Supplementary Table S2: DLCO abnormality in relation to spirometry findings

	Total N = 42*	DLCO abnormality	
		Mild	Mod/severe
Spirometry normal	31	19	6
Spirometry abnormal	11	4	7
Odds ratio, 95% Confidence Interval		0.3, 0.08-1.5	7.2 1.5-33
p-value		0.18	**0.019**

*One patient with an abnormal spirometry did not undergo DLCO test. Significant p-values are in **bold**

Supplementary Table S3: Association of pulmonary function test abnormalities with features of dyskeratosis congenita

	Number of Patients	Abnormal spirometry		Abnormal DLCO			Moderate / severe DCLO ± abnormal Spirometry
		Obstructive	Restrictive	Any	Mild	Moderate/ Severe	
Total number	43	2	10	36	23	13	18
Number of triad features							
0-1	20	2	5	18	14	4	8
2-3	23	0	5	18	9	9	10
p-value		0.2	1.0	0.4	0.07	0.2	1.0
Bone marrow failure							
None/moderate	25	0	3	20	15	5	6
Severe	18	2	7	16	8	8	12
p-value		0.2	0.06	0.7	0.4	0.1	**0.01**
Disease inheritance*							
AR, XLR, *TINF2*	25	1	7	21	12	9	12
AD non-*TINF2*	14	1	3	12	9	3	5
p-value		1.0	0.7	1.0	0.5	0.5	0.5
HH/RS vs Others							
HH/RS	8	1	4	8	4	4	6
Others	35	1	6	28	19	9	12
p-value		0.3	0.07	0.3	1.0	0.2	0.05
Telomere length Z-score in patients with normal versus abnormal PFTs*							
p-value		0.7	0.09			0.9	0.6

*Patients with genes unknown (n=4) and with no data on telomere length (n=1) were excluded from the analysis.

Abbreviations: DLCO, diffusion of lungs for carbon monoxide; HH, Hoyeraal-Hreidarsson syndrome; RS, Revesz syndrome; HH, Hoyeraal-Hreidarsson syndrome; RS, Revesz syndrome; PFT, pulmonary function tests

Significant p-values are in **bold**

Supplementary Table S4: Characteristics of patients who did and did not undergo hematopoietic cell transplantation (HCT)

	HCT	No HCT	p-value
Number	14	32	
DC inheritance			
XLR, AR, TINF2	12	15	**0.02**
AD non-TINF2	1	13	
unknown	1	4	
Telomere length Z-score	-5.7 (-7.2 - -1.4)	-3.3 (-6.7 - -1.2)	**0.008**
Severe BMF (at PFT)	14	8	**<0.0001**
Age at HCT, years	12.9 (3.2 – 29)	NA	
Pulmonary disease	7	5	**0.027**
Age at pulmonary disease, years	15 (11-22)	56 (21-65)	**0.009**
Interval from HCT to pulmonary disease	4.7 (0.7 – 12)		
Interval from pulmonary disease to death or lung transplant	2.1 (0.96 – 3.45)	4.1, 4.2	**0.007**
Number died*	8	4	**0.003**
Age at death*	17 (13 – 29)	50 (19 – 69)	**0.007**
Age alive at last follow-up	18 (8 – 32)	29 (10 – 59)	**0.02**

*Includes all patients who died irrespective of cause.

All ages and interval are in years; values are median (range)

Abbreviations: DC, dyskeratosis congenita; XLR, X-linked recessive; AR, autosomal recessive; AD, autosomal dominant; HCT, hematopoietic cell transplantation; BMF, bone marrow failure; PFT, pulmonary function tests

Significant p-values are in **bold**