Fighting for a cure.
Connecting & helping patients.
Empowering research.

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Pachyonychia Congenita Project
PO Box 17850
Holladay, Utah 84117
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May 7-8, 2019
Chicago, IL

International Pachyonychia Congenita Consortium (IPCC) Symposium

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#HeartandSole #PCisUnderOurFeet #pachyonychia
Pachyonychia Congenita Project

Founded in 2003, PC Project connects patients, researchers, clinicians and industry professionals throughout the world in a united effort to help those with Pachyonychia Congenita. PC Project maintains the International PC Research Registry (IPCRR) that provides free genetic testing and currently serves over 2,000 patients in nearly sixty countries. PC Project is dedicated to helping PC patients navigate life with a debilitating disease, all the while working to find effective treatments and ultimately, a cure.

International PC Consortium (IPCC)

Sponsored by PC Project, the IPCC is an open membership group for all scientists, physicians and industry partners with an interest in Pachyonychia Congenita patients and/or research in keratin disorders. All members agree to work together to develop effective PC therapeutics. A quarterly newsletter edited by Prof. Edel O’Toole provides IPCC members with information on recent publications, clinical trials and studies, research grants and other opportunities. To join the IPCC, email info@pachyonychia.org.

Medical & Scientific Advisory Board (MSAB)

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*Pierre Coulombe
Tracy Funk
*Philip Gard
*David Hansen
*Alain Hovnanian
Peter Hull

Roger Kaspar
Birgit Lane
Sancy Leachman
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Edel O’Toole
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Michael Polydefkis
Dennis Roop
*Frances Smith
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* Also serve on the IPCC Genetic & Steering Committee

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International PC Consortium

Ask Us About
*Data Access
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*Physician Referrals
*Publications
*Quarterly Newsletter
*Sample Access
*Grants

International PC Research Registry

Ask Us About
*Clinical Studies
*Clinical Trials
*Fact Based Data
*Free Genetic Testing
*Publications
*Questionnaire and Images
*Support Services for Patients or Physicians
L’Oreal where he was Group Leader and eventually Vice General Manager of a newly created Galderma Research entity, which he helped to establish. His first post in the Pharmaceutical Industry was with Pfizer where he specialized in the design of antifungal and anti bacterial drugs. Dr. Shroot earned his B.Sc. in Chemistry and Ph.D. in Organic Chemistry at Glasgow University, Scotland, UK. After three years of Postdoctoral Research in the Laboratory of Professor R.B Woodward he embarked on his career in the Industry as described above. He is the author and co-author of over 260 research publications and abstracts and the inventor of over 100 patents.

Eugene J. Sullivan, MD
Clinical and Regulatory Consultant
Palvella Therapeutics
Wayne, PA

Eugene J. Sullivan, MD is an independent consultant in the area of strategic regulatory and clinical drug development at EJS Consulting, LLC. Dr. Sullivan completed training in Internal Medicine at the Medical College of Virginia and in Pulmonary and Critical Care Medicine at the University of Colorado Health Sciences Center, and was a member of the Staff of the Department of Pulmonary and Critical Care Medicine at the Cleveland Clinic Foundation. Following this, he was employed at the US Food and Drug Administration where he served as a medical reviewer, medical team leader, and Deputy Director of the Division of Pulmonary and Allergy Products in the FDA’s Center for Drug Evaluation and Research. Dr. Sullivan has also held various positions in the pharmaceutical industry, including Chief Medical Officer at United Therapeutics (UT), and Chief Medical and Scientific Officer and Chief Product Strategy Officer at Insmed (current).

