AI Algorithm Predicts Risk of FH with High Accuracy

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A new machine learning algorithm was highly accurate in determining whether a patient is likely to have a cholesterol-raising genetic disease that can cause early heart problems, according to the results of a study conducted by researchers at the Stanford University School of Medicine.

The algorithm was 88 percent accurate in identifying familial hypercholesterolemia (FH) in one data sample and 85 percent accurate in another.

In the study published in *npj Digital Medicine*, Joshua Knowles, M.D., Ph.D., assistant professor of cardiovascular medicine at Stanford, and his research team created an algorithm using data from Stanford's FH clinic to learn what distinguishes an FH patient in an electronic health record (EHR).

The algorithm was trained to pick up on a combination of family history, current prescriptions, lipid levels, lab tests and more to understand what signals the disease.

The foundation of the algorithm was built using data from 197 patients who had FH and 6,590 patients who did not, so the program could learn the difference between positive and negative results.

When the algorithm was trained, the research team initially ran it on a set of roughly 70,000 new de-identified patient records. The team reviewed 100 patient charts from the patients flagged and found that the algorithm had detected patients who had FH with 88 percent accuracy.

Knowles and his partner, Nigam Shah, MBBS, Ph.D., associate professor of medicine and biomedical data science at Stanford, collaborated with Geisinger Healthcare System to further test the algorithm.

The algorithm was tested on 466 patients with FH and 5,000 patients without FH, and the predictions came back with 85 percent accuracy.

Shah said that him and Knowles knew that a lot of the Geisinger patients had a confirmed FH diagnosis with genetic sequencing.

"So that's how we convinced ourselves that yes, this indeed works," he said.

FH is an underdiagnosed genetic condition that leads to an increased risk of coronary artery disease if untreated. A patient with FH faces 10 times the risk of heart disease than someone with normal cholesterol. The condition can lead to death or a heart attack, and there are clear benefits of timely management, yet it is estimated that less than 10 percent of those with FH in the U.S. have been diagnosed.

Early diagnosis and treatment of FH can neutralize the threat of the condition. And one diagnosis could help multiple people because FH is genetic, making it likely that other relatives have it too.

Lead author Juan Banda, Ph.D., former research scientist at Stanford, wrote that when the algorithm is applied broadly to screen FH, it is possible to identify thousands of undiagnosed patients with the condition. This could lead to more effective therapy and screening of their families, Banda wrote.