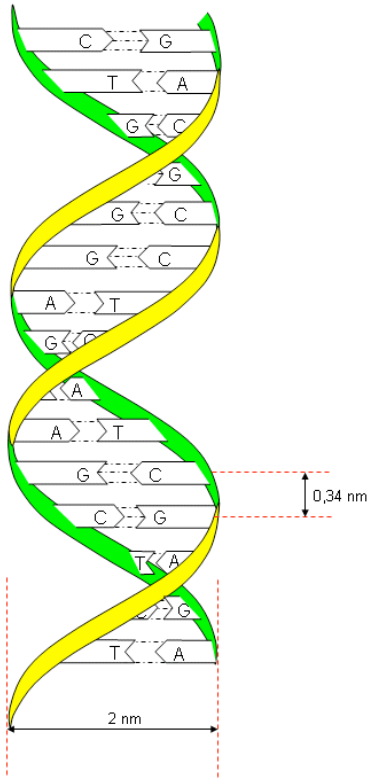
2. Genes and proteins

- The sequence as a model of DNA
- Genes: from Mendel to molecular biology
- The genetic code
- A translation algorithm
- Implementing the genetic code
- Algorithms + data structures = programs
- The algorithm design trade-off
- **DNA sequencing**
- Whole genome sequencing
- How to find genes?

DNA sequencing

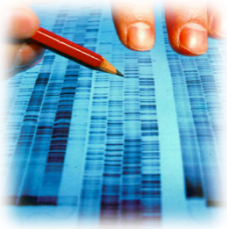
DNA sequencing

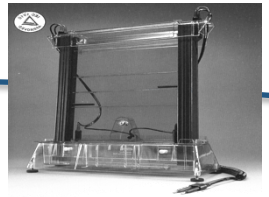


…
GATCACCTCACTACGGGTCAGGGGAA
GGAAAGGGGAACTGAGAGATTTGCAG
TGTGAGAAGCAGTCCCAGGAGTTAGA
AGTAGTGGCTCCATGACTCACAAATT
AACTTCCCTTTCAGGCAGGGCTTCTT
ATTTTCCTTAGCATCCCTGTCTTGAT
CCCAGCCTGCTCAGACCCCTGCCTCT
CACTGCAAGATGTGCTTGAGTATGAG
AGTCAGGAATGTTACTTCTCAGAGGC
GCCAAATGGCAGTTGTCACAGGGTCA
TCATAGAGGGTATATGTTTACTGCAC
TGGGCTCTGAGGCTTGCTTGTGAAGA
AACAGAAGCTAAGGGATCCAGGGAGT
CCCAACTTAGAGAGTCCCACAGGCCC
ACACTCTGGTTCTGTTGGCAGGAAAA
TTTGGCTGAATTGGGGCAGGAAGTTG
TGTAACAAAACGATTACATCCATTTT
TGCAAGGCAAGAGTGAGCTATTCACC
TCCATGTTGGTGATATTTTTTGCCAT
ATAAGCAGCTAATTCCTTTCAGTAAT
TCTACTCTAAACTAGTCTTAATGTGA
CTTCTATATAAATTCTGAACTGAATA
ATTTTGGGAACGTTGTAAAAA…

Sequencing is a so-called “exponential technology”

- First sequences obtained in the early 70’s
- Next Generation Sequencers (NGS) around 2008

