Next-Generation Sequencing Data Analysis

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A Practical Guide to the Highly Dynamic Area of Massively Parallel Sequencing

The development of genome and transcriptome sequencing technologies has led to a paradigm shift in life science research and disease diagnosis and prevention. Scientists are now able to see how human diseases and phenotypic changes are connected to DNA mutation, polymorphism, genome structure, and epigenomic abnormality. **Next-Generation Sequencing Data Analysis** shows how next-generation sequencing (NGS) technologies are applied to transform nearly all aspects of biological research.

The book walks readers through the multiple stages of NGS data generation and analysis in an easy-to-follow fashion. It covers every step in each stage, from the planning stage of experimental design, sample processing, sequencing strategy formulation, the early stage of base calling, reads quality check and data preprocessing to the intermediate stage of mapping reads to a reference genome and normalization to more advanced stages specific to each application. All major applications of NGS are covered, including:

- RNA-seq: mRNA-seq and small RNA-seq
- Genotyping and variant discovery through genome re-sequencing
- *De novo* genome assembly
- ChIP-seq to study DNA–protein interaction
- Methylated DNA sequencing on epigenetic regulation
- Metagenome analysis through community genome shotgun sequencing

Before detailing the analytic steps for each of these applications, the book presents the ins and outs of the most widely used NGS platforms, with side-by-side comparisons of key technical aspects. This helps practitioners decide which platform to use for a particular project. The book also offers a perspective on the development of DNA sequencing technologies, from Sanger to future-generation sequencing technologies.

The book discusses concepts and principles that underlie each analytic step, along with software tools for implementation. It highlights key features of the tools while omitting tedious details to provide an easy-to-follow guide for practitioners in life sciences, bioinformatics, and biostatistics. In addition, references to detailed descriptions of the tools are given for further reading if needed. The accompanying website for the book provides step-by-step, real-world examples of how to apply the tools covered in the text to research projects. All the tools are freely available to academic users.

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