Notes:
Tuesday, May 7, 2019

12:00 pm  LUNCH, PC Project Welcome & Overview
Janice Schwartz

Building the Foundation for Successful PC Studies
Session chair: Edel O'Toole, MD, PhD, FRP

12:25 - 12:50  Integrating the Keratinopathies
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2:50 - 3:10  Next generation ASOs for treatment of PC
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3:15 - 3:25  Opening a window into skin structure and
function
Michael Conneely, PhD

Liat Samuelov, MD
Vice chair department of Dermatology
Tel-Aviv Medical Center
Tel-Aviv, Israel

Dr. Liat Samuelov received her MD degree from the Sackler faculty of
medicine, Tel-Aviv University and graduated Magna cum Laude in
2008. She specialized in Dermatology at the Tel-Aviv Medical center in
2009-2014. During her residency she spent almost a year at the lab of
Prof. Ralf Paus, University of Luebeck, Germany (2010-2011) performing
hair follicle research as a part of a Minerva scholarship. Following
her residency, she performed a research fellowship at the Dermatology
department lab of Prof. Richard Gallo at the University of California
San Diego (UCSD) and between 2016-2017 she completed a Pediatric
Dermatology fellowship with Prof. Amy Paller and Dr. Anthony Mancini
at Lurie Childrens’ Hospital affiliated to Northwestern University, Chi-
icago. She currently serves as the vice chair of the department of Derma-
tology, Tel-Aviv Medical Center. She has co-authored over 25 scientific
publications and several book chapters. Her research focuses on genet-
ics of skin diseases, hair disorders, Pediatric dermatology and atopic
dermatitis.

Braham Shroot, PhD
Chief Scientific Officer
Palvella Therapeutics
Wayne, PA

Dr. Shroot has over 35 years hands-on experience in Drug Research and
Development specializing in Skin disorders including Acne, Rosacea,
Psoriasis, Atopic Dermatitis Fungal Infections, Chronic Ulcers and nov-
el Aesthetic Biological Products. Dr. Schroot is currently focusing on
unmet medical needs Orphan Genodermatoses in the role of Chief Sci-
entific Officer of Palvella Therapeutics. He was the former CEO Signum
BioSciences/ Argyle Therapeutics, a private biotechnology company
dedicated to developing small molecule therapeutics for dermatological
applications. Previously CSO of Barrier Therapeutics, Inc, which was
acquired by Stiefel Laboratories Inc., in August 2008. Dr. Shroot was
CSO and Vice President of R&D for DFB Pharmaceuticals, Inc., a fully
integrated private specialty pharmaceutical company with a focus on the
skin, wound care, and surgical markets. In addition Dr Shroot had
scientific responsibility for another DFB company, DPT Laboratories a
contract research and manufacturing company with a focus on semi-
solid topical formulations. Before DFB, Dr. Shroot was employed by
minimized cell adhesion, increased inflammation and itch, using mouse and keratinocyte models. In one recent project, we work toward compound-based approaches to normalize formation of normal keratin networks. We posit that localized or systemic application of such compounds improve EBS and related keratinopathies such as PC. Since 2015, I am coordinating a German Research Council-funded priority program (SPP1782) of 22 PIs who study the role of epithelial intercellular junctions in mechanotransduction and chemical signaling.

**Edel O'Toole, MD, PhD, FRCP**
Centre Lead and Professor of Molecular Dermatology, Blizard Institute, Queen Mary University of London and Consultant Dermatologist, Royal London Hospital, London, London, England UK

Edel O’Toole is a clinical academic at Barts and the London School of Medicine and Dentistry with an active research group working on rare and common genetic skin disease biology and honorary consultant dermatologist at the Royal London Hospital, Barts Health NHS Trust. She trained in Medicine at University College, Galway, Ireland, followed by general medical and dermatology training in Dublin and London. She was a Howard Hughes Medical Institute Post-Doctoral Fellow with David Woodley at Northwestern University in Chicago from 1994–1998. Her specialist clinical interests are ichthyosis and palmoplantar keratodermas. She is the current clinical lead for the British Association of Dermatologists Dermatology and Genetic Medicine network. She is Chair of the Medical Advisory Board of the Ichthyosis Support Group, is on the steering committee of Pachyonychia Project and is actively involved in 100K Genomes, a gene discovery project within the NHS.

