Common disease prevention and cancer screening would benefit from genomic risk assessment

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Many of the most common causes of death are due to diseases whose onset could be significantly slowed down or whose prognosis could be improved by identifying with increasing accuracy individuals at high risk. In the current system, a considerable number of high-risk individuals cannot be identified in time, as currently available tools measure genetic risk inadequately or not at all.

The findings, published today in *Nature Medicine*, demonstrate that genomic information could be used to improve the selective prevention of cardiac diseases and diabetes, as well as cancer screening. The results are based on the FinnGen research project, which encompasses more than 135,000 Finnish voluntary donors of biobank samples.

The study focused on five common diseases: coronary heart disease, type 2 diabetes, atrial fibrillation, breast cancer and prostate cancer.

Previous studies have identified numerous genetic risk factors for each of these diseases. In this study, the data pertaining to all of these individual risk factors was combined into what are known as genome-wide polygenic risk scores. These scores were calculated for all the 135,000 study subjects, for each of the five diseases.

"In terms of cardiovascular diseases and diabetes, genomic information alone can identify individuals who have a lifetime risk of more than 60% of developing these diseases, which means that most of them will develop these diseases at some point of their lives," says the principal investigator of the study, Professor Samuli Ripatti from the University of Helsinki.

The research group also combined genetic risk data with currently known risk factors and clinical risk calculators. Adding genomic information improved the accuracy of current risk estimation approaches.

"Our findings show that the genetic risk profile was a significant factor in predicting the onset of all five diseases studied. A particular benefit was seen in the identification of individuals who develop diseases at a younger age than on average," says Nina Mars, doctor of medical science at the Institute for Molecular Medicine Finland (FIMM) of the University of Helsinki, who carried out the study.

"Personalised risk calculation engenders opportunities that are important to healthcare. Risk assessment that utilises genomic information could be employed in, for example, determining the age when breast and prostate cancer screening begins. One option is to have those with a elevated genetic risk already undergo screening earlier than instructed in the current screening

recommendations", Mars states.

"A study that combines genomic and health data in such an extensive dataset is exceptional even on the global scale. From the perspective of our healthcare system, it's great to have been able to study Finnish individuals, making the results also directly applicable to Finns," says Aarno Palotie, scientific director of the FinnGen research project.

More information: et al, Polygenic and clinical risk scores and their impact on age at onset and prediction of cardiometabolic diseases and common cancers, *Nature Medicine* (2020). DOI: 10.1038/s41591-020-0800-0

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