Efficient simulation of introgression, admixture and local ancestry

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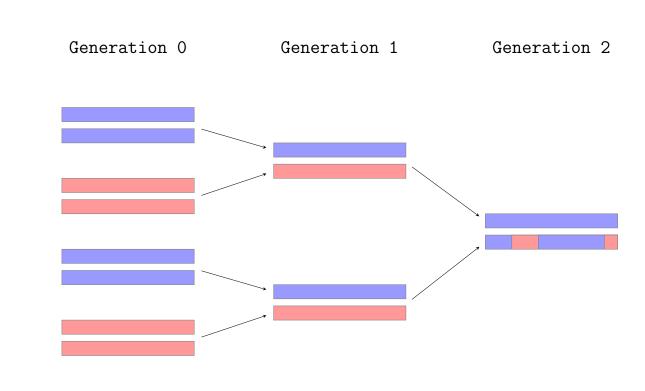
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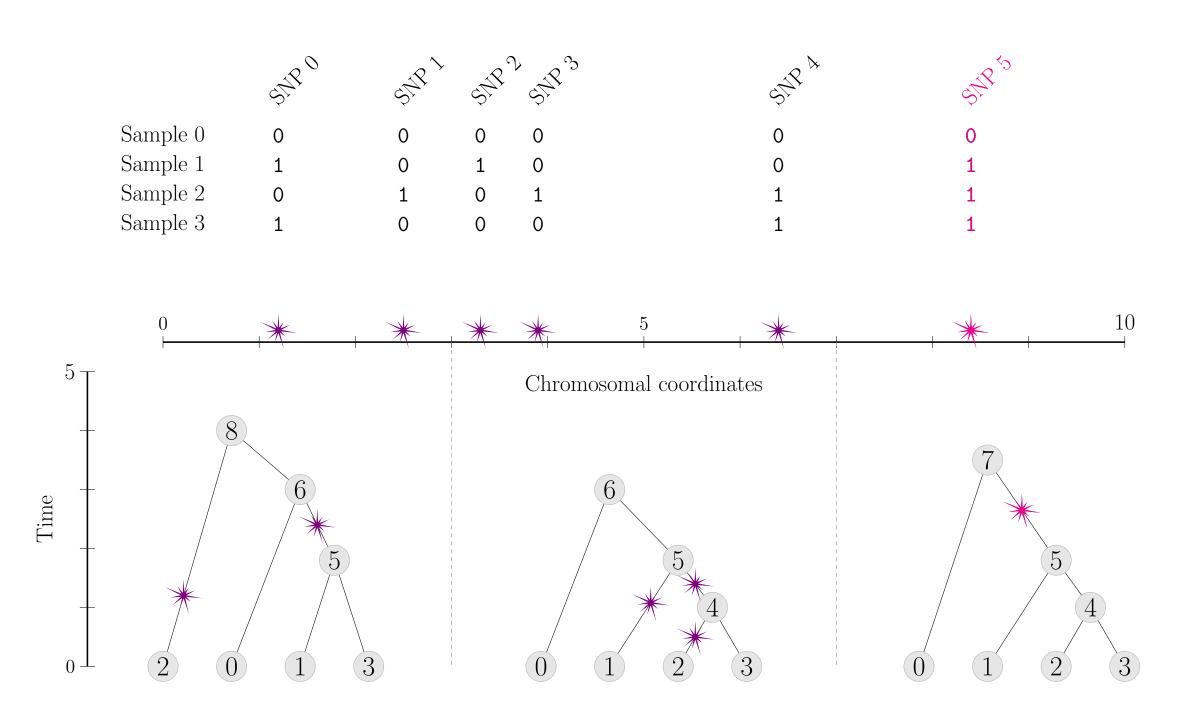
Introduction

To assess the performance of methods in population genetics, we often wish to simulate realistic genetic datasets while retaining detailed information about the history of the simulated genomes. This poster briefly describes how we can efficiently simulate genetic information while also keeping information about the ancestral population that particular genomic segments have been inherited from, often called the local ancestry of the sample. In all pictures here, we represent ancestries with colours.

Section 1 introduces the tree sequence [1], a data structure that is capable of encoding of a complete genealogy for a sample of chromosomes at each chromosomal location. Section 2 shows how local ancestry information can be stored and extracted from tree sequences. Section 3 outlines a method for simulating such information using recent advances implemented in the tree sequence simulation software magnine [1] and SLiM [2]. Section 4 demonstrates the performance of this method. Section 5 provides further information for the interested viewer.

