



# IPCC News


Jul-Aug-Sep 2004


Volume 1, Issue 3


## International Pachyonychia Congenita Consortium


2386 EAST HERITAGE WAY, SUITE B, SALT LAKE CITY, UT 84109  
PHONE: 877-628-7300 FAX: 877-628-7399 EMAIL: INFO@PC-PROJECT.ORG

### THANKS IPCC members for your on-going support for PC Project ..for example

 Referrals continue for new IPCC members (see list).

 Roger Kaspar has offered on-going consultation and an opportunity to speak with those at OOPD (Office of Orphan Product Development).

 Len Milstone and Phil Fleckman met with 10 PC patients at the FIRST Conference in July in Kansas City, KS.

 Irwin McLean and Frances Smith hosted 19 PC patients at a PC Patient Support Meeting in Dundee, Scotland, Sep 3-4. Sancy Leachman, Colin Munro, Edel O'Toole, and Maurice van Steensel gave clinician support. The meeting was an enormous success!

**JID UPDATE** Our great appreciation to authors who have submitted their papers on time! The paper **Clinical and Pathological Features of Pachyonychia Congenita** is in final preparation. Following our meeting in Dundee, Scotland, we are delighted to announce additional collaboration with Birgit Lane. Birgit's state-of-the-art lab will assist the analysis of PC tissues for this paper. Although our new schedule is a few months later for publication, we believe the quality of this publication warrants this extra time and care. Sancy will be in contact with each author for final editing and adjust deadlines.

**COLLABORATION.** Sancy Leachman set up an important conference call in mid-June to facilitate continued collaboration among grant recipients. THANK YOU to all participants for being so generous and open and willing to share.

#### 2004 GRANT UPDATES

**Philip Fleckman: National Registry for Ichthyosis and related disorders.** We were just advised that FIRST and GeneDx have joined us in support of this effort.

**Rudolph Leube: Creation of Cellular Models for PC.** Dr. Leube has provided excellent collaborative input as an active participant in the post-award grant review and the conference call.

**W.H. Irwin McLean & Frances Smith: Drug discovery for the treatment of Pachyonychia Congenita.** A specialist, Dr Yiwei Zhao, from China, has been recruited to work with Frances Smith on this project.

**Leonard Milstone: Inactivation of keratin 6a by sequence-specific Gene Targeting.** "I now have my clones of mutant luciferase CHO cells ready to test, using the upstream K6a triplex target site." Dr. Milstone initiated collaborations with several others

and has been given a supplemental grant to involve Dr. Michael Seidman (of the NIH) who will make new oligos for this project.

#### **Dennis Roop & Jiang Chen: Generating an inducible mouse model for Pachyonychia Congenita.**

Update from Dr. Chen: "The most difficult part of the construction of the targeting vector has been completed earlier this month. Right now, I am in the middle of selecting clones in which the wild type MK6a is replaced by partially humanized N171K-HK6a. Then, I only need to insert the neo cassette, before the first targeting vector is complete."

**Maurice A.M. van Steensel: Gap junction regulation of keratin expression.** Update from Dr. van Steensel includes this overview of status (1) All mutants of interest are cloned; (2) Confocal laser scanning microscopy expertise gained; (3) First test runs with fusion proteins in 3T3 cells performed; (4) Novel gap junction gene mutation identified, clinical studies performed and published. Functional characterization of the mutation in progress; (5) Cultures of keratinocytes, HaCATs and HeLa started" Dr. van Steensel also reported a new clinical

POP trial of novel pharmaceutical agent which may be of interest for PC.

#### **Sancy Leachman: PC Project Core Resource Grant.**

Activities this quarter included a Patient Support Meeting in Kansas City, KS (July 2004) and a Patient Support Meeting in Dundee, Scotland (Sep 2004), a number of physician referrals for patient's special needs, continued Registry consultations and genetic counseling.

**IRB APPROVAL** was given Sep 20 to the PC Project Registry so that we can take biopsies as well as receive and store biopsies, tissue blocks, and glass slides.

**2005 IPCC MEETING  
ST. LOUIS  
(prior to SID)  
IPCC Dinner & Social  
May 3, 2005**

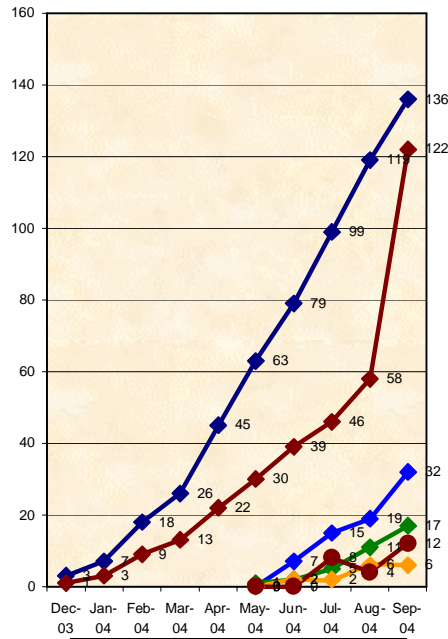
**IPCC Sessions  
9:30 to 4:00  
May 4, 2005**

Excellent sessions on PC for physicians and scientists will be presented throughout the day on May 4th.

