Translating Genetic Sequencing into the Clinic

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Public Health Genetics

DNA Basics

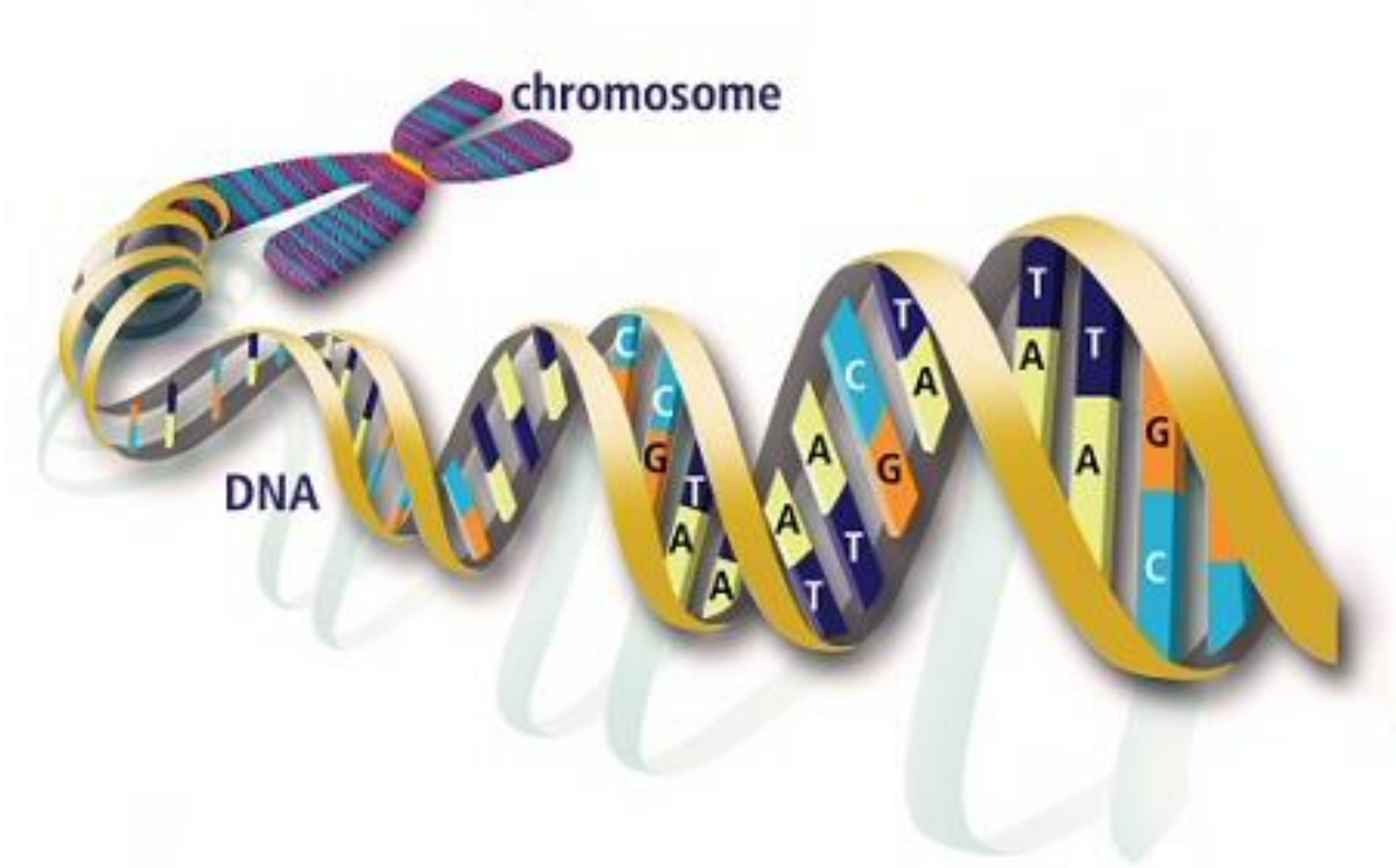
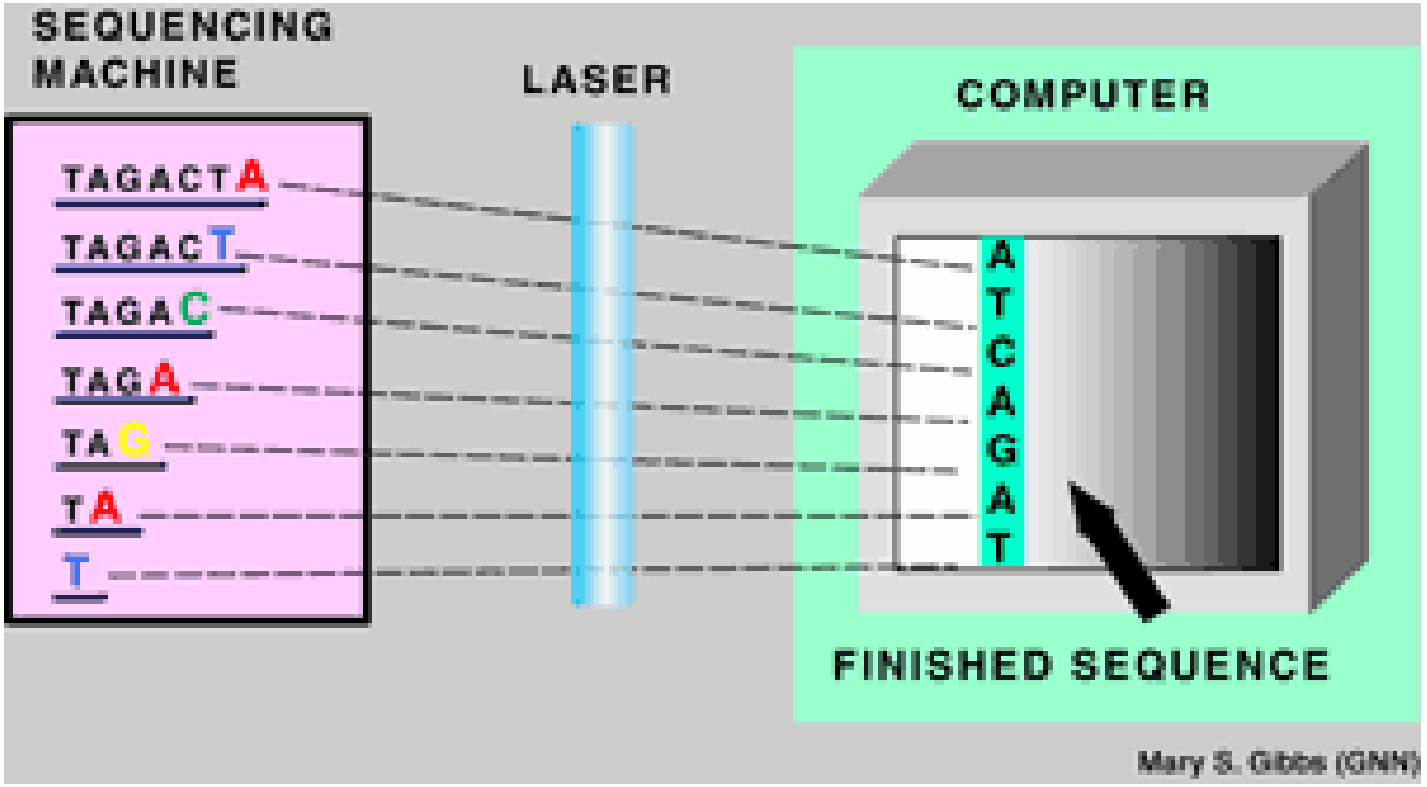


