

Appendix 3: The two Alu elements spanning the breakpoints of the exon 1 deletion.

Genomic position	Sequence
Promoter region Alu: X:18697079-18696802	ttttttttttggaggcggagtcactctgtcgcccaggctggagtgcagtgtcgtgat ctgagctcactgcaacctctgcctcccgggtcacaccattctcctgcctcagcctccc gagtagctgggactacaggcgcctgccacctcgccggctaatttttgtatttttagttg agacgggggttcaccgtgttagccaggatgggtctgatctcctgacctcgtgatccgcc cgcctcggcctcccaaagtgtctaggattacaggcttgagccaccgtgcccgcc
Intron 1 region Alu: X:18692624-18692330	ttttttttttggagaatttcactctgttgcccaggctggagtgcagtggcgtgatctcg gctcaccacaacctccatctcccgggtcaagcgattctcctgcctcaggctcccag tggctgggattacagacgtgtgccaccacgcccggatcattttgtaatttagtagag acgggggttctccatgttgccaggctgggtctgaactccgacctcaggtgatctgcc tgctcggcctcccaaagtgtgggattacaggcatgagccaccgcacctggc