

Preface

Coalescent theory has gone from an obscure corner of population genetics to a central concept for anybody that studies variation at the sequence level.

Besides filling the obvious need for a book on this subject, it is also our wish to present this theory in a straightforward and elementary manner that could dispel the misconception that coalescent theory is inherently very difficult and needs a strong mathematical background to understand it. The key issues needed for data analysis require only basic combinatorics and probability theory. Despite the present prominence of coalescent theory, it also belongs to the future. From an application point of view, human evolution and association mapping/fine scale mapping are two areas that are bound to grow enormously in the next few years. And to make optimal use of the coming flood of data, theoretical advances will be needed. There are areas where present theory fails (or is impractically slow) in the presence of real data and if empirical researchers are to use coalescent based method, there are plenty of challenges for the theoretician both in modelling and in improvement of simulation algorithms.

The present book is definitely not exhaustive, but is only meant to provide a good basis for further study. Chapter 1 provides the basics for understanding the assumptions behind and derivation of the basic coalescent model. Chapter 2 introduces the models of alleles and sequences and associated mutation processes, and Chapter 3 gives some examples of quantities that can be calculated on coalescent genealogies. The basic coalescent is naive for many applications and Chapter 4 and 5 relax the assumptions of the basic coalescent to allow for population size changes, population structure, various forms of selection, and recombination. Chapter 6 changes the emphasis from describing the coalescence structure to inferring parameters from data using knowledge on this structure. In Chapter 7 and 8 two areas of much current interest and where coalescent theory is likely to play a major role are introduced. Chapter 7 discusses the usage of coalescence theory in the field of linkage disequilibrium mapping that aims at locating genes underlying common diseases. Chapter 8 relates the potential and use of coalescence theory to human evolution. In an appendix a brief introduction to the web based tools developed in connection with this book are presented. The collection of tools can be found at <http://www.coalescent.dk>.