



# Pachyonychia Congenita Project

*Fighting for a cure. Connecting & helping patients. Empowering research.*

## ANNUAL REPORT 2013

### **IPCC and IPCRR**

To move forward to find effective treatments for those suffering from Pachyonychia Congenita, PC Project actively supports two major functions:

1. The International Pachyonychia Congenita Consortium (IPCC)
2. The International Pachyonychia Congenita Research Registry (IPCRR).

**International Pachyonychia Congenita Consortium (IPCC).** The IPCC is a group of physicians and scientists who collaborate in research for PC. Unique because it is organized by PC Project, rather than around a university, specific researcher, company or medical organization. Therefore, the focus is always on moving research forward for PC patients.

An IPCC Newsletter was published 4 times in 2013. On Feb 14-17, 2013 more than 50 physicians and researchers met at the Tenth Annual IPCC Symposium in Park City, UT. The introductory lecture showed all we have learned since 2004 about PC through the participation in the IPCRR of hundreds of PC patients. Pierre Coulombe discussed mouse models showing phenotype (callus/blisters) on paws and how they are being used to develop new treatments. Also dis-



10th Annual International Pachyonychia Congenita Consortium (IPCC) Symposium February 14-17, 2013 Park City, Utah

cussed was how microarray work with patient biopsies is revealing the relationship between various keratins with the hope of finding new approaches for treatment.

Throughout 2013 PC Project hosted monthly IPCC Genetics Team web meetings for members of the IPCC Genetics team to discuss specific cases which need additional diagnosis to direct the genetic testing. PC Project sponsors a grant with the University of Dundee for genetic testing of those diagnosed with PC who participate in the IPCRR (registry).

One goal of the IPCC is to have formal clinical studies for PC. In Oct 2013, the IND filed with the US FDA and the IRB filed with Stanford University were approved for the topical rapamycin clinical trial. The topical formulation has been developed to avoid side effects found with oral rapamycin in an earlier study. It has been developed through an agreement with Pfizer and TransDerm, Inc. The 15-patient trial will be held at Stanford University under the direction of Joyce Teng, MD.



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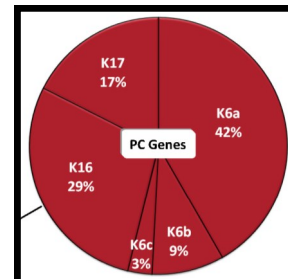
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**International Pachyonychia Congenita Research Registry (IPCRR).** The patient registry is the key to patient services. Through the registry each patient has an opportunity to contribute important data about their personal condition, receive a 30-60 minute telephone consultation with a qualified dermatologist and have free genetic testing completed with a saliva sample using a kit provided by PC Project and sent direct to the laboratory.

The patient growth of PC Project from 2012-2013 is shown on the chart below. Of the total individuals registered with PC Project as of Sept

PC GROWTH	2012	2013
Individuals	1192	1319
Families	644	726

2013, 623 in 371 families are now fully registered with us. This means their questionnaires, photos and physician notes are complete. Of these individuals 507 have been genetically confirmed to have PC (see chart at the right to see the mutation break down).

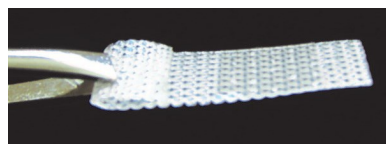


Three PCers attended the Atlantic Derm Conference on Apr 13, 2013 in Washington DC and represented the PC community. About 600 doctors visited with the patients and got to see three different PC mutations. The PCers report that the doctors were very enthusiastic, asked lots of questions, and were impressed with the brochures and organization of PC Project.

The 12th annual PC Patient Support Meeting hosted by our biotech partner, TransDerm was held on Jun 20-22, in Santa Cruz, California. In attendance were 40 PCers and a total of 93 participants. Over \$4,475 in scholarship funding was awarded. As part of the meeting, IRB approval was obtained so that patients could try the micro needles now produced under standards for human trial. Three delivery options were evaluated (1) dissolvable microneedles, (2) the Tri-M laser and (3) a cream.



An initial FDA meeting has been held for the sdTD101. The purpose of the trial is to prove (a) safety (b) effectiveness to stop PC and (c) patient-friendly (pain free) delivery.



