Dyskeratosis congenita is a rare genetic form of bone marrow failure, the inability of the marrow to produce sufficient blood cells. Dyskeratosis is Latin and means the irreversible degeneration of skin tissue, and congenita means inborn. First described in the medical literature in 1906, dyskeratosis congenita was originally thought to be a skin disease that also affects the nails and the mouth. Only later in the sixties was it realized that patients with these skin changes almost always develop bone marrow failure. Thus, for the last 40 years or so, the bone marrow failure syndrome dyskeratosis congenita was diagnosed when patients presented with the triad of abnormal skin, malformation (dystrophy) of the nails, and white, thickened patches on the mucous membranes of the mouth (oral leukoplakia). The skin changes may be present before the development of bone marrow failure. Bone marrow failure is usually diagnosed by the low number of circulating blood cells including red blood cells, white blood cells, and platelets. Additional findings in patients with dyskeratosis congenita may include short stature, eye and tooth abnormalities, thin and early graying of the hair, lung (pulmonary) disease, liver disease, gut abnormalities, bone thinning (osteoporosis), infertility, learning difficulties, and delays in reaching developmental milestones. An increased incidence of leukemia and cancer has also been documented. The prevalence or incidence of dyskeratosis congenita is difficult to assess. In a population of patients with bone marrow failure about 2-5% of patients have bone marrow failure due to dyskeratosis congenita. In patients with pulmonary fibrosis similarly 2-5% are thought to be due to mutations in TERC or TERT. In families with an increased frequency of bone marrow failure and/or lung disease, dyskeratosis congenita should be excluded as a possible cause of their disease.