

## About the National Human Genome Research Institute

In 1989, the National Institutes of Health (NIH) established the National Human Genome Research Institute (NHGRI) (then called the National Center for Human Genome Research) with one bold goal: to sequence the 3 billion DNA letters that make up the human genetic instruction book. With its partners, NHGRI led the International Human Genome Project (HGP) to completion in April 2003. Thus, the history of the HGP, genomics and NHGRI are inextricably intertwined.

Completion of the HGP was a magnificent achievement, but it was just the first step toward fulfilling the goal of improving human health through genomic research. In its 24-year history, NHGRI has actively investigated the basis for inherited susceptibility to numerous diseases as well as seeking to understand the tiny fraction of the genome -- one-tenth of a percent across the 3-billion-base genome -- that distinguishes one human from another.

The enterprise, with overall funding of approximately half a billion dollars in fiscal year 2013, is guided by a series of overlapping five-year strategic plans. The most recent one, [\*Charting a course for genomic medicine from base pairs to bedside\*](#), articulates a new vision for the future of genomics research and charts the path towards an era of genomic medicine. Published in 2011, the plan envisions scientists being able to identify the genetic bases of most single-gene disorders and gaining new insights into multi-gene disorders in the next decade. The goals are more accurate diagnoses, new drug targets and the development of practical treatments for many who currently lack therapeutic options.

While the strategic plan describes areas of research focus for the entire genomics community, NHGRI is organized into three functional programs to implement the plan, including: the Office of the Director, which provides guidance to scientific programs and oversees the general operation of the institute; the Extramural Research Program, which issues grants to institutions and individuals; and the Division of Intramural Research, which is home to the institute's in-house, genetics and genomic research laboratories.

The four divisions of NHGRI's Extramural Research Program award and administer grants for basic genomic research and technology development, as well as major activities such as large-scale genome sequencing; efforts to move genomic technologies and approaches into clinical applications and care; and research related to societal issues relevant to genomics research, including incorporating and extending the activities of the institute's Ethical, Legal and Social Implications (ELSI) research program. Under these broad categories are 24 different research programs (<http://www.genome.gov/27534285>). The National Advisory Council for Human Genome Research helps determine program priorities for NHGRI and performs second-level peer review for grant applications when it meets three times a year.

The Division of Intramural Research conducts research to unravel the genetic basis of human disease. Fifty investigators in the division's seven branches undertake high-risk efforts using genomic sequence data from humans and other species to pinpoint potential disease-causing changes in genes (<http://www.genome.gov/10000010>). Genes studied by NHGRI's researchers have been implicated in cancer, diabetes, premature aging, hereditary deafness, and various neurological, developmental, metabolic and immunological disorders. The scientific director leads the division – which is located on the main NIH campus in Bethesda, Md., the Bayview campus in Baltimore, and the Twinbrook complex in Rockville, Md. -- with input from its Board of Scientific Counselors. The clinical director oversees clinical research.

To support its intramural and extramural research efforts, NHGRI has developed vast databases, intricate technologies and innovative methods to accelerate genome research and its application to human health. NHGRI also supports the training of investigators, as well as the dissemination of genome information to the public and to health professionals.

Challenges remain, but the fundamental goals have not changed since the beginning of the Human Genome Project and the creation of the institute. Genomics and related large-scale biological studies will, in ways not previously available, lead to a profound understanding of human biology and disease, to unimagined advances in medical science and to powerful new ways for improving human health.