A toddler with a mysterious abdomen inflammation endures 100 surgeries. His symptoms continue. Physicians remove his entire colon. Still, no answers. Determined to gather more information to help the boy, they use next-generation sequencing to take a genomic assessment of his DNA. There, they find the problem—and the solution. Hidden in a DNA mutation.

This Pulitzer-prize winning story was published in the Milwaukee Journal Sentinel about Nic Volker, a recent case of a rare disease diagnosed through genomic sequencing. Over the last half-century, more than 2,000 clinical tests based on genetic information have been developed. Most of these focus on one to a few specific genes, to see if a particular variant is present or absent in a given position. This strategy is effective in a majority of cases where there is a clear link between a patient’s symptoms and a suspected diagnosis. But sometimes, as in Nic’s case, other genes and genetic variants are to blame. Enter whole-genome sequencing—a map of the entire human genome—powered by next-generation sequencing technology. It emerged two years ago as a service for physicians.

Whole-genome sequencing is quickly gaining recognition for its potential in the world of rare diseases, where physicians are challenged with identifying a disorder based on symptoms that don’t quite fit with a known disease. When this happens, whole-genome sequencing can provide big-picture information about genetic makeup, enabling physicians to make more informed decisions. In some cases, this has really made a difference for patients—so significantly, that insurance companies have reimbursed for the procedure.

Access to this information can end diagnostic odysseys and offer physicians, patients, and their loved ones answers. Still, it’s a challenge to evaluate and understand genetic variation. Which genes should be examined, and how can we be sure? How can information be managed to protect privacy? Plus, finding an answer does not mean finding a cure. Nonetheless, patients report that even the worst news can result in better decision-making—such as avoiding additional invasive procedures that would not be successful. As many families suffering with rare disease know, sometimes an answer is relief in itself.

Despite the current sequencing challenges, there are patients benefiting from whole genome sequencing right now. While not every test yields a conclusive answer, it’s a helpful option for physicians, clinical laboratories, and patients like Nic Volker, who are working together to solve the mysteries behind human diseases.

Learn More
To learn more about genome sequencing, visit www.everygenome.com