New software tool could help doctors diagnose genetic diseases

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A user-friendly open-source web tool for analyzing WES and WGS in the diagnosis of patients with Mendelian disorders. Credit: Cardenas et al.

An open-source software tool called Mendel,MD could help doctors

analyze patients' genetic data in order to diagnose diseases caused by mutations. Developed by Raony Cardenas and colleagues at Universidade Federal de Minas Gerais, Brazil, the tool is presented in a new study in *PLOS Computational Biology*.

Diseases caused by genetic mutations can be challenging to diagnose. Sequencing of a patient's entire genome or exome—the part of the genome used to build proteins—now offers an effective strategy to pinpoint culprit mutations and make an accurate diagnosis. However, the software needed to analyze these sequences is often costly or too complex for many doctors to use.

To address these issues, Cardenas' team developed Mendel,MD specifically for easy use by physicians, free of charge. Users upload a patient's whole genome or exome sequence via a web-based interface, and the sequence is analyzed and filtered using various computational tools and databases of disease-causing mutations. The result is a list of candidate mutations that can be clinically investigated to arrive at a final diagnosis.

The researchers validated Mendel,MD using previously-published clinical cases. They also had it tested by researchers and students at their own university, as well as at GENE - Núcleo de Genética Médica, Brazil, and the Children's University Hospital in Dublin, Ireland. The results suggest that Mendel,MD is reliable, simple, and efficient in identifying disease-causing mutations in patients.

"We designed the software to be simple and intuitive enough to be used directly by physicians, even those who are not proficient in bioinformatics," says study co-author Sérgio Pena. "We expect Mendel,MD to be adopted in other research centers and laboratories around the world."

More information: G. C. C. L. Cardenas R, D. Linhares N, L. Ferreira R, Pena SDJ (2017) Mendel,MD: A user-friendly open-source web tool for analyzing WES and WGS in the diagnosis of patients with Mendelian disorders. *PLoS Comput Biol* 13(6): e1005520. doi.org/10.1371/journal.pcbi.1005520