

# Scientists use genetic sequencing to identify and treat unknown disease

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A collaborative team of scientists and physicians at the Medical College of Wisconsin and Children's Hospital of Wisconsin uses genetic sequencing to identify and treat an unknown disease.

For the one of the first times in medical history, researchers and physicians at The Medical College of Wisconsin and Children's Hospital of Wisconsin sequenced all the genes in a boy's DNA to identify a previously-unknown mutation. The team was able not only to identify the mutation, but to develop a treatment plan using a cord blood transplant, and stop the course of the disease.

This accomplishment is published in the December 19, 2010 online edition of *Genetics in Medicine*.

At the age of 3, Nicholas Volker of Monona, Wisconsin, had already endured more than 100 surgeries, but was progressively getting sicker. His [intestines](#) continued to swell and more abscesses formed, and doctors concluded they were dealing with an unknown cause of disease.

Dr. Alan D. Mayer, assistant professor of Pediatrics at the Medical College, and pediatric gastroenterologist with Children's Hospital, decided to look for the genetic source of the disease.

"Exhaustive efforts to reach a diagnosis revealed numerous abnormalities in Nick's immune system, but none pointed to a specific disease," Dr. Mayer explained. "So we decided the next logical step was

to sequence his entire exome."

Researchers examined all 20,000 of his genes looking for the cause of this rare disease. After three months poring over data, the researchers identified a unique mutation in one gene. They confirmed that mutation to be responsible for Nick's previously-undocumented form of bowel disease, which is part of a broader XIAP deficiency.

The team then performed a blood cord transplant in June using [stem cells](#) from a matched, healthy donor. Five months later, Nick is home and eating a healthy diet for the first time.

"There has been no return of the [bowel disease](#)," said Dr. David A. Margolis, the program director for the Blood and Marrow Transplant Program at Children's Hospital, and associate professor of pediatrics at the Medical College. "At this point, he is a transplant recipient and his current treatment focuses on maintaining his health with the challenges posed by the transplant."

The Medical College of Wisconsin and Children's Hospital of Wisconsin are developing a new strategy as well as formal policies and procedures to guide the approach to future cases in which [genetic sequencing](#) will be used as a diagnostic tool.

"We are confident that genomic sequencing will have a growing role in establishing the correct diagnosis for patients and, most importantly, improving outcomes," said Dr. Jacob, the Warren P. Knowles Professor of Molecular Genetics, director of the Medical College's Human and Molecular Genetics Center and professor of physiology.

**More information:** Making a Definitive Diagnosis: Successful clinical application of whole exome sequencing in a child with intractable inflammatory bowel disease. Lead authors are Elizabeth Worthey, Ph.

D., and Alan N. Mayer, M.D. Ph.D. Collaborators at Children's Hospital and the Medical College are David A. Margolis, M.D., James Verbsky, M.D., Ph.D., Howard J. Jacob, Ph. D., and David Dimmock, M.D.

<http://journals.lww.com/geneticsinmedicine/pages/default.aspx>

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