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Perspective

Major Structural Variations between the Genome Project and Genome Mapping

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INTRODUCTION

Genome mapping is the process of creating a genetic map that assigns DNA fragments to chromosomes. This map does not exist when a genome is first investigated. The map improves with scientific progress and is complete when the species' genomic DNA sequencing is completed during this process and for the investigation of strain differences, the fragments are identified by small tags. These could be genetic markers (PCR products) or DNA-cutting enzymes with a unique sequence-dependent pattern. The ordering is derived from genetic observations (recombinant frequency) for these markers in the first case or a computational integration of the fingerprinting data in the second case. The term "mapping" has two distinct but related meanings.

Different mapping methods are distinguished. Physical mapping is when traditional genetic techniques are combined with modern molecular biology techniques to achieve the same goal as genetic mapping.

Physical genetic mapping

A restriction enzyme cuts the DNA during physical mapping. Electrophoresis is used to separate the DNA fragments after they have been cut. By analyzing the fingerprints, the resulting pattern of DNA migration (*i.e.*, its genetic fingerprint) is used to identify what stretch of DNA is in the clone. Contigs (large regions of genomes) are assembled into overlapping DNA stretches using either automated (FPC) or manual (Pathfinders) methods. A good selection of clones can now be made to efficiently sequence the clones to determine the DNA sequence of the organism under study. The macro restriction is a type of physical mapping in which high molecular weight DNA is digested with a restriction enzyme with a few restriction sites. There are alternative methods for determining how DNA in a group of clones overlaps without sequencing the clones completely. Once the map is established, the clones can be used to efficiently contain large stretches of the genome. This method of mapping is more precise than genetic mapping. Independent approaches such as *in situ* hybridization can be used to map genes before complete sequencing. Mapping refers to the process of identifying a genetic element that appears to be responsible for a disease. The search is referred to as "fine-mapping" of a gene if the locus in which it is performed is already significantly constrained. This data comes from studies of disease manifestations in large families (genetic linkage) or populations-based genetic association studies.

DESCRIPTION

Genome research project

Genome projects are scientific endeavors that seek to determine an organism's entire genome sequence (be it an animal, a plant, a fungus, a bacterium, an archaean, a protist, or a virus). The genome sequence of any organism necessitates the determination of the DNA sequences for each of the organism's chromosomes. A genome project will attempt to map the sequence of a chromosome in bacteria, which typically have only one. With 22 pairs of autosomes and two sex chromosomes, humans will require 46 separate chromosome sequences to represent the entire genome.

Assemble of genome sequences

The process of assembling a genome involves taking a large number of short DNA sequences. All of these were generated by a shotgun sequencing project and reassembled to form a representation of the original chromosomes from which the DNA originated. The entire DNA from a source (usually a single organism, anything from a bacterium to a mammal) is first fragmented into millions of small pieces in a shotgun sequencing project. These fragments are then "read" by automated sequencing machines capable of reading up to 900 nucleotides or bases at a time. (AGCT stands for adenine, guanine, cytosine, and thymine, the four bases.) A genome assembly algorithm works by aligning all of the pieces and detecting all of the places where two of the short sequences, or reads, overlap. These overlapping reads can be combined, and the process can continue.

Genome assembly is a very difficult computational problem that is exacerbated by the fact that many genomes contain large

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numbers of identical sequences known as repeats. These repeats can be thousands of nucleotides long and occur in thousands of different locations, particularly in large plant and animal genomes.

The resulting (draught) genome sequence is created by joining the information sequenced contigs and then using linking information to build scaffolds.

The process of attaching biological information to sequences in the genome is known as an annotation. It consists of two major steps:

- Identifying genomic elements. Gene prediction is a process.
- Adding biological data to these elements.

Automatic annotation tools attempt to accomplish all of this through computer analysis, as opposed to manual annotation, which requires human expertise. In an ideal world, these approaches would coexist and complement one another in the same annotation pipeline.

The most fundamental level of annotation involves using BLAST to find similarities and then annotating genomes based on that. However, more and more additional information is being added to the annotation platform these days. The additional information enables manual annotators to deconvolute discrepancies between genes that have been annotated the same way. Through their Subsystems approach, some databases use genome context information, similarity scores, experimental data, and integrations of other resources to provide genome annotations. Other databases, such as Ensembl, rely on both curated data sources and a variety of software tools in their automated genome annotation pipeline.

Structural annotation entails identifying genomic elements.

- Coding region.
- Gene structure.
- Regulatory motif distribution.

The process of attaching biological information to genomic elements is known as a functional annotation.

- Biochemical function.
- Biological function.
- Regulated and interacted with.
- Imagination.

These steps may include biological experiments as well as silky analysis.

To allow scientists to view and share genome annotations, several software tools have been developed.

The human genome project's next major challenge will be genome annotation. Now that the human and several model organism genome sequences are nearly complete. Identifying the locations of genes and other genetic control elements is frequently referred to as defining the biological "parts list" for an organism's assembly and normal operation. Scientists are still at an early stage in the process of delineating this pans list and understanding how all the parts "fit together".

CONCLUSION

Genome annotation is an active research area that involves a variety of organizations in the life science community that publish the results of their efforts in publicly available biological databases accessible *via* the web and other electronic means. Here is an alphabetical list of ongoing projects genome annotation related projects:

- DNA elements encyclopedia (ENCODE).
- Ensemble.
- Entrez gene
- Gene ontology consortium.
- GencR IF.
- Uniprot.
- Vertebrate and genome annotation project (Vega).