Human Genome Sequencing

Generating a Reference Genome Sequence (e.g., Human Genome Project)
- Break genome into large fragments and insert into clones
- Order clones
- Break individual clones into small pieces
- Generate thousands of sequence reads and assemble sequence of clone
- Assemble sequences of overlapping clones to establish reference sequence

Reference Sequence

Generating a Person’s Genome Sequence (e.g., Circa ~2016)
- Break genome into small pieces
- Generate millions of sequence reads
- Align sequence reads to established reference sequence
- Deduce starting sequence and identify differences from reference sequence

Reference Sequence

....TATGCGATGCGTATTTCGTAAA....