Meeting Schedule Quick Reference

Tuesday
Session 1  Presentations & Biographical Sketches
Session 2  Horizons in PC Research

Session 2  Translational horizons

Wednesday
Session 3  Presentations & Biographical Sketches
Session 3  Scanning the PC pain and itch horizons
Guest Lecture  Howard Chang
Session 4  Towards new horizons

Attendee Contact Information
Poster Summaries

Meeting Sponsors

Pachyonychia Congenita Project
National Institutes of Health
The Wellcome Trust

Pachyonychia Congenita Project
Fighting for a cure. Connecting & helping patients. Empowering research.

PC Project
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www.pachyonychia.org
12:00 PM  Lunch and welcome reception

**Session 1**  **Horizons in PC research**
1:30 PM  Irwin McLean, PC Project/University of Dundee, Scotland UK
*Broadening our horizons to tackle the problem of keratoderma*

2:00 PM  **Keynote Lecture** - Edel O’Toole, Queen Mary University of London, England UK
*The clinical horizon - similarities and differences between PC and other PPKs*

2:45 PM  **Keynote Lecture** - Eli Sprecher, Tel Aviv Sourasky Medical Center, Tel Aviv, Israel
*The molecular horizon - the genes and proteins involved in PC and PPKs*

3:30 PM  Break

**Session 2**  **Translational horizons**
4:00 PM  Pierre Coulombe, Johns Hopkins Bloomberg School of Public Health, Maryland USA
*Mouse models of PC and PPK*

4:30 PM  Dennis Roop, University of Colorado Denver, Colorado USA
*Cellular reprogramming and gene editing therapy in genodermatology*

5:00 PM  Wesley Kaupinen, Palvella Therapeutics, Pennsylvania USA
*Topical Rapamycin: Update on PC Project & Palvella Therapeutics collaboration to rapidly
advance towards the next clinical study*

5:30 PM  Bishr Omary, Univeristy of Michigan Ann Arbor, Michigan USA
*Small molecule approaches to treating pathogenic keratin aggregation*

6:45 PM  Meet in Martin Luther King Jr Lobby of Convention Center for transport to dinner

7:00 PM  **IPCC Conference Dinner**
Portland City Grill, 111 SW Fifth Avenue, 30th Floor
8:00 AM  Breakfast buffet

**Session 3**  Scanning the PC pain and itch horizons
9:00 AM  Michael Caterina, Johns Hopkins Medicine, MD, USA
*Pain in PC and other PPKs: Clinical and Basic Science Insights.*

9:30 AM  Thomas Magin, University of Leipzig, Germany
*From inflammation to therapy: novel insights from cell biology*

10:00 AM  Break

10:15 AM  **Special Guest Lecture - Howard Chang, Stanford University, California USA**

*How does palmoplantar epidermis know that it is palmoplantar?*
*Molecular mechanisms controlling development & maintenance of ridged skin*

11:00 AM  Break

**Session 4**  Towards new horizons - brainstorming session
11:30 AM  Group discussion
Themes:  Pain in PC
Therapeutic tractability of PPKs - what are the low-hanging fruit?
Drug repurposing in PPKs - genes and screens
Oligonucleotide therapeutics for PPKs
2018 meeting - onwards and upwards

12:30 PM  Box lunch available and discussion continues

1:00 PM  Close of meeting
Irwin McLean was born in Northern Ireland, did his BSc and PhD degrees at The Queen's University of Belfast and is Professor of Genetic Medicine at The University of Dundee, Scotland. Irwin and his collaborators have identified a large number of skin disease genes, especially in the field of keratinizing disorders, with about 300 publications this area. Irwin also has close links with the National Health Service and holds Honorary Consultant Clinical Scientist positions in both Human Genetics and in Dermatology. He is an elected Fellow of the Royal Society of Edinburgh (2005), a Fellow of the Academy of Medical Sciences (2009), a Fellow of the Royal Society (2014) and a Member of the Academia Europaea (2016). He has a long-standing interest in development of therapy for pachyonychia congenita (PC) and epidermolysis bullosa simplex (EBS) and holds program-level grants from The Wellcome Trust, Medical Research Council and industrial sources to support this research. Irwin has recently taken on the role of Chief Executive Officer and Chief Scientific Officer of Pachyonychia Congenita (PC) Project.