Let us know if you'd need travel assistance, or you'd like to submit an abstract or suggest someone for a presentation. Please contact us at IPCC@PC-Project.org or call 877-638-7300.

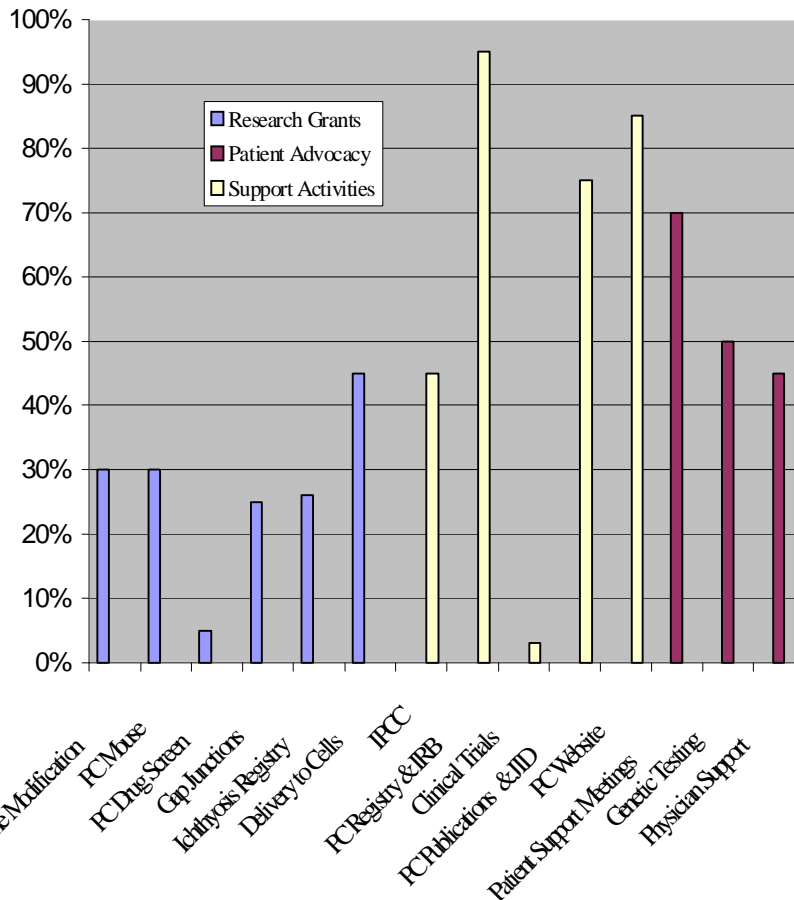
## Location of PC Patients

Australia 13	USA - MA 1
Brazil 1	USA - MD 1
Canada 6	USA - MI 4
England 17	USA - MN 5
Finland 2	USA - NY 13
France 4	USA - OH 7
Italy 1	USA - OR 3
Netherlands 8	USA - PA 2
Sweden 6	USA - SC 3
USA - CA 6	USA - SD 1
USA - CT 2	USA - TX 9
USA - FL 1	USA - UT 6
USA - IL 5	USA - VA 1
USA - IN 1	USA - WA 3
USA - LA 1	Wales 1



IPCRR Statistics

- ◆ 136 Individuals identified with PC
- ◆ 122 Registered with PC Project
- ◆ 32 Questionnaires Completed
- ◆ 17 Genetic Test Results Available
- ◆ 6 Waiting Test Results
- ◆ 12 Waiting for Consultation



**PUBLICATION INCENTIVE BONUS.** The first publication bonus award has been made to Frances Smith for her article on PC which will be published in an upcoming issue of the BJD. We encourage all IPCC members to publish PC data and to let us know of your publications.

## IPCC MEMBERS

\*new this quarter

\*\*PC Project Grant award

Sancy Leachman, M.D. Ph.D.  
Organizer & Chair

Sherri Bale, Ph.D.

Ralph R. Bradley, M.D.

Mario R. Capecchi, Ph.D.

\*\* Jiang Chen, M.D.

John J. DiGiovanna, M.D.

Ervin Epstein, M.D.

\*\* Philip Fleckman, M.D.

C. David Hansen, M.D.

Olga Igoucheva, Ph.D.

Aleksej Kansky, M.D.

Roger Kaspar, Ph.D.

\*Paul Khavari

Gerald G. Krueger, M.D.

Markus Landthaler, Ph.D.

Rudolph Leube, M.D.

Alfred S. Lewin, Ph.D.

\*\* Irwin McLean, Ph.D., D.Sc.

\*\* Leonard M. Milstone, M.D.

\* Colin Munro, M.D.

\* Edel O'Toole, M.D.

\*\* Dennis R. Roop, Ph.D.

\*\* Michael Seidman, Ph.D.

\*\* Frances Smith, Ph.D.

Bhaskar Thyagarajan, Ph.D.

\*\* Maurice van Steensel, M.D.

Pauline Wong, Ph.D.

Kyonggeun Yoon, Ph.D.

**A gift for you!** In appreciation of your support for PC, and in support of DEBRA UK, we're happy to send you this special CD. You'll definitely recognize one of the 'super stars.' Enjoy!