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Title: Modeling Satellite DNA Organization

Abstract:

Repetitive DNAs comprise large portions of eukaryotic genomes. Satellite DNAs (satDNAs) are abundant tandemly repeated DNA sequences found near centromeres, telomeres, and on sex chromosomes. SatDNAs originate through polymerase slippage, recombination between repeat elements, or TE-mediated mechanisms. Arrays of satDNA repeats are highly dynamic over short periods of evolutionary time: they vary in copy number and organization through unequal exchange, and other processes. SatDNA array expansion is thought to decrease organismal fitness but the relative importance of processes shaping satDNA evolution in natural populations is poorly understood. Population genetics studies have primarily focused on studying estimating copy number variation in satDNA arrays, due in part to limits in empirical data, as the repetitive nature of satDNAs make them difficult to study in detail. Recent advances in DNA sequencing now make it possible to infer satDNA organization at the sequence level, providing a richer source of empirical information. Here we provide a novel population genetics approach to study sequence variation in satDNA arrays. We simulate the effects of mutation, unequal exchange, gene conversion, drift, and selection on satDNA array sequence, structure, and organization in populations in a forward simulation framework. We designed a new probabilistic model for unequal exchange and gene conversion that takes into account sequence divergence between monomers in the repeat array. We have identified summary statistics that capture the variation in repetitive satDNA arrays independent of copy number. We use Bayesian inference and regression approaches to infer recombination rate and gene conversion from simulated data and empirical data from a natural population of *Drosophila melanogaster*. We show that our approach could be useful for understanding how mutation, recombination and drift shape satDNA evolution under neutral evolution and selection.