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

10:30-11:00 am  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)

1:45-4:30 pm  Concurrent Meetings
1) Family Support Group Discussions
2) Scientific Collaboration Opportunities

4:30-5:00 pm  Meeting Summary

5:00 pm  Adjourn