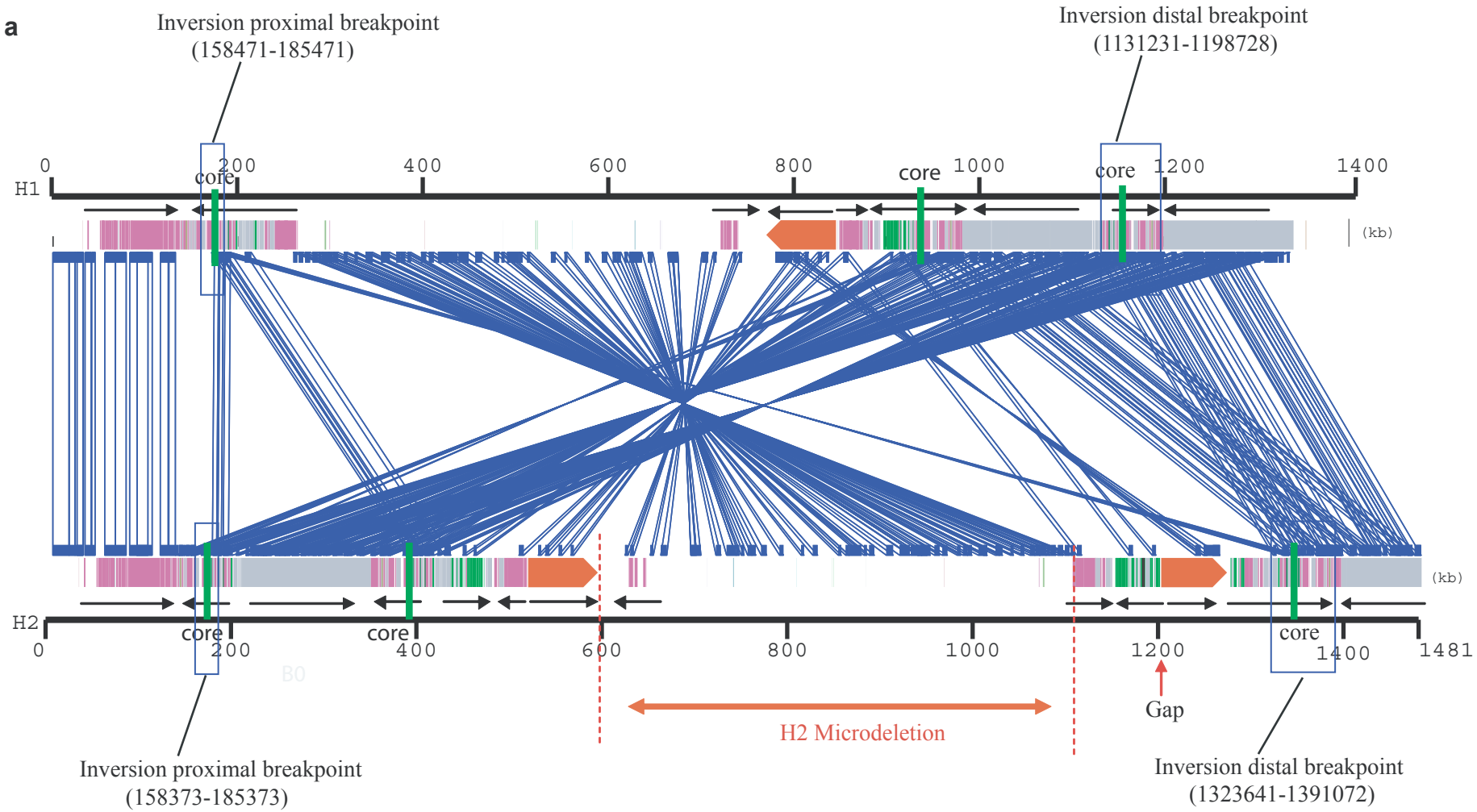

Supplementary Figure 1: Pairwise sequence comparison of chr17q21.31 region among primates.
Sequences (~1.5 Mbp) from human H1, human H2, chimpanzee and orangutan orthologous regions were compared using miropeats (Parsons, 1999) and two-way_mirror.pl (Bailey et al., unpublished). Human duplication structures (colored blocks) were annotated according to a database of curated duplicons (Jiang et al., 2007).
(a) Human H1 vs. H2 sequence comparison revealed that the core segmental duplications (green bars) mapped within the inversion breakpoints (blue box) between H1 and H2 haplotypes. In order to define the inversion breakpoints, we repeatmasked both H1 and H2 haplotype assemblies and compared the sequences using default settings (s -30). We examined each of the four breakpoints in turn by examining the furthest extent of directly oriented sequence between the two haplotypes and the furthest extent of inverted oriented sequence. For each comparison, we required that the majority of consecutive seed alignments be collinear. We defined the breakpoint intervals based on this intersection for each breakpoint. Once intervals had been identified, we further refined the breakpoint by constructing 4 pairwise alignments based on the intervals and requiring at least 99.5% sequence identity. The intervals, thus defined, are indicated by blue boxes. The inversion places larger blocks of sequence homology flanking the 17q21.31 microdeletion region, including H2-specific segmental duplications (red block arrows) in direct orientation (which are single copy in H1). High-density array CGH maps one of the microdeletion breakpoints (red broken lines) to the H2-specific segmental duplication.