

IPCC NEWS

International Pachyonychia Congenita Consortium

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

-  Referrals for new IPCC members (see list).
-  SiRNA's ready for testing on cells.
-  Sample release forms received from Sherri Bale.
-  PC photos received from Phil Fleckman, Irwin McLean, Jerry Kreuger.
-  Dennis Roop made PC Project visitors most welcome at the SID meeting (including a tour of the 'poster session').
-  Len Milstone (and Phil Fleckman) have made possible a special PC patient session at the July F.I.R.S.T. conference.
-  Roger Kaspar, Sherri Bale and Bhaskar Thyagarajan all provided helpful suggestions this quarter.
-  While presenting a paper in China, Jiang Chen contacted researchers there in regard to PC Project and their research (see recent JID articles).
-  John DiGiovanna provided information and contacts which will be essential as we move toward clinical trials.
-  Irwin McLean, Frances Smith are hosting a PC Patient Meeting in Dundee, Scotland in Sep 2004.
-  Sancy Leachman and David Hansen hosted the first PC Project patient discussion meeting. The video can be viewed at <http://www.pachyonychia.org/Events.html#PastEvents>

2004 Grants

The PC Project Grant Awards were announced on April 28, 2004 in Providence, RI prior to the SID meeting.

Details are on the **Researchers** section of the PC Project website. Grants selected are:

Philip Fleckman, M.D.
University of Washington.
Project Title: National Registry for Ichthyosis and related disorders.

W.H. Irwin McLean, Ph.D., D.Sc. & Frances Smith, Ph.D. University of Dundee, Scotland.
Project Title: Drug discovery for the treatment of Pachyonychia Congenita.

Leonard Milstone, M.D.
Yale University.
Project Title: Inactivation of keratin 6a by sequence-specific Gene Targeting.

Dennis Roop, Ph.D. & Jiang Chen, M.D. Baylor College of Medicine.
Project Title: Generating an inducible mouse model for Pachyonychia Congenita.

Maurice A.M. van Steensel, M.D.
University Hospital Maastricht, The Netherlands.
Project Title: Gap junction regulation of keratin expression.

Additional funding was established for PC Project Core Resource needs in coordinating clinical, genetic and patient-related activities in support of PC patients and researchers.
Sancy Leachman, M.D., Ph.D. University of Utah will administer these funds.

Five Stars

to all grant awardees for their prompt and positive



response to post-award communications with results as follows —

- (1) Progress goals have been set with the first target date 1 August 2004. Progress reports on each grant will be published quarterly in the IPCC newsletter.
- (2) Post-award reviews are being completed by Drs. Ervin Epstein, Rudolph Leube, Maurice Seidman and others to further our collaborative efforts and enhance grant potential.
- (3) On-going collaboration including sharing of reagents, discussion regarding setting up the best screening assay and exchange of other ideas among the IPCC members.

Sancy has scheduled a conference call in mid-June to facilitate continued collaboration. If you want to be part of this 'telephonic meeting,' contact PC Project at 877-628-7300 or email Mary and/or Sancy.

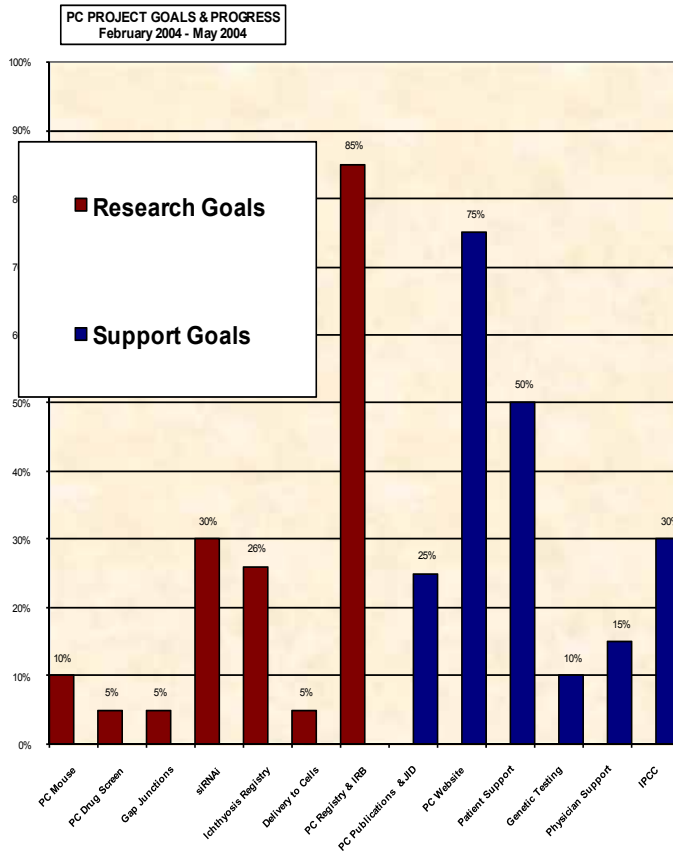
JID—July 1 Deadline

- I. **Introduction (pp. 3-5)** [Sancy Leachman](#)
- II. **Clinical and Pathological Features of Pachyonychia Congenita** [Philip Fleckman](#)
- III. **Therapeutic Approaches to Pachyonychia Congenita (pp.16-20)** [Leonard Milstone](#)
- IV. **The Strength of the HUGO Database** [Irwin McLean](#)
- V. **The Genetic Basis of Pachyonychia Congenita (pp. 24-33)** [Frances Smith](#)
- VI. **Mouse Models in Pre-clinical Trials for PC** [Jiang Chen](#)
- VII. **Genetic & Pharmacological Approaches to Correction, Modification or Regulation of Gene Expression in Autosomal Dominant genetic Disease** [Alfred Lewin](#)
- VIII. **Goal Oriented Integration of Academia and Industry** [Roger Kaspar](#)
- IX. **Advertisement to become a part of IPCC (p. 60)**

When you are contacted to be a co-author or reviewer/reader — please accept and help with this important effort. Sancy says "I am characteristically 'shooting for the top' with this publication! I think we can make this the best resource ever published for dermatologists and scientists interested in the area of PC and I think we should go for it."

NEXT IPCC MEETING
May 3, 2005—New Orleans
 (prior to the 2005 SID Meeting)
 IPCC Welcome Dinner—May 2, 2005

We're on the web at
www.PC-Project.org



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.
- 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.
- ** 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.

◆ 79 PC Cases Identified by Project

◆ 39 on Website Message Boards

◆ 23 Requested Questionnaire

◆ 7 Completed Questionnaires

◆ 2 Completed Genetic Testing

Location of PC Patients

- Australia 8
- Canada 6
- England 9
- France 3
- Sweden 9
- The Netherlands 1
- USA - CA 1
- USA - FL 1
- USA - IL 6
- USA - IN 1
- USA - MA 1
- USA - MI 4
- USA - MN 5
- USA - NY 8
- USA - OH 7
- USA - PA 1
- USA - SD 1
- USA - TX 2
- USA - UT 6
- USA - WA 3
- Wales 1

IRB Approval & IPCRR begins

On April 12, 2004, PC Project received approval from Western IRB for the International PC Research Registry (IPCRR). Registration has begun!

We are pleased that in the first 3 weeks, **23** patients have completed initial screening and received the Questionnaire. PC Project is arranging for clinician review & validation as well as for genetic testing when requested by the participants.

When researchers need information or participants, we plan to be ready with quality data.