Case highlights polygenic risk in severe hypertriglyceridemia

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(HealthDay)—Polygenic risk can mimic major monogenic mutation in

severe hypertriglyceridemia, according to a case report published online July 25 in the *Annals of Internal Medicine*.

Priska Stahel, Ph.D., from the University of Toronto, and colleagues developed a score based on the 14 loci (28 alleles) that most affect triglyceride levels to examine whether a high polygenic risk for hypertriglyceridemia can mimic a major monogenic mutation. A 22-year-old male presented with severe hypertriglyceridemia despite overall excellent health, favorable lifestyle, and no other identifiable cause of the condition. In light of an extensive family history of cardiovascular disease, he underwent fasting plasma lipid testing, and the results revealed marked hypertriglyceridemia and markedly elevated total cholesterol levels. The patient had minimal alcohol intake and there was no evidence that diet was contributing to his lipid profile. He underwent gene sequencing of 69 genes most likely to explain his lipid profile.

The researchers identified no major mutations that are known to cause severe hypertriglyceridemia. The patient had *E3/E2 APOE* genotype, and no copy number variants were observed. Many common polymorphisms were identified that produced a triglyceride polygenic risk score of 19 (>99th percentile) and a low-density lipoprotein cholesterol polygenic risk score of 15 (92nd percentile).

"This case highlights the need for physicians to consider a polygenic cause for patients with severe hypertriglyceridemia," the authors write.

More information: Abstract/Full Text (subscription or payment may be required)