PC PROJECT SPONSORED EVENTS 2012

International PC Consortium (IPCC)  
9th Annual Symposium  
May 9, 2012 prior to SID  
Raleigh, North Carolina  
ALL INVITED—please pre-register at www.pachyonychia.org  
(see page 5 for program outline)

PC Patient Support Meetings  
Roissy, France—June 8-9, 2012  
IPCC members are invited to participate.  
For details or to register, please email info@pachyonychia.org/

Pachyonychia Congenita Awareness Day Around the World  
Because PC is such a rare disease, patients often feel isolated and are unaware of the ‘community’ of patients, researchers and physicians who are part of the PC coalition. This awareness day event involves patients in more than 50 counties. The purpose is to raise funds, and raise awareness and understanding of PC for the individual patient and the communities in which they reside.  
If you can help with an event (race, chess tournament, etc.) please let us know. We’d like everyone to do something.

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Mark Your Calendar Now

10th Int’l Pachyonychia Congenita Consortium (IPCC) Symposium  
February 14-17, 2013  
Yarrow Hotel, Park City, Utah

This is an essential meeting for the IPCC and all who want to participate in the research and clinical studies moving forward.

Accomplishments—Collaboration  
Research Discoveries  
Goals—Grant Awards

Ten Years — look at all we’ve done!  
Yet, still miles to go before we sleep.
UPDATE ON TRANSDERM ACTIVITIES—Roger Kaspar

Clinical trial. Our goal, working with the IPCC, is to be ready for the next siRNA clinical trial by the beginning of 2013. With support from a recently awarded SBIR, Agilent is synthesizing a self-delivery version of TD101 under GMP conditions. This will be administered to patients using dissolvable microneedle arrays. To this end, we have recently installed a clean room facility and are working with compliance consultants to put in place the necessary SOPs to be able to manufacture GMP microneedle arrays at TransDerm.

RNA profiling. Using biopsies provided by PC Project, we have isolated total RNA from involved and uninvolved plantar biopsies. This RNA was analyzed for global mRNA and miRNA expression. A number of genes were identified that are differentially expressed in PC skin, which has been confirmed by RT-qPCR and immunohistochemistry.

Nucleic acid delivery. Together with colleagues at Yale and Sciton, we have evaluated the ability of siRNAs to penetrate the stratum corneum barrier following treatment with a YAG/Er laser and distribute to the live layers of the epidermis. The laser has the ability to generate a define grid of holes of desired depth to allow passage of large, charged biomolecules such as siRNAs. This represents an alternative, patient-friendly way to traverse the stratum corneum barrier.

Each of these topics will be addressed at the upcoming IPCC meeting in Raleigh, where we hope to see many of you. We also want to announce that TransDerm will be participating in PC Awareness Day. We will be hosting a mountain bike duathlon event (run/bike/run race) entitled the ThickSkin Duathlon in Santa Cruz on June 2nd. If any of you are in the area, please come and join us.

The Dermatogenetics conference, sponsored by Nature Genetics, was held Feb 2012 in Miami, FL. A number of IPCC members gave presentations, including the following:

Keynote Lecture

EXPLORING THE THERAPEUTIC POTENTIAL OF INDUCED PLURIPOTENT STEM CELLS FOR INHERITED SKIN DISEASES—Dennis Roop

The therapeutic potential of induced pluripotent stem cells (iPSc) for the treatment of inherited skin blistering diseases is enormous since this procedure enables the delivery of truly personalized medical treatment. The generation of iPSc from the same patient would not only potentially avoid the complication of immune rejection, but also provide a source of rejuvenated adult stem cells that are most likely exhausted as a result of unsuccessful attempts to repair blistered tissues. However, before iPSc can be safely used to treat these patients, several obstacles must be overcome. First, iPSc must be generated using non-genetic methods to avoid the risk of developing cancer due to the random integration of viral vectors. Second, protocols must be developed for the efficient differentiation of iPSc into a keratinocyte lineage. Third, the reprogramming of skin cells into iPSc involves the resetting of gene expression patterns, as well as methylation signatures, throughout the genome. Therefore, it will be essential to determine the genetic stability and histocompatibility of iPSc-derived cells and demonstrate that these cells are stable and able to repair the skin in an in vivo environment without immune rejection. Fourth, efficient and safe methods must be developed for the genetic correction of genes that are defective in these patients.