**Dennis Roop, PhD**
Professor of Dermatology and Director, Charles C. Gates Center for Regenerative, University of Colorado, Aurora, CO

Dennis R. Roop, PhD, is the founding Director of the Charles C. Gates Center for Regenerative Medicine. He is Professor of Dermatology and holds the Charles C. Gates Chair of Regenerative Medicine. He is also founding Director of the Gates Biomanufacturing Facility, a state-of-the-art current Good Manufacturing Practices (cGMP) facility, which opened in April, 2015 and manufactures US Food and Drug Administration (FDA) approved investigational regenerative medicine products for early phase clinical trials.

<table>
<thead>
<tr>
<th>Time</th>
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<tr>
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Session Chairs: Amy Paller, MD & Roger Kaspar, PhD

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John D. Doux, MD

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C. David Hansen, MD

12:30 pm  Close of meeting: box lunches available

12:45 pm  PC Project MSAB Meeting

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**David P. Kelsell, PhD**
Professor of Molecular Genetics, Blizard Institute, Queen Mary University of London
Whitechapel, London UK

My research largely focuses on understanding the genetic and disease mechanisms underlying monogenic skin disorders with a particular interest in keratodermas linked to cardiomyopathy, hearing loss and oesophageal cancer. My group is mainly funded by the Medical Research Council, British Heart Foundation and Cancer Research UK. I was awarded the Chanel-CERIES award for skin research in 2016. I am currently the first non-clinical president of the European Society of Dermatological Research.

**E. Birgitte Lane, PhD, FRSE, FMedSci**
Chief Scientist
Skin Research Institute of Singapore
Singapore

Birgit Lane is the Chief Scientist of the Skin Research Institute of Singapore. She has worked in Singapore for 14 years, building 2 institutes and championing skin research. She led her team to discoveries of causative mutations in EBS, PC and other genetic skin diseases, and currently works on understanding how keratin filaments function, what are the consequences of their failure, and how such knowledge can lead to strategies for therapy.

**Thomas M. Magin, PhD**
Department Chair,
Cell and Developmental Biology
University of Leipzig
Leipzig, Germany

I am chair of Cell and Developmental Biology and head of department at the University of Leipzig. We investigate the function of keratin proteins during epidermal differentiation, regeneration and disease, with a focus on Epidermolysis Bullosa Simplex (EBS). We work toward understanding how keratin mutations lead to diverse pathology, such as di-
Christopher Bunick performs structural biology research on proteins important to the normal function of human skin as well as those involved in skin disease pathogenesis. The Bunick laboratory uses a combination of x-ray crystallography and electron microscopy to examine the molecular mechanisms involved in skin barrier integrity and intermediate filament assembly. The goal is to translate these structural discoveries into new therapeutics for patients with skin disease.

Dr. Caterina received his MD and PhD from Johns Hopkins School of Medicine, then went on to a postdoctoral fellowship at UCSF, where he participated in the discovery of the capsaicin receptor TRPV1. His laboratory at Johns Hopkins is focused on molecular and cellular mechanisms underlying neuropathic and inflammatory pain sensation, using the mouse as a model system. Recent work has been focused on mechanisms that contribute to pain in mouse models of hereditary palmoplantar keratodermas.

Michael Conneely obtained his PhD in Physics from the University of Dundee where he focused on the application of ultrasound for the delivery of therapeutics throughout the body. For the past four years he has worked as part of the Hickerson/McLean team developing state-of-the-art ex vivo skin models for use in the evaluation of delivery and efficacy of therapeutic compounds. The most recent aspect of this work involves...
a collaboration with the National Phenotypic Screening Centre, a world-class facility utilising the latest in robotic and automated platforms, to design a multi-well skin culture system for use in high-content screening (HCS) applications. Dr Conneely is also engaged in several other activities including: adapting novel histology techniques for use with skin tissue, enabling new ways of investigating skin structure and responses to stimuli; and working towards improving patient comfort in their day-to-day lives by coordinating a collaboration with colleagues in Physics and Engineering evaluating the feasibility and design of cooled footwear.

