

1163-92-967

Lauren A Sugden* (sugdenl@duq.edu). *Mining genomic data for patterns of adaptive mutations, genes, and pathways*. Preliminary report.

Interpreting the signatures of human evolution in modern-day genomic data is a persistent challenge that requires ever more sophisticated mathematical approaches. I will describe here a framework for detecting patterns of adaptation at the single mutation level, and up to the level of genes and gene pathways. I will first describe a probabilistic model that leverages the correlated patterns left by a mutation spreading rapidly through a population, as well as a hidden Markov model that can improve sensitivity and provide measures of uncertainty. I will discuss recent efforts to build on this model to incorporate patterns of adaptation on larger genomic scales, in order to assess the adaptive force experienced by a gene or pathway as a functional unit. I will demonstrate the ability of the initial probabilistic model to identify well-known adaptive mutations in well-studied populations, as well as its usefulness in proposing plausible targets of selection in the ‡Khomani San hunter gatherer population of southern Africa, an example of a historically underrepresented population in genomic studies. I will then share some preliminary data concerning the hidden Markov model integration of multiple scales, and the benefits derived from this approach. (Received September 14, 2020)