(NRARE)2: THE GENETICS OF UNCOMMON SKIN DISORDERS IN POPULATION ISOLATES

Eli Sprecher

The Middle Eastern population is a puzzle of numerous small ethnic subgroups that have lived for centuries in a state of genetic isolation one relative to the other. This situation has led to the emergence of a unique spectrum of population-specific mutations and disorders. The deciphering of the mo-
CELL THERAPIES FOR INHERITED BLISTERING SKIN DISEASES  John McGrath

Developing new experimental medicine treatments for inherited blistering skin diseases is challenging. Approaches involving gene, protein, cell and drug therapies (or combinations thereof) are all being pursued, although clinical trials and involvement of patients is perhaps most advanced for cell therapy interventions. First-in-man trials of intradermal injections of allogeneic fibroblasts in subjects with recessive dystrophic epidermolysis bullosa (RDEB) were first reported in 2008. Injections of allogeneic fibroblasts into wound margins may also promote faster wound closure although some studies have demonstrated that saline can promote similar rates of wound healing. As well as allogeneic fibroblast cell therapy, a clinical trial of third party bone marrow transplantation following myeloablation has also been reported in 7 children with RDEB. Although 2 children died, increased collagen VII was demonstrated in the skin that persisted for more than 2 years (follow-up ongoing) with functional improvement in the skin (increased negative pressure suction cup blister times) as well as increased quality of life metrics. Although this trial clearly indicated a clinical benefit from bone marrow transplantation, there remain several unanswered questions, including the nature of the reparative cells and the mechanism of recruitment into damaged skin and mucous membranes.

One other fascinating experimental model relates to the phenomenon of revertant mosaicism, whereby a patch of keratinocytes can undergo spontaneous genetic correction. One translational research objective is to try to harness this “natural gene therapy”, either through the application of revertant cell skin grafting, or by generating iPS cells which avoid the need for additional correction of the inherent germline genetic defect underlying the skin blistering.

KERATIN AGGREGATES IN EPIDERMOLYSIS BULLOSA SIMPLEX (DOWLING.MEARA): CAUSE OR EFFECT OF THE EBS PATHOLOGY? Birgitte Lane

People with severe epidermolysis bullosa simplex (Dowling Meara type, or DM-EBS), caused by dominant mutations in basal cell keratins K5 or K14, have very fragile skin that blisters easily upon mechanical stress. There is no cure for this disorder, and gene therapy approaches will be costly due to the rarity of the disorder and the diversity of mutations. Our lab is seeking more generic approaches to reducing the skin fragility by targeting the downstream consequences of the keratin mutations. Using EBS-mimetic cell lines and cell culture stress assays to generate disease model systems, pathogenic mechanisms can be studied ahead of direct patient testing. We have repeatedly observed that DM-EBS cells are in a constitutively stressed state, seen as activation of stress-associated indicator responses in the cells. The physiological stress is evident even when cells are not directly under mechanical stress, and it appears to be fundamental to the pathology of EBS. The possible origins of this stress are being investigated.

DM-EBS keratinocytes often show misfolded keratin aggregates in their cytoplasm, whereas milder forms of EBS tend not to. It is debated whether these keratin aggregates, the hallmark of DM-EBS, directly contribute to the worse pathology of DM-EBS over other milder EBS subtypes. Upon mapping the behaviour and biochemistry of these protein aggregates against stress-induced signalling pathway activity in affected cells, data obtained suggest that the link between the prevalent Dowling-Meara EBS mutations (involving Arg125 in K14) and the clinical phenotype seen in patients (clustered and spreading blistering) may lie in the combined action of the keratin mutation together with the activation of stress signals normally involved in wound healing. If the stress signaling is prevented, affected keratinocytes can be partially rescued from the disease-associated stress state. This approach may provide the sought-after generic therapy solution for EBS.
All interested researchers are invited to join the IPCC symposium, and are invited to the hosted dinner, breakfast and luncheon. The registration fee of $125 is waived for all who pre-register at www.surveymonkey.com/s/IPCC2012 so please take a minute and register. Details on the dinner time and location will be sent to those registered.

