

ФИНАНСИРОВАНИЕ РАБОТЫ

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В работе не было экспериментов с участием животных или людей.

КОНФЛИКТ ИНТЕРЕСОВ

Авторы заявляют об отсутствии конфликта интересов.

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New in Centromere Genomics: Lessons from the First T2T Human Genome Assembly

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With the recent development of long-read sequencing technologies, it is now possible for the first time to read a complete gapless sequence of the human genome. The result was the first T2T (telomere-to-telomere) genomic assembly, published by an international consortium of scientists in 2022. The most significant contribution of the new assembly were the centromeric regions consisting of highly repetitive satellite DNA. In this review, we will briefly list the major achievements of the T2T consortium related to centromeres and take a closer look at the unexpected findings of cytogenetic magnitude that analysis of first assembled human centromeres has brought, such as the “split” centromeres of chromosomes 3 and 4, mega-inversion in the active centromere array of chromosome 1, haplotypic epialleles in the centromere of X chromosome and the macro-repeats found in several centromeres.

Keywords: alpha satellite, centromere, HOR-haplotype, inversion, macro-repeats, centromeric epialleles, T2T-consortium