



## Thursday February 14

- 4:00 pm Registration – Yarrow Hotel Lobby  
 6:30 pm Hosted Social Hour – Mountain View Room, Yarrow Hotel  
 7:00 pm Dinner – Mountain View Room, Yarrow Hotel  
*Welcome* Pierre Coulombe, Dennis Roop, Mary Schwartz (Meeting Co-chairs)  
*Keynote Introduction* Sherri Bale, GeneDx, Gaithersburg, MD, USA  
*Keynote Speaker* Sharon Terry, Genetic Alliance, Washington DC, USA

## Friday February 15

- 7:00 am **Breakfast buffet**—Mountain View Room  
**Introduction** Meeting Sessions - Ivers, Hearst & Kearns Room  
 7:50-8:10 am *20/20 Vision: PC Project and the IPCC*  
 Eli Sprecher, Chair, International PC Consortium (IPCC), Tel Aviv, Israel  
 Mary Schwartz, Pachyonychia Congenita Project, Salt Lake City, UT, USA  
 8:10-8:15 am *IPCRR: Case Studies*  
 8:15-8:30 am *Pachyonychia Congenita: What We Know 2013*  
 Edel O'Toole, Barts and the London School of Medicine and Dentistry,  
 Queen Mary University of London, London, England UK

### Session 1 – Preclinical

- Chair* Dennis Roop, University of Colorado, Aurora, CO, USA  
 8:35-8:50 am *Genetics of Pachyonychia Congenita*  
 Frances J.D. Smith, University of Dundee, Dundee, Scotland UK  
 8:55-9:15 am *Palmoplantar keratoderma: A role for exaggerated alarmin response?*  
 Pierre A. Coulombe, Johns Hopkins University, Baltimore, MD, USA  
 9:20-9:40 am *Mouse models of keratoderma and for optimizing siRNA delivery*  
 W. H. Irwin McLean, University of Dundee, Dundee, Scotland UK  
 9:45-10:15 am **Break**  
 10:20-10:35 am *TRPV channels make skin sensational*  
 Michael J. Caterina, Johns Hopkins University, Baltimore, MD, USA  
 10:40-10:55 am *Itchy Skin: Scratching for an explanation*  
 Gil Yosipovitch, Wake Forest Health Sciences, Winston-Salem, NC, USA  
 11:00-11:15 am *Intravital visualization of mutant keratin 6a inhibition by siRNA in mouse skin*  
 Robyn P. Hickerson, TransDerm, Santa Cruz, CA, USA  
 11:20-11:35 am *RNA profiling of PC skin*  
 Yu-An Cao, TransDerm, Santa Cruz, CA, USA  
 11:40-11:55 am *Corneocyte proteomic profiles - possible application to skin disease?*  
 Robert H. Rice, University of California Davis, Davis, CA, USA  
 12:00-1:00 pm **Working Lunch**—Mountain View Room  
 Wyatt Felt, Ryan Butler, BYU Capstone Team Members, Provo, UT, USA  
 Nathan Schwartz, PC patient, Salt Lake City, UT, USA

### Afternoon

- Recreation/free time: Ski or alternate recreation**  
 5:30 pm Social hour – Skyline Bar, Yarrow Hotel (no host)  
 6:15 pm Bus leaves for dinner (bus will begin loading at 6:00 pm)  
 7:00 pm Dinner – downtown Salt Lake City (no alcohol served at site)  
*Remarks* Carl Baker, NIH/NIAMS

## Saturday February 16

- 7:00 am **Breakfast buffet**—Mountain View Room
- Introduction** Meeting Sessions - Ivers, Hearst & Kearns Room
- 7:50-8:00 am *Vision 2020*  
Eli Sprecher, Chair, International PC Consortium (IPCC), Tel Aviv, Israel  
Mary Schwartz, Pachyonychia Congenita Project, Utah, USA
- Session 2 – Clinical**
- Chair* C. David Hansen, University of Utah, Salt Lake City, UT, USA
- 8:05-8:20 am *Dyskeratosis congenita: a rare but highly informative telomere biology disorder*  
Sharon A. Savage, NIH-NCI, Rockville, MD, USA
- 8:25-8:40 am *Connexin-mediated skin disease*  
Maurice A. M. van Steensel, University Hospital, Maastricht, The Netherlands
- 8:45-9:00 am *A Soft Story*  
Eli Sprecher, Tel Aviv Sourasky Medical Center, Tel Aviv, Israel
- 9:05-9:20 am *Topical Rapamycin for genetic skin disease and transepidermal delivery*  
Joyce M. Teng, Stanford University, Palo Alto, CA, USA
- 9:25-9:40 am *PCQoL - A Quality of Life Assessment Measure for Pachyonychia Congenita*  
Mariam Abbas, University of Saskatchewan, Saskatchewan, Canada
- 9:45-10:00 am *Comparative study of high resolution multifrequency ultrasound of the plantar skin*  
Ilan Goldberg, Tel-Aviv Sourasky Medical Center, Tel Aviv, Israel
- 10:05 – 10:30 am **Break**
- 10:35 – 10:55 am *Regulatory requirements for human clinical trials*  
Ajit Simh, Shiba Biotechnology Consultants, La Jolla, CA, USA
- 11:00 – 11:20 am *Preclinical and clinical development of an antisense for skin scarring*  
Nicholas M. Dean, Nicholas Dean Consulting, Encinitas, CA, USA
- 11:25 – 11:55 am *Upcoming sd101 and topical rapamycin clinical trials*  
Tycho J. Speaker, Roger Kaspar, TransDerm, Inc, Santa Cruz, CA, USA
- 12:00-1:00 pm **Working Lunch**—Mountain View Room
- Introduction* E, Birgit Lane, Institute of Medical Biology, Singapore
- Speaker* John D. Doux, Palo Alto Investors, California, USA
- Afternoon** **Recreation/free time: Ski or alternate recreation**
- 4:00-6:00 pm** *Discussion Session: Planning Goals for IPCC - 2013 Forward*  
*Chair* Leonard M. Milstone, Yale University, New Haven, CT, USA  
Open to all; attendance optional
- 6:00 pm Social hour—Skyline Bar, Yarrow Hotel (no host)
- 6:30 pm Leave hotel for Payday Lift, Park City Mountain Resort
- 7:00 pm Sleigh Ride to dinner
- 7:30 pm Dinner at Snowed Inn, Park City, UT

## Sunday Feb 17

- 9:30 am Bus leaves for Homestead Resort, Midway, UT
- 10:00-12:15 pm Closing Brunch at Homestead Resort
- Remarks* Sancy Leachman, University of Utah, Salt Lake City, Utah, USA