System can recognize Mendelian genetic diseases before clinical teams

December 16 2019, by Paul Govern

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Mendelian diseases, those caused by a single deleterious genetic variant, often present as constellations of clinical features.

In a previous study, Lisa Bastarache, MS, Joshua Denny, MD, MS, and colleagues demonstrated a computational method, phenotype risk scoring, or PheRS, that uses hospital billing codes to identify likely Mendelian disease cases in an electronic health record (EHR)

population. In that study, comparison of risk scores with genotypes revealed previously unknown deleterious variants and suggested a potential expanded role for genetic testing in the clinic.

Back with a new study in the *Journal of the American Medical Informatics Association*, the investigators confirm cases of 16 different Mendelian diseases within a de-identified EHR group of 2.5 million patients and proceed to compare four variations on PheRS.

Not uncommonly, PheRS can recognize Mendelian disease cases before clinical teams do, the study shows. All methods served well to distinguish cases from controls; performance tended to improve when billing codes were augmented with clinical lab results.

More information: Lisa Bastarache et al. Improving the phenotype risk score as a scalable approach to identifying patients with Mendelian disease, *Journal of the American Medical Informatics Association* (2019). DOI: 10.1093/jamia/ocz179

Provided by Vanderbilt University