

Supporting Information

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SI Text

Telomere length in PBL was analyzed in three families with *TERT* mutations (*TERT* A202T, *TERT* K570N, and *TERT* R889X) whose pedigrees are shown in Fig. S1. The actual telomere length of leukocytes, granulocytes, and lymphocytes was determined and is shown in Tables S1–S3. Specifically, Fig. S1A shows the pedigree of *TERT* A202T family whose telomere length is shown in Table S1.

Fig. S1B shows the pedigree of *TERT* K570N family whose telomere length is shown in Table S2, and Fig. S1C shows the pedigree of *TERT* R889X family whose telomere length is shown in Table S3.

The calculated actual telomere lengths of B6 and *Tert*^{+/-}/g(17) mice were compared and demonstrated a 54% reduction in TL in *Tert*^{+/-}/g(17) mice (Fig. S2), a finding that is consistent with the results generated by flow cytometry-FISH.

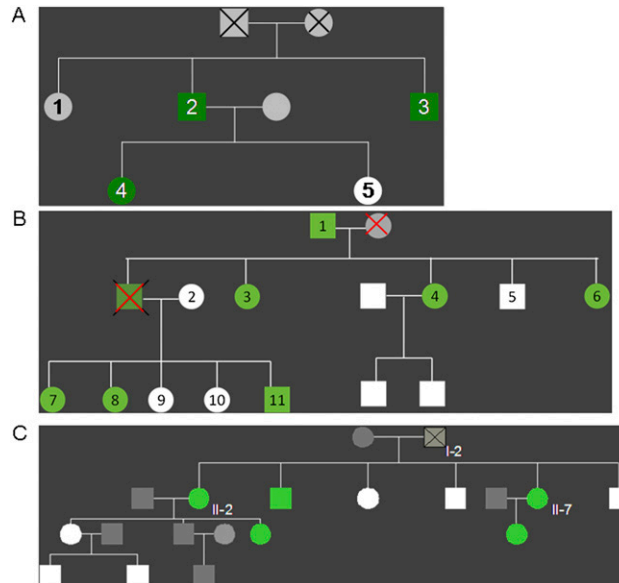


Fig. S1. (A) Pedigree of family carrying a *TERT* A202T mutation. Individuals in green tested positive for the mutation; individuals in white are noncarriers; and individuals in gray were not tested. (B) Pedigree of family carrying a *TERT* K570N mutation. Individuals in green are mutation carriers; individuals in white are noncarriers; and individuals in gray were not tested. (C) Pedigree of family carrying a *TERT* R889X mutation. Individuals in green are mutation carriers; individuals in white are noncarriers; and individuals in gray were not tested.

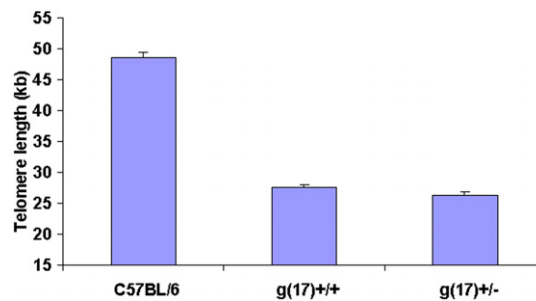


Fig. S2. The actual telomere lengths of splenocytes from C57BL/6, *Tert*^{+/+}/g(17), and *Tert*^{+/-}/g(17) mice were determined as described by Ding et al. (1) and Baerlocher et al. (2).

1. Ding H, et al. (2004) Regulation of murine telomere length by Rtel: An essential gene encoding a helicase-like protein. *Cell* 117:873–886.
2. Baerlocher GM, Vulto I, de Jong G, Lansdorp PM (2006) Flow cytometry and FISH to measure the average length of telomeres (flow FISH). *Nat Protoc* 1:2365–2376.

Table S1. Telomere length (TL) in *TERT* A202T family

Patient no.	Leukocyte TL observed	Granulocyte TL observed	Lymphocyte TL observed
1	6.5 kb (<10th percentile)	7.4 kb (normal range)	5.5 kb (<10th percentile)
2	4.8 kb (<first percentile)	6.1 kb (<10th percentile)	4.7 kb (<10th percentile)
3	6.1 kb (<10th percentile)	6.6 kb (<10th percentile)	5.0 kb (<10th percentile)
4	7.3 kb (<10th percentile)	7.3 kb (<10th percentile)	7.4 (<10th percentile)
5	8.3 kb (normal range)	8.5 kb (normal range)	8.2 kb (normal range)

Table S2. Telomere length (TL) in *TERT* K570N family

Patient no.	Leukocytes TL	Granulocyte TL	Lymphocyte TL
1	3.9 kb (<first percentile)	4.9 kb (<first percentile)	3.6 kb (<first percentile)
2	6.2 kb (<10th percentile)	7.0 kb (normal range)	5.6 kb (normal range)
3	3.3 kb (<first percentile)	3.9 kb (<first percentile)	3.3 kb (<first percentile)
4	3.9 kb (<first percentile)	5.3 kb (<first percentile)	3.7 kb (<first percentile)
5	***	4.8 kb (<10th percentile)	***
6	4.1 kb (<first percentile)	5.4 kb (<first percentile)	4.0 kb (<first percentile)
7	5.6 kb (<first percentile)	6.7 kb (<10th percentile)	5.2 kb (<first percentile)
8	4.6 kb (<first percentile)	4.5 kb (<first percentile)	4.4 kb (<first percentile)
9	8.1 kb (normal range)	8.3 kb (normal range)	8.0 kb (normal range)
10	6.3 kb (<10th percentile)	7.1 kb (<10th percentile)	6.2 kb (<10th percentile)
11	3.1 kb (<first percentile)	3.8 kb (<first percentile)	3.1 kb (<first percentile)

***Not to be determined.

Table S3. Telomere length (TL) in *TERT* R889x family

Patient no.	Leukocytes TL	Granulocyte TL	Lymphocyte TL
II-2	4.2 kb (<first percentile)	5.1 kb (<first percentile)	3.8 kb (<first percentile)
II-3	4.5 kb (<first percentile)	4.9 kb (<first percentile)	3.6 kb (<first percentile)
II-4	6.1 kb (<10th percentile)	6.4 kb (<10th percentile)	5.8 kb (normal range)
II-5	6.1 kb (<10th percentile)	6.6 kb (<10th percentile)	5.8 kb (normal range)
II-7	5.1 kb (<first percentile)	5.0 kb (<first percentile)	5.1 kb (<first percentile)
III-1	5.9 kb (<10th percentile)	6.2 kb (<10th percentile)	5.6 kb (<10th percentile)
III-5	4.5 kb (<first percentile)	5.0 kb (<first percentile)	4.2 kb (<first percentile)
III-6	6.3 kb (<first percentile)	6.3 kb (<first percentile)	6.3 kb (<first percentile)
IV-1	6.9 kb (<10th percentile)	6.9 kb (<10th percentile)	6.9 kb (<10th percentile)
IV-2	7.4 kb (normal range)	7.3 kb (<10th percentile)	7.4 kb (normal range)