SUMMER CLIMB WITH NEW HORIZONS AND DISCOVERIES

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 (IPCRR), 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!