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Automatic Reporting System for Transcriptome Indel Identification and Annotation Based on Snapshot of Next-Generation Sequencing Reads Alignment

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Abstract : The analysis of Indel for RNA sequencing of clinical samples is easily affected by sequencing experiment errors and software selection. In order to improve the efficiency and accuracy of analysis, we developed an automatic reporting system for Indel recognition and annotation based on image snapshot of transcriptome reads alignment. This system includes sequence local-assembly and realignment, target point snapshot, and image-based recognition processes. We integrated high-confidence Indel dataset from several known databases as a training set to improve the accuracy of image processing and added a bioinformatical processing module to annotate and filter Indel artifacts. Subsequently, the system will automatically generate data, including data quality levels and images results report. Sanger sequencing verification of the reference Indel mutation of cell line NA12878 showed that the process can achieve 83% sensitivity and 96% specificity. Analysis of the collected clinical samples showed that the interpretation accuracy of the process was equivalent to that of manual inspection, and the processing efficiency showed a significant improvement. This work shows the feasibility of accurate Indel analysis of clinical next-generation sequencing (NGS) transcriptome. This result may be useful for RNA study for clinical samples with microsatellite instability in immunotherapy in the future.

Keywords : automatic reporting, indel, next-generation sequencing, NGS, transcriptome
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