2015 Patient Support Meeting—Newark, NJ

The meeting was held June 14-16, 2015 at the Marriott Renaissance Airport Hotel. At the meeting, there were 60 PCers plus family members, physicians and scientists for a total of 115 attendees. The meeting theme was working together to make a difference. No single bee can make honey. It takes a hive! This meeting used a hive of PCers participating to make a wonderful meeting.

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

Presentations

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

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

Panels & Discussions

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

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

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

Special Thanks to our Medical & Scientific Experts

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

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

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

OVERAL RATING 4.4 on a scale of 5

<table>
<thead>
<tr>
<th>Rating</th>
<th>Score</th>
</tr>
</thead>
<tbody>
<tr>
<td>Better than other meetings I attended</td>
<td>6/19</td>
</tr>
<tr>
<td>Same as other meetings I attended</td>
<td>11/19</td>
</tr>
<tr>
<td>Not as good as other meetings I attended</td>
<td>2/19</td>
</tr>
</tbody>
</table>

SCHEDULE

<table>
<thead>
<tr>
<th>Rating</th>
<th>Score</th>
</tr>
</thead>
<tbody>
<tr>
<td>Excellent</td>
<td>27/35</td>
</tr>
<tr>
<td>Okay</td>
<td>8/35</td>
</tr>
</tbody>
</table>

PRESENTATIONS

<table>
<thead>
<tr>
<th>Rating</th>
<th>Score</th>
</tr>
</thead>
<tbody>
<tr>
<td>Helpful</td>
<td>34/35</td>
</tr>
<tr>
<td>Not helpful</td>
<td>1/35</td>
</tr>
</tbody>
</table>

PATIENT DISCUSSION

<table>
<thead>
<tr>
<th>Rating</th>
<th>Score</th>
</tr>
</thead>
<tbody>
<tr>
<td>Helpful</td>
<td>30/35</td>
</tr>
<tr>
<td>Not Helpful</td>
<td>5/35</td>
</tr>
</tbody>
</table>

PSM COSTS & FREQUENCY

<table>
<thead>
<tr>
<th>Rating</th>
<th>Score</th>
</tr>
</thead>
<tbody>
<tr>
<td>Continue As Now *</td>
<td>19/35</td>
</tr>
<tr>
<td>Costs paid fully by meeting fees</td>
<td>1/35</td>
</tr>
<tr>
<td>Use Funds Only for research</td>
<td>2/35</td>
</tr>
<tr>
<td>More Often (Every Year in US)</td>
<td>8/35</td>
</tr>
<tr>
<td>Less Often (Every 3-4 years in US)</td>
<td>5/35</td>
</tr>
</tbody>
</table>

WHAT LIKE BEST

<table>
<thead>
<tr>
<th>Likes</th>
<th>Score</th>
</tr>
</thead>
<tbody>
<tr>
<td>Connect with other patients</td>
<td>17/32</td>
</tr>
<tr>
<td>Connect with staff, researchers, doctors</td>
<td>15/32</td>
</tr>
</tbody>
</table>

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

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

Typical Positive Comments

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

Suggestions to improve

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

No one puts pressure on you to show your pc or to talk.
You should definitely come. Everyone was nice and supportive. Everyone is like a big family there.
The atmosphere is supportive and the meeting is educational.
Just do it! You will be welcome and accepted. These people understand like no one else can. A very informative and positive experience.
Please join us. We can get one step closer to finding a cure.

2386 East Heritage Way, Ste B, Salt Lake City, UT 84109 · www.pachyonychia.org · Phone 877-628-7300 · Email: info@pachyonychia.org
**What’s Next?**
At the end of the recent Patient Support Meeting this question was asked. It surprised us because we are not at a ‘stopping’ point. So, in our mind, the question really is ‘What is on-going?’

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

**Continuing Goals for PC**

**IPCCR**—We need to enroll every PCer; numbers do matter!

- **Questionnaire**—to gather accurate data from those with PC
- **Genetic Testing**—provided free so every one with PC can know their own specific gene/mutation and this data can drive research forward
- **Publications**—to introduce PC (an ultra rare disorder) and educate physicians and scientists with accurate information

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

**Clinical Studies**—we have completed two Phase 1b Clinical Trials through the FDA:

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

We have many small studies on-going at all times involving a few patients in each study. Every study teaches us and leads us to additional studies. Since many of these are ‘off label’ studies, the results are not published, but the information is used for each step forward.

3. Statins. 4 patients with mixed results.
5. Topical Pain formulation. Studies in 3 or 4 centers will begin shortly.
6. PC tooth study. Please send any teeth that are pulled or baby teeth that fall out.
7. Imaging, biopsy and tape stripping studies are all on-going.

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

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

**PC Project New Goals**

**Quality Medical Care for Every PC Patient**—with over 2000 rare skin diseases, we cannot expect medical experts to know about PC which is one of the rarest of the rare skin disorders. We must begin an aggressive Educational Outreach program including

- **Continuing Medical Education** (CME credit) for physicians and other medical staff to teach about PC. The costs for this range from $50,000 to $300,000 and up.
- **Grand Rounds** with multiple PC patients teaching doctors. The costs vary with each institution where these are held.
- **PC Natural History** which we will begin to compile. By updating your information in the registry regularly, you will help us create a PC Natural History — without the $million dollars$ otherwise needed.

Please help us build a PC Natural History by updating your IPCRR information. So far 42/690 have updated — that’s a start!

⇒ Adults—once every year
⇒ Age 6-18—twice every year
⇒ Under 6—four times a year
⇒ Under 1—every month

Please help us with this project.

**PC Project Staff**

Another is to add Staff Members to PC Project to achieve all our goals!
PC AWARENESS
June is PC Awareness month and we are beginning to hear about events we would like to share with you. We hope you will take these ideas or come up with others and raise awareness in your local area in June or anytime you can. Please let us know what you do.

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

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

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

Victoria Barbier put donation boxes at local newsagent stands.

Annamarie Baulmer is selling special PC Nail wraps.

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

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

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

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

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

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

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

Andreas Berroth, PhD. PC Fellow at Stanford University