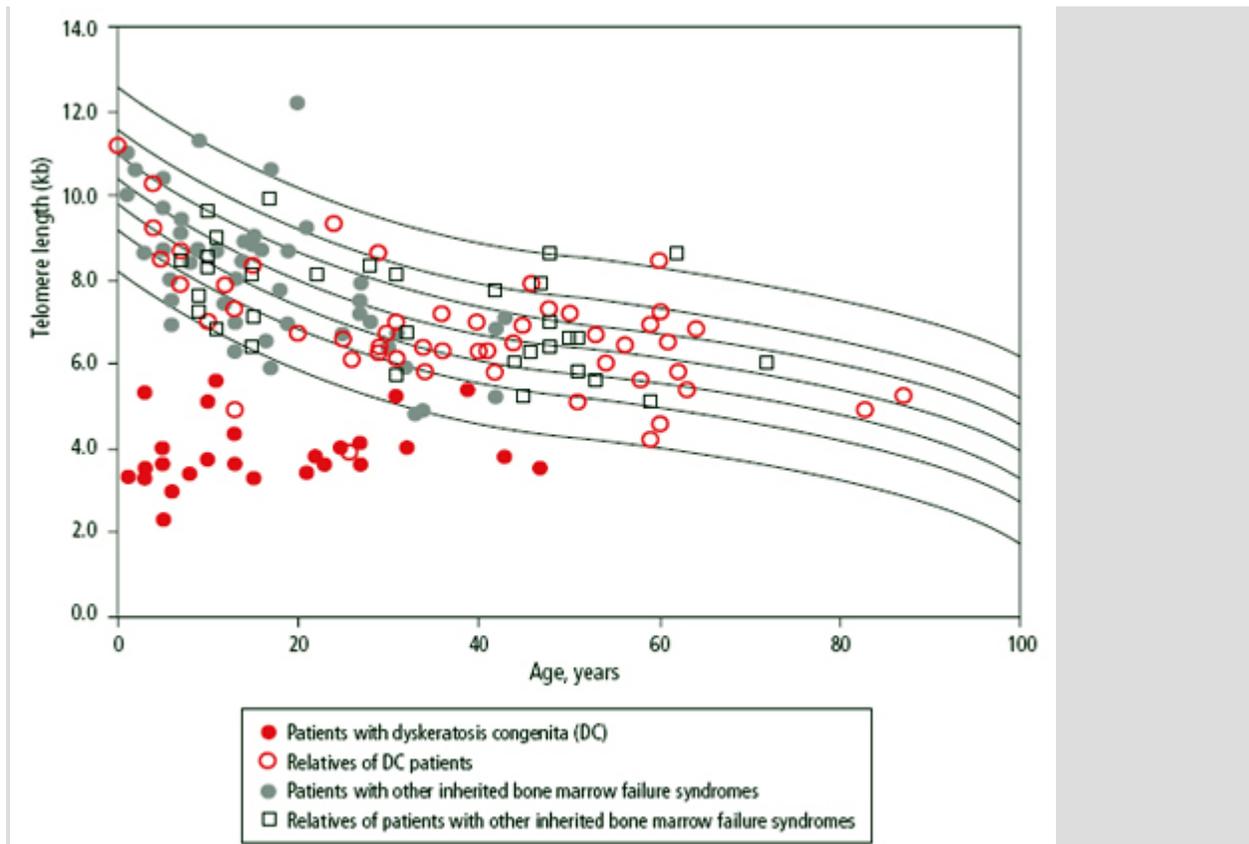


## Dyskeratosis Congenita: First NIH Clinical Research Workshop

In September, the first NIH Clinical Research Workshop on Dyskeratosis Congenita (DC) was held. Organized by **Sharon A. Savage, M.D.**, of the Clinical Genetics Branch (CGB), and cosponsored by the NIH Office of Rare Diseases, the meeting focused on bringing clinicians and scientists studying DC together with affected patients and families in order to share recent findings, explore opportunities for future research, and empower patients with DC and their families to organize a support group.

DC is an inherited bone marrow failure and cancer predisposition syndrome characterized by exceedingly short germline telomeres. About half of the patients with this multisystem disorder have mutations in telomere biology genes (see figure 1). Patients with DC are being studied in NCI's Inherited Bone Marrow Failure Syndromes study (<http://marrowfailure.cancer.gov>). Recent advances in understanding the molecular pathogenesis of DC include the discovery of new causative genes and the development of a diagnostic test. These developments, coupled with the need for a DC-specific family support group expressed by families participating in the NCI study, prompted this workshop.



**Figure 1.** Telomere length measurement by flow-FISH (flow cytometry with multicolorfluorescence *in situ* hybridization) of leukocyte subsets is sensitive and specific for dyskeratosis congenita (DC). The 1st percentile (bottom line) and the 99th percentile (top line) were derived from 400 controls.

**Mark H. Greene, M.D.**, Chief of CGB, welcomed 80 participants, including clinicians, scientists, and 42 family members representing 17 families. Presentations by **Blanche P. Alter, M.D., M.P.H.**, **Neelam Giri, M.D.** (both of CGB), Dr. Savage, and other investigators from the United States and Europe covered clinical findings, diagnosis, management, and genetic characterization of DC. Patients and families met with representatives from several other family support and advocacy groups and created a mission statement, action plan, and board of directors. Additional information about the DC family support group can be found at [www.dcoutreach.com](http://www.dcoutreach.com).

The workshop resulted in the formation of collaborative studies involving the clinical, scientific, and patient communities. The research strategies will be key in advancing scientific understanding of the molecular pathogenesis

of DC and telomere biology, as well as improving therapeutic options for patients with DC. A future meeting will focus on developing consensus treatment guidelines.

—June A. Peters, M.S., C.G.C.