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LINE-1 Elements in Structural Variation and Disease
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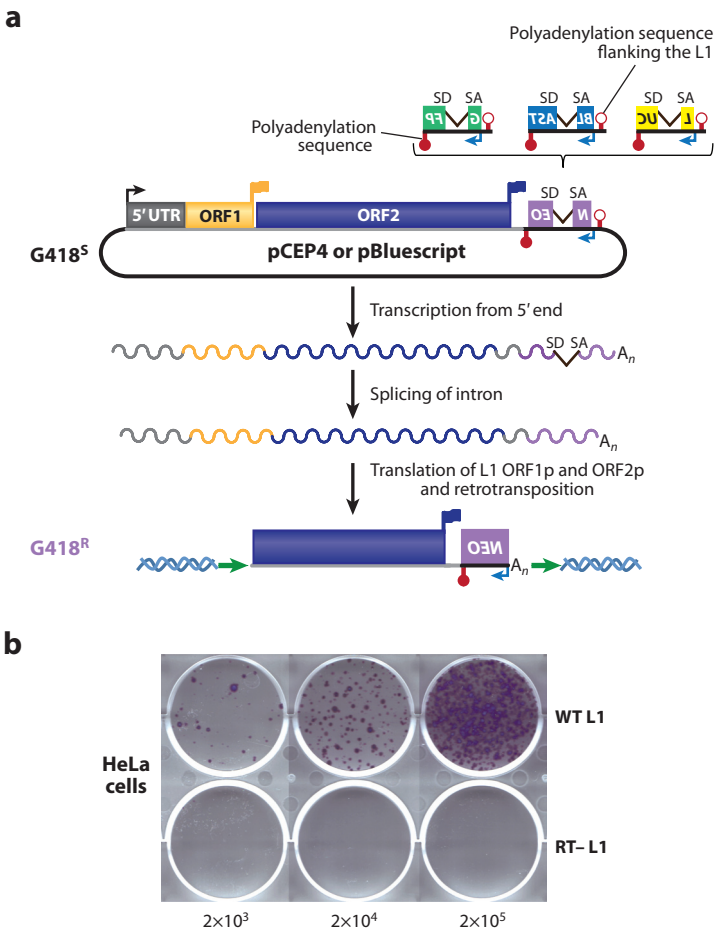

Supplemental Figure 1

A cultured cell assay to detect LINE-1 retrotransposition: (*a*) Candidate active human L1s (1, 2, 7) are tagged in their 3′ untranslated region (UTR) with an indicator cassette designed to detect retrotransposition events in cultured cells. The selectable/screenable markers [e.g., NEO (5, 7), GFP (9), blasticidin (8), and luciferase (12)] are in the opposite transcriptional orientation compared with the L1 and contain their own promoters (*backward blue arrow*) and polyadenylation sequences (*upside-down filled red lollipop*); they also contain an intron in the same transcriptional orientation as the L1 [splice donor (SD) and splice acceptor (SA), respectively]. This arrangement ensures that the reporter gene (e.g., *NEO*) will become activated only upon a successful round of retrotransposition (*bottom*). Flags represent epitope tags that can be placed on ORF1p and/or ORF2p (4, 6), and open lollipops represent the polyadenylation sequences flanking the L1s. Details regarding the assay can be found in Supplemental References 7 and 11. (*b*) Representative results of an L1 retrotransposition assay in cultured HeLa cells. WT is an active L1 allele, L1.3 [accession number L19088 (3)]. RT− is a retrotransposition-defective control containing a missense mutation (D702A) in the reverse transcriptase domain of ORF2p (10). The number of HeLa cells transfected in each experiment is depicted below the wells. The assay was performed by C.R. Beck.

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