Tuesday 25th April 2017
1:30 PM Irwin McLean, PC Project/University of Dundee, Scotland, UK
Broadening our horizons to tackle the problem of keratoderma
Edel O’Toole obtained her medical degree at University College Galway and subsequently trained in Medicine and Dermatology in Galway and Dublin, Ireland. She acquired expertise in keratinocyte biology at Northwestern University, Chicago, where she was a Dermatology Foundation and then a Howard Hughes Medical Institute Physician Post-doctoral Fellow with David Woodley from 1994-1998. She completed her clinical training in Dermatology in London and became a Senior Lecturer in the Centre for Cutaneous Research at Barts and the London School of Medicine and Dentistry in 2001. She was promoted to Professor of Molecular Dermatology in 2008. She was Chair of the British Society for Investigative Dermatology from 2009-2011 and was awarded the Parkes-Weber medal by the Royal College of Physicians (London) in 2012. In 2015, she became Centre Lead of the Centre for Cell Biology and Cutaneous Research, Blizard Institute, Barts and the London. She has diverse research interests including keratinocyte and cancer biology, the basement membrane zone, genetic skin diseases and stem cells. Her clinical interests include the keratodermas and ichthyosis. She is on the medical and scientific advisory board of Pachyonychia Project and the Ichthyosis Support Group.

Tuesday 25th April 2017
2:00 PM  **Keynote Lecture** - Edel O’Toole, Queen Mary University of London, England, UK
*The clinical horizon - similarities and differences between PC and other PPKs*
Eli Sprecher was born in Waterloo, Belgium. He moved to Israel in 1981, received his MD and PhD degrees from the Hebrew University of Jerusalem. He specialized in dermatology at the Rambam Medical Center and spent a post-doc fellowship at Thomas Jefferson University, Philadelphia. He is now Chair of the Department of Dermatology at the Tel Aviv Medical Center and Professor of Dermatology at the Tel Aviv University. He has published over 200 peer-reviewed publications and mentored over 40 students. His research interest is in the molecular genetics of skin diseases.

Tuesday 25th April 2017
2:45 PM  **Keynote Lecture** - Eli Sprecher, Tel Aviv Sourasky Medical Center, Tel Aviv, Israel
*The molecular horizon - the genes and proteins involved in PC and PPKs*
Pierre A. Coulombe, a native of Montréal, Québec, Canada, serves as the E.V. McCollum Professor and Chair of the Department of Biochemistry and Molecular Biology at the Bloomberg School of Public Health of the Johns Hopkins University since 2008. He is jointly appointed in the Departments of Dermatology, Oncology, and Biological Chemistry at the Johns Hopkins School of Medicine, and is a member of the Sidney Kimmel Comprehensive Cancer Center. As of August 1, 2017, Dr. Coulombe will move to the University of Michigan as the Carl Huber Professor and Chair of the Department of Cell and Developmental Biology in the School of Medicine. He has played key roles in the identification of the first intermediate filament-based disease, in defining the vital role of mechanical support fulfilled by keratin filaments in epithelial cells, and in the discovery of novel, non-canonical functions for keratin proteins. Recently, his laboratory has been studying the role of keratin proteins in regulating inflammatory and immune responses in skin epithelia, with direct implications for the pathogenesis of several types of complex disease traits including cancer. Because of his focus on keratins 6, 16, and 17, Dr. Coulombe’s research has direct relevance to pachyonychia congenita and related disorders. In addition to research and discovery efforts, Dr. Coulombe has been very active in the education and training of graduate students and postdoctoral fellows, and also in faculty recruitment and mentoring.

Tuesday 25th April 2017
4:00 PM Pierre Coulombe, Johns Hopkins Bloomberg School of Public Health, MD, USA

*Mouse models of PC and PPK*
**Dennis R. Roop** received a B.A. degree in Biology from Berea College, Berea, KY in 1969 and a M.S. and Ph.D. in Microbiology from the University of Tennessee, Knoxville, TN in 1972 and 1977, respectively. Following a postdoctoral fellowship at Baylor College of Medicine, Houston, TX (1977 to 1980), he was recruited to the National Cancer Institute, National Institutes of Health, Bethesda, MD, where he rose to the rank of Senior Investigator. In 1988, he was recruited back to Baylor College of Medicine, where he held the positions of Professor of Molecular and Cellular Biology and Dermatology and Director of the Center for Cutaneous Molecular Biology until the end of 2006. In January, 2007, he was recruited to the University of Colorado Anschutz Medical Campus as 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 also serves as 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. He has authored more than 300 peer-reviewed articles. His research program has been continuously funded by the National Institute of Arthritis and Musculoskeletal and Skin Diseases, and the National Cancer Institute since 1989.

**Tuesday 25th April 2017**

4:30 PM  Dennis Roop, University of Colorado Denver, CO, USA

*Cellular reprogramming and gene editing therapy in genodermatology*
Wesley Kaupinen is the Founder and CEO of Palvella Therapeutics, a specialty biopharmaceutical company centered on relentlessly serving individuals suffering from rare diseases that is currently developing a novel topical formulation of sirolimus for the treatment of pachyonychia congenita and other rare skin diseases. Prior to founding Palvella, Wes was Senior Vice President, Corporate Development and Commercialization at Insmed, a publicly traded biopharma company focused on the treatment of rare lung diseases where he had responsibility for leading the corporate development and commercial functions, including patient advocacy, marketing, and business development. Wes previously held roles with venture capital firms Apax Partners and Quaker Partners where he was involved as a founding investor or Board member for several entrepreneurial pharmaceutical and medical device companies. Mr. Kaupinen has an MBA from The Wharton School where he was the recipient of the William L. Kissick M.D. Scholarship Award, and a B.A. in Economics from The University of Virginia.