Pierre A. Coulombe, PhD
G. Carl Huber Professor and Chair
Department of Cell and Developmental Biology
University of Michigan Medical School
Ann Arbor, MI

A native of Montréal, Canada, Dr. Coulombe serves as the G. Carl Huber Professor and Chair of the Cell and Developmental Biology at the University of Michigan Medical School. He holds appointments in the Department of Dermatology and the Comprehensive Cancer Center at the same institution. Dr. Coulombe joined U-M in 2017 after spending 25 years on faculty at Johns Hopkins University. He has had a long-standing interest in studying the genetic basis and mechanistic underpinnings of keratin mutation-based disorders, with a focus on epidermolysis bullosa simplex and pachyonychia congenita.

Mark P. de Souza, PhD
CEO
FIBRX Derm, Inc.
Berkeley, CA

Mark is an entrepreneur in the rare disease and dermatology space. He has cofounded and/or served as CEO or Executive Chairman of several biotech companies, including Chromaderm (melasma), Lotus Tissue Repair and FIBRX Derm (Dystrophic Epidermolysis Bullosa), PellePharm (Basal Cell Nevus Syndrome), and NFlection Therapeutics (Neurofibromatosis).

John D. Doux, MD
Analyst
Palo Alto Investors
Palo Alto, CA

John Doux obtained a BS with distinction and an MD from Stanford University, where he was a Howard Hughes Medical Institute Fellow performing and publishing research in the laboratory of Dr. David Woodley. He is board certified in dermatology following completion of training at Brigham and Women's Hospital and Stanford Medical Center, after which he maintained a solo clinical practice in medical and surgical dermatology for seventeen years. He also completed an MBA at the Wharton School of Business where he was a Palmer Scholar. Since 2004 he has also served as an analyst at Palo Alto Investors, an investment firm specializing in healthcare.

David A. Giljohann, PhD
CEO
Exicure
Skokie, IL

Dr. Giljohann has served as CEO of Exicure since 2013. Dr. Giljohann obtained his Ph.D. in 2009 from Northwestern University under the direction of Dr. Chad A. Mirkin where he developed oligonucleotide-modified nanoparticles, including NanoFlare™, and Spherical Nucleic Acid (SNA™) constructs. Dr. Giljohann has contributed to over 25 manuscripts and over 100 patents and applications.

C. David Hansen, MD
Dermatology, Professor
University of Utah
Salt Lake City, UT

David Hansen completed his residency training in dermatology at the University of Michigan then returned to the University of Utah where he joined the clinical faculty in dermatology. He is a board certified dermatologist focused on the treatment of general dermatology. He has a particular emphasis in the management of acne, eczema, psoriasis, and skin cancer. His research interests are in genetic skin disorders including Pachyonychia Congenita. In 2004, he participated in the first PC symposium involving many of the scientists who continue to work with
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PC Project. Since then, he has been privileged to continue to associate with PC Project, helping with patient consultations, patient support meetings, taking numerous biopsies and collaborating with other medical, scientific and industry professionals. He is a member of the Medical and Scientific Advisory Board, PC Project’s Board of Trustees and is currently the PI for PC Project’s International PC Research Registry.

Robyn P. Hickerson, PhD
Principal Investigator, School of Life Sciences
University of Dundee
Dundee, Scotland UK

Robyn’s primary research focuses on drug discovery for rare genetic skin disorders. A major arm of this program is focused on nucleic acid-based therapeutics – specifically the development of strategies to deliver these potential therapeutics. Her team has developed novel and state-of-the-art ex vivo human skin models required for evaluation of delivery and efficacy. With programs primarily focused on antisense therapeutics through a partnership with WAVE Life Sciences, the primary goal is to bring these molecules to the clinic within the next few years. In addition to nucleic acid-based therapeutics, her group is also interested in developing small molecules to treat genetic skin disease. To this end, her team is engaged in various collaborative projects with the Drug Discovery Unit and the National Phenotypic Screening Centre, both within the University of Dundee.

