Genomics and Human Health

Genetics and genomics knowledge and tools are facilitating biomedical research across NIH. Two decades ago, during the completion of the Human Genome project, the National Human Genome Research Institute (NHGRI) alone funded 95% of NIH’s human genomics research. With the human reference genome in hand, genomics informs nearly all aspects of biomedical research, and this has spurred dramatic growth in the funding of genomic approaches across other NIH Institutes and Centers—funding 90% of the research today. Rapid, large-scale, and low-cost DNA sequencing is now a mainstream tool for researching the leading causes of death for Americans, such as cancer and heart disease.

In 2009, NIH added the Research, Condition, and Disease Categorization (RCDC) process to categorize and report the amount of funding in more than 280 reported categories of disease, condition, or research area. From Fiscal Year (FY) 2008 to FY 2020, NIH’s funding of human genome research has grown by over $3 billion, or from 4% of NIH’s total budget to over 10% of NIH’s budget.
Human Genetics & Genomics Research Programs at NIH Institutes and Centers

- The All of Us Research Program, funded by the Innovation Fund in the 21st Century Cures Act, serves to gather genomic and other health data from over one million participants from around the United States. Researchers can use these data to discover genetic and other factors that contribute to illness and find improved ways to diagnose and treat disease.2
- The Trans-Omics for Precision Medicine program (TOPMed), is seeking to improve our understanding of the biological processes that underlie heart, lung, blood, and sleep disorders. Supported by the National Heart, Lung, and Blood Institute (NHLBI), the project has sequenced over 90,000 individual genomes from people with diverse backgrounds.3
- The Ultra-Rare Gene Therapy (URGenT) program, funded by the National Institute of Neurological Disorders and Stroke (NINDS), supports the development of gene-based therapies for ultra-rare neurological diseases. There are approximately 7,000 known rare and ultra-rare diseases, affecting 30 million people in the U.S. Many are life-threatening and few have FDA-approved treatments, but the majority are single-gene disorders, making them excellent candidates for gene therapy.4 URGenT aims to accelerate the development of therapies for these diseases.
- Under the Human Microbiome Project, scientists have applied DNA sequencing and computational tools for characterizing the community of microbes living on and in healthy adults and those with specific microbiome-associated diseases, such as inflammatory bowel disease and prediabetes. The information generated by this project is now available to researchers worldwide to investigate the influence of microbes on human development, physiology, immunity, and nutrition.5

How Can Congress Support Research?

Congress can support the next genetics and genomics advances by continuing to fund basic biomedical research across NIH. While genetics and genomics is integrated into many areas of human health, there is still much to learn about genomic variation across all populations. More research is needed to understand the genetic underpinnings of different diseases and to better understand how to translate genomics discoveries into clinical care advances.