



Pachyonychia Congenita News Brief

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Project

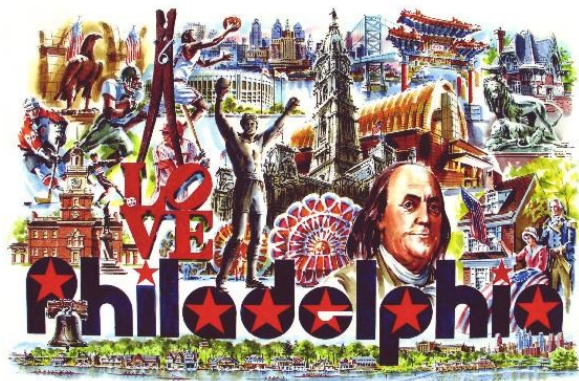
PATIENT SUPPORT MEETING— AUG 5, 6 & 7 IN PHILADELPHIA 2011

We are very excited about the upcoming Patient Support Meeting to be held August 5, 6, & 7 in Philadelphia. This is a great opportunity for you to meet other PCers and share your experience. You'll also be able to meet scientists and dermatologists specializing in keratin disorders including PC.

We have negotiated special room rates for those attending the meeting. The meeting will be held at the Philadelphia Airport Marriott. Rates are \$99 per night. You can make your reservation on-line or by calling 1-888-236-2427. In order to receive the special rate, you need to give them group code **peppepa**. To make reservations online, use the Events link on the dropdown list from the Patients page at www.pachyonychia.org. You must reserve your hotel room before July 15, 2011 to receive the special rate.

SCHOLARSHIP DEADLINE APRIL 30, 2011

Scholarships are available to those in need of financial assistance to attend the conference. The scholarship can cover up to \$500 for travel and meeting fees. If you are interested in scholarship funds please (1) register online or by returning the attached paper form (2) fill out the scholarship application either online or by returning the scholarship application attached to this newsletter. For paper applications, mail the form to PC Project 2386 E Heritage Way, Suite B, Salt Lake City, UT 84109.



GOALS FOR 2011-2012.

The newly re-organized International PC Consortium of physicians and scientists have set three goals for the coming year.

1. Publicize the disorder to the medical community and to the public. This will help you as you seek medical assistance, as you try to qualify for disability permits, as you talk with school officials. To properly publicize the disorder we need the help of PC patients.
2. Publish 'Best Practice' guidelines for PC. What works? What doesn't? What should a physician do to help those with PC? There are guidelines for care available for many disorders. There are no guidelines for PC. So, a physician has to guess what he/she thinks may be helpful. We want to make facts available so that a physician can have proper guidelines, understand the specific mechanisms of PC and better help PC patients.
3. Multi-center Clinical Studies. Only those who know their specific mutation through genetic testing will be able to participate in upcoming studies.

NATIONAL TV SHOW FEATURES DAILY LIFE OF PCER IN FRANCE

Sylvie Cierpucha was featured on a national TV show in France showing her daily life with PC (crawling to avoid painful standing, kneeling to fix dinner...) This is powerful footage. We are working to translate the audio and make this available in English.

HELP US—We need pictures and video that we can use in publicizing PC and making PC a known disorder. People with breast cancer march, wear pink ribbons and generally say 'help me!' Those with PC can do a lot for one another by sharing photos and video of themselves which we can use in publicity (faces, daily life, smiles, as well as crutches, wheelchairs, etc.)

Contact us at info@pachyonychia.org or call us 1-877-628-7300 to discuss how you can participate.

NEW CLASSIFICATION—NO PCI OR PC II

The Phenotypic and Molecular Genetic Features of Pachyonychia Congenita W.H.I. McLean, C.D. Hansen, M.J. Eliason, F.J.D. Smith. *J Invest Dermatol.* 2011 (May) This is one of 9 articles in the May JID and is the best short reference article for understanding PC. All of these articles are posted on the bibliography at www.pachyonychia.org (search 2010-2011).

“Pachyonychia congenita (PC) is an autosomal dominant genodermatosis caused by heterozygous mutations in any one of the genes encoding the differentiation-specific keratins K6a, K6b, K16, or K17. The main clinical features of the condition include painful and highly debilitating plantar keratoderma, hypertrophic nail dystrophy, oral leukokeratosis, and a variety of epidermal cysts. Although the condition has previously been subdivided into PC-1 and PC-2 subtypes, the phenotypic characterization of 1,000 mutation-verified PC patients enrolled in the International PC Research Registry, coordinated by the patient advocacy group PC Project, shows that there is considerable overlap between these subtypes. **Thus, a new genotypic nomenclature is proposed**, in which PC-6a represents a patient carrying a mutation in the K6a gene, etc. Although a rare disorder, PC represents a good model for therapy development, and international efforts are ongoing to develop and deliver siRNA, gene, correction, small molecule, and other strategies to treat this painful, disabling skin condition. The special relationship between PC Project and the PC research community has greatly accelerated the development pathway from gene identification to clinical trials in only a few years and represents a paradigm of hope for other orphan diseases.”

INT’L PACHYONYCHIA CONGENITA RESEARCH REGISTRY (IPCRR)

If you hope to have treatments and a cure for yourself or for future generations, the IPCRR registry is the first step. Please take 30 minutes to fill out the questionnaire, have the free telephone consultation with a dermatologist, and get the referral for genetic testing provided at no cost to you (a \$2000 to \$2500 value!).

