GENOMIC INVERSIONS

The first draft of the human genome sequence was released on 2001. It was ameliorated continuously and in 2013 the hg38 was released. Then the coupling of PacBio and Nanopore sequencing technologies allowed the sequencing of telomere to telomere of all human chromosome, with an incredible precision. This approach can solve the many problems created by repeated sequences, including centromeric and pericentromeric satellites and segmental duplications (SD).

The group of Eichler compared the sequence of 41 genomes against the highly accurate T2T-CHM13 sequence and against the hg38 release (Genom Biol[1]). The comparative analysis was able to detect, among others, novel inversions in the pericentromeric region of chromosomes 1 and 7 (almost inaccessible to classical sequencing), and in SD-rich regions at 15q25.2, 16p11.2, 16q22.1-23.1, and 22q11.21. Of note is the fact that the organization of some of these inversions was the minor allele in the hg38.

1-https://genomebiology.biomedcentral.com/articles/10.1186/s13059-023-02919-8