Science of genome-sequencing marks 10 years

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A decade after completion of the Human Genome Project on April 14, 2003, a top official of the National Institutes of Health surveyed the rarefied view from that mountaintop:

Admitting, "we have a long way to go to deliver on the promise of genomic medicine," Eric D. Green, director of the institute devoted to this research, still stressed that progress in some areas has been "amazing."

Scientists have obtained the full genetic script of a woman's unborn baby, and the federal government is already preparing for a future in which all babies will have their genomes sequenced at birth.

Doctors now know the genetic underpinnings of almost 5,000 rare diseases, more than twice as many as a decade ago.

We have learned that humans are more than 99.9 percent alike in their DNA, and yet so vast is the script - a sequence of almost 3.2 billion chemical bases - that our genome is "astonishing in the depth and breadth of its variation," Green said.

Researchers have also overturned a major foundation of genetics by showing that large portions of the script, up to 80 percent once dismissed as "junk DNA," actually turn out to have specific biological functions.

To a degree few had expected in the 1990s, the speed of genome

sequencing has increased and the cost has dropped. Green pointed out that the first human genome took six to eight years to complete at a cost of roughly $1 billion. On the day it was finished in 2003, scientists were already able to sequence the second human in about three to four months at a cost of $30 million to $50 million.

Today, sequencing a genome takes just one or two days and costs about $5,000; what was once a national scientific quest is now well on its way to becoming a common procedure like an MRI.

And then there are the patients such as Wisconsin youngster Nic Volker, whose genes were sequenced in 2009 and used to diagnose and treat a disease that had never been seen before. Across the country, there has been just a trickle of similar success stories.

However, information from the genetic script is already allowing doctors to treat different cancers with greater precision. By sequencing cancer patients, doctors can figure out what medications will and will not work for them.

Progress has been especially rapid in Wisconsin, where gene-sequencing came of age in 2009, when doctors and scientists sequenced Nic, a Monona boy suffering from an extremely rare intestinal disease.

Since the Volker case, the two institutions involved, Medical College of Wisconsin and Children's Hospital of Wisconsin, have expanded their collaboration, forming a first-of-its-kind sequencing clinic for kids with baffling, undiagnosed illnesses.

Howard Jacob, who has directed the sequencing efforts as head of the Medical College's human and molecular genetics center, has begun seeking support for an ambitious $60 million institute that would include clinical and research programs, data storage, analysis experts and

businesses that serve genomic medicine. The plan is still in the early stages.

Across the United States, other early adopters have brought sequencing closer to mainstream medicine.

Last year, Partners HealthCare System, the largest hospital chain in New England, introduced a DNA sequencing program.

Last fall, Kansas City-based Children's Mercy Hospitals and Clinics announced that it had begun sequencing the genomes of newborn babies with serious illnesses and planned to offer the same services to other hospitals around the country.

At about the same time, the Translational Genomics Research Institute, or TGen, in Phoenix announced that it had started a center for rare childhood disorders that would use genome sequencing to discover the causes of the disorders.

In October, the federal government's Undiagnosed Diseases Program announced a $145 million expansion plan to include the development of half a dozen or so regional centers around the country.

The rapid emergence of genomic medicine has also led to a host of start-up companies and to partnerships among businesses, hospitals and universities.

Scientists and ethicists have long warned that use of our full genetic scripts will raise a host of ethical issues, a likelihood the federal government has anticipated. In addition to all of the money going to research, millions of dollars are being spent to investigate the ethical, legal and social implications of this new world in which our genes are becoming a crucial medical tool.