

# International Pachyonychia Congenita Consortium (IPCC) Symposium



## Innovations and Transformations

June 28-29, 2021

A virtual meeting



# Monday, June 28, 2021

**8:00-11:00 MDT**

8:00 am *Welcome from PC Project*

**Janice Schwartz, Executive Director, PC Project**

## **The Innovation Day**

**Chair: Eli Sprecher MD, PhD**

08:05 am *How Desmosomes Help Drive Tissue Form and Function*

**Kathleen Green, PhD**, Northwestern University Feinberg School of Medicine

08:20 am *Mouse PPK models*

**David P. Kelsell, PhD**, Queen Mary University of London

08:35 am *A rationale for the interplay between expression and function for intermediate filament proteins*

**Pierre A. Coulombe, PhD**, University of Michigan

08:50 am *Spatial transcriptomics of iRhom2 mouse model paw skin*

**Diana Blaydon, PhD**, Queen Mary University of London

09:05 am *Mitochondria and lysosomes in PC pathogenesis*

**Sonja Lehmann**, RWTH Aachen University

09:20 am *Coffee/Tea/Comfort break*

09:35 am *Biomechanical analysis of the keratin cytoskeleton and its role in cellular mechano-sensing*

**John Connelly, PhD**, Queen Mary University of London

09:50 am *ASPRV1 mutations cause dominantly inherited ichthyosis*

**Lynn Boyden, PhD**, Yale University School of Medicine

10:05 am *Structural modeling guided understanding of genotype-phenotype correlation in PC*

**Joyce Teng, MD, PhD, and Christopher Bunick, MD, PhD**, Stanford University and Yale University

10:20 am *Open discussion Basic research – Hypes and Gaps*

**Moderator: David Kelsell PhD**, Queen Mary University of London

10:35 am *Open discussion: Between clinical and basic research: bio-banking*

**Moderator: Eli Sprecher MD, PhD**, Tel Aviv Medical Center

11:00 am *Wrap-up*

**Janice Schwartz**



## Tuesday, June 29, 2021

### 8:00-11:00 MDT

8:00 am *Welcome from PC Project*  
**Janice Schwartz, Executive Director, PC Project**

### The Transformation Day

Chair: **Edel O'Toole, MD, PhD, FRCPI, FRCP**

08:05 am *Revisiting pachyonychia congenita: a case cohort study in 815 patients*

**Liat Samuelov, MD**, Tel Aviv Medical Center

08:20 am *Phenotypic and pathogenetic heterogeneity in desmoglein 1 deficiencies*

**Akiharu Kubo, MD**, Keio University School of Medicine

08:35 am *Prevalence and characterisation of itch in PC*

**Lloyd Steele**, Academic Clinical Fellow in Dermatology at Barts Health NHS Trust, London

08:45 am *Patient-reported quality of life differentials in PC management*

**Albert G. Wu, MD Candidate**, New York Medical College and **Shari Lipner, MD, PhD**, Weill Cornell Medicine, Department of Dermatology

09:00 am *Co-existence of PC and hidradenitis suppurativa: more than a coincidence.*

**Mor Pavlovsky, MD**, Tel Aviv Medical Center

09:15 am *Coffee/Tea/Comfort break*

09:30 am *Toward a chemical compound-based therapy for EBS*

**Thomas M. Magin, PhD**, Leipzig University

09:45 am *A new case series of Olmsted syndrome subjects confirms EGFR activation and that targeted systemic EGFR inhibition shows remarkable efficiency and acceptable side effects.*

**Alain Hovnanian, MD, PhD**, Imagine Institute, Necker Hospital, Paris

10:00 am *Are fully-modified therapeutic siRNAs the answer for PC?*

**Roger Kaspar, PhD**, Ayni Therapeutics

10:15 am *Phase 2/3 Trial – Next steps and what's been learned for future clinical studies for PC*

**Wes Kaupinen**, Palvella Therapeutics

10:35 am *Open discussion. Clinical research in PC – in/out-comes*

**Moderator: Joyce Teng, MD, PhD**, Stanford University

11:00 am *Wrap-up*

**Janice Schwartz**

