If you were one of the more than 50 specialists attending the 10th Annual IPCC Symposium, you know that the meeting was extraordinary both for the quality of presentations and the camaraderie* at the meeting.

Opening remarks were offered by meeting co-chairs, Pierre Coulombe and Dennis Roop. Pierre encouraged attendees to participate and contribute in the discussions. Dennis gave a bit of statistical history pointing out that more than 80% of those who were part of the first IPCC meeting in 2004 are still active participants.

Sharon Terry, keynote speaker for the conference set the bar high, challenging all to share and exchange information and looking to the time when research will be conducted as a community effort. Sharon also mentioned the new registry platform being developed by Genetic Alliance which will be available for the International PC Research Registry in the next months so that the data can be collected online. (Also, see page 2 in this issue for the announcement of the Genetic Alliance Disease InfoSearch.)

The program is available at www.pachyonychia.org so that you can locate and follow-up on specific presentations of interest. Also, a meeting review is being prepared by Maurice van Steensel (with other attendees contributing) and will be published when available.

*When I checked the spelling, I found that camaraderie and comradery both refer to the spirit of goodwill and fellowship that exists between friends. That absolutely was the tone of the conference.

HOW IS THE IPCC DOING IN MEETING GOALS?
A poster displayed at the 2013 meeting showed the progress 2004-2013 as IPCC members continue to set and meet annual goals. Following is a brief summary of progress on the 2012 goals.

2012 GOAL 1—PUBLIC AWARENESS
to include TV shows, professional articles in lay press and scientific journals.

— A professionally prepared 6-minute video on Pachyonychia Congenita is now on YouTube at 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 some recent publications by IPCC leaders is included in this Newsletter. In her remarks, Sancy Leachman (who was active with the IPCC 2004-2010) reviewed the large increase in publications over the last years through the work of the IPCC members. Of special note was the effort by Irwin McLean and Roger Kaspar in heading the preparation of the May 2011 JID articles on PC. Our thanks to each author who has pre-
pared and published articles on PC since 2004 that include genetically confirmed and accurate data.

— 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 Atlantic Derm Conference in April 2013. In 2012, PC Project participated at the Philadelphia Dermatologic Society Conference in and the Stanford Conference where three CA universities joined together for the event.

2012 Goal 2—Clinical Studies
Completion of planned randomized/controlled trials.

— The siRNA (sdTD101) clinical trial is on track with drug manufacturing completed, and the initial tox study conducted by TransDerm. Applications for Orphan Drug Status have been filed with the FDA and the EU. Materials are being submitted to the FDA for a May 1 pre-IND meeting. Len Milstone (USA) and Alan Irvin (EU-Ireland) have agreed to help with the clinical trial.

— The multi-center statin study MRC grant was not funded. We greatly appreciate the work of Edel O’Toole in that effort. Data from three small studies (Saskatchewan/4 patients, Israel/2 patients and Alberta/3 patients) are being reviewed. Although there is some indication that some patients have reduced pain levels measured by increased walking time before pain is unbearable, the data needs further evaluation. Discussions are continuing on how to move forward with statin studies.

— 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 will begin shortly. Pfizer has agreed to provide TransDerm with rapamycin for use with PC patients. A pre-IND meeting with the FDA is scheduled for April 10. Joyce Teng and Jean Tang (Stanford dermatology) have experience with topical rapa for other rare disorders and will administer the PC clinical trial.

2012 Goal 3—Diagnosis
Development of a practical algorithm for the clinical and molecular diagnosis of PC and other similar disorders.

— Although not yet written or published, a great deal of data has been gathered. Eli Sprecher plans to complete this goal in 2013.

IPCC 2013 Goals
There is a strong desire to continue the public awareness and publication efforts. The main emphasis for 2013 will be on clinical trials.

2013 Goal 2—Clinical Studies
Completion of planned randomized/controlled trials. This is the major goal for the next year for the IPCC. In order to achieve this level of clinical trial, resources will be directed to placebo-controlled, randomized studies or efforts directly related to this type of trial.

2013—Goal 2 Assessment Tools
— Develop a set of assessment tools which can be utilized in various clinical trials. A project has begun to develop these tools and to reach a consensus that these tools will be used in each study/trial so that we have consistent evaluation of various treatments. The specific clinical endpoints may differ from study to study, but a pool of PC validated assessment tools will be available to investigators for use in evaluating outcomes in PC.