Alain Hovnanian, MD, PhD
Professor of Medical Genetics and Director of INSERM
Imagine Institute, Necker Hospital
Paris, France

Prof. Alain Hovnanian’s laboratory is based at the Imagine Institute for genetic diseases at Necker hospital in Paris. It is affiliated to the National Institute of Health and Medicinal Research (INSERM) and Paris Descartes University. His central theme of research is the genetics of severe skin diseases with high unmet medical needs. Specifically, the laboratory focuses on the study and the development of new treatments for epidermolysis bullosa, Netherton syndrome and severe palmoplantar keratoderma such as Pachyonychia Congenita and Olmsted syndrome. Hovnanian’s and Keith Choate’s laboratories from Yale University have recently identified « PERP » as a new gene for Olmsted syndrome and for erythrokeratoderma, another rare and heterogenous genetic skin condition. PERP is a component of desmosomes, which are specific structures which hold cells together in the epidermis. Finding a new

Biographies of Presenters

Christopher G. Bunick, MD, PhD
Assistant Professor of Dermatology
Yale University Department of Dermatology
New Haven, CT

Christopher Bunick performs structural biology research on proteins important to the normal function of human skin as well as those involved in skin disease pathogenesis. The Bunick laboratory uses a combination of x-ray crystallography and electron microscopy to examine the molecular mechanisms involved in skin barrier integrity and intermediate filament assembly. The goal is to translate these structural discoveries into new therapeutics for patients with skin disease.

Michael J. Caterina, MD, PhD
Professor of Neurosurgery
Johns Hopkins School of Medicine
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Michael Conneely, PhD
Postdoctoral researcher
University of Dundee
Dundee, Scotland UK

Michael Conneely obtained his PhD in Physics from the University of Dundee where he focused on the application of ultrasound for the delivery of therapeutics throughout the body. For the past four years he has worked as part of the Hickerson/McLean team developing state-of-the-art ex vivo skin models for use in the evaluation of delivery and efficacy of therapeutic compounds. The most recent aspect of this work involves
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Edel O’Toole, MD, PhD, FRCP
Centre Lead and Professor of Molecular Dermatol-
ogy, Bzizard Institute, Queen Mary University of
London and Consultant Dermatologist, Royal Lon-
don Hospital, London, London, England UK

Edel O’Toole is a clinical academic at Barts and the London School of
Medicine and Dentistry with an active research group working on rare
and common genetic skin disease biology and honorary consultant der-
matologist at the Royal London Hospital, Barts Health NHS Trust. She
trained in Medicine at University College, Galway, Ireland, followed by
general medical and dermatology training in Dublin and London. She
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**Liat Samuelov, MD**  
Vice chair department of Dermatology  
Tel-Aviv Medical Center  
Tel-Aviv, Israel

Dr. Liat Samuelov received her MD degree from the Sackler faculty of medicine, Tel-Aviv University and graduated Magna cum Laude in 2008. She specialized in Dermatology at the Tel-Aviv Medical center in 2009-2014. During her residency she spent almost a year at the lab of Prof. Ralf Paus, University of Luebeck, Germany (2010-2011) performing hair follicle research as a part of a Minerva scholarship. Following her residency, she performed a research fellowship at the Dermatology department lab of Prof. Richard Gallo at the University of California San Diego (UCSD) and between 2016-2017 she completed a Pediatric Dermatology fellowship with Prof. Amy Paller and Dr. Anthony Mancini at Lurie Childrens’ Hospital affiliated to Northwestern University, Chicago. She currently serves as the vice chair of the department of Dermatology, Tel-Aviv Medical Center. She has co-authored over 25 scientific publications and several book chapters. Her research focuses on genetics of skin diseases, hair disorders, Pediatric dermatology and atopic dermatitis.

