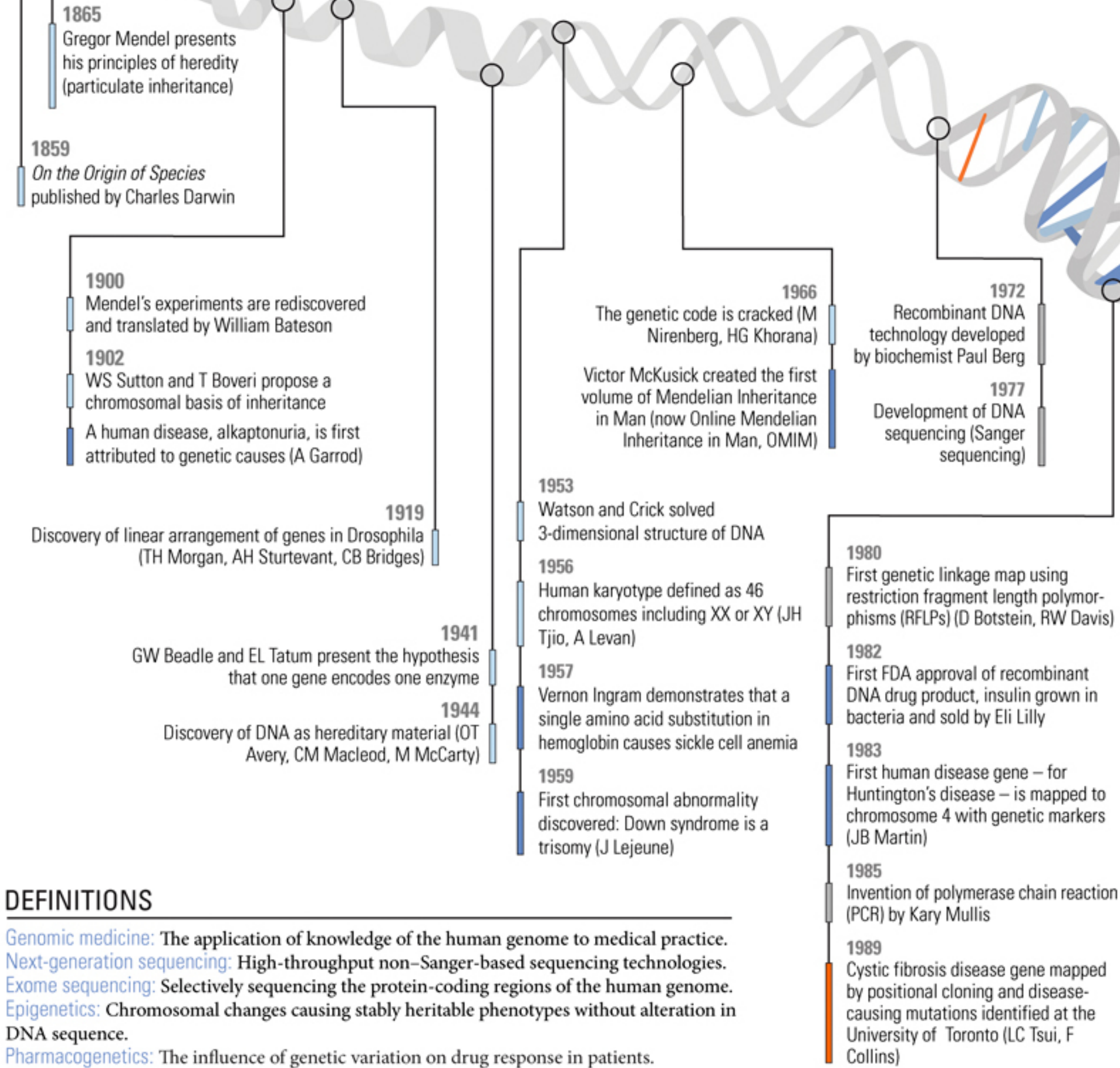


MEDICAL GENETICS OVERVIEW

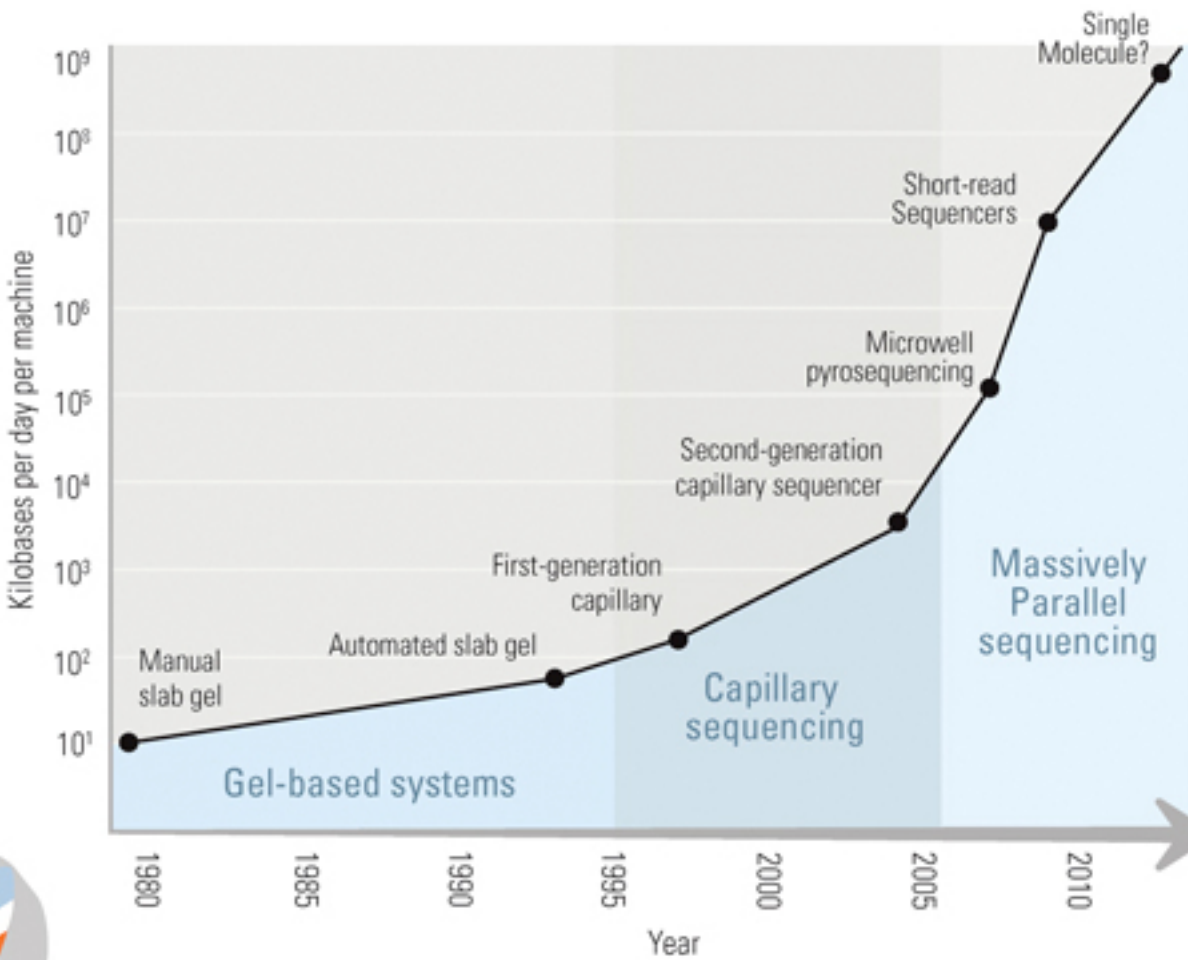
The era of genomic medicine is now coming to fruition on the shoulders of 19th and 20th century achievements in basic genetics, genetic and genomic technologies, and medical genetics. With the cost of sequencing an individual's complete genome decreasing rapidly as DNA sequencing capabilities improve exponentially, these technologies are on the verge of evolving from research platforms to clinical tools. Researchers in the IMS are working at the forefront of several trends in genomic medicine: next-generation sequencing technologies, human genetic variation, rare disease genetics, pharmacogenomics, epigenomics, and defining the ethical considerations for a new paradigm in how medicine is delivered.



DEFINITIONS

- Genomic medicine:** The application of knowledge of the human genome to medical practice.
- Next-generation sequencing:** High-throughput non-Sanger-based sequencing technologies.
- Exome sequencing:** Selectively sequencing the protein-coding regions of the human genome.
- Epigenetics:** Chromosomal changes causing stably heritable phenotypes without alteration in DNA sequence.
- Pharmacogenetics:** The influence of genetic variation on drug response in patients.

IMPROVEMENTS IN THE RATE OF DNA SEQUENCING



- 2000**
First draft of human genome announced by competing private and public projects (JC Venter, F Collins)
- 2004**
Commencement of the human epigenome project
- 2004**
Discovery of copy-number variations as a source of human sequence variation at The Centre for Advanced Genomics (TCAG)
- 2005**
The HapMap project is complete: a database of human sequence variations
- 2008**
Appearance of massively parallel next-generation DNA sequencing
- 2008**
Genomewide map of DNA methylation patterns appears (M Weber, D Schübeler)
- 2008**
First human genome completed using massively parallel next-generation DNA sequencing (JM Rothberg)
- 2009**
Appearance of single molecule sequencing

LEGEND

- Basic genetics milestone
- DNA technology milestone
- Medical genetics milestones
- U of T Achievements