IPCC Member Wins Award on Worldwide Rare Disease Day
February 28, 2013 was Rare Disease Day in the USA. This is an international movement. We were pleased to learn that IPCC member, Maurice van Steensel was awarded the Rare Diseases Angel in the Netherlands. This is awarded every year to one volunteer and one professional who dedicate themselves to helping people with rare diseases. He was nominated by the CMTC-OVM foundation who represent people with cutis marmorata congenita and other vascular malformation disorders.
ONE STOP SHOPPING FOR ALL DISEASES: GENETIC ALLIANCE LAUNCHES DISEASE INFOSEARCH

Disease InfoSearch (www.DiseaseInfoSearch.org), a one-stop shop for information and engagement on more than 13,000 conditions, launched today. Disease InfoSearch connects healthcare providers, researchers and the general public with support groups, relevant and timely peer-reviewed articles, open and appropriate IRB-approved clinical trials, and general disease information. Each condition page also features an interactive tool that enables individuals to contribute to biomedical research by securely sharing their health information.

For the past 26 years, Genetic Alliance has connected individuals to advocacy and support organizations. The release of this new online database marks a dramatic expansion in the information available for both common and rare diseases. Genetic Alliance takes a novel approach to the management of disease-specific information, calling on experts—disease advocacy organizations—to provide current, quality information on their respective conditions. This tailored information is supplemented with resources from quality federal databases, distilled through consumer-tested algorithms, and presented in an accessible interface.

Disease InfoSearch can now be used by the public to answer questions after a family member’s diagnosis, find a clinical trial, and more; by healthcare providers to offer point-of-diagnosis referral to support groups; and by the research community to identify disease advocacy organizations with registries and biobanks, and to facilitate collaboration with other organizations doing similar work.

“Compiling these resources in a single location makes it significantly easier for healthcare providers, researchers and the public to find the information they need about thousands of conditions,” said Sharon Terry, President and CEO of Genetic Alliance. For more information, please visit www.DiseaseInfoSearch.org and watch our video http://youtu.be/BzUZxQWuKvk.

PC PATIENT SUPPORT MEETING AND 2ND THICKSKIN DUATHLON JUNE 20-22, 2013 SANTA CRUZ, CALIFORNIA

PC Patient Support Meetings are filled with scientific discovery in a personal and emotional setting. If you have never attended a PC PSM, we urge you to come. If you have been before, we know you want to attend.

The ThickSkin Duathlon sponsored by TransDerm, Inc. is a major fundraising event for PC research. We invite all interested physicians and scientists to contact PC Project for details and how to participate in the PSM and/or in the Duathlon.

RECENT RELEVANT PUBLICATIONS BY IPCC MEMBERS


We also report three ‘case study’ articles published in the last quarter as Pachyonychia Congenita without genetic confirmation. We ask all reviewers to please encourage authors to obtain free genetic testing sponsored by PC Project before publishing cases as “Pachyonychia Congenita.” Often these cases are not PC at all and should be published properly.
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Eli Sprecher, MD, PhD, IPCC Chair

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Diagnostics and Genetics Team

Webmeeting 1st Wednesday monthly

Frances J.D. Smith, Chair

Members —

C. David Hansen, Leonard M. Milstone, Edel O’Toole, Eli Sprecher, Maurice van Steensel

Research and Clinical Trials Team

Annual IPCC Meeting, Webmeetings and Individual Collaborations

Roger L. Kaspar, Chair

The purpose of this team is to focus specifically on clinical trial development and assist one another in moving lab research forward to delivery to patients. The team membership fluctuates depending on specific needs and focus. Outside consultants are often involved with IPCC members where special expertise is needed. Many IPCC researchers have ongoing projects to develop therapeutics for PC. These are often featured at the annual IPCC meetings.

