

**Supplemental Figure S4:** Multiple sequence alignment and schematic overview of breakpoint analysis. **A.** In green, the LINE HAL1ME element, in blue the LTR gypsy element and in yellow the regions of microhomology. Oligo(G)n tracts are in bold underscored. **B.** In green, the SINE *AluJb* element, in blue the DNA Tigger16b element and in yellow the regions of microhomology. Inverted sequences are framed and A phased repeat is in bold.

**A. c.585+2223\_3326+5684del**

Proximal	TTTCTACTGT <b>CCT</b> TGTGTAGGACTCTTTTCCTAATATCTTCTCCCTGGAA <b>JAGGCTGACT</b>
Junction	TTTCTACTGTGGTTGTGTAGGACTCTTTTCCTAATATCTTCTCCCTGGAA <b>JAGGCTGACT</b>
Distal	CCTCACCTTTCTCCTGAGTCTTAACTCTCACTATTCCCTTCCCCTGAGGAA <b>AACGATTT--</b>
Proximal	GTGAATTTTGTGA <b>AGGGCAAAACATGTTGACT</b> CTCA <b>GGCTTTACTTTAGGTGGACATGG</b>
Junction	GTGAATTTTGTGA <b>AGGGCAAAACATGTTGACT</b> TCCATCTGAATTTTCTCTGCTACTTTT
Distal	CTCCAGTACA <b>TGTA--TGATAAAGGCCTGTACT</b> TCCATCTGAATTTTCTCTGCTACTTTT
Proximal	TGAGAAGCACTCCCCTTCC <b>ACCTTGCCAAAATAAAGTGGGCCATATTGTGAATGCAGAG</b>
Junction	CTCACTCAGCCAGACACGTA <b>ACCT</b> CTCAAGAGTTATACGCTGCA <b>TCAGTTCAAGCTCTGT</b>
Distal	CTCACTCAGCCAGACACGTA <b>ACCT</b> CTCAAGAGTTATACGCTGCA <b>TCAGTTCAAGCTCTGT</b>

**B. c.1487+1134\_3741-2607delins15**

Proximal	TGCAACCTTGGCTTCCTGGGCTCCACTTCAAGCTATCCTGCCACCTCAGCCTGCCAGGTA
Junction	TGCAACCTTGGCTTCCTGGGCTCCACTTCAAGCTATCCTGCCACCTCAGCCTGCCAGGTA
Distal	TACCCATAATGTGCCTAAAGTAGTAATAACAGACCTAAACCATTTTGTGTACCAGTG <b>TTT</b>
Proximal	GCTAGGACTACAGGTACATGCC----- <b>ACTGTGCATGA</b>
Junction	GCTAGGACTACAGGTACATGCC <b>CCTTGGATTGTACCT</b> GTTTCCCCTTGGATTGCTGTGGA
Distal	<b>CCATGGAAA</b> <b>AATTCACTCAAAA</b> ----- <b>GTTTCCCCTTGGATTGCTGTGGA</b>
Proximal	CT <b>CATTT</b> TTGTATTTTTTTGTAGAG <b>ATGGGGTCTCCCAT</b> CTTGCCCAAGCTGATCTTGA
Junction	CACATTTCTACA--TCACAGGGAGAATGGGGACTCTATGCTAGAAGCAGTAGAATCATT
Distal	CACATTTCTACA--TCACAGGGAGAATGGGGACTCTATGCTAGAAGCAGTAGAATCATT