



Pachyonychia Congenita Project

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

ANNUAL REPORT 2012

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 have agreed to collaborate in research for PC. It is unique because it is organized by PC Project, a patient advocacy organization, rather than around a university, specific researcher or company or medical organization. Therefore, the focus is always on moving research forward for PC patients.

The IPCC Newsletter was published 3 times in 2012. On May 8-9, 2012 the Ninth Annual IPCC Symposium was held in Raleigh, NC.

PC Project hosts monthly IPCC Genetics Team web meetings where members of the IPCC Genetics team meet and 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 of the goals set by the IPCC is to have formal clinical studies for Pachyonychia Congenita. After years of effort, in 2012, Pfizer gave approval to TransDerm for development of topical rapamycin for PC and donated \$500,000 of the drug compound to the scientists to find a way to deliver the drug to the patients in a topical form to avoid the side effects associated with oral administration of the drug. The required toxicity testing for this study was completed in 2012.

Eight 2012 publications about PC by IPCC members include one article by Alan Irvine in May 2012 JID (Journal of Investigative Dermatology) which highlights the IPCRR (registry).

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.

Those participating in the IPCRR receive regular PC NewsBrief email publications. They are also eligible for scholarship funding for the annual Patient Support meetings. During 2012, there were two Patient Support Meetings. The first, co-sponsored by Le Coeur au Pied Association was held near Paris, France in June 2012. A second European meeting was held in October 2012 in Edinburgh, Scotland with 49 PCers and a total of about 100 attendees for the largest ever gathering in Europe of sufferers of this extremely rare genetic disease.



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Clinical studies continue to be carried out for PC research. Eight patients in Utah and Colorado donated biopsies from affected plantar skin, unaffected plantar skin and/or a hip area. The cells were used in projects at Johns Hopkins, University of Colorado, TransDerm and other collaborating universities including a study on PC nerves and pain, microarray analysis to determine related genes as targets for PC treatments, stem cell research and other research needs. A study with UCSF has begun to conduct histopathology on PC cysts especially for type PC-K17. A dozen specimens were gathered during 2012 and additional samples will be collected.

PC Project was selected to participate with the CAPSTONE mechanical engineering project at Brigham Young University (BYU) in Provo, Utah. The students spend spend 10 hours/week under the direction of a professor to develop a shoe that would alleviate pain in walking for PC patients. The project's \$20,000 cost was met by a \$10,000 matching grant award from BYU and a \$10,000 match from PC Project sponsors.

Several web meetings for patients were held in 2012 with those in the IPCRR. For back-to-school our PC kids got together on 28 September 2012 for a Kids Club Web Meeting and shared tips on how to handle teasing and keep comfortable at school. A special meeting for PC-K17 was also held in the fall of 2012 and will continue in 2013.

FUNDRAISING

To help raise funds to support the activities of the IPCC and IPCRR, for the first time in 2012, PC Project sponsored June PC Awareness. Volunteers around the world held individual events. Events were held in 10 countries. This was amazing and empowering for everyone involved!



As part of PC Awareness 2012, a press conference was held at TransDerm in Santa Cruz to kick off the first ThickSkin Duathlon to raise funds for PC research. Five PC patients attended the press conference and a video was filmed by HealthLine TV for special PC spotlights on public television and YouTube.

The 2nd Annual PC Charity Golf Tournament was held 18 August 2013. There were nearly 25 sponsors and 139 golfers! Many indicated they will repeat in 2013.

Quick Summary—2012 Fundraising

| | | |
|-------------------------|-------------|---------------------------------|
| 2012 ThickSkin Duathlon | \$56,167.95 | |
| 2012 Awareness Events | \$49,329.88 | Total \$113,570 raised 100% for |
| 2012 Golf Tournament | \$ 8,071.76 | Patient Support and Research |

With the \$2-for-\$1 match, \$340,709 was raised in 2012 for PC Project services.

GRANTS

PC Project was one of the organizations selected to participate in the National Rare Disease Registry (NIH/ORDR) GRDR. This Grant award was not quite as expected inasmuch as no funds were provided. PC Project received the right to share our data in the GRDR and the data was submitted before the end of 2012.



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AN R13 Grant in the amount of \$35,000 was awarded PC Project by NIH/NIAMS and NIH/ORDR as partial funding for the 10th Annual IPCC scientific meeting scheduled for February 2013 in Park City, Utah.

MISCELLANEOUS MEETINGS

22-24 October 2012 Rare Disease US Conference. The conference was co-sponsored by NORD and DIA and held in Washington, D. C. Fortunately, PC Project received a scholarship award and Mary Schwartz, PC Project Director, and Roger Kaspar, CEO of TransDerm, PC Project's biotech partner were able to attend.

28-30 November 2012 Partnering For Cures Conference. The conference was sponsored by Michael Milken Faster Cures organization and held in New York City, NY. Mary Schwartz, PC Project Director, and Roger Kaspar, CEO of TransDerm, PC Project's biotech partner were able to attend.

30 November Dermatological Conference. As part of PC Project's Educational Outreach program, four PC patients met over 200 physicians at an educational event to provide residents and established physicians the chance to learn about specific rare skin diseases not routinely seen.

Board of Trustees. Board of Trustees meetings were held 10 May 2010, 10 September 2010, and 3 December 2010. 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, Chair (PC Project Director)
Janice N. Schwartz, Secretary (PC Project Patient Advocate)
Lex L. Udy, PhD (Scientist and CEO; retired)
Craig T. Vincent, Esq. (Attorney)

FINANCIAL Summary 2012

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

ATTACHMENTS

Audited Financial Statement 2012 (attached)
IRS Form 990 (attached)