down very complicated information into understandable concepts and ideas. Once again we were very impressed. It meant so much that these very busy and talented scientists were spending so much time and resources to help our family and others with PC.

We hope that by learning more and more through these conferences and growing our knowledge that this will help my wife and I better explain and work with our son as he grows up with PC.

Growing up not knowing about PC, not even having something to call it until having my son and being connected with PC Project, it was so impactful to meet others with the same condition. Through breakout sessions we were able to meet other conference attendees with the same mutation, as well as many others with other variations but similar PC symptoms. We talked with others about how they go about their daily lives and what we have done in regards to PC treatment. It was nice to have similar things in common and be able to talk openly about it and have a mutual understanding. It was also nice to hear from others with small children and pick their brains about suggestions for working on toddler feet who can’t sit still. We were surprised with how many families and children were in attendance. It was so nice to talk with those in similar situations to ours.

Connecting with others was one of the most beneficial parts of the conference. Being a part of the Facebook PC Patient chat group is also very helpful and allows us to provide/receive input on numerous PC related issues. It was nice to return home and continue some of the dialogs that were started at the conference.

The future seems bright in regards to research, funding, networking, and ultimately finding a cure. We are blessed to have this support network and look forward to the next conference. Thanks for all of the support and information!
RIDE LONDON 100 for PC Awareness

James Wright decided to do a last minute fundraising event as he completed the RideLondon100 marathon. His wife, Kate Mellor Wright, PCer, completed the 46 mile ride. Their ride raised over £1,400 for PC research. Great job James & Kate! Learn more about his ride at https://www.justgiving.com/fundraising/ridelondon-100pc

Let us know what you have been doing in your own area to raise awareness.

Giving Tuesday

In 2016, PC Project participated in Giving Tuesday and managed to raise an amazing $50,000, which was our target goal. We decided to participate in Giving Tuesday again this year and this will take place on Tuesday, November 29th. We will need your help again and so please watch for the emails and social media posts throughout November. In addition, we would love it if you could create some cool social media posts for this event and would ask that your posts are emailed to us at info@pachyonychia.org no later than October 16th.

PC Reaching Across the World

PC Project was represented at the World Congress of Pediatric Dermatology in Chicago, Illinois, July 6-9, 2017. A few of the dermatologists that stopped by our booth had PC patients who needed help and were excited that there was such an organization as PC Project. We aim to bring awareness across the globe and to reach out to those who feel isolated. It is important to bring hope to those in need of support and it is why we do what we do.

PC Staff Grows

We are delighted to announce that Christabelle (Belle) Goh, PhD has joined the team as the Patient Support Officer (for Asia Pacific) and is based in Singapore.

Belle received her PhD from the University of Dundee under the supervision of Prof. Irwin McLean from 2013-2015, where she learnt and developed useful research skills. For the past few years, she has been working in the Institute of Medical Biology, Singapore, as a therapeutics scientist for diseases of the skin. Starting September 2017, she will be based in Singapore in order to give PC Project a greater reach to the rest of the world. With Belle on board, there will now be around the clock access to PC Project. She is very excited to join the PC Project family!
ENDPOINT STUDY FOR FDA - WE NEED YOUR PARTICIPATION

Your input is needed to gain a better understanding of clinical endpoints and outcome measures for those living with PC. Respond at https://goo.gl/5wWBsh

At the Salt Lake City Patient Support Meeting this past summer, we held a PC Patient Focus Group led by Attorney James Valentine and Dr. Robyn Hickerson. As promised, James and his team assessed the input from PC patients and their caregivers to identify possible outcomes to study in trials for PC. As a result, the team developed a follow-up survey for all PC patients to help us understand which existing outcome measures/endpoints make sense for this PC community. It is intended to get important input which will help ensure future therapy for PC, address the symptoms and burdens of the condition that are most important to patients. The feedback will also help ensure clinical trials for PC capture this information in a way that is useful for decisions to be made by patients and their health care providers, in terms of treatment.

What we need now is for all PC patients in the PC Project registry to complete the survey before Monday, Oct. 30th. This survey is for PC patients only, not caregivers or family members. PC Project will de-identify the survey so that the survey will be anonymous for the researchers. We ask that you please complete the survey at https://goo.gl/5wWBsh on a computer rather than on a mobile device.

PATIENT SUPPORT MEETING
As many of you may know, we alternate the Patient Support Meetings so in odd years they are held in the USA and even years they are held in Europe. The 2018 Patient Support Meeting will be held in Dundee, Scotland in October 2018. The exact dates, registration and additional information will be available early next year.
**PC Awareness Stars**

**Soe Mattijsen's** (a PCer) family raised over $1150 for PC. On September 16, Soe's family held a birthday/anniversary party. Soe's mom turned 55, Soe's dad turned 65 and it was also their 25th wedding anniversary. Instead of gifts they asked for money to donate to PC project.

**Keith & Rhonda Ashworth's** granddaughter, Coraline Jennings has PC. They held a yard sale and raised over $900.

**Robert & Merilyn Bramer** have been making monthly donations to PC Project since January 2010. Here is what they did to make 100% of their donation go to PC Project:

"We discovered recently that donations through third party organizations to PC Project have started keeping a percentage of the donation. We found another way to make sure 100% of the donation goes to the PC Project. Through bill pay from our credit union/bank, we can set up a one time or recurring donations to PC Project. This donation method can also be accomplished through some credit cards. Each banking institution will have it's own method, but direct donations can be done. We are grandparents of a PC child. Supporting the research of the PC Project is very important to us. Please consider the method of your donations to support the PC Project."

**Tom & Timmy Baker** shared their PC story for Jeans for Genes Day at www.walesonline.co.uk/news/wales-news/dad-sons-rare-skin-condition-13650774

PC Project recently received a grant from Jeans for Genes to make a webpage and book for children. We will need PCers and family members help. We would love children's photos and stories. We will send out more information soon by email and in the next newsletter.

**PC Mailbox**

"My dermatologist put me in touch with PC Project. Mine was a mysterious case - might be PC or something else entirely. The people at PC Project are the kindest, most big-hearted people I've ever met. Turns out I don't have PC, but something new, discovered through testing via PC Project! I finally know what I have! Thank you!"