If you have questions or need more information, please contact info@pachyonychia.org.

**International Pachyonychia Congenita Consortium (IPCC) Program 2012**

**WEDNESDAY—May 9**
7:00 am—7:45 am BREAKFAST
7:55 Welcome—Eli Sprecher, IPCC Chair

**Section Chair: Dennis R. Roop, University of Colorado Anschutz Medical Campus**
8:00 Introduction of Keynote Speaker
8:05 *Harnessing Stem Cell Gene Biology to Combat Genodermatoses*
Jakub Tolar, University of Minnesota, Minneapolis, Minnesota, USA

**Section 1: Diagnostics and Genetics**
8:45 *New approaches to the diagnostics of genodermatoses*
Gabriele Richard, GeneRx, Inc., Gaithersburg, Maryland, USA
9:00 *Update on Pachyonychia Congenita Genetics*
Francis J.D. Smith, Epithelial Genetics Group, University of Dundee, Dundee, Scotland UK
9:15 *Frizzled 6 Gene*
Reinhard G. Betz, University of Bonn, Germany

**Section Chair: E Birgit Lane, Institute of Medical Biology, Singapore**

**Section 2: Research Studies**
9:30 *Review of the GO Delivery Grant*
Christopher H. Contag, Stanford University, California, USA
9:50 *Keratinocytes and Pain*
Michael J. Caterina, Johns Hopkins School of Medicine, Maryland, USA

10:10 BREAK

10:30 *Nucleoside-modified messenger RNA for gene therapy*
Katalin Kariko and Drew Weissman, University of Pennsylvania
10:50 *Update on Pachyonychia Congenita Research at TransDerm*
Roger L. Kaspar, Robyn P. Hickerson, Tycho Speaker
11:30 *K16 Null Mice*
Juliane C. Kellner, Pierre A. Coulombe, Johns Hopkins Medical Institutions
11:50 *Keratins, Pachyonychia Congenita and Research Advances*
W. H. Irwin McLean, Epithelial Genetics Group, University of Dundee, Scotland UK

12:15 HOSTED LUNCH

**Section Chair: To be Announced**

**Section 3: PC Physician Network**
1:15 *Trials and Tribulations*
John A. McGrath, St John's Institute of Dermatology, Kings College London, England UK
1:35 *Towards Differential Diagnosis: What We Have Learned From The IPCCR*
Edd A. O'Toole, Barts & the London, Queen Mary's, London, England UK
1:55 *Developing Best Practices For Pachyonychia Congenita*
Ilan Goldberg, Dermatology Department, Tel-Aviv Sourasky Medical Center, Israel
2:15 *Painless Biopsies for PC Planar Keratoderma*
Leonard M. Milstone, Yale School of Medicine, New Haven, Connecticut, USA
2:25 *DISCUSSION: Moving forward with clinical studies for Pachyonychia Congenita*
Eli Sprecher, Tel Aviv Sourasky Medical Center, Israel
INTERNATIONAL PACHYONYCHIA CONGENITA CONSORTIUM (IPCC)

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The IPCC members have agreed to work together to develop PC therapies.

DIAGNOSTICS AND GENETICS TEAM — FRANCES J.D. SMITH, PhD, Chair

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PHYSICIAN NETWORK — ELI SPRECHER, AMY S. PALLER, MAURICE VAN STEENSEL – Co-chairs


* Parcipated in the 2011 PC Treatment and Care Survey

Open Membership: Physicians and Scientists interested in collaborating for PC Therapies

PC Awareness Day June 2, 2012—PC is an ultra rare disease. Patients feel isolated, and are often unaware of the ‘community’ of patients, researchers and physicians who are part of the PC coalition. This 2012 Awareness Day event involves patients and supporters in more than 50 counties uniting to raise funds and also raise awareness of Pachyonychia Congenita for the individual patient and for the communities in which they reside. The Duathlon sponsored by TransDerm scientists is one of over 50 events which will take place June 2.

