Smartphone app diagnoses genetic diseases with a snapshot

[When Omar Abdul-Rahman, a clinical geneticist at the University of Mississippi Medical Center, had to provide a diagnosis for a young boy with distinct facial features and intellectual disabilities, he] turned to a new tool in a geneticist's arsenal, an app based on facial recognition software that helps identify genetic conditions based on facial features. The app suggested, with a high degree of certainty, a condition he had considered [and a test] confirmed that the boy had Mowat-Wilson syndrome….
Face2Gene, the tool Abdul-Rahman used, was created by the Boston startup, FDNA. The company uses facial recognition software to aid clinical diagnoses of thousands of genetic conditions, such as Sotos syndrome, Kabuki syndrome, and Down syndrome.

…

[T]here are more than 7,000 other genetic conditions that aren't always so easy [to diagnose]. The founders of FDNA…thought it might be possible to use similar techniques to match facial characteristics to genetic conditions.
…

Face2Gene's user pool has grown quickly since the app was released in 2014—more than 65 percent of clinical geneticists worldwide use it—and as it grows, the tool becomes more robust.

face gene map infographic v
When a geneticist confirms the diagnosis of an uploaded
photo, the app incorporates that in its database, creating a
sort of crowdsourced loop. Credit: Smithsonian.

The GLP aggregated and excerpted this blog/article to reflect the diversity of news, opinion, and analysis. Read full, original post: This App Uses Facial Recognition Software to Help Identify Genetic Conditions