Dyskeratosis Congenita
The classic telomere biology disorder

Clinical Features
• “Diagnostic triad”: nail dystrophy (A), reticular pigmentation of the skin (B), and oral leukoplakia (C)
  Not all patients will present with all of these features
• Patients have very high rates of bone marrow failure, including myelodysplastic syndrome and immunodeficiency
• Increased risk of pulmonary fibrosis and liver abnormalities
• Increased risk of cancers, including leukemias and squamous cell cancers of the head, neck and anogenital regions. Patients may have other medical complications that may include esophageal, urethral, or lacrimal duct stenosis, avascular necrosis of the hips or shoulders, developmental delay
• Symptoms typically present within the first decade of life, although less severe cases can present in adulthood
• Symptom severity ranges from mild to the more clinically severe Hoyeraal-Hreidarsson and Revesz syndromes

Diagnosis and Lab Tests
• Based on clinical triad and other signs and symptoms
• Very short telomere length in white blood cells or leukocytes
• Mutations in DKC1, TERC, TERT, NOP10, TINF2, WRAP53, NHP2, and CTC1

Treatment and Management
• An individualized multidisciplinary treatment plan should be established early on
• Options for bone marrow failure may include:
  - Anabolic steroids
  - Growth factors (filgrastim and erythropoietin)
  - Hematopoietic stem cell transplantation tailored for DC patients
  - Regular comprehensive medical evaluation including, at a minimum:
    - quarterly CBC
    - annual bone marrow biopsy, pulmonary function test, gynecological exam, endocrine and hormonal evaluation
    - bi-annual ENT and dental exam

Ongoing Research
• Discovery of the genetic causes
• Understanding the molecular biology of the genes associated with DC
• Use of induced pluripotent stem cells as models of DC and telomere biology disorders
• Identifying optimal hematopoietic stem cell transplantation regimens
• Optimizing clinical management

Dyskeratosis Congenita Outreach, Inc.  www.dcoutreach.com
Formed in 2008, our organization exists to provide support services and information to families affected by DC (Dyskeratosis Congenita). We aim to educate medical providers and encourage the medical community to conduct research on what causes and how to treat this often fatal disease.

Leadership
Our volunteer board consists of patients and family members affected by DC. We meet monthly to plan events, produce education materials and discuss how best to meet the needs of our growing community.

Medical Advice
A five-member rotating medical advisory board, comprised of specialists from the National Cancer Institute, Harvard Medical School, Children’s Hospital of Philadelphia, University of Minnesota, and the University of Sao Paolo (Brazil) works alongside us to promote awareness in the medical arena as well as give counsel to individual patients.

Family Support
Each month the board hosts a virtual support meeting via Skype to talk with and answer questions from members of the DC community. Every other month a member of our medical advisory board participates in that meeting to give first-hand information to those in attendance. DC Outreach members are in regular e-mail and phone communication with newly-diagnosed patients and those going through difficult medical procedures. In October 2012, DCO’s second bi-annual family weekend was held at Camp Sunshine in Maine. These long weekends provide a place for our families to meet, share experiences with each other, as well as attend lectures by well known doctors and researchers in the DC community.

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May you never be alone