AI accurately diagnoses genetic condition from facial photographs

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A Yale School of Medicine team reports in a new study that an artificial intelligence (AI) model was able to reliably diagnose people living with

Marfan syndrome from a simple facial photograph.

Marfan syndrome is a genetic disorder, affecting about 1 in 3,000 people, which impacts the body's connective tissues. "Patients living with Marfan syndrome are usually very tall and thin," said John Elefteriades, MD, professor of surgery at Yale School of Medicine and senior author of the study. "They have long faces and are prone to spine and joint issues. However, many are not diagnosed."

Marfan syndrome increases the risk for aortic dissection, where the aorta splits suddenly after becoming enlarged. "It is often lethal, and when the patient survives, urgent surgery is needed," Elefteriades said. "Being able to identify individuals from a photograph with AI will enhance diagnosis and enable protective therapies."

In a pilot study recently published in *Heliyon*, researchers assembled 672 facial photographs of people with and without Marfan syndrome.

A Convolutional Neural Network was trained on 80% of the photographs, then asked to identify the other 20% as Marfan or non-Marfan faces. The model successfully distinguished between Marfan and non-Marfan faces with 98.5% accuracy.

Researchers say they plan to make the tool available online in the future. "We are planning to extend this work beyond this initial pilot project," said Elefteriades. "We anticipate that many individuals may self-test once we put the test online."

"Yale School of Medicine faculty and students are leading the way in developing novel applications of AI to recognize and diagnose diseases, including rare diseases, earlier when we can have the greatest impact," said Nancy J. Brown, MD, dean of Yale School of Medicine.

More information: Danny Saksenberg et al, Pilot study exploring artificial intelligence for facial-image-based diagnosis of Marfan syndrome, *Heliyon* (2024). DOI: 10.1016/j.heliyon.2024.e33858