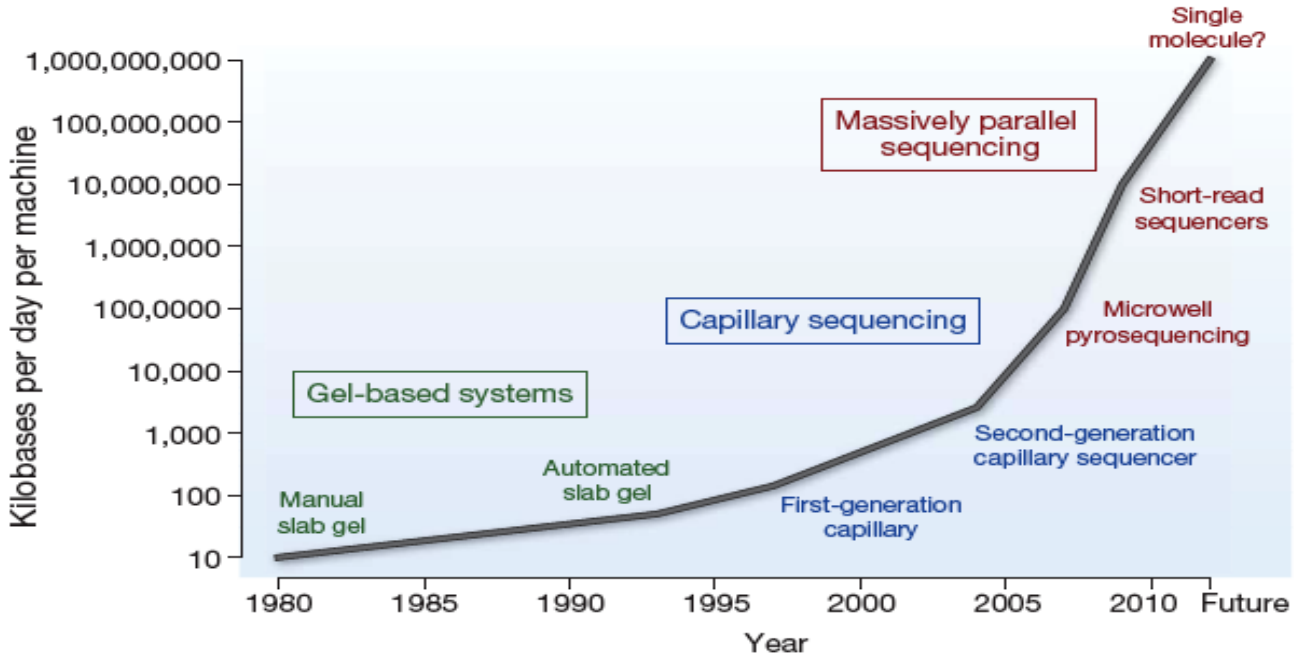




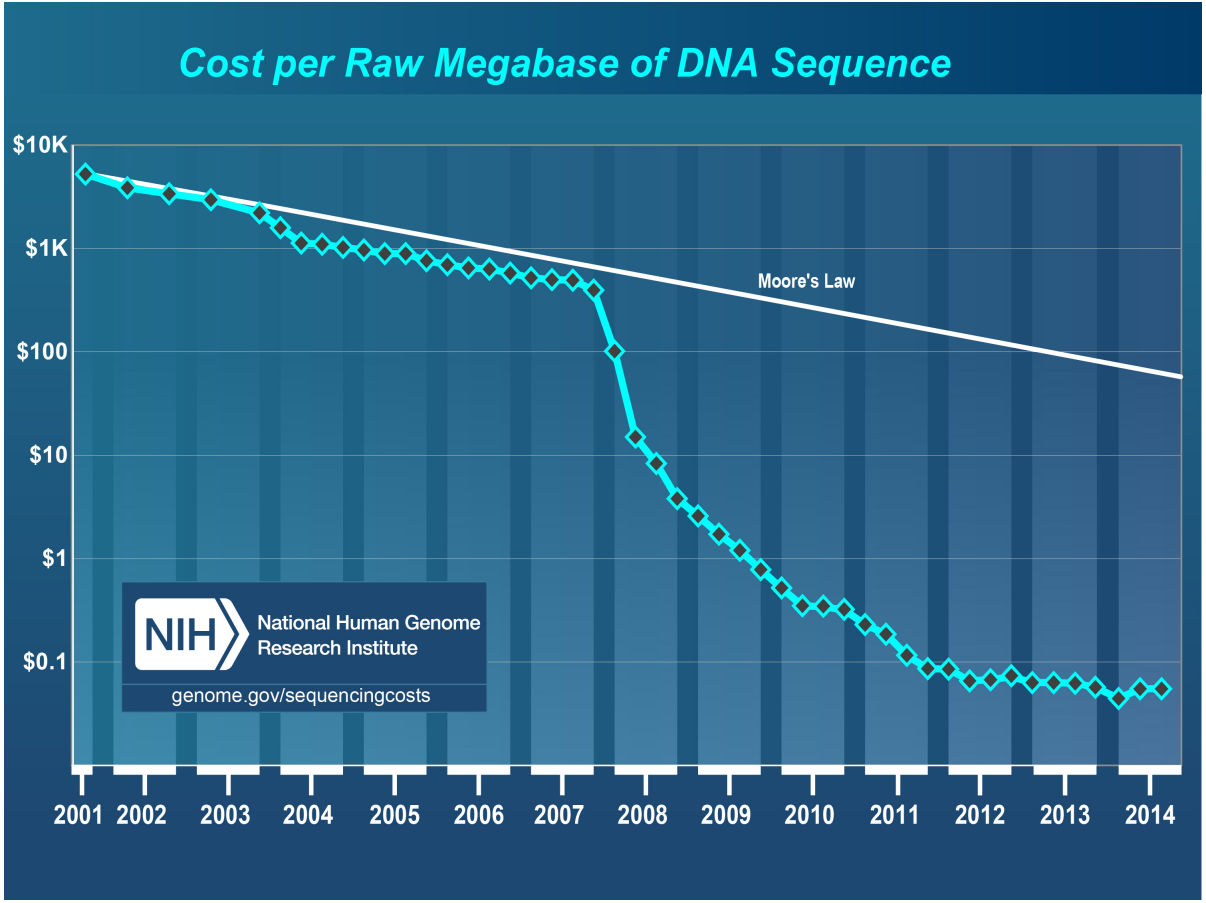
1990: 10^3 bases/day

2000: 10^6 bases/day

2010: 10^9 bases/day



“The cancer genome”, Michael R. Stratton, Peter J. Campbell, P. Andrew Futreal, *Nature*, Vol. 458, Avril 2009



http://www.genome.gov/sequencingcosts/

Next Generation Sequencers generate “reads”

