RECOMBINATION BETWEEN ACROCENTRIC CHROMOSOMES

Human acrocentric chromosomes possess distinct traits: (i) in metaphase spreads, they often form satellite associations due to their active involvement of ribosomal DNA in nucleoli formation; (ii) acrocentric chromosomes 13 and 21, as well as 14 and 22, share highly homologous alpha satellite DNA; (iii) on occasion, these chromosomes fuse together to create Robertsonian translocations.

Homologous sequences between centromere and/or short arm are shared by these acrocentric chromosomes and are believed to contribute to the occurrence of heterologous short arm exchanges.

In a recent Nature article, Guarracino et al.[1] took advantage of the recently published T2T sequence of all human chromosomes, to investigate the underlying factors behind these unique characteristics. They discovered “the presence of regions in which most contigs appear nearly identical between heterologous acrocentric chromosomes” and observed a high frequency of heterologous exchanges in these regions. These exchanges provide a comprehensive explanation for the occurrence of Robertsonian fusions, thereby confirming hypotheses proposed by cytogeneticists long ago.

1- https://www.nature.com/articles/s41586-023-05976-y