Physician Network

Quarterly Webmeeting, Annual IPCC Meeting

Led by the physicians on the PC Project MSAB, this network is open to all who treat or are interested in treating patients with Pachyonychia Congenita. The quarterly meetings are recorded for viewing online at any time and present current data and discussions on research and on treatments for PC.
NEW PC WEBSITE
We invite you to visit the newly designed PC Project website at www.pachyonychia.org. We hope you will find a great deal of helpful information on PC for professionals. The site also offers easy access to request consultations, case reviews or to refer patients to the PC registry for genetic testing and many other support services. All services are provided at no cost to patients, family or physicians.

IPCC Physician Quarterly Meeting August 7
12 noon MDT
Please participate in the next IPCC Physician Quarterly Webmeeting which is open to physicians interested in treating patients with PC and related disorders.

The August IPCC Webmeeting will review a number of conditions which have overlapping phenotypic features. Case reports will be presented by David Hansen, Leonard Milstone, Edel O’Toole, Eli Sprecher, and Maurice van Steensel (all members of the IPCC Genetics Team). Frances Smith will provide an overview of current genetic findings. We will also report on several upcoming clinical studies which are moving forward.

Don’t miss this opportunity. Links to the meeting will be on the website or you may email us to reserve a place at the live meeting.

PC AWARENESS AROUND THE WORLD 2013
Please donate a bit of time or some funds during June to help us spread PC Awareness around the world. If each IPCC member gave only 15 minutes to some PC Awareness effort, what a difference that would make! And please let us hear how you help increase PC awareness in 2013.
BEYOND MECHANICS

Keratins have been traditionally regarded as responsible for maintaining cell structural integrity and for conferring resilience to epithelial cells. Over the past years, it has become clear that they may play many more roles. Among these, their involvement in the regulation of inflammatory processes may be of paramount importance to our understanding of the clinical manifestations of keratinopathies such as the pain, redness and in some cases, systemic signs associated with some keratin disorders. It may also explain well-known but still puzzling anecdotal observations such as cases of epidermolysis bullosa simplex erroneously but successfully treated with corticosteroids.

A paper by Thomas Magin and colleagues recently shed further light on the relationship between inflammation and keratins. Using a KRT1 knockout mouse model, they were able to show that KRT1 deficiency is associated with a marked impairment in epidermal integrity with a concomitant increase in the expression of a number of inflammatory mediators.

Among these, IL-18 was of special interest as depletion of IL-18 partially rescued Krt1(-/-) mice. IL-18 release was shown to be mediated by caspase-1 in a Krt1-dependent manner. These data therefore provide a mechanistic basis for the well-known association of epidermal ichthyosis and other keratin disorders with clinical and pathological evidence of inflammation and epidermal barrier compromise.


IIID & DUNDEE CONFERENCES

We are fortunate to have many of the world’s leading keratin experts as part of our IPCC team. IPCC members were hosts and featured speakers at these important recent meetings held in Edinburgh and Dundee, Scotland in May 2013.

RECENT PUBLICATIONS


Abstract

Inherited keratinizing disorders are caused by mutations in the genes encoding cornified cell envelope proteins, enzymes and their inhibitors, adhesion molecules, cytoskeletal proteins and others in the epidermis. These molecules are known to regulate differentiation, proliferation and cell adhesions. Intriguingly, some keratinizing disorders show blistering skin lesions, while some inherited blistering disorders show abnormal keratinization. Therefore, hereditary keratinizing and blistering diseases are closely related and show overlapping genetic backgrounds.

In this review, we overviewed keratinizing and blistering disorders in terms of overlapping of the two disease groups. Gene mutations in desmosomal components cause striate keratoderma, Naxos disease, epidermolytic palmo-plantar keratoderma and plakophilin deficiency, which first show skin fragility and blisters and later hyperkeratosis. Gene mutations in hemidesmosomal components cause various forms of epidermolysis bullosa, some of which show hyperkeratosis on the nails, palms and soles, in addition to blister formation.

Diseases with gene mutations in calcium pump proteins are Darier disease and Hailey-Hailey disease, which show clinicopathological overlaps and develop both keratinizing and blistering skin lesions. Finally, gene mutations in epidermal keratins cause epidermolysis bullosa simplex, epidermolytic ichthyosis, superficial epidermolytic ichthyosis, epidermolytic palmo-plantar keratoderma and pachyonychia congenita/focal palmo-plantar keratoderma, which show thickening of the palms and soles with underlying blister formation.