Image source: http://www.odec.ca/projects/2006/bach6k2/background.htm

Next Generation Sequencing (NGS)



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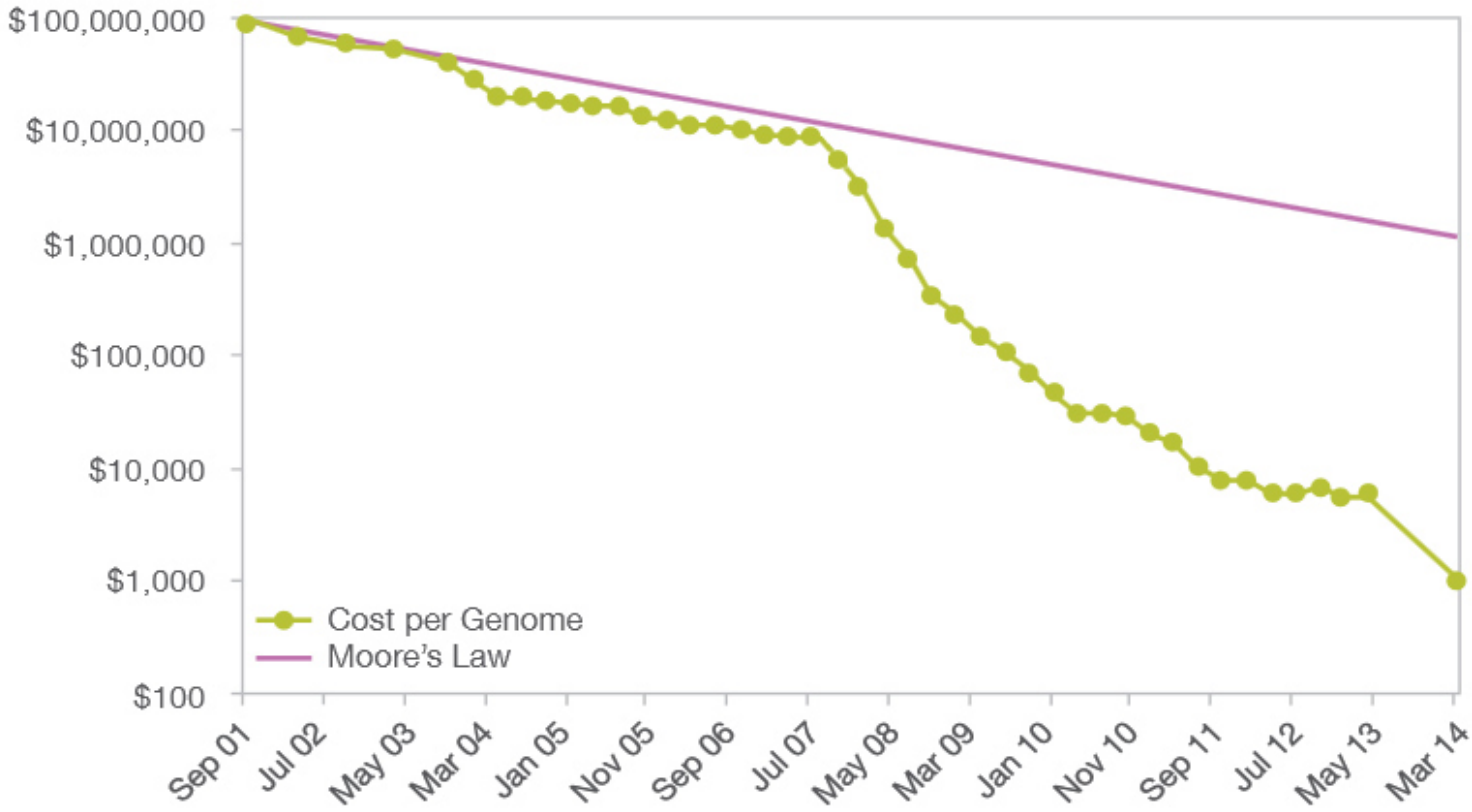

Next Generation Sequencing (NGS)

Uniqueness in 0.1% of sequence compared to any two people

Generates a raw sequence (whole genome or exome)

Comparing individual sequence to reference sequence teases out the variation

Clinical Sequencing Exploratory Research

*Moving the **Genome** Into the **Clinic***

377 Researchers
21 Institutions
1 Consortium

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www.genome.gov/CSER
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Value of clinical sequencing

- Disease diagnosis
- Clinical treatment (i.e., personalized cancer therapies)
- Prevention decisions
- Reproductive decision making (i.e., carrier status information)

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Barriers to translation

- Payer reimbursements

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Other potential barriers

- Best practices evidence gap
  - Standardizing variant annotation processes
  - Harmonizing outcome measures
- Heterogeneity in primary indications for utilizing clinical sequencing
- Regulatory oversight challenges

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Time's Up!

- **About you:**
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  - **Quick bio:** Ragan holds a BS in Exercise Physiology from Auburn University, previously researching genetic underpinnings of type 2 diabetes, and an MS in Genetic Epidemiology from UW, investigating gene-environment interactions in patients using warfarin. She is a doctoral student in Public Health Genetics. Recognizing a myriad of factors exist in the implementation of clinical sequencing into healthcare, her research interests are at the intersection of health economics, clinical validation, and health policy.