



Aalto-yliopisto  
Perustieteiden  
korkeakoulu

# Optimising the use of genetic testing in prevention of CHD using decision programming

*Helmi Hankimaa*

*14.10.2020*

Advisor: *Olli Herrala*

Supervisor: *Fabricio Oliveira*

The document can be stored and made available to the public on the open internet pages of Aalto University.  
All other rights are reserved.

# Context

The majority of CHD events occur within the population that is *not* classified to be at high risk. Targeted genetic screening has been shown by to improve classification Tikkanen et al. (2013).

Genetic testing has been found cost-effective for some cardiovascular diseases. For preventative statin treatment of CHDs, there are contradicting results. A study by Hynninen Y, Linna M, Vilkkumaa E (2019) found genetic testing to be cost-effective using dynamic programming.

# Objective

The aim is to optimise the targeting and the use of genetic testing with traditional risk factors in preventing CHDs and thus assess the cost-effectiveness of genetic testing.

Mathematically, the optimisation will be done by maximising the expected value of net monetary benefit (NMB).

$$NMB = \text{health outcomes} \cdot \lambda - \text{costs}$$

where health outcomes are measured in quality-adjusted life-years, and  $\lambda$  is the societal willingness-to-pay threshold.

# Objective

- *Who to test?*
- *Which test to use and in which order?*
  - *Options: traditional risk score and genetic risk score.*
- *Who to treat with preventative care?*

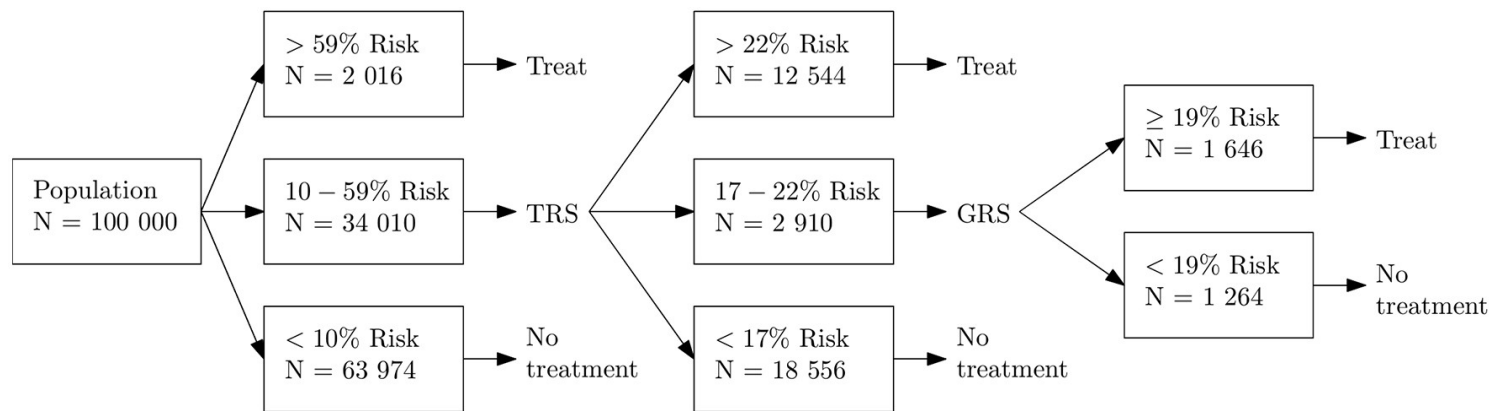


Figure from "Value of genetic testing in the prevention of coronary heart disease events" by Hynninen Y, Linna M, Vilkkumaa E, 2019, PLoS ONE 14(1): e0210010. <https://doi.org/10.1371/journal.pone.021>, page 16.

# Limits

- The research will solve the same optimisation problem as is solved in the study “Value of genetic testing in the prevention of coronary heart disease events” by Hynninen Y, Linna M and Vilkkumaa E (2019)
- Some limits of the model: we consider a 10-year time horizon, the health of a patient is represented by a binary variable and the health is assumed to be static during the testing period.

# Method

A decision programming model will be constructed to optimise this problem.

The decision programming model will be implemented using the Julia code language, using the DecisionProgramming.jl package.

The same data is used as in the study by Hynninen Y, Linna M, Vilkkumaa E (2019) where the same problem was optimised using dynamic programming.

# Literature and references

- Salo, A., Andelmin, J., & Oliveira, F. (2019). Decision Programming for Multi-Stage Optimization under Uncertainty. *arXiv preprint arXiv:1910.09196*.
- Hynninen Y, Linna M, Vilkkumaa E (2019) Value of genetic testing in the prevention of coronary heart disease events. *PLoS ONE* 14(1): e0210010.  
<https://doi.org/10.1371/journal.pone.0210010>
- Jarmul J, Pletcher MJ, Hassmiller Lich K, Wheeler SB, Weinberger M, Avery CL, et al. Cardiovascular genetic risk testing for targeting statin therapy in the primary prevention of atherosclerotic cardiovascular disease. *Circ Cardiovasc Qual Outcomes*. 2018; 11(4):e004171. <https://doi.org/10.1161/CIRCOUTCOMES.117.004171> PMID: 29650716
- Tikkanen E, Havulinna AS, Palotie A, Salomaa V, Ripatti S. Genetic risk prediction and a 2-stage risk screening strategy for coronary heart disease. *Arterioscler Thromb Vasc Biol*. 2013; 33(9):2261–6.  
<https://doi.org/10.1161/ATVBAHA.112.301120> PMID: 23599444

# Key dates

- Presentation of topic 14.10.2020
- Studying decision programming October 2020
- Development of optimisation model October-November 2020
- Writing thesis October-December 2020
- Presentation of results 2.12.2020