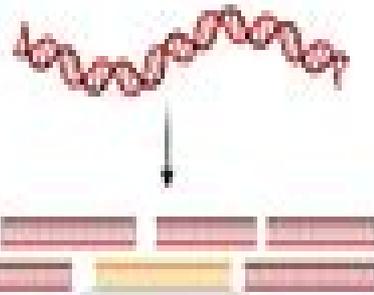


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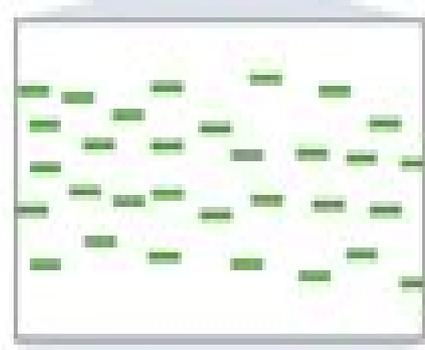
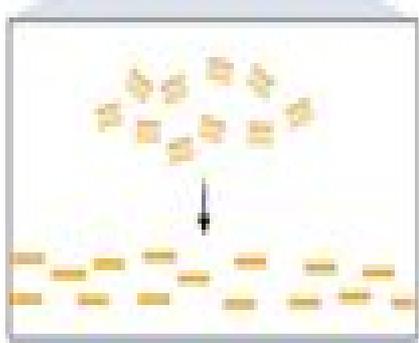
Reference Genome

Individual Genome



1 Break genome into large fragments and clone

1 Break genome into small fragments



2 Break individual clone into small fragments

2 Generate millions of sequence reads

3 Generate thousands of sequence reads

4 Assemble sequence reads for each clone

3 Align sequence reads into a reference genome

Reference genome

Individual genome



Techniques For The Analysis Of Complex Genomes

Tao Wei



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Complex Genome Analysis with High-throughput Sequencing Data Xin Li,2020 The genomes of most eukaryotes are large and complex The presence of large amounts of non coding sequences is a general property of the genomes of complex eukaryotes High throughput sequencing is increasingly important for the study of complex genomes In this dissertation we focus on two computational problems for high throughput sequence data analysis including detecting circular RNA and calling structural variations especially deletions Circular RNA or circRNA is a kind of non coding RNA which consists of a circular configuration through a typical 5 to 3 phosphodiester bond by non canonical splicing CircRNA was originally thought as the byproduct from the process of mis splicing and considered to be of low abundance Recently however circRNA is considered as a new class of functional molecule and the importance of circRNA in gene regulation and their biological functions in some human diseases have started to be recognized In this research work we propose two algorithms to detect potential circRNA In order to improve the performance of running time we design an algorithm called CircMarker to find circRNA by creating k mer table rather than conventional reads mapping Furthermore we develop an algorithm named CircDBG by taking advantage of the information from both reads and annotated genome to create de Bruijn graph for circRNA detection which improves the accuracy and sensitivity Structural variation SV which ranges from 50 bp to 3 Mb in size is an important type of genetic variations Deletion is a type of SV in which a part of a chromosome or a sequence of DNA is lost during DNA replication In this research work we develop a new method called EigenDel for detecting genomic deletions EigenDel first takes advantage of discordant read pairs and clipped reads to get initial deletion candidates Then EigenDel clusters similar deletion candidates together and calls true deletions from each cluster by using unsupervised learning method EigenDel outperforms other major methods in terms of balancing accuracy and sensitivity as well as reducing bias Our results in this dissertation show that sequencing data can be used to study complex genomes by using effective computational approaches

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