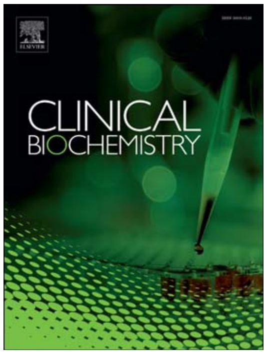

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Bioinformatics analysis of circulating cell-free DNA sequencing data

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Abstract

The discovery of cell-free DNA molecules in plasma has opened up numerous opportunities in noninvasive diagnosis. Cell-free DNA molecules have become increasingly recognized as promising biomarkers for detection and management of many diseases. The advent of next generation sequencing has provided unprecedented opportunities to scrutinize the characteristics of cell-free DNA molecules in plasma in a genome-wide fashion and at single-base resolution. Consequently, clinical applications of circulating cell-free DNA analysis have not only revolutionized noninvasive prenatal diagnosis but also facilitated cancer detection and monitoring toward an era of blood-based personalized medicine. With the remarkably increasing throughput and lowering cost of next generation sequencing, bioinformatics analysis becomes increasingly demanding to understand the large amount of data generated by these sequencing platforms.