



allelica

Enabling the next generation
of clinical genomics

THE POLYGENIC RISK SCORE COMPANY



Allelica Office in Rome, Italy

Founded in 2017 by a team of researchers from the Wellcome Trust Center for Human Genetics in Oxford, UK and University of Rome La Sapienza, Italy, Allelica was established with the goal of advancing genomic medicine to reduce the onset and burden of common disease.

Today Allelica is working with leading health systems and genetics labs in the United States and across Europe to advance chronic disease prevention through widened application of the Polygenic Risk Score (PRS) in healthcare.

STREAMLINING POLYGENIC RISK SCORE DEVELOPMENT & IMPLEMENTATION

PRS identify the contribution to a patient's risk that comes from their genes. Allelica has created a secure, trusted and easy to use platform which equips genetics labs and research groups to compute existing PRS and generate new scores using a suite of the most powerful algorithms and bioinformatics tools. Our platform has been used to develop and publish top-ranking PRS for coronary artery disease, breast cancer, prostate cancer, and a range of other diseases.

SOFTWARE AS A SERVICE FOR PRS

Allelica's Software as a Service (SaaS) is a cloud or on-premises solution for comprehensive PRS analysis. The platform enables clinical laboratories and researchers to calculate individual PRS, produce automated reports, and build new PRS according to the "Polygenic Risk Score Reporting Standards" developed by the NHGRI Clinical Genome Resource (ClinGen), Complex Disease Working Group and the Polygenic Score Catalog (PGS). The SaaS is made up of 3 modules: PREDICT, DISCOVER, and VALIDATE.

Platform features

- HIPAA & GDPR Compliant
- CE-marked medical device
- Available on the cloud or on-premises
- API integration for direct report uploading
- Compatible with raw microarray or Low coverage WGS data

THE PREDICT MODULE

CALCULATE & REPORT PRS FOR CHRONIC DISEASE

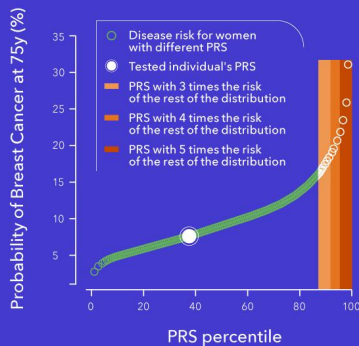
The PREDICT module is a resource for clinical laboratories and healthcare providers to build personalized PRS reports for a range of complex diseases. Our end-to-end solution starts with a simple file upload of patient genotype data and delivers a patient's customized genetic risk by leveraging the UK Biobank data with our own analytics. Our technology accounts for a patient's ancestry by utilizing the genetically diverse 1000 Genomes Project resource.



Generate automated, highly personalized PRS reports with clinical grade risk prediction.

- + Ancestry-specific risk prediction
- + Top-performing PRS
- + Lifetime genetic risk and absolute risk models
- + Customizable and white label report

Comparison of disease risk and PRS



Trait	min	Genetic Risk	max	Absolute Risk Including PRS and Clinical risk factors
Coronary Artery Disease		High		0% 5% 5% 7.5% 7.5% 20% >20% NORMAL BORDERLINE INTERMEDIATE HIGH Absolute 10 year risk thresholds per AHA guidelines
Type 2 Diabetes		Low		0% 6% 6% 20% >20% LOW INTERMEDIATE HIGH Absolute 10 year risk per ARIC score
Breast Cancer		Average		0% 20% >20% Lifetime absolute risk thresholds per ACA guidelines
Early Menopause		High		0% 5% 5% 15% 15% 50% >50% LOW INTERMEDIATE HIGH VERY HIGH Probability of menopause before age 45

This is an example report and not an exhaustive list of PRSs and integrated models that Allelica performs

THE DISCOVER + VALIDATE MODULES

DEVELOP & TEST POWERFUL NEW PRS

Use the DISCOVER Module to build your own PRS from genetic data and summary statistics from a Genome Wide Association Study (GWAS) by running LDPred2, SCT, PRScs, SBayesR, lassosum, Support vector machine and Clumping & Thresholding in parallel. Validate your new PRS on an independent dataset and investigate its applicability to a new population with the VALIDATE module.

Together, the DISCOVER + VALIDATE Modules enable researchers to construct new PRS using their own datasets or the resources of the UK Biobank.

The outputs of these modules provide a set of PRS and a quantification of their predictive performance. Having the ability to choose the best PRS from different methods allows users to align their research to [Polygenic Risk Score Reporting Standards](#) published in Nature (2021).

1

Select datasets for new PRS development

2

Run the top seven algorithms in parallel

3

Test the predictive power of the PRS across different populations

CORONARY ARTERY DISEASE

Reference	Khera et al 2018	Bolli et al 2021
AUC	0.818	0.822
3 fold risk	10.5%	21%

BREAST CANCER

Reference	Mavaddat et al 2018	Allelica
AUC	0.679	0.708
3 fold risk	1%	8%

PROSTATE CANCER

Reference	Schumacher et al 2018	Allelica
AUC	0.826	0.848
3 fold risk	30%	30%



Schedule a free PRS consultation now: allelica.com/contactus/