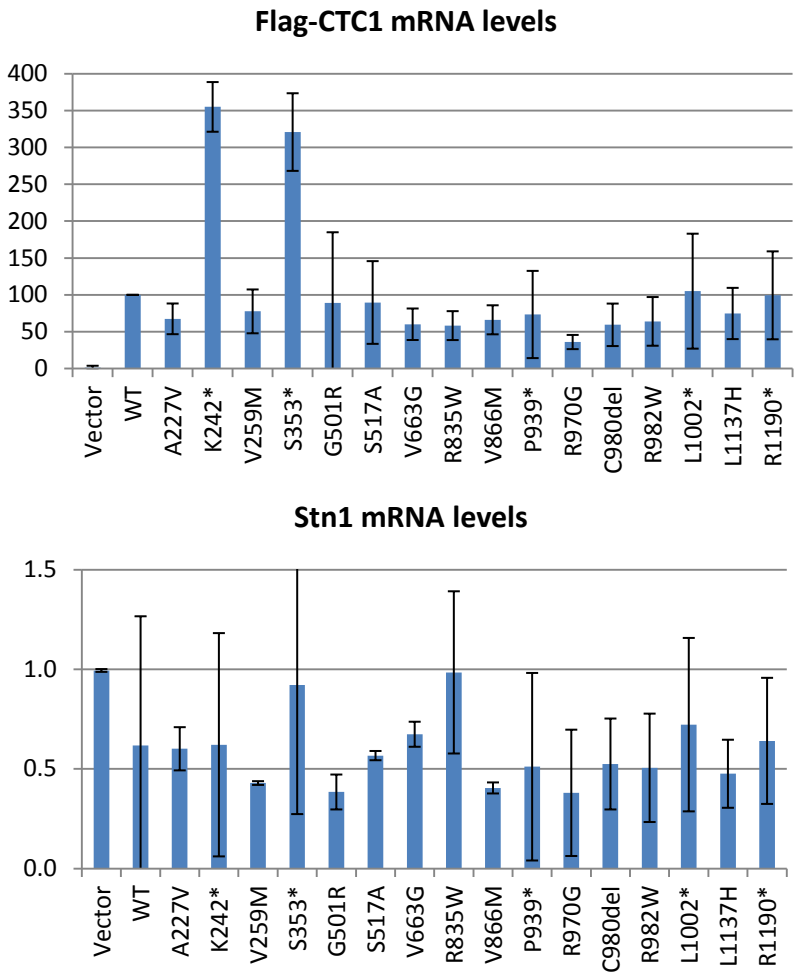
Functional characterization of human CTC1 mutations reveals novel mechanisms responsible for the pathogenesis of the telomere disease Coats plus

Peili Gu[1] and Sandy Chang[1,2,*]

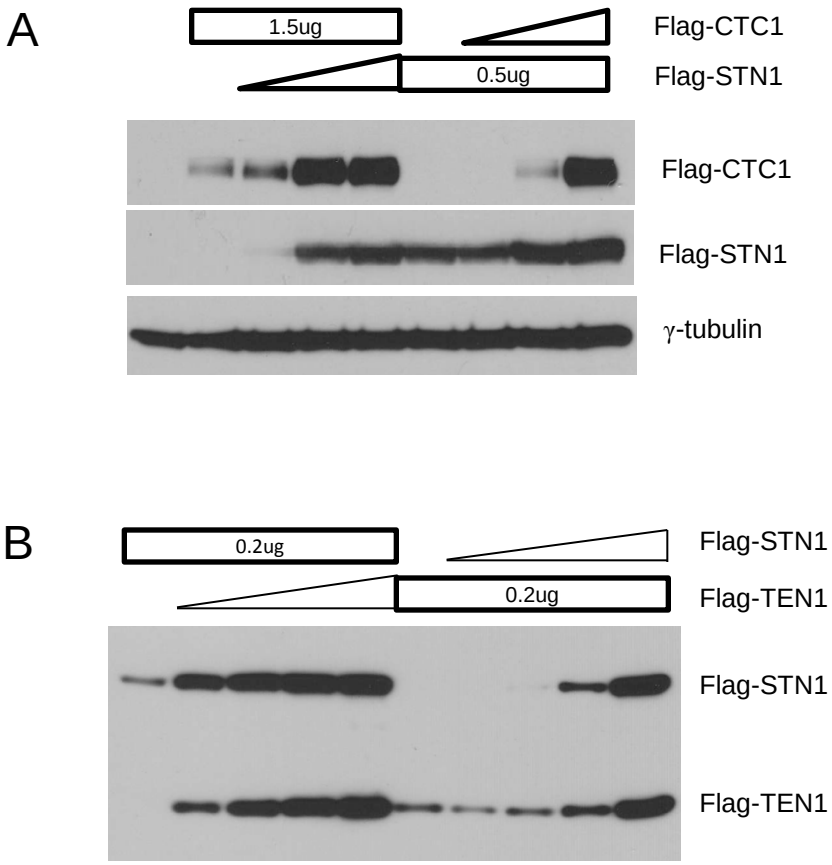
[1]Department of Laboratory Medicine and [2]Pathology
Yale University School of Medicine
330 Cedar St.
New Haven, CT 06520

Supplementary Figure 1. RT-PCR analysis of Flag-tagged WT and mutant CTC1 expression (top) and endogenous STN1 expression (bottom) in reconstituted *CTC1*$^{-/-}$ MEFs. The forward primer used for Flag-CTC1 corresponds to the Flag sequence and the reverse primer corresponds to the mouse CTC1 cDNA. Error bar was derived from two independent cell lines.

Supplementary Figure 2. Stabilization of CTC1, STN1 and TEN1 requires CST complex formation. A. Flag-CTC1 and Flag-STN1 mutually stabilize each other. B. STN1 and TEN1 mutually stabilize each other. In (A), fixed amount of Flag-CTC1 (1.5ug) was added in 293T cells along with increasing concentrations of Flag-STN1 (0, 0.02, 0.1, 0.5ug) (left). We also fixed Flag-STN1 concentration at 0.5ug with increasing concentrations of Flag-CTC1 added (0, 0.06, 0.3, 1.5ug)(right). For (B), the Flag-STN1 concentration was fixed at 0.2ug, and increasing concentrations of Flag-TEN1 added (0, 0.025, 0.05, 0.1, 0.2ug)(left). The reciprocal experiment was also performed (right). The expressed proteins were detected with anti-Flag antibody.



Gu and Chang, Supplemental Figure 1



Gu and Chang, Supplemental Figure 2