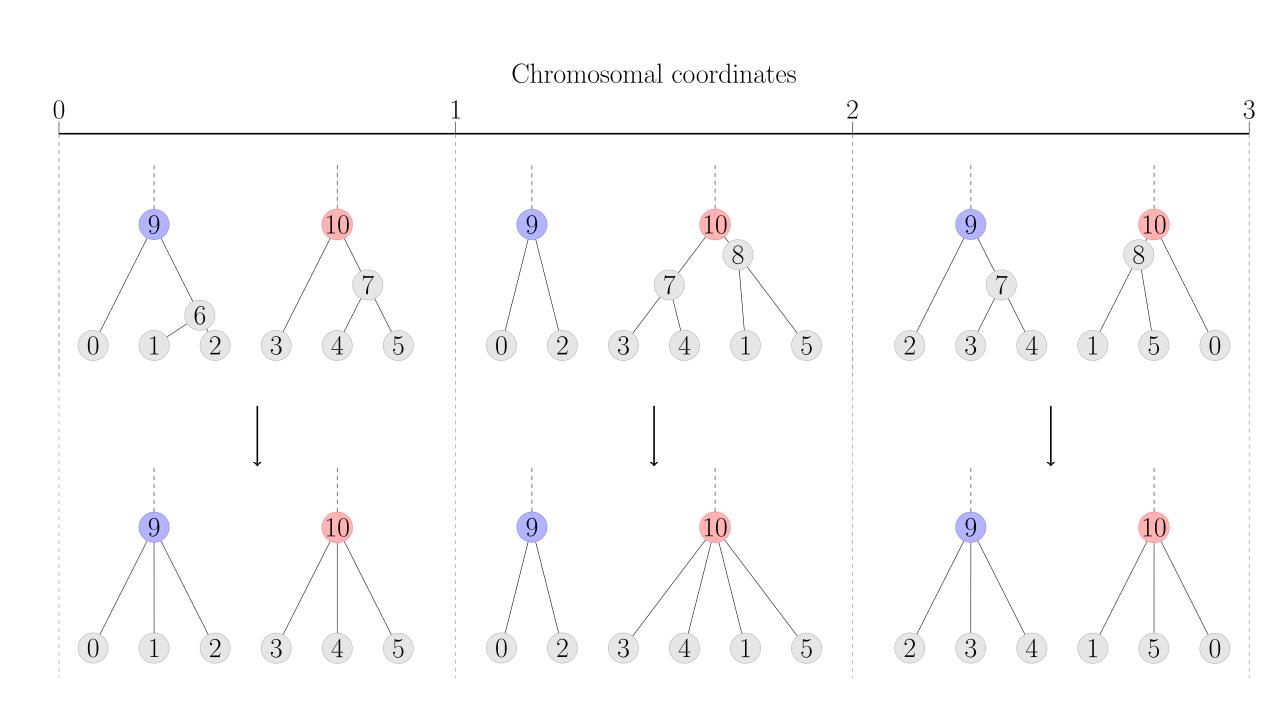


1. Tree sequences



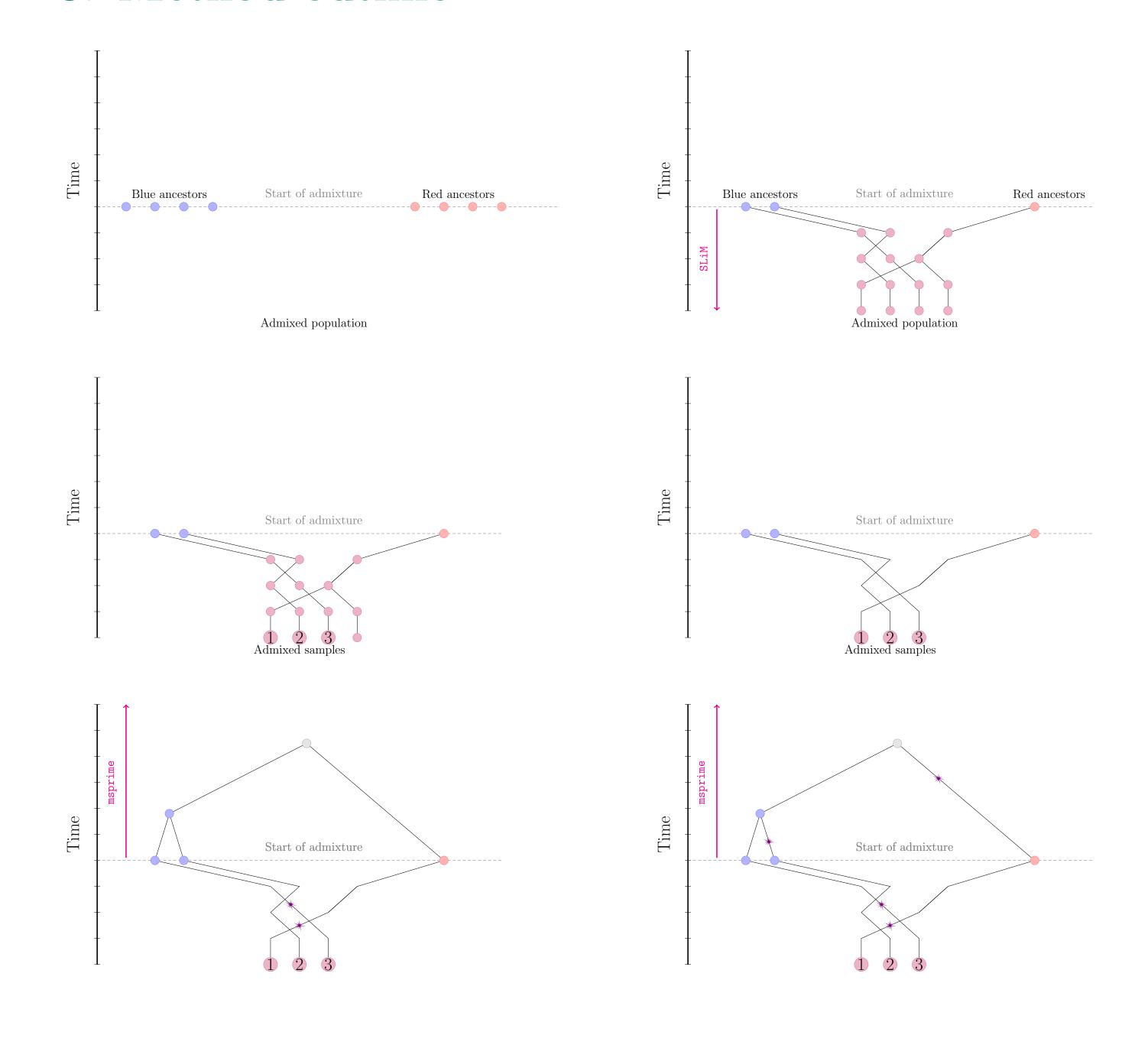
A tree sequence [1] consists of: nodes, which represent the alleles held by different chromosomes in the history of the sample; edges, which represent genealogical relationships between the alleles; and mutations, which encode full haplotype data for each samples. The tskit package encodes tree sequences as a compact set of tables that can be efficiently created, queried and modified. See [1, 2, 3] for more information.

2. Local ancestry in tree sequences



By assigning population labels to the nodes that correspond to ancestors of the sample, tree sequences can store the sample's local ancestry. The branch joining a sample node to an ancestral node shows its ancestry. Extracting this information efficiently is challenging due to correlations in genealogical structure between samples, and across chromosomes; in an upcoming paper, we will describe an algorithm that enables us to do this.

3. Method outline



4. Method performance

	Missing data	Run time	File size	Selection
msprime	4.0%	6 sec	9 Mb	No
msprime + full ARG	0.0%	53 sec	1700 Mb	No
SLiM	0.0%	> 1 hr	41 Mb	Yes
slime	0.0%	86 sec	39 Mb	Yes

To illustrate the power of our method, slime [3], we simulated a toy demographic scenario inspired by the history of Neanderthal introgression into the Eurasian population. We simulated 200 chromosomes of length 50 Mb from 100 present-day Eurasian individuals, assuming a 2% introgression of Neanderthals into Eurasians 2500 generations ago. For simplicity, we assumed a constant effective population sizes of 5000 individuals, a uniform recombination rate of 1×10^{-8} bp per generation, a uniform mutation rate of 1×10^{-8} bp per generation and neutral variation. However, note that all of these methods can deal with more complexity than this. In particular, both slime and SLiM are capable of simulating under selection.

5. Acknowledgements, references and further information

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[1] Kelleher, J., et al. (2016). Efficient Coalescent Simulation and Genealogical Analysis for Large Sample Sizes. PLOS Computational Biology, 12(5).

[2] Galloway, J., et al. (2018). Tree-sequence recording in SLiM opens new horizons for forward-time simulation of whole genomes. Molecular Ecology Resources, (November 2018), 552–566.







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