ABSTRACT:
High-throughput sequencing of RNA (RNA-Seq) is becoming widely applied in the biomedical and clinical realms. This technology generates an enormous amount of data that captures the gene expression landscape of the genome globally. Many aspects of gene regulation can be examined using RNA-Seq and related techniques. Here, we will focus on single-nucleotide analysis and address the challenges and opportunities in this area. We will introduce newly developed methods ranging from read mapping to segregation of genetic variants and RNA editing sites. Applications of these methods to study RNA editing in a large number of human samples will be presented. RNA editing is an important post-transcriptional mechanism where a single nucleotide in the RNA can be modified and converted into a different nucleotide, thus diversifying the expression of the genome. These applications highlight the value of the methods in revealing novel evolutionary and regulatory patterns of RNA editing.

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