

chr3:34666617-34666653

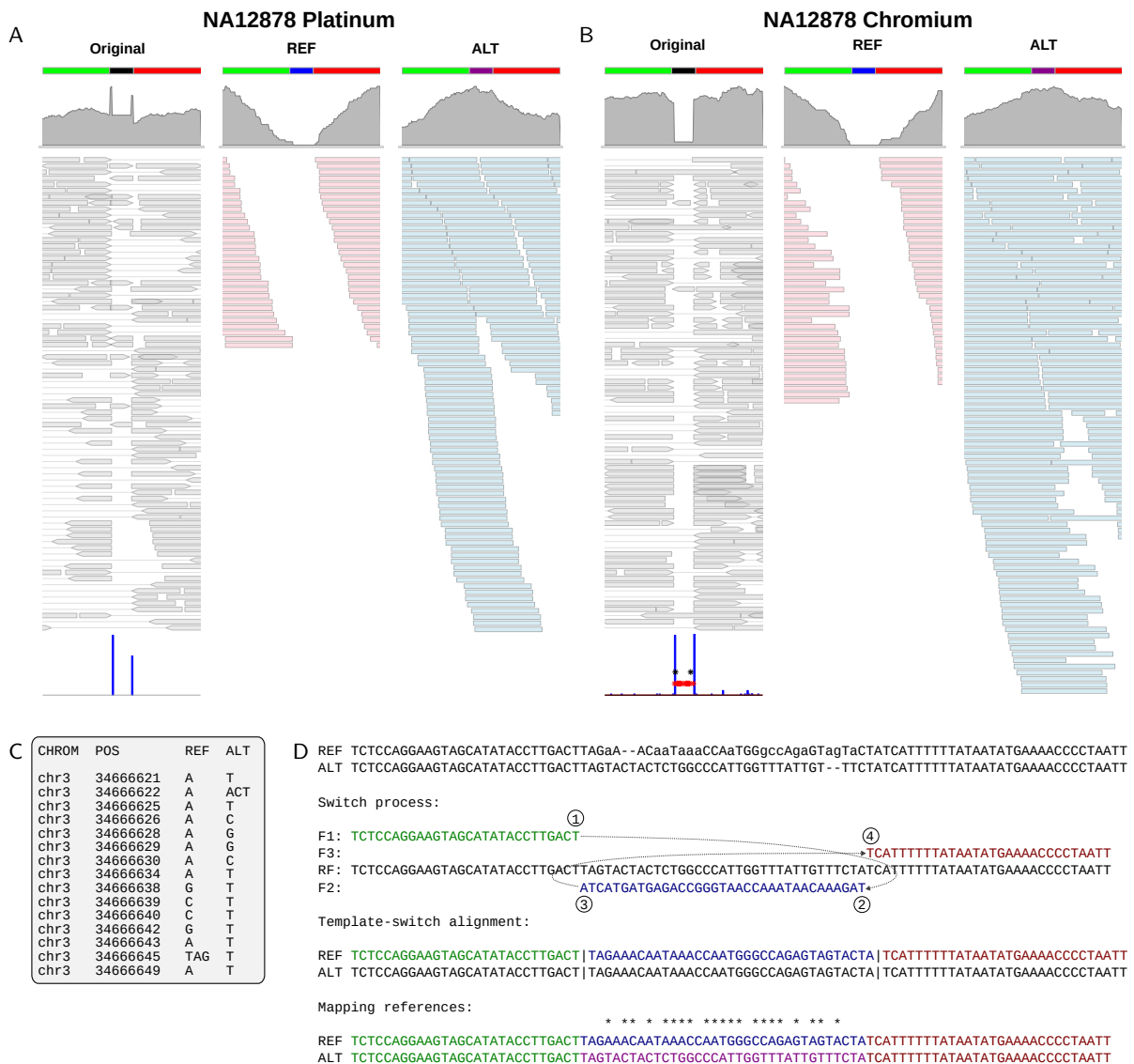


Fig S9. A homozygous TSM event identified in NA12878 with short-read data. **(A)** Platinum data (left) and **(B)** Chromium data (left) have excess of soft clips (bottom, blue bars) and atypical patterns in the mapping coverage (top, in gray). **(C)** Fifteen variants fully explaining the differences were called in the Chromium data while no variants were indicated in this region in the Platinum data or by Ebert et al. **(D)** De novo assembly of the reads creates two locally highly dissimilar haplotypes (top). All differences 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 on the ALT haplotype only **(A,B;** middle, right). Data were visualised with Gviz (Hahne and Ivanek 2016).