Preface

Why you should read this book

My mind's-eye title for this book is *A Fireside Chat about Molecular Population Genetics*. Winters are cold here in Canada, and I like to sit back by a fire and warm up to the alluring collection of ideas in molecular population genetics. I'd like you to, as well. If you live somewhere hot, then substitute the fireplace and cushions with a shady tree and hammock. Let this book be your hot chocolate or your lemonade. That sums up my outlook for this *Primer*.

So, what are these alluring ideas? Here are a few savory questions that molecular population genetics aims to answer. What are the genomic inner-workings of adaptations and how do we see them in DNA? How much of evolution is actually driven by natural selection versus something else? How does the ebb and flow in the abundance of individuals over time get marked onto chromosomes to record this history? Molecular population genetics is the main way that researchers apply theory to data to answer questions like these. It provides the way to learn about how evolution works and how it shapes species by looking at DNA. It lets us understand the logic of how mutations originate to then change in abundance in populations to potentially get locked-in as DNA sequence divergence between species. This crucial role in modern science stems in no small part from the mainstreaming of population genomic sequencing technologies that reinforce the ever-growing relevance of molecular population genetics to diverse problems in biology. All of this makes it important for you to start on your way to learning about molecular population genetics.

I presume that you are a novice to studying population genetics from a molecular angle. Welcome! I also presume that you would like your first foray into mol-pop-gen to be friendly and unintimidating, while nevertheless enlightening you about how we can think about evolution in a genetic way. That is what I aim to do with this book. This *Primer* is not, however, an all-encompassing text in population genetics. I provide only limited detail of classic population genetics theory. Fortunately, many excellent resources for classic population genetics theory are readily available, from the introductory to the advanced. I *do* expect that you have basic knowledge about genetics, molecular biology, and evolutionary principles. If you are rusty, take heart, as I provide quick reminders about critical basic background at the relevant points. My reason for minimizing coverage of both basic biology and classic population genetics is so that we can jump right in to the portion of population genetics that deals explicitly with molecular data and ideas. I take DNA sequence as our starting point, and then I bring in key concepts from classic theory as we need them.

There are many ways that I could have chosen to organize the material in this book. I settled on a way that makes sense to me and that I hope will make sense to you. I start with the tangible and build toward the more abstract concepts, then bring it all home with integrative case studies. I start by jumping in with the nuts and bolts of molecular population genetics: what is genetic variation at the molecular level and how do we

measure it? With these concrete metrics in hand (Chapters 1–3), I then take a pause for a moment to step back and deal with the inevitable questions: why are we doing this? What is the context? What does it mean to see new mutations and polymorphisms and DNA differences between species? What should we *expect* to see? This gives us a chance to connect ideas of neutral molecular evolution (Chapter 4), the pistons and motors in this tenuous mechanical analogy, to those nuts and bolts of mutation and genetic variation. With this new firm footing in both practical and conceptual topics, I then drive into some more challenging but important concepts relating to genealogies in genome evolution (Chapter 5), including the basics of coalescent theory and phylogenies, and to recombination and linkage disequilibrium (Chapter 6). That wraps up the groundwork.

The next phase of the book explores the theme of confronting neutral models with data (Chapters 7–8). What happens when the real world does not match the convenient predictions of the standard neutral model? How should we anticipate selection and demography to perturb molecular signatures in genomes? How can we detect it? As a capstone to having worked through and built up all these views toward thinking about evolution in populations with molecular data, I end with a series of case studies that integrate many of the ideas and approaches from throughout the book (Chapter 9). I hope that this flow works and helps you to get the ideas. I also hope that it keeps you engaged to see why so many people have been enchanted by the genetics of evolution for over 100 years and why molecular population genetics, in particular, remains such a flourishing discipline today.

The biochemical details of DNA mixed together with mathematical models of evolutionary change, spiced up with the lifestyle realities of wild organisms, makes for a rich and complex scientific soup. For our purposes here, we focus on the essentials of molecular population genetics. We will zoom in and out to think about single nucleotides and individual gene loci and whole genomes; I will take for granted that "genomics" is simply an integrated part of modern molecular population genetics.

My intention with this book is to provide a concise and readable introduction to the important ideas and caveats for understanding evolution at the molecular level. Whether your interest is the adaptive process or the historical demography of populations, there is something to chew on in these pages. To give you a peek at some of the complexities and the emphasis of current research in this field, I also include some intriguing teasers for topical advanced issues. However, this book does not aim to detail thoroughly the nuances of advanced topics in molecular population genetics. I also mostly avoid details about specific analysis software and sequencing technologies, given how rapidly these techniques turn over; I take it for granted that there are robust ways to identify and quantify differences in DNA sequences. Instead, I hope that undergraduates, junior graduate students, and practicing scientists from cognate fields hoping to gain a basic understanding and appreciation for molecular population genetics will all find this *Primer* valuable for that simple goal stated in the top sentence of this paragraph.

Different traditions of study emphasize to greater degrees different portions of population genetic phenomena. In the immortal words of Christopher Wallace, "It's all good." For example, you may be familiar with the focus on the history and timing of demographic dynamics of species by phylogeographers and molecular anthropologists to learn about the history of migration and population size change over time. Or perhaps the molecular phylogeneticist's emphasis on delineating species trees in evolutionary history to understand the relationships among taxa. Or the selectionist's aim to localize recent molecular targets of adaptation in genomes. In the chapters that follow, we will consider

all these views, acknowledging each as an important facet of the dynamics of DNA change in populations from mutational origin to fixed divergence between species. They are all good ways to help us appreciate from the inside what Charles Darwin appreciated from the outside, how "from so simple a beginning endless forms most beautiful and most wonderful have been, and are being, evolved."