IPCC Physician Network Webinar
August 1, 2012—12 noon MDT
Eli Sprecher will present and discuss
Inherited nail disorders
All physicians are invited.
To register, please email info@pachyonychia.org

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TENTH
International Pachyonychia Congenita Consortium (IPCC)
SYMPOSIUM
February 14-17, 2013
Yarrow Hotel, Park City, Utah
Please contact info@pachyonychia.org to submit abstracts or apply for travel stipends

GOALS FOR THE IPCC 2012—2013
1. Public awareness. This will include TV shows; professional articles in lay press and scientific journals
2. Completion of planned randomized/controlled trials
3. Development of a practical algorithm for the clinical and molecular diagnosis of PC and other similar disorders

The IPCC 10th Annual Symposium will be held in Utah on Feb 14-17, 2013 where IPCC members will present advances in PC research from numerous projects currently being carried out.

Image from Pachyonychia Congenita skin biopsy courtesy of TransDerm, Inc.
INT’L PC CONSORTIUM SYMPOSIUM MAY 8-9, 2012 RALEIGH, NC

We were encouraged by the positive comments provided in our post-meeting survey which reflected high satisfaction with the quality of presentations at the 2012 meeting, as well as a definite sense of collaboration and community among the participants.

The participants met the challenge of a shortened program to provide crisp, clear, focused discussions of many leading research approaches. A summary of some of the presentations is included here, and a full meeting report will be published.

May 9
Keynote Address by Jakub Tolar
(University of Minnesota) outlined the remarkable results achieved in his work with stem cell transplantation in children suffering from dystrophic epidermolysis bullosa.

In the Diagnostics and Genetics session, Gabrielle Richard (GeneDx) presented the advances in diagnostic approaches and the challenges for inherited skin disorders.

Frances Smith (University of Dundee) outlined the protocol and findings from the International Pachyonychia Congenita Research Registry (IPCRR) which has enrolled more than 1000 patients, over 500 with genetic test results including more than 85 specific PC mutations. Several rare cases of homozygosity of dominant mutations in PC were illustrated as well as typical cases for each of the PC types including PC-K6a, PC-K6b, PC-K16 and PC-K17.

Findings from RNA profiling studies were shared by Roger Kaspar (TransDerm). RNA was isolated from biopsies from affected and non-affected areas of the sole and analyzed for differences in mRNA and miRNA expression patterns. Of particular interest, keratin 9 (KRT9) expression was nearly absent in affected PC lesions. KRT9 appeared to be reciprocally expressed with respect to PC gene expression (i.e., KRT6a, 6b, 16 and 17) suggesting potential therapeutic strategies.

The Research Studies session began with a summary by Christopher Contag (Stanford University) of progress in developing patient-friendly delivery of nucleic acids through research conducted under the GO Delivery! Grant (NIH/NIAMS ARRA).

May 8. The IPCC members and guests enjoyed a walking tour of Raleigh and were hosted at a dinner on Tuesday evening at the Second Empire Restaurant in Raleigh, NC. Informal discussions are often some of the meeting highlights and build the comaraderie and fellowship evident in the IPCC.

At the IPCC Dinner—May 8, 2012
Second Empire Restaurant, Raleigh, NC
implicating keratinocytes in cutaneous sensation and the potential roles of ion channels of the transient receptor potential (TRP) family in this multicellular process including recently obtained transgenic mouse data directly supporting a role for keratinocytes in the perception of pain.

Irwin McLean (University of Dundee) dubbed some recent advances as “The Year of the Mouse” and outlined the various new options available for studies using recently available mice.

Robyn Hickerson (TransDerm, Inc.) pointed out that although siRNAs have exquisite sensitivity and potency, translation to the clinic has been hampered due to the lack of efficient delivery systems that allow penetration through the stratum corneum outer barrier. Use of the Er:YAG laser is showing the potential to facilitate delivery of nucleic acid-based therapeutics targeting previously untreatable genetic skin disorders, such as PC.

Tycho Speaker (TransDerm, Inc.) showed real-time non-invasive imaging by confocal fluorescent microscopy of siRNA delivery and diffusion from loaded water-soluble polyvinyl alcohol microneedle arrays. The capacity to visualize both depot and released drug opens new doors in efforts to optimize delivery systems for PC therapeutics.

Dress Weissman (University of Pennsylvania) showed that in vitro-transcribed mRNA has great therapeutic potential to transiently express encoded protein without the adverse effects associated with viral- and DNA-based deliveries or direct protein supplementation.

