

SUPPLEMENTARY MATERIAL

Table 1S. Subject characteristics.

Country/Sex	N	Telomere Length (kb) (2.5 th - 97.5 th percentile)	Age (years) (2.5 th - 97.5 th percentile)	Period*
France/females	61	6.5 (5.5 – 7.6)	59 (37 – 75)	1998
France/males	124	6.4 (5.3 – 7.5)	57 (38 – 74)	1998
Denmark/females	1159	6.3 (4.7 – 8.0)	64 (22 – 102)	1996-2005
Denmark/males	568	6.4 (4.7 – 8.0)	55 (21 – 102)	1996-2005
Israel/females	207	7.5 (6.5 – 8.7)	30 (29 – 32)	1990
Israel/males	413	7.3 (6.1 – 8.6)	30 (29 – 32)	1990
USA/females	3187	6.8 (5.5 – 8.2)	62 (32 – 83)	1992-1996
USA/males	2539	6.6 (5.4 – 7.9)	62 (33 – 84)	1992-1996
Italy/females	284	6.0 (4.3 – 7.7)	60 (24 – 101)	2004
Italy/males	264	6.1 (4.6 – 7.6)	54 (23 – 96)	2004
UK/females	3200	7.0 (5.8 – 8.4)	49 (22 – 71)	1998
UK/males	314	6.6 (5.4 – 8.0)	47 (21 – 76)	1998

Table 2S. Characteristics of patients with dyskeratosis congenita and their relatives.

Characteristics	DC	Relatives
Number (male:female)	23 (16:7)	45 (18:27)*
Age (years)		
Mean±SD	30.8±15.4	38.4±17.2
Median (range)	30.3 (7.4-71)	40.6 (7.7-69.4)
LTL (kb)		
Mean±SD	4.99±0.79	6.49±0.71
Median	4.66 (3.83-6.55)	6.55 (4.51-7.86)

DC, dyskeratosis congenita; LTL, leukocyte telomere length; *6 relatives of DC patients for whom the causative mutation is unknown displayed mean=6.96 kb, median=7.2 kb (range=5.59-7.86 kb).

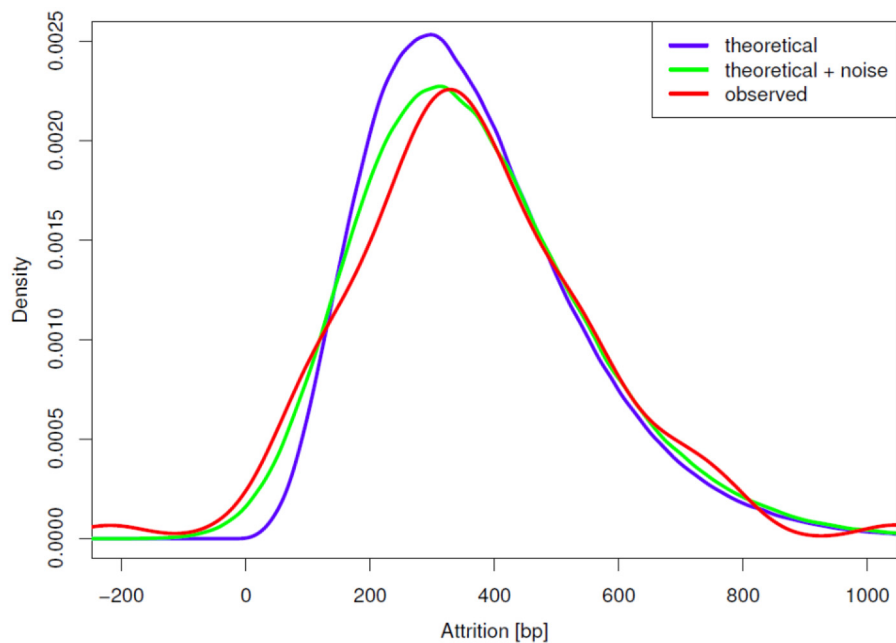


Figure S1. Observed LTL attrition distributions after approximately 12 (5th - 95th percentile: 11.2-17.8) years along with theoretical distributions (one with- and one without measurement error). The observed LTL attrition distribution is from whites of European descent [1]. The theoretical distribution is based on assumed yearly independent gamma distributed attrition (in bp) with shape parameter 0.4 and scale parameter 75 (these values have been chosen such that the 10-year attrition distribution has mean 300 bp and SD 150 bp). The theoretical curve is based on the actual follow-up times observed in the study [1] (each observation time is turned into 5000 simulations, from which the theoretical curve is found by kernel density estimation). The theoretical curve with noise is generated by further adding normally distributed noise with mean zero and SD 74. The choice of SD 74 is based on the estimated intra-assay SD for the study (1).

REFERENCES

1. Chen, W, et al. Longitudinal versus cross-sectional evaluations of leukocyte telomere length dynamics: age-dependent telomere shortening is the rule. *J Gerontol A Biol Sci Med Sci.* 2011; 66: 312-319.