

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 CG TTGTGTAGGACTCTTTTCCTAATATCTTCTCCCTGGAAAGGGCTGACT
Junction	TTTCTACTGTGGTTGTGTAGGACTCTTTTCCTAATATCTTCTCCCTGGAAAGGGCTGACT
Distal	CCTCACCTTTCTCCTGAGTCTTAACTCTCACTATTCCCTTCCCCTGAGGAACGATTT--

Proximal	GTGAATTTTG TGTA AGGGGCAAAACATGT TG ACTCTCA CG CTTTACTTTA GGT CG ACAT CG
Junction	GTGAATTTTG TGTA AGGGGCAAAACATGT TG ACTTCCATCTGAATTTTCTCTGCTACTTTT
Distal	CTCCAGTACA TGTA --TGATAAAGGCC TGT ACTTCCATCTGAATTTTCTCTGCTACTTTT

Proximal	TGAGAAGCACTCCCCCTTCC ACCT TGCCAAAATAAAGTGGGCCATATTGTGAATGCAGAG
Junction	CTCACTCAGCCAGACACGTA ACCT CTCAAGAGTTATACGCTGCA TC AGTTCAAGCTCTGT
Distal	CTCACTCAGCCAGACACGTA ACCT CTCAAGAGTTATACGCTGCA TC AGTTCAAGCTCTGT

B. c.1487+1134_3741-2607delins15

Proximal	TGCAACCTTGGCTTCCTGGGCTCCACTTCAAGCTATCCTGCCACCTCAGCCTGCCAGGTA
Junction	TGCAACCTTGGCTTCCTGGGCTCCACTTCAAGCTATCCTGCCACCTCAGCCTGCCAGGTA
Distal	TACCCATAATGTGCCTAAAGTAGTAATAACAGACCTAAACCATTTTGTGTACCAGTG TTT

Proximal	GCTAGGACTACAGGTACATGCC----- ACTGTGCATGA
Junction	GCTAGGACTACAGGTACATGCC CCTTGGATTGTACCT GTTTCCCCTTGGATTGCTGTGGA
Distal	CCATGGAAA AATTCACTCAAAA -----GTTTCCCCTTGGATTGCTGTGGA

Proximal	CT CATT TTTGTATTTTTTTGTAGAG ATGGGG TCTCCCCAT CTTG CCCCAAGCTGATCTTGA
Junction	CACATTTCTACA--TCACAGGGAGA ATGGGG ACTCTATGCTAGAAAGCAGTAGAATCATTA
Distal	CACATTTCTACA--TCACAGGGAGA ATGGGG ACTCTATGCTAGAAAGCAGTAGAATCATTA