



Fig S7. Genotyping of the TSM candidate loci found with short-read data. **(A)** The 211 candidate TSM loci found in NA12878 Platinum data passing the sequence-based filtering. Ratio of REF and ALT allele mapping coverage ($r = \text{depth}_{\text{REF}}/\text{depth}_{\text{ALT}}$) reflects the genotype: $r \approx 1$ (and thus $\log_{10}(r) \approx 0$) for a heterozygote; $r \ll 1$ and $r \gg 1$ for the two types of homozygotes. The Log10-ratios agree for the NA12878 Platinum and Chromium datasets, and 105 and 96 loci (of the total 211) are called heterozygous and homozygous for ALT by both datasets. **(B)** Of the 755 candidate TSM loci found in NA12878 Chromium data, 438 are called homozygous for REF by both methods. **(C)** The 176 loci found in both short-read datasets. **(D)** The 67 loci found in both short-read datasets but not in HaplotypeSV dataset.