Juliane Lessard and Pierre Coulombe (Johns Hopkins Medical Institutions) presented findings from a recently developed K16 knock-out mouse. In this study, although inherited in a recessive fashion, the knockout of Krt16 in mice consistently causes oral lesions as well as PPK-like hyperkeratotic calluses on Krt16/-/- front and hind paws, which severely compromise the animals’ ability to walk.

The Physician Network session began with John McGrath (Guy’s & St. Thomas’ Hospital, King’s College) recognition of the obstacles encountered in developing therapeutics for rare disorders. He encouraged participants to continue past the obstacles. A short video clip of the effect a clinical study can have in improving the quality of life for a patient was shared. Edel O’Toole (Barts & the London, Queen Mary’s) presented a variety of clinical cases showing the similarities and differences for various skin disorders like PC. She provided a ‘test’ for the participants. See page 4 for a special contest using this ’test’.

Ilan Goldberg (Sourasky Medical Center Tel Aviv) reported on the statistical analysis of the PC patient survey on “best practices for treatment of PC.” Patients reported mechanical and surgical treatments as the most effective conventional treatment for all investigated manifestations, while most of the available conventional medical treatments, including topical or systemic retinoids, antibiotics and antifungal agents, were found to be only slightly to non-effective at all. Univariate analysis was used to determine the relationships between each explanatory variable and the treatment effect outcome variables (multivariate models were found not to be stable due to the small sample size).

During the last year, plantar biopsies have been taken from PC patients. Observers have noted remarkable different pain experience for patients. Leonard Milstone (Yale University) was asked to summarize his ‘pain free’ biopsy technique which he explained with ‘The Three T’s’ of Time, Tremor and Talk.’
Eli Sprecher (Tel Aviv Sourasky Medical Center and IPCC Chair 2011-2013) outlined the achievements of the IPCC during 2011-2012 and set forth the 2012-2013 goals for the IPCC. He emphasized the importance of the 10th Annual IPCC Symposium to be held February 14-17, 2013 in Park City, Utah where many of the studies now being conducted will be reported.

CONTEST—can you identify which of the patients below have Pachyonychia Congenita?
To enter contest, print this page, mark those with PC, and mail (or scan and email) to PC Project.
First Prize — $250 (first entry with all correct answers)
All entries will be recognized and receive a prize from PC Project.
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All physicians and scientists willing to collaborate to help find treatments and a cure for Pachyonychia Congenita are invited to actively join the IPCC. During the next months, we will update the IPCC membership list to build further collaborations and identify physicians available for clinical studies. We will contact IPCC members to determine their status (active or mailing list only.) Then, we will add links from our website to highlight special interests and expertise of active IPCC members. We will also create a listing of resources and reagents available to active IPCC members. We hope you will want to be an active IPCC participant in 2012-2013.

* Participated in 2011 PC Treatment and Care Survey
GOALS FOR THE IPCC

At the May 2012 IPCC meeting, several goals for the 2012-2013 year were outlined:

1. Public Awareness. This will include TV shows, professional articles in lay press and scientific journals.
2. Completion of planned randomized/controlled trials.
3. Development of a practical algorithm for the clinical and molecular diagnosis of PC and other similar disorders.

How are we doing? Here is a brief summary of progress and problems in each goal area.

GOAL 1—PUBLIC AWARENESS

A professionally prepared 6-minute video on Pachyonychia Congenita is now on YouTube. http://goo.gl/tD9Am
This video is also on the PC website and available on dvd on request. A 3-minute version is being distributed to several hundred public television stations in the USA as part of the HealthlineTV series of shorts between programs. Please let us know if this airs in your area.

A list of publications by IPCC leaders is included in this Newsletter. A lay review article has been accepted for publication in the Jan-Feb 2013 issue of the Journal of the Dermatology Nurses' Association. The JDNA is a bi-monthly publication provided as a member benefit by the Dermatology Nurses Association. It is available both in print and in its entirety on the Internet. Several major articles on PC (including an updated review article, a ‘best practices article, and other PC-related research articles) are all in final preparation or submitted.

