Polygenic risk scores could improve colorectal cancer screening

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Rates of colorectal cancer are high despite widespread adoption of screening programs in many high-income European countries. Such programs tend to use a one-size-fits-all approach where most people are

screened starting from the same age, and no individual factors are considered in organized population screening. Now, based on one of the largest genomics studies on the topic to date, researchers from Finland have outlined how common genetic factors could be used to identify individuals at high risk of developing the disease and hence improve current colorectal screening strategies.

Max Tamlander, MD at the Institute for Molecular Medicine Finland (FIMM) of the University of Helsinki, will present to the annual conference of the European Society of Human Genetics today (Monday 12 June) his team's work on the development of a polygenic risk score (PRS) specifically for colorectal cancer. A PRS summarizes the combined impact of an individual's genetic risk factors for a disease into a single score. This allows an estimation of an individual's likely disease risk and the identification of those who might benefit from earlier screening.

The researchers used data from the FinnGen study, a collection of health and genome data from more than 400,000 Finnish individuals, for which they calculated a genome-wide PRS for colorectal cancer. "A challenge of many prior PRS studies is that they have been performed in smaller datasets that are not representative of the general population, but in this study we used epidemiological and statistical approaches to calibrate our estimates with that population," Dr. Tamlander explains.

Most cases of colorectal cancer occur in individuals who do not have a family history of the disease or any other known strong risk factors. PRSs offer a new way of assessing risk in these individuals, which until now has been based on attained age alone. The researchers' results also show that a PRS could be useful in the assessment of future colorectal cancer risk after a colonoscopy, the current gold-standard screening method, and identify those individuals who would potentially benefit from more frequent surveillance.

In Finland, screening for colorectal cancer in the general population currently starts at age 60; in some other European countries it starts earlier, from 50. The researchers found that, based on the current screening age 60 in Finland, individuals with a high PRS as compared to those with a low PRS could start screening at up to 16 years apart. For example, women and men at the top 1% of the PRS already had equivalent risks at ages 48.7 and 49.8, respectively. "This indicates that a colorectal cancer-specific PRS would be able to define more appropriate ages to start screening for individuals based on their genetic risk," says Dr. Tamlander.

As the cost of genotyping continues to fall, PRS-based approaches may become a feasible way to guide population-wide screening. "Millions of individuals already have their genomes available in large-scale biobanking initiatives," says Dr. Tamlander. "For example, the FinnGen biobank study already contains the genomic data of over 7% of all Finns, and this will soon increase to around 10%. One very useful aspect of PRSs is that genetic data extracted from a single sample can be used over the course of life to calculate risk scores for many common diseases, including the most common cancers."

Further clinical studies as well as data on cost-effectiveness and the effective communication of risk information will be needed before large-scale implementation of the colorectal PRS, say the researchers. Another problem is that, to date, PRSs have been mainly developed in individuals of European descent, and therefore may not be valid for people of other ancestries.

"However, our findings are well in line with other studies on PRSs in breast cancer, another common cancer with organized population-level screening. For breast cancer, large clinical trials are currently underway to evaluate the performance of personalized breast cancer screening, and their results will help us to understand the implications of genome-

guided, risk-based screening for colorectal cancer, as well as other diseases," Dr. Tamlander continues.

"In the future, risk-based approaches considering genetic factors alongside other relevant risk factors have potential for personalizing recommendations regarding how we could most effectively screen for colorectal cancer," Dr. Tamlander will conclude.

Professor Alexandre Reymond, chair of the conference, said, "Recognizing the individuals who are at risk is fundamental if our health systems want to truly embrace personalized health in the future."

More information: Abstract no. C20.2 Genome-wide polygenic risk scores substantially impact colorectal neoplasm risk with implications for stratified screening, The European Human Genetics Conference, 2023.eshg.org/

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