The first phase of our cyst study has been completed. Samples from 12 individuals with genetically confirmed PC provided the following interesting results: 1 PC-K16 patient with 1 steatocystoma. 9 PC-K17 patients with 3 steatocystomas and 6 epidermal inclusion cysts. 1 PC-K6a patient with 4 verrucous cysts with isthmic and follicular germinative differentiation. 1 PC-K6a patient with 1 'unremarkable skin'. A prospective study of PC cysts is now being planned.

A new publication "Best Treatment Practices for Pachyonychia Congenita" appeared in the Journal of the European Academy of Dermatology and Venereology in 2013. The article was made possible by participation in an online survey by 125 of the patients registered in the IPCRR.



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PC Project participated in the BYU CAPSTONE mechanical engineering project. The PC Shoe team gave an excellent presentation at the concluding event which featured numerous mechanical engineering and health projects. The orthotic brace available from Toad Medical reduces pressure by **33%**. With an extra two-layer foam cushion, the pressure is reduced by **77%**. The cost of the brace including the orthotics fittings is about \$6,400. We do not know whether insurance covers these costs.

In Jun 2013 PC project launched a new website with many new features. One of the most important is the PC research registry form, online version. There is a life with PC section where PCer's can share their story.

Several web meetings for patients have been held in 2013 with those in the IPCRR. A PC-K17 meeting started to be held monthly.

## Quick Summary—2013 Fundraising

2013 ThickSkin Duathlon	\$27,889	Total <b>\$39,326</b> raised 100% for Patient Support and Research
2013 Awareness Events	\$ 3,644	
2013 Golf Tournament	\$ 7,793	

With the \$2-for-\$1 match, **\$117,978** was raised in 2013 for PC Project services.

**FUNDRAISING.** Events were held to raise funds to support the activities of the IPCC and IPCRR. The second annual PC Project sponsored June Awareness Month. Volunteers around the world held individual events. Some of the events held were: a barefoot challenge, employer match, coffee morning in the UK, family donations, garage sale, co-worker donations at a UK hospital, rummage sale, Beaune challenge.



As part of PC Awareness 2013, the second ThickSkin Duathlon to raise funds for PC research was held at TransDerm in Santa Cruz to end the Patient Support Meeting.

The 3rd Annual PC Charity Golf Tournament was held August 24, 2013 at the beautiful golf course in Midway Utah. The weather wasn't looking promising, but it turned out to be a gorgeous day. Nearly 15 sponsors and 128 golfers supported the event.

**MISCELLANEOUS.** In June 2013, PC Project was recognized at the University of Dundee, Scotland Graduation ceremony with an Honorary Doctorate degree awarded to PC Project Director, Mary Schwartz. As a complete surprise to Mary, five PC patients were also invited to Dundee and recognized at the university graduation events. At the time of graduation, a new facility was opened at the McLean/Smith lab. This new unit will provide whole exome screening for PC samples.





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**MISCELLANEOUS MEETINGS.** PC projects representatives were able to attend various meetings throughout the year.

**Sep 7-10 CSD Development Day and the American Academy of Dermatology Legislative Conference.** We were fortunate to be sponsored by AAD and have full cost paid to attend this conference in Washington, DC.

**Oct 7-9 Rare Disease US Conference.** The conference was co-sponsored by NORD and DIA and held in Bethesda, Maryland. Fortunately, PC Project received a scholarship award to attend the meeting.

**Oct 18-20 PeDRA Annual Conference.** Held in Chicago, Illinois. Mary Schwartz was a guest speaker and presented.

**Nov 3-5 Partnering For Cures Conference.** The conference was sponsored by Michael Milken Faster Cures organization and held in New York City, NY.

**Nov 6 NIAMS Coalition Meeting.**

**Board of Trustees.** Board of Trustees meetings were held April 26, Aug 23, and Dec 6 2013. Board of Trustee members are:

C. David Hansen, MD (Dermatologist)  
Roger L. Kaspar, PhD (Scientist and CEO)  
F. Dwight Marchant, Treasurer (Bank Executive)  
David R. Owens, Esq. (Attorney)  
Mary E. Schwartz, (PC Project Director)  
Janice N. Schwartz, Secretary (PC Project Patient Advocate)  
Lex L. Udy, PhD, Chair (Scientist and CEO; retired)  
Craig T. Vincent, Esq. (Attorney)

## **FINANCIAL Summary 2013**

Total income: \$641,267  
Total Program Expenses: \$376,490  
Total Administrative Expenses: \$55,258  
Total Fundraising Expenses: \$28,022  
End of Year Net Assets: \$326,620