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

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