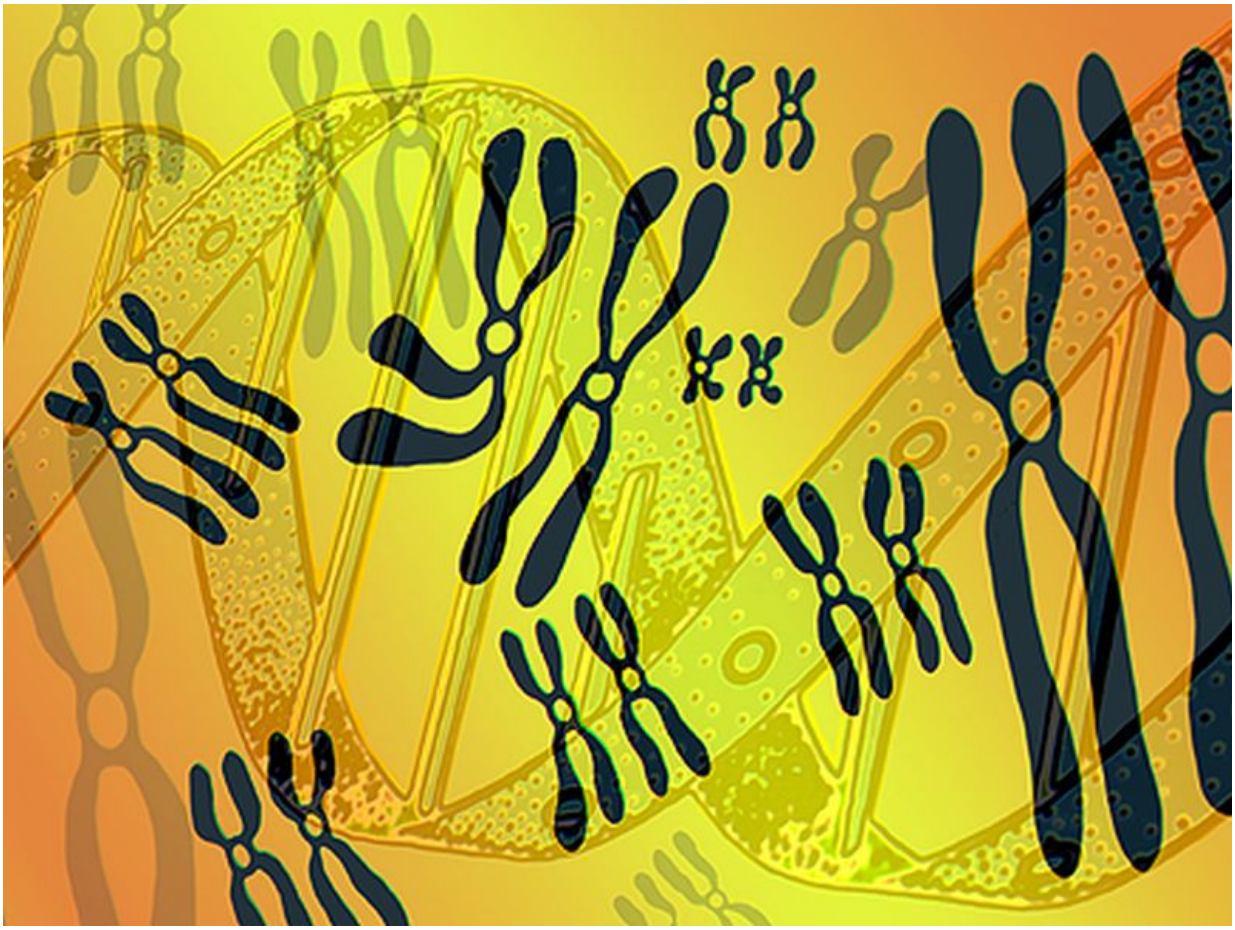
MedicalXpress

Functionally ID'd variants in HNF1A linked to diabetes risk

December 5 2016



(HealthDay)—Functionally characterized variants in hepatocyte nuclear

factor-1a (*HNF1A*) genes, which are associated with maturity-onset diabetes of the young (MODY3), are strongly associated with diabetes, according to a study published online Nov. 29 in *Diabetes*.

Laeya Abdoli Najmi, from the University of Bergen in Norway, and colleagues examined whether functional classification of *HNF1A* rare coding variants can inform models of diabetes risk prediction in the general population. They assessed the effect of 27 *HNF1A* variants identified in well-phenotyped populations (4,115 individuals).

The researchers found that 11 of the variants were classified by bioinformatics tools as likely pathogenic, and they showed no correlation with diabetes (odds ratio, 2.02; 95 percent confidence interval, 0.73 to 5.60). In the general population, there was a strong correlation with diabetes for a different set of 11 variants that reduced transcriptional activity of HNF-1A to less than 60 percent of normal (odds ratio, 5.04; 95 percent confidence interval, 1.99 to 12.80). In functional investigations, 0.44 percent of the population were found to carry *HNF1A* variants that result in increased risk for developing diabetes.

"These results suggest that functional characterization of variants within MODY genes may overcome the limitations of bioinformatics tools for the purposes of pre-symptomatic diabetes risk prediction in the general population," the authors write.

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