In general, responsible proteins for diseases developing both keratinizing and blistering conditions are adhesion molecules, calcium pump proteins and keratins, but not connexins, cornified cell envelope proteins, enzymes or inhibitors. It is still unknown how particular keratinizing diseases develop blisters and vice versa.

Abstract
Pachyonychia congenita (PC) is a rare genodermatosis caused by mutations in any of the four genes KRT6A, KRT6B, KRT16, or KRT17, which can lead to dystrophic, thickened nails and focal palmoplantar keratoderma, among other manifestations. Although classically subdivided into two major variants, PC-1 (Jadassohn-Lewandowski syndrome) and PC-2 (Jackson-Lawler syndrome), according to the localization of the mutations in the KRT6A/KRT16 or KRT6B/KRT17 genes, respectively, a classification system based on the mutant gene (PC-K6a, PC-K6b, PC-K16 and PC-K17) has been recently proposed. We report a 2-year-old female patient with a history of thickened and discolored nails, small cystic papulonodules on the central face, dry, unruly and curly hair, slight palmoplantar hyperkeratosis, and nasal teeth. Both her father and paternal grandfather presented onychodystrophy, palmoplantar keratoderma, and previous excision of "sebaceous" cysts. Molecular genetic analysis of the patient revealed a missense mutation (c.1163T>C) in heterozygosity in exon 6 of the KRT17 gene, confirming the diagnosis of PC-2 (Jackson-Lawler type), or PC-17. We conclude that PC is a relatively easy and consistent clinical diagnosis, but a high index of suspicion is required if the diagnosis is to be made correctly. With this case, the authors intend to draw attention to this condition and the role of the dermatologist in the diagnosis. 


Abstract
Focal palmoplantar keratoderma (PPK) with severe pain is a hallmark of pachyonychia congenita, a rare autosomal dominant disorder involving PPK and hypertrophic nail dystrophy. Some families present focal PPK with either minimal or no nail changes. Dominant-negative mutations in any of the four identified keratin genes, KRT6A, KRT6B, KRT16 or KRT17, lead to pachyonychia congenita. However, the majority of families with focal PPK showing minimal or no nail changes do not harbor mutations in these genes. Recently, mutations of the equivalent residue have been reported in a variety of inherited diseases, including neurodegenerative diseases, corneal dystrophy and skin disorders, suggesting that this residue is vital to keratin function.


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DIAGNOSTICS AND GENETICS TEAM
Webmeeting 1st Wednesday monthly
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Members —
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Led by the physicians on the PC Project MSAB, this network is open to all who treat or are interested in treating patients with Pachyonychia Congenita. The quarterly meetings are recorded for viewing online at any time and present current data and discussions on research and on treatments for PC.

1 These physicians provide patient consultations for the International PC Research Registry (IPCRR) sponsored by PC Project
Abnormal differentiation and aberrant pigmentation have been known to often occur concomitantly, suggesting a mechanistic link between keratins and the regulation of skin pigmentation. Human diseases exemplifying this association includes epidermolysis bullosa simplex with mottled pigmentation, mostly associated with a recurrent mutation affecting the head domain of KRT5; Naegeli syndrome due to mutations in KRT5; and Dowling-Degos disease (DDD), featuring reticular pigmentation of the flexures, sometimes associated with erosive dermatitis of those regions. Heterozygous mutations resulting in haploinsufficiency for KRT5 have been shown to cause some but not all cases of DDD. How do loss-of-function mutations in KRT5 impair the normal process of epidermal pigmentation remains an enigma.

In a recent paper published online in the American Journal of Human Genetics, Li et al report that in the absence of mutations in KRT5, generalized DDD can be caused by mutations in POFUT1, which encodes protein O-fucosyltransferase 1, which is responsible for adding O-linked fucose to EGF-like repeats of Notch receptors. Notch signaling pathway plays an important role in the regulation of melanocyte development and function. Of particular interest, morpholino knockdown of pofut1 in zebrafish replicated abnormal melanin distribution observed in DDD individuals, establishing a role for this enzyme in the regulation of skin pigmentation. Finally, POFUT1 knockdown was found to reduce the expression of KRT5 in HaCaT cells, suggesting that phenotypic overlap in individuals carrying mutations in either KRT5 or POFUT1 may be due to the fact that both gene products are part of a common physiological pathway.

