

First NIH Dyskeratosis Congenita Clinical Research Workshop

September 19, 2008

- 7:30-8:00 am **Registration and Continental Breakfast**
- 8:00-8:10 am **Welcome and Meeting Overview**
- 8:10-8:40 am **Keynote Address: History and Characterization of Dyskeratosis Congenita**
Professor Inderjeet Dokal, Barts and The London School of Medicine and Dentistry
- 8:45-10:30 am **Clinical Findings in Dyskeratosis Congenita**
- **Description of the NCI cohort:** Dr. Sharon Savage, NCI, NIH
 - **Telomere length as a diagnostic test:** Dr. Blanche Alter, NCI, NIH
 - **Oral Findings:** Dr. Demetrio Domingo, NIDCR, NIH
 - **Ocular Findings:** Dr. Ekaterina Tsilou, NEI, NIH
 - **Central Nervous System Findings:** Dr. Neelam Giri, NCI, NIH
 - **Pulmonary fibrosis:** Dr. Mary Armanios, Johns Hopkins University
 - **Skin Findings and Skin Cancer Prevention:** Dr. Ed Cowen, NIH
- 10:30-11:00am **Break**
- 11:00-12:00 pm **Genetic characterization of DC**
- **Genetic Testing:** Dr. Sharon Savage, NCI, NIH
 - **Studies of Dyskerin, the DKC1 Gene:** Dr. Judy Wong, University of British Columbia
 - **Telomerase Mutations in DC:** Dr. Neal Young, NHLBI, NIH
 - **Functional Characterization of *TINF2* mutations:** Dr. Geraldine Aubert, British Columbia Cancer Research Center
- 12:00-12:45 pm **Management of DC**
- **Medical Therapies and Surveillance:** Dr. Blanche Alter, NCI, NIH
 - **Bone Marrow Transplantation:** Dr. Paul Orchard, University of Minnesota
 - **Head and Neck Cancer Screening:** Dr. Carter Van Waes, NIDCD, NIH
- 12:45-1:45 pm **Lunch** (on your own, there is a cafeteria in the building)
- 1:45-4:30pm **Concurrent Meetings**
- 1) Family Support Group Discussions
 - 2) Scientific Collaboration Opportunities
- 4:30-5:00 pm **Meeting Summary**
- 5:00pm **Adjourn**