CASE STUDY

Polygenic risk scores enable cost-effective prevention of coronary heart disease

Key findings

- Coupling polygenic risk scores (PRS) with traditional risk factors is a promising, cost-effective method for targeted statin treatment in the prevention of coronary heart disease.

- The results of the study suggest that generating polygenic risk scores for patients whose risk level is average according to traditional risk factors results in the highest expected net benefit.

- Compared with the use of traditional risk factors only, the optimal use of polygenic risk score data in a population of 100,000 would decrease the total costs of a preventative intervention program for coronary heart disease by approximately $300,000 in a 10-year follow-up period, while increasing health outcomes by 1.7 quality-adjusted life-years (QALYs).

Background

Coronary heart disease (CHD) remains the leading cause of death globally, accounting for more than 7 million deaths per year. In order to help ensure that preventative interventions such as statin treatment will be effective, it is important to obtain reliable prognostic information on patients' state of health. Current population screening methods that rely only on traditional clinical indicators and family history for CHD risk are missing the average-risk patients who experience more than half of all CHD events. Polygenic risk scores can potentially detect important characteristics overlooked by conventional population health screening programs.

Evidence on the health and economic value of polygenic risk scores across various targeted interventions has been limited and often ambiguous. The reproducible evidence presented here demonstrates the role polygenic risk scores can play in the prevention of CHD-associated morbidity.

ThermoFisher
SCIENTIFIC

Methodology

Experimental design: Hypothetical patients were segmented for testing based on a model informed by data from the Institute for Molecular Medicine Finland (FIMM), national health care registers, and published literature.

Population: 100,000 patients (age 45 or older, women and men from Finnish population).

Time horizon: 10 years.

Willingness-to-pay threshold: The optimized strategy (blue in the figure below) had a 100% probability of being cost-effective and improving health outcomes at and above a willingness-to-pay threshold of $30.5K/QALY.

Approach

The cost benefit of multiple patient management strategies for using traditional and polygenic risk scores in the prevention of CHD-associated outcomes through tailored statin therapy was evaluated. The figure below illustrates the optimal approach, leading to tailored statin treatment for a moderate-risk group.

Example result: In a Finnish population of 100,000 patients aged 45 or older, polygenic risk scores would be expected to be generated for the 2,910 patients whose risk level is between 17% and 22% according to traditional risk factors, decreasing the total costs of a preventative intervention program by approximately $300,000 and increasing the health outcome by 1.7 QALYs compared to the use of traditional risk factors alone.

Conclusion

If targeted optimally, the use of polygenic risk scores along with traditional risk factors is cost-beneficial in the prevention of coronary heart disease.

"… genetic testing is a promisingly cost-effective technology in the prevention of CHD, compared to the optimal use of traditional risk factors only …" [1]

Optimized strategy for patients with moderate risk for CHD
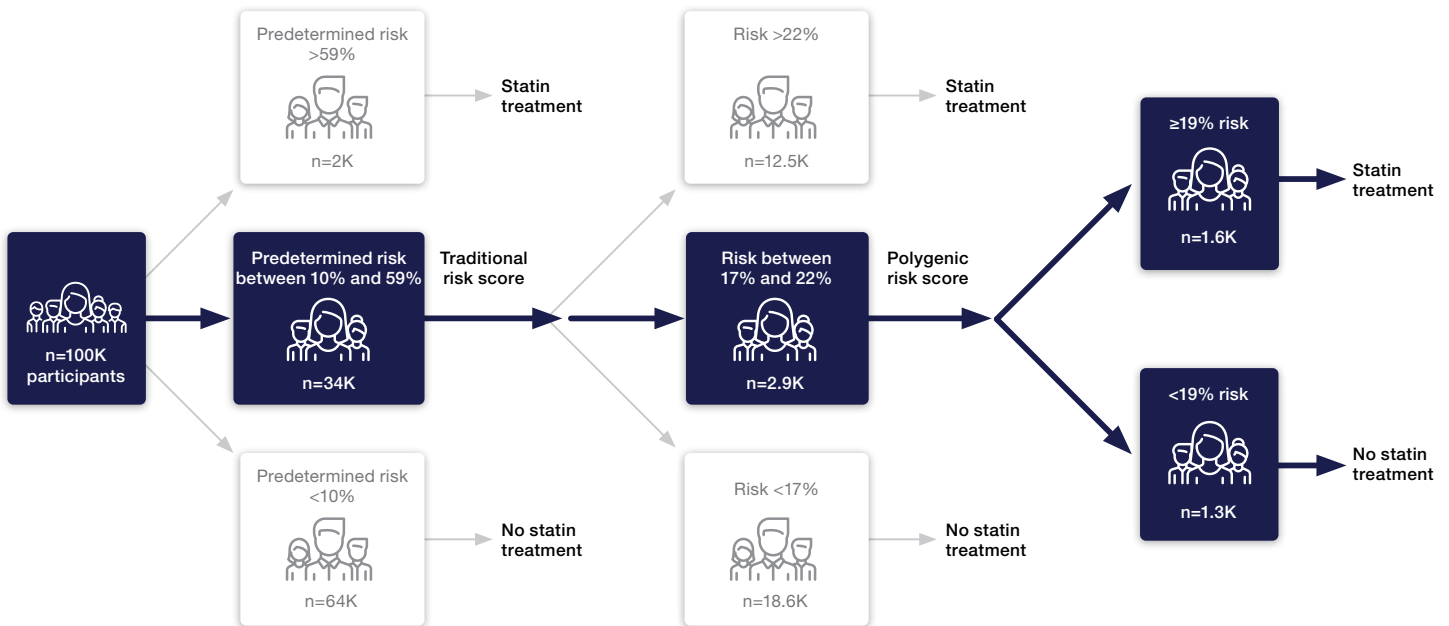
Reference

1. Hynninen Y, Linna M, Vilkkumaa E (2019) Value of genetic testing in the prevention of coronary heart disease events. *PLoS ONE* 14(1): e0210010.

Find out more about PRS at
thermofisher.com/predictive-genomics

Thermo Fisher
SCIENTIFIC