**Braham Shroot, PhD**  
Chief Scientific Officer  
Palvella Therapeutics  
Wayne, PA

Dr. Shroot has over 35 years hands-on experience in Drug Research and Development specializing in Skin disorders including Acne, Rosacea, Psoriasis, Atopic Dermatitis Fungal Infections, Chronic Ulcers and novel Aesthetic Biological Products. Dr. Schroot is currently focusing on unmet medical needs Orphan Genodermatoses in the role of Chief Scientific Officer of Palvella Therapeutics. He was the former CEO Signum BioSciences/ Argyle Therapeutics, a private biotechnology company dedicated to developing small molecule therapeutics for dermatological applications. Previously CSO of Barrier Therapeutics, Inc, which was acquired by Stiefel Laboratories Inc., in August 2008. Dr. Shroot was CSO and Vice President of R&D for DFB Pharmaceuticals, Inc., a fully integrated private specialty pharmaceutical company with a focus on the skin, wound care, and surgical markets. In addition Dr Shroot had scientific responsibility for another DFB company, DPT Laboratories a contract research and manufacturing company with a focus on semi-solid topical formulations. Before DFB, Dr. Shroot was employed by
L’Oreal where he was Group Leader and eventually Vice General Manager of a newly created Galderma Research entity, which he helped to establish. His first post in the Pharmaceutical Industry was with Pfizer where he specialized in the design of antifungal and antibacterial drugs. Dr. Shroot earned his B.Sc. in Chemistry and Ph.D. in Organic Chemistry at Glasgow University, Scotland, UK. After three years of Postdoctoral Research in the Laboratory of Professor R.B Woodward he embarked on his career in the Industry as described above. He is the author and co-author of over 260 research publications and abstracts and the inventor of over 100 patents.

**Eugene J. Sullivan, MD**
Clinical and Regulatory Consultant
Palvella Therapeutics
Wayne, PA

Eugene J. Sullivan, MD is an independent consultant in the area of strategic regulatory and clinical drug development at EJS Consulting, LLC. Dr. Sullivan completed training in Internal Medicine at the Medical College of Virginia and in Pulmonary and Critical Care Medicine at the University of Colorado Health Sciences Center, and was a member of the Staff of the Department of Pulmonary and Critical Care Medicine at the Cleveland Clinic Foundation. Following this, he was employed at the US Food and Drug Administration where he served as a medical reviewer, medical team leader, and Deputy Director of the Division of Pulmonary and Allergy Products in the FDA’s Center for Drug Evaluation and Research. Dr. Sullivan has also held various positions in the pharmaceutical industry, including Chief Medical Officer at United Therapeutics (UT), and Chief Medical and Scientific Officer and Chief Product Strategy Officer at Insmed (current).

Notes:
Pachyonychia Congenita Project

Founded in 2003, PC Project connects patients, researchers, clinicians and industry professionals throughout the world in a united effort to help those with Pachyonychia Congenita. PC Project maintains the International PC Research Registry (IPCRR) that provides free genetic testing and currently serves over 2,000 patients in nearly sixty countries. PC Project is dedicated to helping PC patients navigate life with a debilitating disease, all the while working to find effective treatments and ultimately, a cure.

International PC Consortium (IPCC)

Sponsored by PC Project, the IPCC is an open membership group for all scientists, physicians and industry partners with an interest in Pachyonychia Congenita patients and/or research in keratin disorders. All members agree to work together to develop effective PC therapeutics. A quarterly newsletter edited by Prof. Edel O'Toole provides IPCC members with information on recent publications, clinical trials and studies, research grants and other opportunities. To join the IPCC, email info@pachyonychia.org.

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International PC Consortium

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International PC Research Registry

Ask Us About
*Clinical Studies
*Clinical Trials
*Fact Based Data
*Free Genetic Testing
*Publications
*Questionnaire and Images
*Support Services for Patients or Physicians

PC data gains
Increased researchers
Patient joins registry
Quality publications
Clinical Trials for PC
Fighting for a cure.
Connecting & helping patients.
Empowering research.

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May 7-8, 2019
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International Pachyonychia Congenita Consortium (IPCC) Symposium

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