ABSTRACT

Several important questions in human evolution center around the relationship between present-day human populations and closely-related but extinct populations like Neandertals. Did these two populations interbreed (a process termed admixture). If so, are there regions in the genomes of present-day humans that trace their ancestry to Neandertals and what are the biological consequences of these regions? Answering these questions requires genome sequence data from present-day populations as well as Neandertals, which have now become available, as well as appropriate statistical models.

In the first part of my talk, I will focus on the question of whether Neandertals and modern humans admixed. I will describe a statistical model to estimate the time of last genetic exchange between the two populations. Applying this model to data from the 1000 Genomes Project in conjunction with a Neandertal genome sequence, we find that admixture between the ancestors of Neandertals and present-day non-Africans occurred less than 100,000 years ago. Given that admixture indeed occurred, I will describe the problem of discovering regions of an individual genome that have Neandertal ancestry. I will present a discriminative statistical model designed for this task that allows us to infer the first map of Neandertal ancestry in modern humans. I will conclude with examples of novel biological insights that can be obtained from this map.