Medical Xpress

Institute presses for greater use of gene sequencing in medicine

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Almost a year after researchers in Wisconsin published a groundbreaking paper describing their use of genetic sequencing to diagnose and treat a 4-year-old boy, a national health agency is shifting its focus to put $416 million into making the secrets of our genetic script part of standard medical practice.

The National Human Genome Research Institute announced the move Tuesday, saying that much of the effort is going toward overcoming the barriers hindering the widespread use of sequencing in hospitals. These include the ethical and analytical challenges of reading the vast flood of data from the human genome.

Doctors and scientists foresee daunting challenges in the thousands of genetic variations they expect to find in every one of us. What software will help them determine which of these variations is causing a specific problem in our blood, kidney or colon? What will doctors do when they are trying to understand one problem in our bodies, but find another, unrelated condition? What information will they tell a child's parents and what will they withhold?

Such questions are coming to the fore a little more than a decade after scientists announced completion of the first draft of the genome, the full picture of our genetic makeup - everything from the color of our hair and eyes to the slight variations in code that cause rare diseases or put us at higher risk of heart disease. All of this information is written into a four-letter code, each letter representing a chemical base, stretching for

3.2 billion base pairs.

It was only last December that doctors and scientists at the Medical College of Wisconsin and Children's Hospital of Wisconsin published their study in the journal Genetics in Medicine, describing one of the first cases in medical history in which all of a person's genes were sequenced and the information used to craft a treatment. The boy, Nicholas Volker, of Monona, Wis., had a mysterious intestinal disease never seen before and was given a transplant of umbilical cord blood. He has experienced periodic health problems since then but has also been well enough to play T-ball and return to school.

In the meantime, the Medical College of Wisconsin has launched a sequencing program for children with unknown illnesses that meet specific criteria, and institutions such as Duke University Medical Center and Partners HealthCare System, the largest hospital chain in New England, have introduced their own programs.

"We still have much to learn about how the human genome works, how it works in health and in illness," said Mark S. Guyer, deputy director of the genome research institute, part of the National Institutes of Health.

The largest share of the money, $319 million, will go to three large sequencing centers: the Broad Institute in Cambridge, Mass.; the Genome Institute at Washington University in St. Louis; and the Human Genome Sequencing Center at the Baylor College of Medicine in Houston. These institutions will use the money to continue basic research into how the genome works, studies examining the role of genes in complex illnesses such as diabetes and heart disease, and other projects such as a genomic study of cancer.

The remainder of the money will go toward programs on: rare, inherited diseases called Mendelian disorders; ethical and logistical issues

expected to arise in bringing genome sequencing to hospitals; and software tools to help doctors sift through the thousands of genetic variations more efficiently.