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Polygenic risk scores identify high-risk individuals in European and Asian ancestry, but less so in African ancestry

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An international research consortium led by the University of Helsinki has performed the largest study to date evaluating transferability of genome-wide polygenic risk scores across ancestries. Polygenic risk

scores (PRS) measure genetic disease susceptibility by combining risk effects across the genome.

The team focused on four common diseases with great public health impact: coronary artery disease, type 2 diabetes, breast and prostate cancer. Polygenic risk scores are well established for all these diseases.

The study combined data across six biobanks or biobank studies with altogether 1 million individuals from Europe, U.S. and Japan. The global ancestry groups studied were European, South Asian, East Asian, and African ancestry. From Finland, more than 250,000 FinnGen study participants were involved.

The results, published in *Cell Genomics*, showed that polygenic risk scores transfer well across European and moderately to South and East Asian populations. The transferability was much poorer to individuals of African ancestry.

"Widely adopting current polygenic risk scores to clinical care could increase existing health disparities. On the other hand, if future studies prioritize diversity, polygenic risk scores have great potential to improve health outcomes across ancestries," said the lead author of the study, Dr. Nina Mars from the Institute for Molecular Medicine Finland (FIMM), University of Helsinki.

The study also evaluated the PRS transferability locally within Finland, where there are well-documented genetic differences between the early-settlement region in the South and West and the late-settlement region in the East and North. Despite this known population substructure, the risk estimates were found to be uniform within Finland.

This large-scale study further emphasizes the pressing need for diversity in genetic studies.

"We and many others have previously shown that polygenic risk scores are powerful tools for identifying high-risk individuals for diseases of great public health impact among individuals with European ancestry. What our results now show is that this is not only true for individuals in Finland, UK, Norway or Massachusetts, but also for individuals with South or East Asian ancestry. However, we need to work hard to advance risk prediction for example for African ancestry individuals to ensure equitable use of genetic prediction for everybody," said Professor Samuli Ripatti from the University of Helsinki who led the study.

The study was conducted as part of a EU H2020 project INTERVENE that aims to develop AI-based integrative risk scores for the next generation of predictive and personalized medicine and test their utility in clinical medicine. The project has currently its first in-person meeting in Berlin this week, when the data analysts meet for a workshop.

More information: Nina Mars et al, Genome-wide risk prediction of common diseases across ancestries in one million people, *Cell Genomics* (2022). DOI: 10.1016/j.xgen.2022.100118

Iftikhar J. Kullo et al, Polygenic scores in biomedical research, *Nature Reviews Genetics* (2022). DOI: 10.1038/s41576-022-00470-z

Provided by University of Helsinki

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