PC is an ultra rare disorder. We need everyone to work together if we hope to succeed in this quest. We have about 1000 in touch with PC Project and yet only about 400 have returned questionnaires. That’s very discouraging to us. Please help us find treatments and a cure! We can do nothing without patient participation.

Thanks to all patients who have participated. Please know that YOU are the reason for the progress we are making and for the remarkable number of articles now being published about PC. Publications are the #1 tool to attract scientific research, funding for PC research and new discoveries about the disorder. It may be nice to have an article published about ‘your case’, it doesn’t push research forward in the same way as articles on multiple cases.

UPDATE YOUR IPCRR IMAGES — if it has been awhile since you sent images for the IPCRR, please don’t hesitate to send new updated images. We especially need pictures of infected feet, nails, cysts — pictures of real problems to help doctors know what to look for and how to help others.

PC WEBSITE--WWW.PACHYONYCHIA.ORG

The website has a wealth of information regarding PC. We offer the most information on Pachyonychia Congenita in the world! If you could help us prepare a tool to assist new patients in finding information on the website or if you have any tips or suggestions for the website, please email us at info@pachyonychia.org.

CARING FOR PC DVD—ADDITIONAL VIDEO NEEDED FOR CYSTS, FHK AND LEUKOKERATOSIS

The Caring for PC dvd illustrates various care techniques used by PC patients. The sections covering (1) keratoderma care (2) nail care include video from PC patients. **We need video and photos from patients** on care for cysts, follicular hyperkeratosis and leukokeratosis. Please contact us if you can help with this project.



If you would like to receive copies of this dvd for yourself, family members, physicians, school personnel or others, please contact PC Project by phone 877-628-7300 or email info@pachyonychia.org.

We want to update this dvd and continue to collect important tips for care. **Please share video or photos of your own PC care techniques.** Please help with this project—you could make such a big difference for another PC patient. Please share your information.

Philadelphia, PA — PC Patient Support Meeting 2011 Registration Form

August 5, 6, & 7, 2011

Philadelphia Airport Marriott

One Arrivals Road, Philadelphia, Pennsylvania 19153

You may complete and mail this form to PC Project or register on-line at www.pachyonychia.org/Events.

If you cannot access the meeting registration, please first complete the website registration.

MEETING FEE received before 15 June 2011 are \$100 for adults, \$75 for ages 13-17 and \$50 through age 12.

The meeting fee for any registration form or payment received after 15 June 2011
are \$110 for adults, \$82.50 for ages 13-17 and \$55 through age 12.

Last Name	First Name
Mailing Address	
Telephone	Email

Please answer the following questions for yourself and for each person who will be attending the meeting with you. If you are registering more than three people, please use additional copies as needed.

<p>First Person Your name as you would like it on your name tag: _____</p> <p>Adult: ___ YES ___ NO</p> <p>Type of Attendee: ___ Has PC ___ Family Member has PC ___ Other: _____</p> <p>Special Food Needs: ___ NO ___ YES (Please indicate) _____</p>
<p>Second Person Your name as you would like it on your name tag: _____</p> <p>Adult: ___ YES ___ NO</p> <p>Type of Attendee: ___ Has PC ___ Family Member has PC ___ Other: _____</p> <p>Special Food Needs: ___ NO ___ YES (Please indicate) _____</p>
<p>Third Person Your name as you would like it on your name tag: _____</p> <p>Adult: ___ YES ___ NO</p> <p>Type of Attendee: ___ Has PC ___ Family Member has PC ___ Other: _____</p> <p>Special Food Needs: ___ NO ___ YES (Please indicate) _____</p>
<p>Additional Comments:</p>

Philadelphia, PA — PC Patient Support Meeting 2011 Scholarship Application Form

PC Patient Support Meetings will be held each year and will alternate between USA (odd years) and UK-Europe (even years). We encourage you to apply for scholarship funding for the meeting closest to your home. To complete this form online go to <http://www.pachyonychia.org/phpQ/fillsurvey.php?sid=16>

Last Name	First Name
Mailing Address	
Telephone	Email

Those answering YES to the following questions will be given special consideration in deciding (a) whether any scholarship funds should be awarded and (b) the amount of the award.

1. This will be my first PC Patient Support Meeting. YES ___ NO ___
2. I have joined the IPCRR. I have already submitted the Questionnaire and Consent Forms to PC Project and I have scheduled or completed the consultation. YES ___ NO ___
3. Statement of Need. I will not be able to attend the Patient Support Meeting unless I receive scholarship funds. YES ___ NO ___

So that as many as possible can use the available funds, we ask that you make your request the minimum amount that you need in order to attend the meeting. Travel stipends will be limited to \$500 per PC patient.

I am applying for funding for :

PC Patient Support Meeting Fee in the amount of _____
 Travel Funds in the amount of _____
TOTAL SCHOLARSHIP FUNDS REQUESTED _____

Please list any others that will be attending the meeting with you, whether or not they have PC and what their relationship is to you:

You must submit a formal typed essay of 300 to 500 words answering "Why I hope to receive scholarship funds to attend the PC Patient Support Meeting ." You may submit the essay in English or in your own language. The essays will be judged by members of the PAB, MSAB, and Board of Trustees who will determine the award amounts. You may submit this form and your essay by mail, or you may scan this form and your essay and send via email. The deadline for scholarship applications is April 30, 2011.