



## Research saves a young boy's life

A little boy named Nic came to Children's Hospital of Wisconsin with an undiagnosed illness that required 100 trips to the operating room, including the removal of his colon – all before he turned 5 years old. He couldn't eat foods without getting sick. His little body couldn't fight off even the most common cold. Time was running out.

A team of physicians including a hematologist, oncologist, geneticist and gastroenterologist reached out to their [research colleagues](#) at [The Medical College of Wisconsin](#) to ask the question: Could sequencing the boy's genome pinpoint the problem and finally lead to a definitive diagnosis and ultimately treatment?

The human genome is the complete set of genes in an individual. The genome is made up of DNA. DNA, made up of smaller units called nucleotides, is the molecule that is the hereditary material in all living cells. It's estimated there are 30,000 genes and more than 3 billion base pairs or combinations of DNA.

Beyond a potential of reaching a diagnosis for Nic, a new possibility was emerging that [individualized genomic sequencing](#) could help other children with rare, undiagnosed illnesses. Up until this point, genomic sequencing was all but limited to the lab. Sequencing Nic's DNA signaled the breakthrough in the progress of translational research from lab to bedside.

Scientists and researchers were able to narrow down a list of 32 genes with variations. From there, they mined further and discovered one



*Milwaukee Journal Sentinel* reporters Kathleen Gallagher and Mark Johnson and photojournalist Gary Porter chronicled Nic and his family's medical odyssey in a three-part series called [One in a Billion: A boy's life, a medical mystery](#). "Somewhere in a pool of 16,000 variations in Nicholas' genetic script," wrote Gallagher and Johnson, "lurks the cause of his disease."

misplaced base in the long chain of Nic's DNA that was changing production of an amino acid. This one misplaced base was causing a life-threatening genetic mutation. The diagnosis: an X-linked lymphoproliferative disorder or XLP. It is an inherited immune system disorder that affects fewer than 1 in a million children. The disorder is found on the X chromosome, only strikes boys and is usually fatal, rendering them unable to survive one of the most common human viruses, Epstein-Barr.

Doctors believed they could treat Nic's identified disease with a bone marrow transplant. He received a transplant in July 2010. Nic finally left the hospital in November – after more than 700 days at Children's Hospital. Nic's latest discoveries? Playing outside and having pizza with his family!

So much has changed for one boy. So much may now change for others.

- Nic's story, including the ground-breaking research and treatment provided by Children's Hospital of Wisconsin and The Medical College of Wisconsin received international media attention. It was published in *Genetics in Medicine*, profiled in a three-part series in the *Milwaukee Journal Sentinel* and on NBC's *The Today Show* in December 2010.
- Children's Research Institute conducted more than 1,000 clinical trials in 2010.
- Last year, Children's Hospital invested more than \$12 million in research to help improve the health of children.
- A study, featured in the *Journal of the American Medical Association* in 2010, discovered high hospitalization rates for children with sickle cell disease. The study was conducted by researchers at Children's Research Institute, The Medical College of Wisconsin and the U.S. Department of Health and Human Services' Agency for Healthcare Research and Quality.
- Doctors at Children's Hospital of Wisconsin are finding ways to treat humans infected with rabies, a disease once considered incurable. Since an experimental treatment known as the Milwaukee Protocol saved the life of 15-year-old Jeanna Giese, from Fond du Lac, Wis., doctors have gone on to refine the protocol and yield even greater results.

