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Isoform quantification in RNA sequencing: Challenges and applications

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Due to alternative splicing, over 90% of human genes have multiple transcript isoforms. Isoforms of the same gene can play distinct or even opposite biological roles. For instance, gene TP53 has an important role in oncology and different cancer types show different expression profiles of its transcript isoforms. Therefore, it is tempting to quantify RNA-Seq experiments at transcript level, rather than at the gene level. However, estimating the expression of individual isoform is intrinsically challenging because different isoforms of a gene usually have a high proportion of genomic overlap. Recently, a number of tools have been developed for RNA-Seq isoform quantification, including RSEM, Cufflinks, eXpress, Tigar2, Kallisto, Salmon and Sailfish. We performed a systematic evaluation on those methods using both simulated dataset and UHRR and HBRR, and furthermore investigated the impact of gene/isoform structures on the accuracy of isoform quantification. Besides, the library size and relative abundance of different isoforms also influence the quantification results. We determined why RNA-Seq is unable to detect less abundant TP53 transcripts and discussed its implications for the general interpretation of RNA-Seq data.

Biography

Shanrong Zhao is working as the Director, Computational Biologist and Bioinformaticist at Pfizer Inc. He has more than 20 years experience in computer science, statistics, genetics and computational biology. He also has deep scientific knowledge in immunology, autoimmune diseases, antibody design and biomarker discovery. He has demonstrated track of records in scientific initiative, innovation, and leadership, including 4 patents, over 20 peer-reviewed publications, and 20 invited talks at international meetings. A recognized Pioneer in the field of RNA-Seq, big data analysis and cloud computing. He is further enthusiastic about using NGS technology, computational approaches and informatics systems to drive drug discovery and biological research.

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