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American Society of Human Genetics

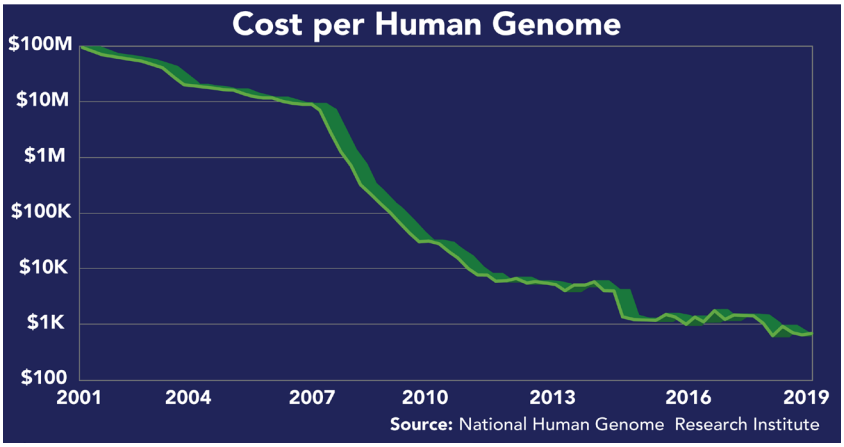
Success Stories
in Human Genetics and Genomics Research

INNOVATION IN DNA SEQUENCING TECHNOLOGIES

DNA sequencing is a standard technique used in genetic research and medicine to read a person's DNA code. Through DNA sequencing, researchers are investigating how the human genome functions and exploring the genetic causes of disease, and this knowledge is being translated into advances in clinical care. Sustained investments in the National Institutes of Health (NIH) have funded remarkable advances in DNA sequencing technologies, enabling new research and clinical applications.

Improvements in DNA sequencing are advancing genetic research and medicine. Beginning with the Human Genome Project, the National Human Genome Research Institute (NHGRI) at the NIH has funded innovative research to develop faster, lower-cost DNA sequencing technologies. This public funding together with private investments has led to the emergence of several companies producing a range of sequencing technologies[1], and the results have been remarkable. Whereas sequencing the first human genome took several years, it now takes just one day and the cost has dropped from approximately $100 million to $1,000.[2]

The availability of efficient DNA sequencing technologies is empowering publicly funded research across biomedicine on an unprecedented scale. It is enabling new approaches for investigating how the genome functions and the significance of genetic differences between people. It is allowing new ways of exploring the causes of cancer and for investigating the genetic factors involved in myriad common and rare diseases. The knowledge gained is being used to inform clinical care, providing new diagnostic tools and guiding treatment.[3]



From Sequencing to Life-Changing Treatment

Elizabeth Davis had suffered from a debilitating, undiagnosable condition her entire life. Doctors at the University of North Carolina referred her to an NIH-funded initiative aimed at moving sequencing tools to clinical application.[4] Analyzing her genome, researchers found a variant in a gene associated with dopa-responsive dystonia, a rare neurological disorder. Upon receiving medication—similar to that given to patients with Parkinson's disease—to treat her disorder, Elizabeth saw immediate improvement. She can now live a relatively normal life.

"Six weeks later, she came back to our clinic, and she left her crutches at home. It was awesome: it's why you do research."

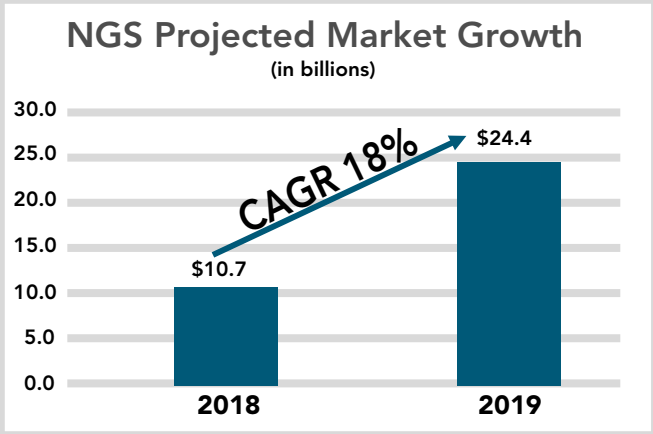
James Evans, MD, PhD
who studied Elizabeth's genome.[4]

Sequencing 1 Million: NIH All of Us Research Program

Launched in 2015, the NIH All of Us program is creating a diverse database of genetic information and other data from at least one million research participants across the U.S. The advent of cheaper, faster, and more efficient sequencing technology has made this historic effort possible. Researchers will have the opportunity to conduct genetics research on a large scale, including the study of diseases and new clinical treatment applications, with the hope of accelerating precision medicine[5]. Precision medicine—factoring lifestyle, environment, and genetics into treatment—has the potential to make treatment more tailored to an individual's needs and save on health care costs.[6]

Genome Sequencing—Drastic Cost Reduction

Sequencing a human genome was once expensive, slow and beyond the capacity of most research programs. Thanks in part to NHGRI-funded research and the emergence of new DNA sequencing companies, since 2008 costs have plummeted as new technologies have become available.[7] One company predicts that the cost to sequence an entire human genome may one day be as low as $100.[8] Such cost reductions facilitate the use of DNA sequencing tools across biomedical research and their integration into standard clinical care.



According to BCC Research, the next-generation sequencing (NGS) market is projected to grow from $10.7 billion in 2018 to $24.4 billion in 2023, with a compound annual growth rate (CAGR) of 18%.

Source: *BCC Research*

Setting a Sequencing World Record

Genetic diseases are the leading cause of death in newborns.[9] Aided by rapid sequencing platforms, researchers at Rady Children's Institute for Genomic Medicine took just 19.5 hours to deliver genetic diagnoses for newborns. Such prompt analysis and diagnosis are critical for saving newborn lives.[10]

"Our hope is that pediatric genomic medicine will one day become routine so that ultimately all children who need it can have access to this life-saving technology"

Stephen Kingsmore, MD, pioneer of faster delivery of genetic test results

How Can Congress Support Research?

Thanks to Congress' commitment to biomedical research funding, there has been a revolution in DNA sequencing. The technology is now being applied beyond human genetics and genomics, such as for investigating the spread and virulence of viruses like SARS-CoV-2, the virus causing COVID-19, rapidly developing new vaccines. Congress can support further technological advances by continuing to prioritize funding for biomedical research.

Additional Resources

All of Us Research Overview
https://allofus.nih.gov/about/all-us-research-program-overview

NIH Cost of Sequencing a Human Genome
https://www.genome.gov/about-genomics/fact-sheets/Sequencing-Human-Genome-cost

References:
ashg.org/advocacy/fact-sheets/

6120 Executive Blvd, Suite 500, Rockville, Maryland 20852 · policy@ashg.org · (301) 634-7300 · www.ashg.org

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