Tuesday 25th April 2017
5:00 PM Wesley Kaupinen, Palvella Therapeutics, PA, USA
   Topical Rapamycin: Update on PC Project & Palvella Therapeutics collaboration to rapidly advance towards the next clinical study
**Bishr Omary** is Professor and Chair of Physiology at the University of Michigan. A major focus of his laboratory for more than 25 years has been to study the regulation, disease association, and function of intermediate filaments in digestive organs, with a major emphasis on keratins and lamins. A more recent focus is aimed at using high throughput drug screening to identify potential therapies for keratinopathies.

Tuesday 25th April 2017
5:30 PM    Bishr Omary, University of Michigan Ann Arbor, MI, USA

*Small molecule approaches to treating pathogenic keratin aggregation*
Michael Caterina, M.D., Ph.D. is the Solomon H. Snyder Professor of Neurosurgery, and Professor of Biological Chemistry and Neuroscience at Johns Hopkins School of Medicine. He is also the Director of the Neurosurgery Pain Research Institute at Johns Hopkins. Dr. Caterina earned his B.S. from Penn State University and M.D. and Ph.D. degrees from Johns Hopkins. During his postdoctoral fellowship at the University of California San Francisco, he discovered the ion channel protein, TRPV1, which is the receptor for capsaicin as well as a sensor of painfully hot temperatures. Dr. Caterina and his colleagues have studied extensively the contributions of heat-stimulated ion channels to heat and pain sensation. They have also discovered that non-neuronal epithelial cells such as keratinocytes can play a role as “first responders” to environmental stimuli and can communicate the presence of these stimuli to the nervous system. Their most recent work, in collaboration with Drs. Pierre Coulombe and Michael Polydefkis at Johns Hopkins, has focused on understanding the mechanisms underlying pain sensation in palmoplantar keratodermas.

Wednesday 26th April 2017
9:00 AM Michael Caterina, Johns Hopkins Medicine, MD, USA
Pain in PC and other PPKs: Clinical and Basic Science Insights.
Thomas Magin is the head of the department of Biology and Professor of Cell and Developmental Biology at the University of Leipzig since 2010. His research investigates molecular mechanisms controlling epithelial differentiation, regeneration and pathogenesis. Specifically, his lab focuses on the function of the epithelial keratin cytoskeleton to understand how keratin mutations contribute to pathomechanisms in keratinopathies and how this knowledge can be exploited to develop molecular therapies. Since 2015, Dr Magin has been coordinating a German Research Council-funded network of 20 PIs who take a multidisciplinary approach to study the role of epithelial intercellular junctions in mechanotransduction and chemical signaling.

Wednesday 26th April 2017
9:30 AM    Thomas Magin, University of Leipzig, Germany
From inflammation to therapy: novel insights from cell biology
Howard Y. Chang M.D., Ph.D. is Director of the Center for Personal Dynamic Regulomes and Professor of Dermatology at Stanford University School of Medicine. Chang earned a Ph.D. in Biology from MIT, M.D. from Harvard Medical School, and completed Dermatology residency and postdoctoral training at Stanford University. His research addresses how large sets of genes are turned on or off together, which is important in normal development, cancer, and aging. Chang discovered a new class of genes, termed long noncoding RNAs, that control gene activity throughout the genome, illuminating a new layer of biological regulation. He has invented new methods for defining the shapes of RNA and DNA genome-wide. The long term goal of his research is to decipher the regulatory information in the genome to benefit human health. Dr. Chang's honors include the Judson Daland Prize of the American Philosophical Society, Howard Hughes Medical Institute Early Career Scientist, the Vilcek Prize for Creative Promise, Alfred Marchionini Research Prize, American Cancer Society Research Scholar Award, Damon Runyon Scholar Award, and elected membership to the American Society for Clinical Investigation. His work was honored by the journal Cell as a Landmark paper over the past 40 years and by Science as “Insight of the decade”.

Wednesday 26th April 2017
10:15 AM  Special Guest Lecture - Howard Chang, Stanford University, CA, USA
How does palmoplantar epidermis know that it is palmoplantar?
Molecular mechanisms controlling development and maintenance of ridged skin
Wednesday 26th April 2017
11:30 AM    Group discussion
Themes:      Pain in PC
             Therapeutic tractability of PPKs - what are the low-hanging fruit?
             Drug repurposing in PPKs - genes and screens
             Oligonucleotide therapeutics for PPKs
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