
Preface

Sean O’Faolain, an Irish writer (1900–1991), wrote that “there is only one admirable form of the imagination: the imagination that is so intense that it creates a new reality, that it makes things happen.” In the biomedical field we are fortunate to be living in a new technological era in which our imagination is ignited by the new DNA sequencing machines. The quantum leap is so remarkable that a new genetic reality is created. It is now up to us scientists to make it come about and change our world of DNA sequence information.

The new genetic revolution is fuelled by Deep Sequencing (or Next Generation Sequencing) apparatuses which, in essence, read billions of nucleotides per reaction. Effectively, when carefully planned, any experimental question that can be translated into reading nucleic acids can be applied.

In our book titled “Deep Sequencing Data Analysis,” under the “Methods in Molecular Biology” series, numerous leading authors contribute to the multifacet deep sequencing data analysis. We start with an introduction to a high-throughput sequencing experimental design and then present a solution for compressing the masses of data generated. Given that most current technologies are based on short reads, accuracy, coverage, and assembly are included in subsequent chapters. For the identification of variants in a given genome we look at interpretation of short reads in the context of the “Exome” information (the coding-gene portion of the genome), then apply it to the identification of disease-causing mutations. The complexity of the genetic sequence, its expression, and interpretation are dealt with as part of the analysis of tandem repeats, editing of microRNAs and alternative mRNA splicing. The closing chapters present novel methods, Chromatin Immunoprecipitation (ChIP-seq) and Reverse Transcriptase Termination Site (RTTS), their statistics and implications thereof.

The new sequencing outputs generated by deep sequencing bring up major computational challenges. The myriad amounts of sequence reads should be collated to build a coherent interpretation. We present an overview of data analysis that aids in making sense out of this information. We discuss several topics in depth presenting the “tip of the iceberg” in terms of available methods. However, we have collected key data analysis procedures that should be of great use for the beginner or savvy bioinformaticians when approaching deep sequencing data analysis.

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