**Clinical Trial Progress**

1. **Topical Rapamycin for PC**
   - Roger Kaspar, CEO of TransDerm, Inc. (Santa Cruz, CA) reports that their Investigational New Drug (IND) application has been filed with the FDA for use of topical rapamycin in Pachyonychia Congenita patients.
   - Kaspar indicated that initial animal studies and a small off-label trial have shown excellent results.
   - The clinical trial will be held under the direction of Joyce Teng, MD, (Stanford University) with a planned enrollment of 15 patients. The trial will run for six months and may begin before the end of 2013 if FDA approval is received by early October.

2. **Targeted siRNA**
   - A second clinical trial is also planned for sdTD101 (an improved version of the drug used in the successful 2007 clinical trial).
   - Orphan Drug status has been granted for sdTD101 in both the USA and the EU.
   - Because injections by hypodermic needles were unbearable in the earlier trial, a new delivery system has been developed using dissolvable microneedles or PADS. The drug, sdTD101, targets a specific mutation—K6aN171K. There are only 17 patients in the world with this mutation. If the trial proves safety and efficacy, the technology developed will be available not only for the other mutations found within PC patients, but for many other disorders which require delivery of large molecules such as siRNA.
HELP WANTED

Consultations. PC Project provides a 30-60 minute telephone consultation with a qualified dermatologist for all patients who join the PC registry. Because we are now receiving from 2-4 new patients each week, we would like to engage additional physicians in this role. We compensate on a per-patient basis. If you have experience with palmo-plantar genodermatoses, please send an email with cv and discuss with us.

Authors. We are always eager to work with authors to provide solid, validated data for publications. Please contact us with your ideas — or contact us and we’ll work with you to comb the IPCRR data for ideas.

PHYSICIAN QUARTERLY MEETING — AUGUST 7

We appreciate all who joined the live webmeeting and many who have requested the link to view the recorded session. A number of conditions were reviewed which have overlapping phenotypic features with PC.

Special thanks to those presenting: David Hansen, Leonard Milstone, Edel O’Toole, Eli Sprecher, and Maurice van Steensel.

The next IPCC Quarterly Physicians webmeeting will be held November 6, 2013 at 12 noon MDT.

GLOBAL RARE DISEASE REGISTRY (GRDR)
The GRDR is a project sponsored by the Office of Rare Disease Research (ORDR) at NIH. Under the direction of Yaffa Rubenstein, patient advocacy groups with no registries were funded to create registries. Those groups with registries, such as PC Project, were invited to share their de-identified data in the GRDR to create a universal rare disease registry for researchers.

PC Project has sponsored the International PC Research Registry (IPCRR) since 2004. The registry now has data (including genetic testing data) on over 500 patients. Another 700 patients have partial information listed with the IPCRR.

PC Project first shared all Common Data Elements (CDEs) from the PC registry with the GRDR in 2012 and updated that data in 2013. Recently the GRDR invited groups to share all de-identified data (not only the specific CDEs) and PC Project submitted that data as of 1 September 2013.

There are many efforts now underway regarding registries as a means of gathering rare disease data. The IPCRR continues to be a resource for information about PC.

IMPROVING PUBLICATIONS: An Appeal to Authors and Editors and Reviewers

We understand the challenge of publishing and appreciate all those interested in preparing articles on PC, other rare disorders and related topics. Nevertheless, it is often heartbreaking for us at PC Project to read current publications which fail completely to include the most basic current information on PC. We hope these suggestions and the publication examples are helpful.

1. Genetic testing is essential. No journal should accept an article claiming to be about a case of PC when no mutation can be identified or when no genetic testing has been performed. Genetic testing is provided at no cost through PC Project to patients everywhere in the world.

2. Current citations are often ignored and old inaccurate citations repeated. All de-identified data from the IPCRR is available to review so that information such as number of cases, common characteristics of each type of PC, etc. can be easily accessed and established.

3. Single-case studies are very prone to error by associating particular characteristics of a single patient/family with a disorder.

4. Unnecessary testing of patients. For example, testing for fungus or for oral cancer is based on findings typical of PC. By a simple referral, the patient could receive genetic testing and support services.

