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Satellite DNAs as modulators of gene expression: a case study of human alpha satellite DNA

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Satellite DNAs are tandemly repeated sequences preferentially assembled into large arrays within constitutive heterochromatin. In many species, including humans, satellite DNA repeats are in addition to heterochromatin dispersed in the vicinity of genes within euchromatin. Here we reveal that such genomic organization enables partial suppression of genes located nearby dispersed satellite DNA repeats. The gene suppression effect is observed at standard physiological conditions but is more pronounced after heat stress when satellite transcription is significantly enhanced, and this novel mode of gene regulation is common to both human alpha satellite DNA and the insect Tribolium castaneum major satellite DNA. Satellite DNA-mediated gene repression is based on enrichment of silent histone marks at dispersed satellite repeats which is guided by satellite DNA transcripts. We show that satellite DNA repeats act as mutagenic units able to move and to insert within or near genes where they serve as nucleation cores for repressed chromatin. Variation in their insertion generates gene expression diversity among individuals which could facilitate adaptive evolution while some insertions could affect proper gene function and cause a disease.