Phylogeny and population genetics: The mutation process on the ancestral line

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We consider a well-known observation at the interface of phylogeny and population genetics: mutation rates estimated via phylogenetic methods tend to be much smaller than direct estimates from pedigree studies. To understand this, we consider the Moran model with two types, mutation, and selection, and investigate the line of descent of a randomly-sampled individual from a contemporary population. We trace this ancestral line back into the distant past, far beyond the most recent common ancestor of the population (thus connecting population genetics to phylogeny) and analyze the mutation process along this line. We use a probabilistic tool, namely the pruned lookdown ancestral selection graph, which consists of the set of potential ancestors of the sampled individual at any given time. A crucial observation is that the mutation process on the ancestral line is not a Markov process by itself, but it becomes Markov when consindering a broader state space. Relative to the neutral case (that is, without selection), we obtain a general bias towards beneficial mutations. These results shed new light on previous analytical findings of Fearnhead (2002).