

Unit 2 - Structural Genomics Part 1

Thursday, September 29, 2022 10:00 AM

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Genomics and Bioinformatics

Genomics and Bioinformatics

- ☐ **Genome:** all DNA content + info in a haploid cell
- ☐ **Bioinformatics:** the info/content of genomes
- What does genomics + bioinformatics involve?
 - Large datasets
 - High-throughput methods
 - Fast data gathering
- Sequences often reported as a FASTA type file
 - Text-based format representing nucleotide or peptide sequences
 - Base pairs or amino acids represented by single-letter codes
 - Start: a single-line description
 - marked by ">"
 - Followed by lines of sequence data

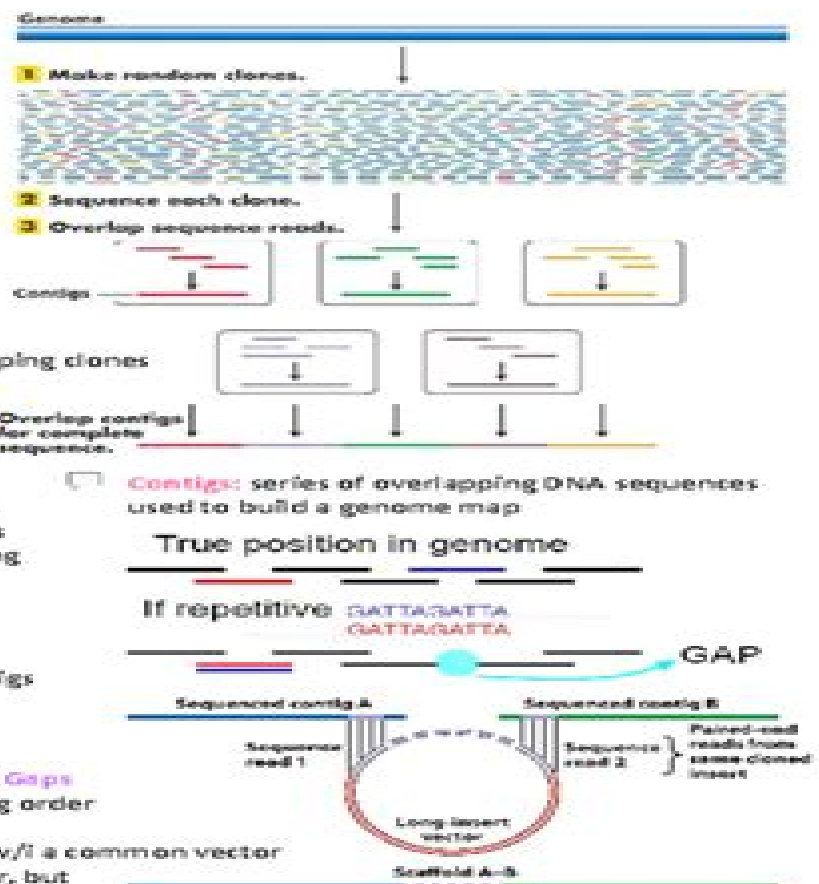
Genome Sequencing

- ★ **Steps of genome sequencing**
 1. Preliminary sequencing
 2. Finishing/Assembly
 3. Annotation → Assigning f²s to genes
- Human genome seq. uses two diff. approaches
 - **Hierarchical genome shotgun (HGS)**
 - Uses random markers
 - Organize (map) segments of DNA
 - Choose minimum number of overlapping clones
 - ◻ Tiling path
 - Sequence
 - **Whole genome shotgun (WGS)**
 - Fragment genome into pieces
 - Sequence all pieces
 - ◻ Completed through auto seq.
 - ◻ Involves making random clones
 - Assemble pieces into **contigs** spanning each chromosome
- Shotgun sequencing end reads
 - Primers from vectors
 - Reads are approx. 600 bp
 - Overlapping reads are assembled into contigs
- Problem: **Repetitive DNA + duplications**
 - Can fit in different places
 - Can make it challenging to assemble
 - Can be spaces where no contigs overlap → **Gaps**
 - More challenging to determine contig order
- Solution: **Paired-End Reads**
 - Recognizing end reads of two diff. contigs w/ a common vector
 - If two contigs are found on the same vector, but

More info on the differences [here](#) and [here](#)

Genomics

- ☐ **Structural genomics:** an assembly of contiguous stretches of chromosomal DNA
- ☐ **Functional genomics:** characterizes the role(s) of transcripts and proteins
 - i.e. Level of expression, biological function
- ☐ **Comparative genomics:** comparing genomes from different organisms
 - Sequence conservation
 - Nucleotide composition bias



Structural Genomics Part 2 Structural Genomics Part

David Kirk



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