Two radio spots have been aired: (1) a radio interview from a station in Indiana was completed as a result of the PC Awareness Day 2012 and (2) In California, a public radio segment was professionally prepared and distributed about PC and PC research efforts.

PC Project continues to participate in grand rounds/conferences. The next opportunity will be the Philadelphia Dermatologic Society Conference on Friday, November 30, 2012. This will be similar to the Stanford Conference where all of the local universities participate.

The multi-center statin study MRC grant was not funded. Discussions are...
continuing within the research team on how to best move this forward. Several small statin studies have been completed and show some promise in reduced pain levels measured by increased walking time before pain is unbearable.

A topical gabapentin study was completed with six patients using an active agent and placebo. The patients did not report any benefit. Other small studies using other topical pain medications are underway. PC Project is seeking to involve neurologists more in our planning since pain is the main focus for all PC patients.

The rapamycin study is now moving forward with the announcement of an agreement from Pfizer to provide TransDerm with rapamycin for use with PC patients. Once the drug is manufactured, the topical will have to be approved by the FDA. Joyce Teng at Stanford has had experience with topical rapa for another rare disorder and will be participating in design of the clinical trial.

**TOWARDS A BETTER GRAND ROUNDS OPPORTUNITY**

We invite IPCC members to create Grand Rounds opportunities in your areas. Here is what PC Project provides:

1. a minimum of 2 patients representing different PC types (such as PC-K6a, PC-K17, etc. We prefer to involve 3 or 4 patients to provide the most comprehensive teaching opportunity.
2. Copies of current select publications on PC from leading journals.
3. History, images and data on the individual patients.
4. Any financial support the patients need for travel, host them for a lunch or dinner and make it a very positive, educational experience for the patients — and those being trained.

PC is a syndrome. Any teaching done with a single PC patient is unavoidably misleading and incomplete. The different types vary greatly and pain (not thickened nails) is the common element in PC.

**GOAL 3—CLINICAL AND MOLECULAR DIAGNOSIS OF PC**

We have the data available to create a practical algorithm to meet this goal — but need an investigator to assist with this. We would like to have this prepared prior to the February 2013 IPCC meeting.

**BEST PRACTICES**

Data from a survey of 125 PC patients has been analyzed, and a publication “Best Treatment Practices for Pachyonychia Congenita” has been prepared by Ilan Goldberg under the direction of Eli Sprecher. The manuscript has been submitted and will be published shortly.

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Everyone involved with PC Project and the IPCC must be an optimist and we must find the opportunities available to us and maximize those options.

Chris Austin, newly named Director of the NIH/NCATS institute, said to the group ‘you are my people.’ He is a neurologist, and one of the immediate goals at PC Project is to add neurologists and other pain specialists to the marvelous core of dermatologists now working with us.

Steven Groft, Director of NIH/ORDR (Office of Rare Disease Research) was available at the meeting, as well as his counterpart Anne Pariser from the FDA/ORD (Office of Rare Diseases).

Excellent, practical information was provided through the 3-day meeting on specific ‘how to’ take a drug from discovery, through the ‘valley’ of ‘death’ to approval at the FDA and, ultimately to patients needing treatments.

The recent bipartisan user fee or PDUFA legislation for the FDA contains language that enhances the patient voice in critical FDA deliberations and accelerates the review process for therapies for rare disorders.

There are currently many excellent options for rare disease development.

**SELECT PUBLICATIONS**

aging atopic dermatitis in adult patients. Semin Cutan Med Surg, 31(3 Suppl), S18-22.


PC Project Medical & Scientific Advisory Board 2012

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Sherri J. Bale, PhD
► C. David Hansen, MD
► Peter R. Hull, MD
Roger L. Kaspar, PhD
E. Birgitte Lane, PhD

John A. McGrath, MD, PhD
W. H. Irwin McLean, DSc, FRSE
► Leonard M. Milstone, MD
► Edel O’Toole, MD, PhD
Amy S. Paller, MD

Dennis R. Roop, PhD
Frances J.D. Smith, PhD
Eli Specher, MD, PhD
Jean Y. Tang, MD, PhD
Maurice van Steensel, MD, PhD

► These physicians provide patient consultations for the Int’l PC Research Registry (IPCRR) sponsored by PC Project

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