



## Pachyonychia Congenita 1 (PC 1) Retreat Agenda

Sponsored by the Pachyonychia Congenita Project

**Goals** Brainstorm to develop a strategy for advancement of a cure for PC 1 using a disease-specific model for this orphan condition  
Discuss development of PC 1 research registry for future use in preparation for clinical trial opportunities  
Introduce newly established PC 1 foundation and potential funding opportunities new incentive-based models of support

**Host** PC Fund represented by Mary and Jan Schwartz

**Organizers** Sancy Leachman  
Patti Champine

**Thursday, February 12, 2004** Arrival in Salt Lake City, UT, Drive to Park City, Check In at Yarrow Hotel

5:00–8:00 pm Welcome by Sancy, Mary and Jan with complimentary beverages in the Pub at the Yarrow Hotel

**Friday, February 13, 2004**

7:00-8:00 am Breakfast buffet for guests, spouses and families in the Summit B Ballroom

**Meeting Location: Summit A Ballroom**

8:00-8:15	Sancy Leachman	Goals of each meeting section, introduction of Seed Grants, Career Development Award and website
8:15-8:45	Philip Fleckman	Brief description of PC 1 and Ichthyosis registry: the next step toward a PC1 clinical trial
8:45-9:05	Frances Smith	Molecular genetics and diagnosis of PC
9:05-9:20	Maurice van Steensel	Gap junctions and Pachyonychia
9:20-9:50	Discussion	PC Registry Questionnaire Part I
9:50-10:20	Break	Review Physical Findings section of Questionnaire with refreshments served outside Summit A Ballroom
10:20-10:50	Discussion	PC Registry Questionnaire Part II
10:50-11:20	Dennis Roop therapy in humans	Cells, Mice or other animals: the fastest track to PC 1
11:20-11:40	Pauline Wong	Insights into the phenotypic complexity of PC through transgenic mouse studies
11:40-11:50	Collect boxed lunches	Boxed lunches will also be available for spouses and families to pick-up outside Summit A Ballroom.
11:50-1:00	Discussion over lunch	Which available model systems are the most promising?
1:00-5:00	Recreation/free time	ski or alternate recreation rentals and lift tickets available
5:00-6:00	Cocktails in Summit B Ballroom (Hosted)	spouses and families welcome

6:00 Dinner in Summit B Ballroom (Hosted) spouses and families welcome  
Introduction to PC Project by Mary & Jan Schwartz assisted by Sam, Nate and Spencer and possibly other 'Schwartz kids'

### Saturday, February 14, 2004

7:00-8:00 am Breakfast buffet for guests, spouses and families in the Summit B Ballroom

#### Meeting Location: Summit A Ballroom

8:00-8:15	Sancy Leachman	Goals for today's session
8:15-8:45	Irwin McLean	Introduce potential options to treat PC 1 (HR, RNAi, ribozymes, transcriptional expression regulation. Which is the most promising and why.
8:45-9:00	Leonard Milstone	Triplex-forming oligonucleotides as potential gene correction or gene knockout therapeutics
9:00-9:15	Alfred Lewin	AAV-vectored ribozyme for treating dominant genetic disease
9:15-9:30	Markus Landthaler	RNAi technology as a tool to discover clinically relevant treatment options
9:30-9:45	Mario Capecchi	Clinical utility of homologous recombination in the treatment of autosomal dominant disorders
9:45-10:00	Olga Igoucheva	Oligonucleotide-mediated gene repair: potential applications to human skin disorders
10:00-10:15	Roger Kaspar	RNA Lassos in the treatment of autosomal dominant disorders
10:15-10:30	Break	Refreshments served outside Summit A Ballroom
10:30-11:00	Bhaskar Thyagarajan	Barriers to "gene therapy" delivery to skin
11:00-11:10	Collect boxed lunches	Boxed lunches will also be available for spouses and families to pick-up outside Summit A Ballroom.
11:10-1:00	Discussion over lunch	Which technologies will permit the earliest possible clinical trial?
1:00-5:15	Recreation/free time	– ski or alternate recreation rentals available
5:15-5:30	Meet in the hotel lobby	for ride to Payday Lift at Park City Mountain Resort Spouses and families invited
5:30-6:00	Sleigh ride to Snowed Inn	for dinner (Hosted)
6:00-8:45	Dinner and return sleigh ride,	transportation back to the Yarrow Hotel

### Sunday, February 15, 2004

9:15 am Meet in the hotel lobby to board shuttle for ride to Midway for brunch, spouses and families welcome

10:00-12:15 Brunch at Simon's Restaurant at Midway and return ride to Yarrow hotel

12:30 Recreation/free time ski passes or alternate recreation rentals and lift tickets available

### Monday, February 16, 2004

6:00-9:00 am Breakfast buffet (in restaurant) or boxed breakfasts to go (requires 24 hr notice to hotel) spouses and families welcome

6:30-9:00 Departure travel

