

BREAKPOINT RESOLUTION OF CHROMOSOMAL REARRANGEMENTS

Here is another paper that utilizes high-fidelity, long-read sequencing to characterize chromosomal rearrangements (1).

“Multiple breakpoints were localized to genomic regions previously recalcitrant to sequencing such as acrocentric p-arms, ribosomal DNA arrays, and telomeric repeats, and involved complex structures such as a deletion-inversion and interchromosomal dispersed duplications”.

1. <https://www.cell.com/cms/10.1016/j.ajhg.2024.10.006/attachment/92a03bbb-42fb-41b5-9720-2709d7756cc1/mmc5.pdf>