

Polygenic Risk Scores

Genetic risk analysis for complex diseases

Understanding Polygenic Risk Scores

Polygenic risk scores (also known as PRS or polygenic scores) are tools for assessing an individual's risk of developing a specific disease. Combining genomic data and statistical modeling, polygenic risk scores have the potential to advance complex disease understanding and precision medicine.

What Can PRS Do?



Individuals

Provide a better understanding of their relative risk of developing a specific condition, empowering them to make corresponding lifestyle adjustments.



Population

Inform population-wide disease analysis, informing clinicians, researchers, and decision-makers about large-scale disease prevalence and risk.



Clinicians & Research

Provide insight into disease, with the potential to inform treatment decisions, drug development, and further disease research directions.

The Value of PRS

Predicting risk for genetic diseases is a challenge of varying degrees of complexity. Unlike monogenic diseases, for which risk predictions are relatively straightforward, polygenic diseases require a tailored approach. PRS is a helpful tool for simplifying the prediction of complex, heritable diseases.

Paired with environmental and lifestyle factors, a validated PRS can provide a clearer disease risk.



Genetic factors
& associated
relative PRS



Environment &
lifestyle



Overall complex
disease risk

PRS In Practice

PRS has potential applications across many common diseases and conditions, including:

- > Coronary artery disease¹
- > Type 2 diabetes^{2,3}
- > Breast cancer⁴
- > Psychiatric conditions, like bipolar disorder^{5,6}
- > Heart failure⁷

Efforts to expand PRS development and application are picking up speed. To date, researchers have developed **more than 3,500 Polygenic Risk Scores, for more than 600 traits, resulting in more than 450 publications.**⁸

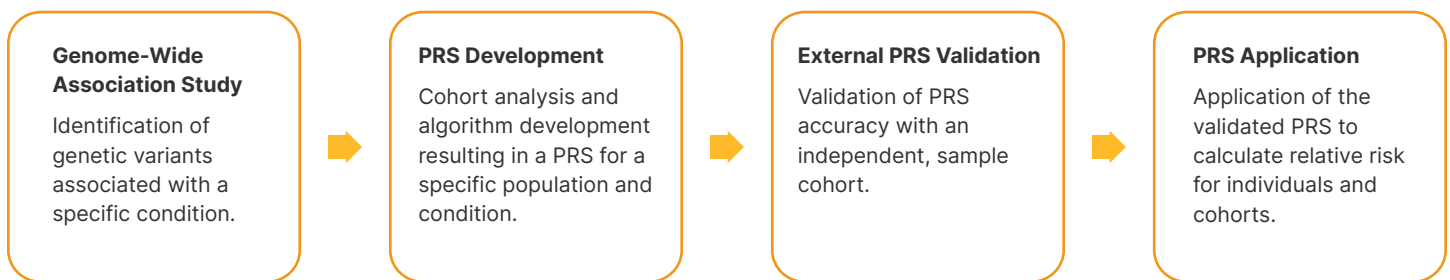


“ ...polygenic risk scoring for various disease states is helpful for everybody and has the power to change the way healthcare providers and patients understand health and communicate about disease risk. ”

Dr. Ernst Schaefer, MD
Chief Medical Officer and Laboratory Director,
Boston Heart Diagnostics

Developing and Validating a PRS Score

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Want to Learn More About Polygenic Risk Scores?

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Learn more at

<https://www.illumina.com/areas-of-interest/complex-disease-genomics/polygenic-risk-scores.html>

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