

chr1:248458096-248458132

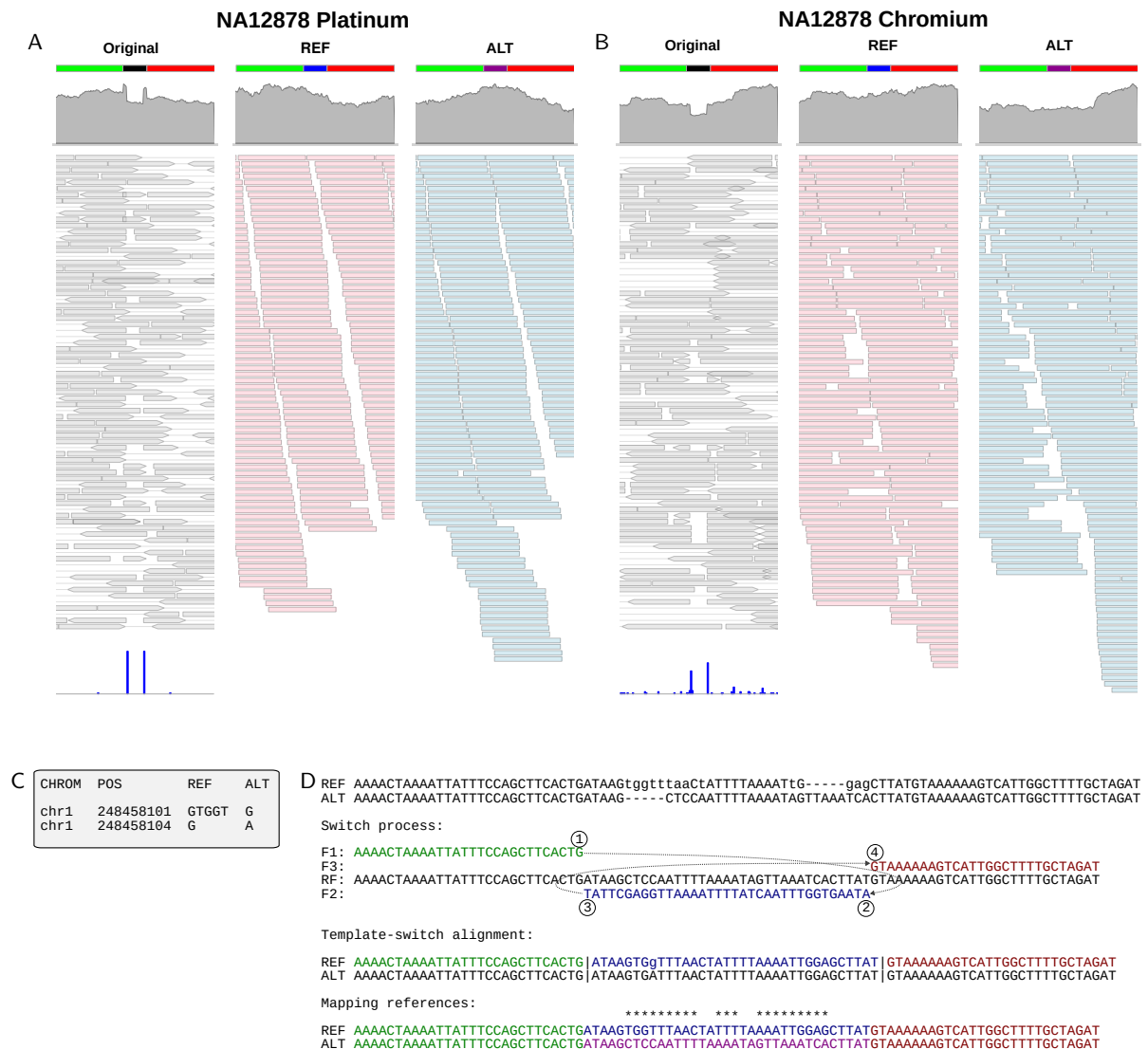


Fig S8. A heterozygous TSM event identified in NA12878 with short-read data. **(A)** Platinum data (left) and **(B)** Chromium data (left) have excess of soft clips (bottom, blue bars) and atypical patterns in the mapping coverage (top, in gray). **(C)** Two variants were called in this region by Ebert et al. (2021) but these fail to explain the haplotype differences. **(D)** De novo assembly of the reads creates two locally highly dissimilar haplotypes (top). All but one difference can be explained with a TSM event, an inversion in place (middle). Using two haplotypes with alternative central parts (blue and magenta; bottom) as the reference, extracted reads map in full length **(A,B;** middle, right) with roughly even coverages. Data were visualised with Gviz (Hahne and Ivanek 2016).