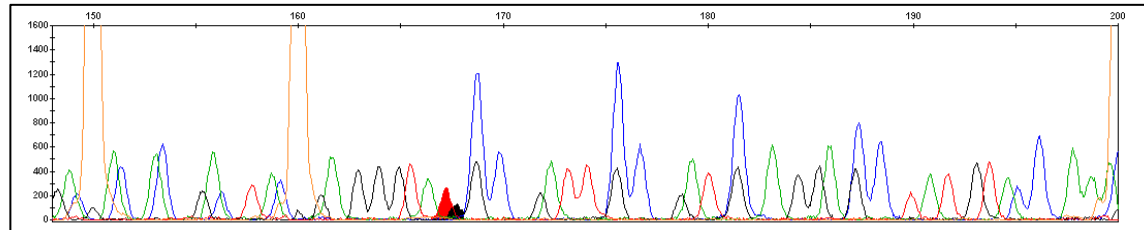


### Additional file 3:

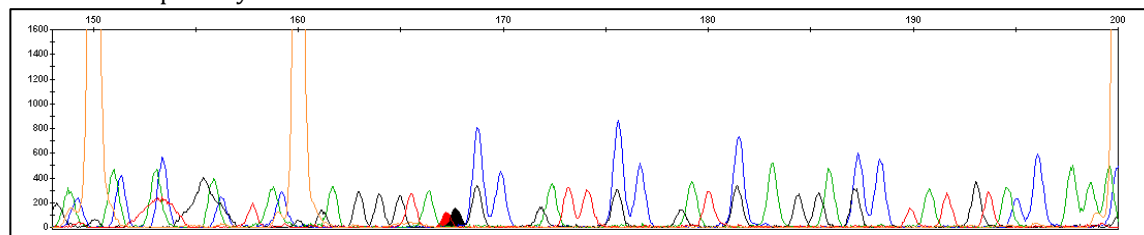
Fragment analysis of c.2311T>C in BC17 (forward)

A loss of the mutant allele could be detected. In this patient, the T (black) represents the mutant allele. This loss of allelic imbalance is in agreement with the MiSeq data. The VAF for cDNA is 26%, or only half of the frequency on gDNA. This is reflected in the ratio of 0.48 for fragm. analysis in this particularly example.

a. cDNA



b. cDNA with puromycin



c. genomic DNA

