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Dawn of the genome era

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The Human Genome Project concluded in 2003, but many of its benefits are only now being realized, according to Alan Guttmacher, director of the National Institutes of Health's Eunice Kennedy Shriver National Institute of Child Health and Human Development.

A decade ago it cost $100 million to sequence a single genome, Guttmacher told a score of Northeastern students and researchers at a recent lecture hosted by the Bouvé College of Health Sciences and the Office of Government Relations. If scientists wanted to uncover any one disease-related gene, he said, the effort would have set them back a staggering $10 billion.

"But then some wonderful things happened," Guttmacher said, explaining that new tools and technologies have reduced the cost of genome sequencing to about $10,000—a price that continues to fall.

This, Guttmacher said, is a human health game-changer: We now know that all of the major causes of mortality among Americans have genetic underpinnings and scientists have even managed to identify the specific genes responsible for some of them.

But there is more work to be done. The market's 21,000 FDA approved drugs target just 2.5 percent of the total human genome, Guttmacher said, and the vast majority of our DNA remains an unexplored territory ripe for investigation.

Guttmacher, a former pediatrician, went on to note that the idea of

sequencing every individual's genome at birth is beginning to look like a reasonable enterprise. This possibility is dictating much of NICD's future endeavors. For example, the revamped National Children's Study, announced earlier this month, seeks to understand how genetics and the environment interact to cause health and disease. The effort will rely heavily on genome sequencing.

Another NICHD initiative, the Human Placenta Project, aims to strip away the mystery surrounding the most poorly understood human organ within the next 10 years. We know the placenta is important for fetal growth and development, Guttmacher said, but it's beginning to look like it's also important to the individual's lifelong health and wellbeing as well as that of the mother.

But the new investigations enabled by genome sequencing also demand new tools and technologies. "The way we conduct science needs to evolve if we're to be successful," said Guttmacher. For instance, we need a new model for data sharing that supports open collaboration and communication, and a new paradigm for transdisciplinary research, which rewards investigators for working together rather than penalizing them. As researchers develop new tools for their own work, they need to be made available to others so scientists can spend their time conducting research rather than preparing to conduct research.

The lecture dovetails with Northeastern's focus on use-inspired research that solves global challenges in health, security, and sustainability. In meetings following the talk, Guttmacher met with several of the university's faculty members, many of whom are already tackling the issues he sees as critical to the future of science.

Provided by Northeastern University