

Novel algorithm better assembles DNA sequences and detects genetic variation

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Bodily and a team of researchers have created a new algorithm that can better identify variations in DNA.

Researchers at BYU have developed a new method for human genome assembly and inadvertently discovered a new way to identify elusive markers for several common genetic disorders.

The novel approach relies on a new [algorithm](#), developed by the researchers, that is more sensitive to detecting specific types of variation in DNA sequences.

"It required a lot of persistence and the outcome wasn't what I had originally planned to investigate," said lead author Paul Bodily, PhD student in computer science. "I just continued down a path, not knowing where it was going to lead, until I found something shiny."

Every person has two copies of the genome in each of their cells, one from each parent. The DNA sequence of the two copies is mostly identical, but there are occasionally places where one of the sequences gets reversed, called an inversion.

Inversions are often biologically significant. Previous research has demonstrated an association between inversions and mental retardation, diabetes, epilepsy, schizophrenia and autism.

The research, published in medical journal *Bioinformatics*, details how the new algorithm outperforms other methods and the unintended discovery of the algorithm's ability to detect inversions.

Previously, inversions have been very difficult to detect, but the researchers believe their method can significantly enhance the process.

"There are diseases, like autism, that we're really uncertain as to what causes them," said senior author Mark Clement, professor in computer science. "To at least have improved tools for detecting a possible cause is important."

The group made the algorithm source freely available to any fellow researchers interested in applying it.

Bodily is still a few years from graduation but anticipates the development of his research to apply the new module on a bigger scale.

"You can make almost any algorithm look good if you craft the right data set, but ours is based on some pretty fundamental theory," said Bodily. "Theory and practice don't often agree, but I have a lot of hope that when applied to the bigger picture, our approach will work well."

Provided by Brigham Young University

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