Genetic diagnosis of rare diseases up with genome sequencing

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(HealthDay)—Use of genomic sequencing yields an increase in genomic diagnoses across a spectrum of rare diseases, according to a study published in the Nov. 11 issue of the *New England Journal of Medicine*.

Damian Smedley, Ph.D., from Genomics England in London, and

colleagues conducted a pilot study involving 4,660 participants from 2,183 families who had 161 disorders covering a broad spectrum of rare diseases.

The researchers observed variation in diagnostic yields among family structures, with the highest yield in family trios (both parents and a proband) and families with larger pedigrees. Disorders likely to have a monogenic cause had much higher diagnostic yields than those likely to have a complex cause (35 versus 11 percent). For intellectual disability, hearing disorders, and vision disorders, diagnostic yields ranged from 40 to 55 percent. Genetic diagnoses were made in 25 percent of the probands. Three new disease genes and 19 new associations were discovered in cohort-wide burden testing across 57,000 genomes. Twenty-five percent of the genetic diagnoses made had immediate ramifications for clinical decision making for patients or their relatives.

"We hope that our findings will assist other health systems in considering the role of genome sequencing in the care of patients with rare diseases," the authors write.

Several authors disclosed financial ties to the biopharmaceutical industry.

More information: Abstract/Full Text

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