5. Inferring that a treatment was effective for a patient even when all that was known was the initial prescription, perhaps a few weeks of treatment and no follow-up. We find most patients discontinue treatments by three months because they are ineffective, but this is almost never reported.

Publications impact patients and patient care. We hope these comments will help to improve the quality of information published about PC.
2013 PUBLICATIONS
Comment—this article has no genetic testing, cites 2004 statistics, incorrectly classifies PC types, fails to cite current publications, implies that treatment was effective although patient was ‘lost to follow-up.’ It is an example of the points on page 3 of this issue.

Abstract—Background: High-variable-frequency ultrasound is used as an imaging tool for various cutaneous disorders. We utilized this tool in pachyonychia congenita (PC) patients, who typically present with plantar hyperkeratosis and often severely debilitating pain, compared to patients with epidermolytic palmoplantar keratoderma (EPPK) and mal de Meleda (MDM). Conclusion: PC patients, as opposed to a group of patients with MDM and EPPK, displayed subepidermal blistering beneath their calluses. This finding may help in...partially explaining plantar pain as part of PC symptomatology.

Comment—based on data on over 600 genetically confirmed PC patients, there is no connection between PC and corneal dystrophy. This case is almost certainly not PC. We urge the authors to obtain complete genetic testing (free through PC Project) and to find an accurate diagnosis so a correction can be published.

Abstract—A fifteen-year-old female with pachyonychia congenita (PC) confirmed by genetic testing presented with pain and a localized subcutaneous swelling on her right back. Biopsy confirmed Ewing’s sarcoma of the posterior 10th rib.

Comment—This is PC-K17 and other cases with this mutation are found in the IPCRR registry.

Abstract: Josef Jadassohn was a pioneering dermatologist who influenced the development of his specialty in many ways.

SELECT RECOMMENDED PUBLICATIONS ON PC
Full text available on PC website.


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Life Without Keratins

What would life without keratins look like? This is the question addressed by Thomas Magin and his colleagues in a paper published last October online in the *Journal of Investigative Dermatology*. The authors of the paper generated two models. In the first of these two models, all keratin type II genes were deleted in the skin in a mosaic pattern. These mice died at 8-12 days of age.

The second model was generated by mating keratin type II negative mice with mice transgenic for K8, allowing for normal morphogenesis of internal epithelia. These mice died upon birth.

In both models, cell cytolysis was observed in basal cells. Desmosomes were fewer and smaller as compared with normal skin, suggesting that keratins are necessary to stabilize desmosomes and ensure cell-cell adhesion. In parallel, acanthosis was marked, suggesting a response to injury, possibly stimulated by intraepidermal blistering.

Of interest, a marked inflammatory response with increased expression of interleukin-1 beta was observed. The new data also suggest that keratins are able to mutually compensate for their malfunction as the phenotype of keratin II deleted mice was significantly more severe than that of single or double keratin knock out mice. In addition, the fact that the patchy keratin II deficient mice were able to survive after birth, suggest that regional and partial restoration of keratin function may be sufficient to mediate a clinically significant therapeutic effect in keratinopathies.


**FDA and IRB Approvals Granted for Topical Rapa Clinical Trial**

During October both the IND filed with the US FDA and the IRB filed with Stanford University were approved making it possible to move forward with the clinical trial.

Finding PC-related keratins in the mTOR pathway led Roger Kaspar, CEO at TransDerm to initiate a study for PC patients using oral rapamycin. (Hickerson RP, Leake D, Pho LN, Leachman SA, Kaspar RL. Rapamycin selectively inhibits expression of an inducible keratin (K6a) in human keratinocytes and improves symptoms in Pachyonychia congenita patients. *J Dermatol Sci.* 2009 Nov;56(2):82-8.) The topical formulation has been developed to avoid the side effects found with oral rapamycin in the earlier study and 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.

PC Project is currently contacting patients in the Int’l PC Research Registry (IPCCR) who have been genetically confirmed for permission to release contact information.

**IPCC 2014 MEETING**

**TUESDAY MAY 6, 2014**

**ALBUQUERQUE, NM**

**ABSTRACTS INVITED**

The organizing committee (Roger Kaspar, W.H. Irwin McLean, Frances Smith and Eli Sprecher) are pleased to invite all interested clinicians and researchers (including young investigators) to submit abstracts on completed research as well as proposals for new research or clinical studies for PC or related disorders. Please send submissions to info@pachyonychia.org.