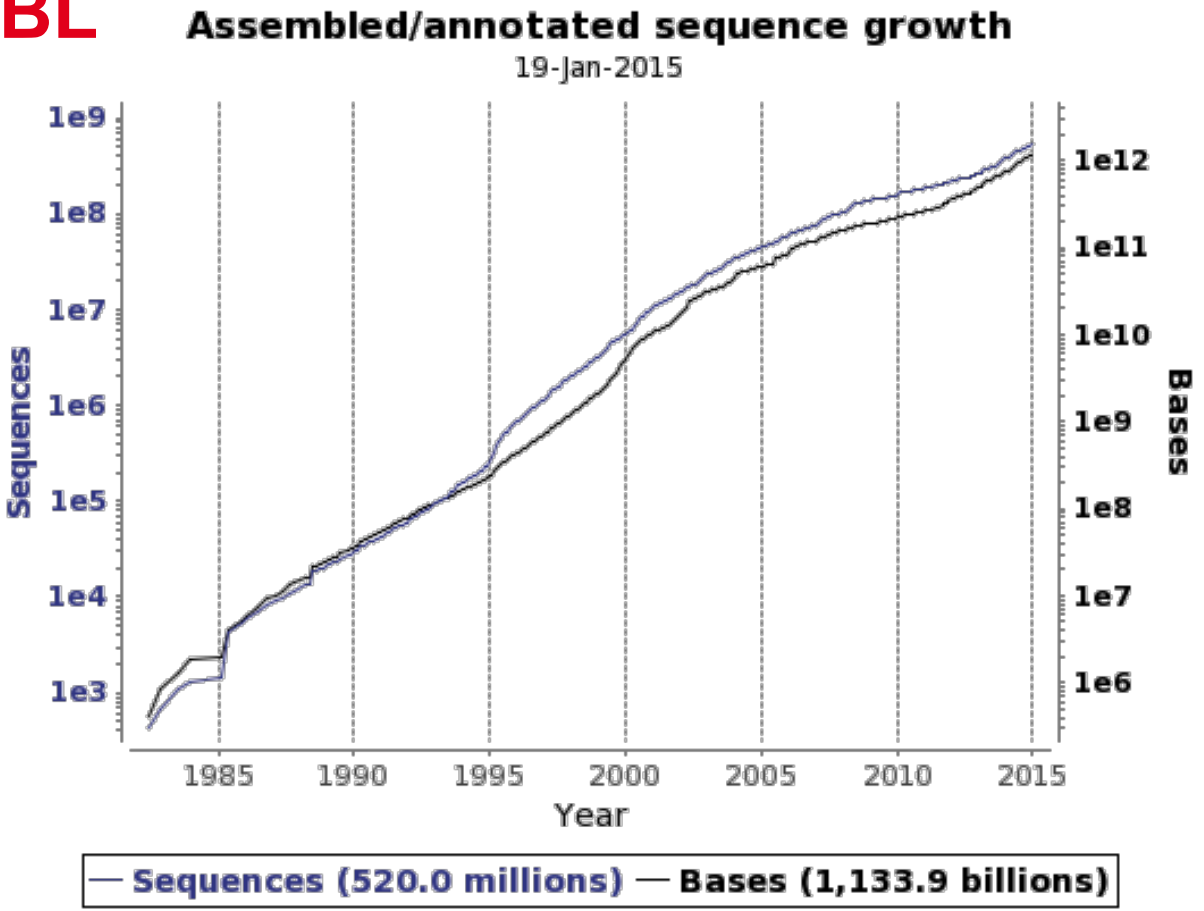
- Raw data
  - Overlapping reads to be assembled
  - Assembly algorithms

- Sequencing depth
  - The number of reads aligned over a given position of the resulting sequence
- Coverage
  - The length of the regions which are covered by reads as a percentage of the expected length of the whole genome

The International Nucleotide Sequence Database Collaboration

- An annotated collection of all publicly available DNA sequences
  - GenBank at NCBI
  - DNA DataBank of Japan (DDBJ)
  - European Molecular Biology Laboratory (EMBL)
- Submission of sequences by labs
- These three organizations exchange data on a daily basis
- http://www.insdc.org/

EMBL


Assembled/annotated sequence growth
19-Jan-2015
Sequences
1e9
1e8
1e7
1e6
1e5
1e4
1e3
Bases
1e12
1e11
1e10
1e9
1e8
1e7
1e6
1985
1990
1995
2000
2005
2010
2015
Year
Sequences (520.0 millions)
Bases (1,133.9 billions)

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