EVOLUTION OF HUMAN CENTROMERS

The first complete sequence of the human genome (telomere-to-telomere, T2T) (1) made use of long-reads technologies. Even these technologies, however, are not yet able to completely resolve the haploid arrangement of the centromeres of a diploid cell line. Indeed, the authors used a complete hydatidiform mole which, being the duplication of a human male haplotype, greatly simplified the task.

Many other human genomes have since been sequenced T2T, but centromeres had to be deliberately excluded from the analysis. As a result, whereas the precise variability of the euchromatic part of the human genome is continuously being refined, no progress has been made regarding the centromeres.

In an article published online in Nature (2), a sequence of the T2T genome of a second hydatidiform mole was published, focusing on the comparison of the centromeres of the two sequences. Additionally, samples of primate centromere sequences were also analyzed. The centromeres have been found to be the most rapidly evolving regions in primates and humans in particular. These results substantiate the evidence obtained from FISH experiments many years ago (3).

1. https://www.science.org/doi/10.1126/science.abj6987?url_ver=Z39.88-2003&rfr_id=ori:rid:crossref.org&rfr_dat=cr_pub%20%200pubmed
2. https://www.nature.com/articles/s41586-024-07278-3
3. https://www.sciencedirect.com/science/article/abs/pii/088875439580048Q?via%3Dihub