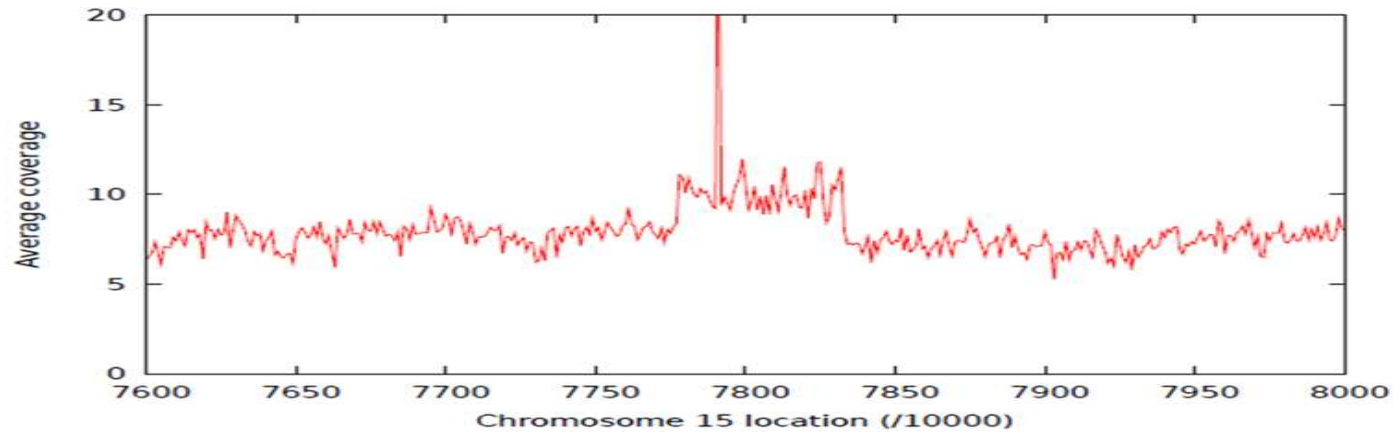


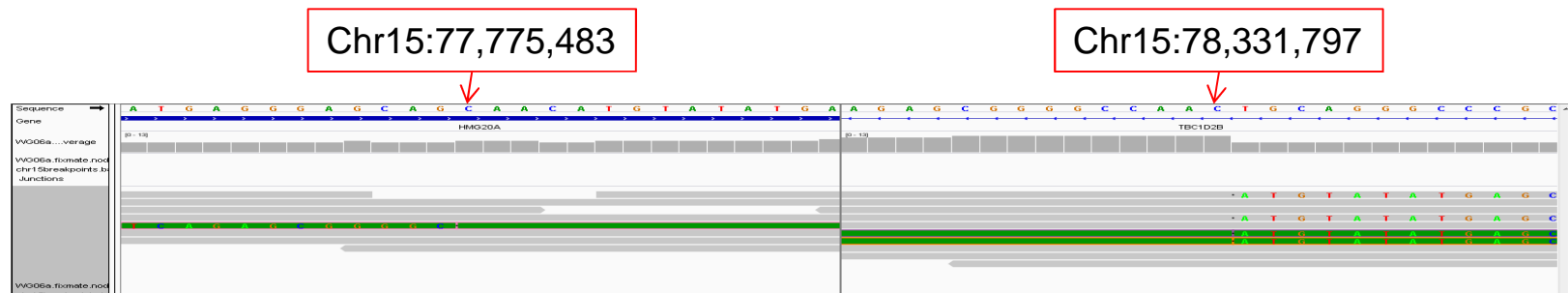
**Figure e-1. A:** Graph showing average read coverage across the region during targeted next generation sequencing. Read count across the region indicates average coverage increases from ~7.5x to ~10x, corresponding to the expected 50% increase in read number for a heterozygous duplication. The tall peak corresponds to a common LTR (long terminal repeat element), the reads have multiple mapping locations within a few mb of this location

**B:** Targeted next generation sequencing (III:16) identified read pairs mapping ~550kb apart indicative of a tandem duplication event, delineating the position of the rearrangement event (chr15:77775483-78331797dup [hg19]).

**A**



**B**



**Table e-1.** The duplicated region in each individual delimited by KaryoStudio, containing 14 RefSeq genes

Case	Duplicated segment position	Cytobands	Genes
II:1	chr15:77628700-78034927	q24.3 q25.1	<i>LOC101929457; LOC105370906; LINGO1; LINGO1-AS1; LINGO1-AS2; LOC105370907; CSPG4P13; DNMT1P9; LOC645752; LOC440292; LOC105370908; ADAMTS7P3; LOC91450; TBC1D2B</i>
II:9	chr15:77628700-78034927	q24.3 q25.1	
III:5	chr15:77622761-78034927	q24.3 q25.1	
III:9	chr15: 77816944-78034927	q24.3 q25.1	
III:16	chr15: 77672118-78034927	q24.3 q25.1	
IV:1	chr15: 77628700-78054525	q24.3 q25.1	
IV:2	chr15: 77495127-78034927	q24.3 q25.1	
IV:8	chr15: 77628700-78034927	q24.3 q25.1	
V:1	chr15: 77495127-78034927	q24.3 q25.1	