*NATURE* | NEWS

# Funds dedicated to personalized genetics

**NIH aims to push genome-sequencing into mainstream medicine.**

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The funds announced today could help DNA-sequencing technologies to reach the clinic.

The US National Institutes of Health has earmarked nearly half a billion dollars for a plan that it hopes will usher in an era of diagnoses and treatments based on genome sequencing.

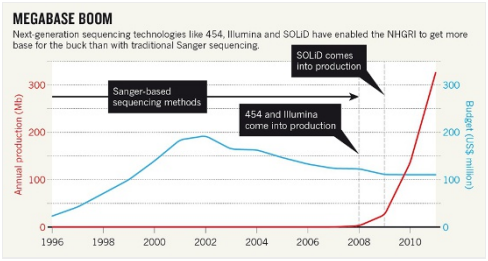
The National Human Genome Research Institute (NHGRI), part of the National Institutes of Health (NIH) in Bethesda, Maryland, today released a breakdown of its funding priorities. The four-year plan expands its flagship Large-Scale Genome Sequencing Program to focus on medical applications "to begin to explore the front edge of genomics, which will move us into genomic medicine", says Eric Green, director of the NHGRI.

That goal is now within striking distance, thanks to recent advances in sequencing technology, says Green. During the past decade, the per-base cost of sequencing has dropped by nearly 500,000-fold, (see Megabase boom, below) raising the possibility of individuals routinely undergoing sequencing for medical purposes. This is already a reality for a small minority (see: Genomes on prescription).

The lion's share of the $461 million announced by the NHGRI, starting at $86 million a year, will be divided amongst existing projects at three large genome-sequencing centres: the Broad Institute in Cambridge, Massachusetts, the Genome Institute at Washington University in St. Louis, Missouri, and the Human Genome Sequencing Center at Baylor College of Medicine in Houston, Texas. The amount of funding for the centres will decrease each year, according to the NHGRI.

These centres, which were all part of the Human Genome Project, will continue their work on projects such as the 1000 Genomes Project, which aims to catalogue human genetic variability, and the Cancer Genome Atlas, which is exploring the genomic changes associated with human cancer.

About $20 million over the four years will help develop genome sequence analysis software that can be used by researchers outside of large sequencing centers who may not have the expertise to process raw sequence data into clinically useful information. The software funds will be distributed in early 2012.




### Mendel and the clinic

About 20% of the funding will be used to establish two new programmes. The first, the Mendelian Disorders Genome Centers, will aim to identify the genetic basis of all Mendelian diseases — those that arise from a mutation in a single gene. Although many Mendelian diseases are rare, they provide a clear look at the function of a given gene in the body. A number of drugs in widespread use today, such as statins, have their origins in Mendelian-disease studies[1].

However, of the estimated 6,000 human Mendelian diseases, the cause of fewer than half is known.But with the reduction in sequencing costs and time, this stands to change. "All the barriers have been ostensibly eliminated by the new sequencing technologies," says Richard Lifton, a principal investigator of the Center for Mendelian Disorders being established by the NHGRI funds at Yale University in New Haven, Connecticut.

The NHGRI money, along with $8 million from the National Heart, Lung, and Blood Institute, will fund the Center for Mendelian Genomics at the University of Washington in Seattle and the Baylor–John Hopkins Center for Mendelian Genetics, a collaboration between the campuses in Houston and in Baltimore, Maryland. All three groups plan to join the International Rare Disease Research Consortium next year (see: Rare-disease project has global ambitions).

In contrast to the targeted Mendelian disease centres, the second new programme, the Clinical Sequencing Exploratory Research Project, is tasked with an open-ended question: what are the medical, ethical and social impacts of using genomic sequencing in a clinical setting?

Projects at five institutions — Baylor College of Medicine, the Brigham and Women's Hospital in Boston, Massachusetts, the Children's Hospital of Philadelphia in Pennsylvania, the University of North Carolina at Chapel Hill, and the University of Washington in Seattle — will each run trials of the effects of clinical sequencing for different diseases.

In Seattle, for example, researchers will test how sequencing the protein-encoding regions of the genome in patients with colon cancer affects people's diagnoses, treatment, mental well-being and finances.

Medical geneticist Gail Jarvik at the University of Washington, a principal investigator on the project, says that the time is right to test the utility of clinical sequencing. "The technology is there, but there is a lot of work to be done on what to do with the technology," she says. There is also the matter of patients' views on the subject. "How do people feel about getting genomic data?" she asks.

Green acknowledges that moving sequencing into the clinic is not going to be simple. "We expect there to be surprising findings, we expect some things not to work," he says. "One of the things our institute can do on behalf of all of NIH is to get data on varying overarching sets of issues around applying genomics to medicine."

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## References

1. Brinkman, R. R., Dubé, M.-P., Rouleau, G. A., Orr, A. C. & Samuels, M. E. *Nature Rev. Genet.* **7**, 249–260 (2006).