The IPCC meetings are held yearly to review ideas and select the most promising research and studies moving forward.

**The Keratin Corner**

IPCC Chairman
Eli Sprecher MD PhD
Tel Aviv Sourasky Medical Center
elisp@tlvmc.gov.il

2386 East Heritage Way, Ste B, Salt Lake City, UT 84109 · www.pachyonychia.org · Phone 877-628-7300 · Email: info@pachyonychia.org
to the clinical trial staff for possible enrollment. The trial will last for six months and will involve seven (7) in person visits at Stanford University.

**PeDRA Hosts Impressive First Conference**

The Pediatric Dermatology Research Alliance (PeDRA) hosted its first conference at The Westin Hotel in Chicago, IL on October 17-19, 2013. The program was well-organized and included a variety of topics intended to lead to more research in pediatric dermatology. Patient advocacy groups were included in the discussions and PC Project was invited to present information on the IPCCRR PC patient registry.

From the PeDRA website (which is currently under development): “Amidst a growing sea of scientific work focused on skin disorders in adults, but only a limited amount in children, pediatric dermatologists in 2012 founded PeDRA, the Pediatric Dermatology Research Alliance. PeDRA hopes to better treat and cure dermatologic diseases in infants, children and adolescents by fostering collaborative clinical, translational, and basic science research. Thanks to generous seed funding from the Society for Pediatric Dermatology (SPD), PeDRA is establishing a multi-site, collaborative network – a platform for high quality clinical investigations and bench research.”

Because attendance was under 100, there was ample opportunity to connect with many of the participants. Also, a ‘speed dating’ version of connecting was an enjoyable adventure enabling attendees to meet many others in a short period of time.

Another excellent part of the meeting were the six-minute audio-only presentations on research proposals. These short presentations carried passion and commitment and were very informative.

We hope to continue contact with PeDRA and develop collaborative projects with PeDRA members.

**Recent Publications**


**PC Cyst Histopathology**

Samples from 12 individuals with genetically confirmed PC provided the following interesting results:

1 PC-K16 patient
   1 steatocystoma

9 PC-K17 patients
   3 steatocystomas
   6 epidermal inclusion cysts

1 PC-K6a patient
   4 verrucous cysts with isthmic and follicular germinative differentiation

1 PC-K6a patient
   1 ‘unremarkable skin’

A prospective study of PC cysts is now being planned. If interested, please contact PC Project.
INTERNATIONAL PACHYONYCHIA CONGENITA CONSORTIUM (IPCC)

Eli Sprecher, MD, PhD, IPCC Chair

The IPCC is an open membership organization for researchers and physicians who have an interest in Pachyonychia Congenita research and who agree to work together to develop PC therapeutics.

IPCC STEERING COMMITTEE 2013 —

The following have been invited to serve as the IPCC Steering Committee to assist PC Project in guiding PC Project collaborations with the IPCC.

W. H. Irwin McLean, Roger L. Kaspar, Frances Smith, Eli Sprecher

DIAGNOSTICS AND GENETICS TEAM

Webmeeting 1st Wednesday monthly
FRANCES J.D. SMITH, Chair
Members —
C. David Hansen, Leonard M. Milstone, Edel O’Toole, Eli Sprecher, Maurice van Steensel

RESEARCH AND CLINICAL TRIALS TEAM

Annual IPCC Meeting, Webmeetings and Individual Collaborations
ROGER L. KASPAR, Chair

The purpose of this team is to focus specifically on clinical trial development and assist one another in moving lab research forward to delivery to patients. The team membership fluctuates depending on specific needs and focus. Outside consultants are often involved with IPCC members where special expertise is needed. Many IPCC researchers have ongoing projects to develop therapeutics for PC. These are often featured at the annual IPCC meetings.

PHYSICIAN NETWORK

Quarterly Webmeetings, Annual IPCC Meeting

Led by the physicians on the PC Project MSAB, this network is open to all who treat or are interested in treating patients with Pachyonychia Congenita. The quarterly meetings are recorded for viewing online at any time and present current data and discussions on research and on treatments for PC.