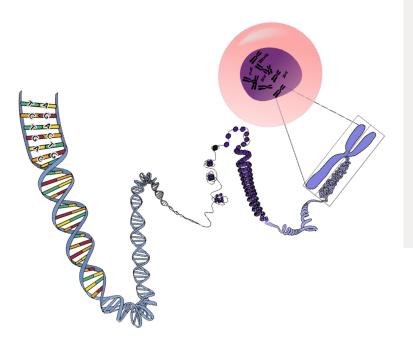
# **BOSTON HEART DIAGNOSTICS**

# **POLYGENIC RISK MAP**

Boston Heart's Polygenic Risk Map test is a genetic test that leverages Polygenic Risk Score (PRS) analysis and reporting. The resulting risk scores provide clinical insights into a variety of important health conditions.

# CLINICAL UTILITY OF THE POLYGENIC RISK MAP

- Risk scores are determined by numerous well-studied genetic variations, and allow for impactful and targeted application of genomic testing to personalized medicine.
- High-risk individuals generally have a substantially increased relative genetic risk for these conditions as compared to low-risk subjects.
- Because lifetime risk of these conditions are also substantially influenced by environment, lifestyle, and medical care, a greater understanding of genetic risks may lead to greater preventive efforts.
- Cost effectiveness studies suggest that providers can reduce cardiovascular events and healthcare system costs by using polygenic risk scores in appropriate patients.<sup>1</sup>
- This type of testing is meant to augment rather than replace standard risk factor assessments for cardiovascular disease, breast cancer, prostate cancer or other conditions.





# POLYGENIC RISK MAP REPORTS

Validated polygenic risk scores are available for many of the most prevalent causes of premature death and morbidity.

- Coronary Artery Disease (CAD) Leading cause of death.
- Atrial Fibrillation 5-fold increase in stroke risk.
- Type 2 Diabetes 3-fold increase in CAD risk.
- **Hypertension** Causes CAD, stroke, and kidney disease.
- **BMI** Increases risk of CAD, diabetes and some cancers.
- **Triglycerides** -Increases risk of CAD and pancreatitis.
- Low HDL Cholesterol Increases risk of CAD.
- **High LDL Cholesterol** Increases risk of CAD.
- Lipoprotein (a) Increases risk of CAD and aortic stenosis.
- Alzheimer's Disease The leading cause of dementia.
- Inflammatory Bowel Disease (IBD) Ulcerative colitis and Crohn's disease risk.
- Breast Cancer A leading cause of cancer in women.
- Osteoporosis A common cause of bone fractures.
- **Early Menopause** Risk of menopause before age 45.
- Prostate Cancer A leading cause of cancer in men.
- Ancestry Based on 26 global populations.



## **ABOUT THE METHODS**

- Uses the Illumina Infinium Global Screening Array technology.
- The analysis assesses over 650,000 DNA markers across the human genome.
- Risk scores are based on over 500.000 subjects from the United Kingdom BioBank and multiple other studies.
- Genetic analyses are performed by Eurofins laboratory partner Clinical Enterprise.
- The risk informatics are provided by Allelica, a leading polygenic risk score company.
- Risk scores provide the lifetime risk of developing the condition of interest.

## ORDERING, REPORTING, AND SAMPLE INFORMATION

### Polygenic Risk Map Panels

### Polygenic Risk Map – Female Complete Order Code:87103 \$ 349

- CAD Atrial fibrillation Type 2 Diabetes Hypertension BMI Triglycerides Low HDL Cholesterol High LDL Cholesterol Lipoprotein (a)
- Alzheimer's disease
   Inflammatory Bowel Disease (IBD)
   Breast cancer
   Osteoporosis
   Early Menopause
   Ancestry

#### Polygenic Risk Map – Male Complete

Order Code:87104

\$ 349

- CAD Atrial fibrillation Type 2 Diabetes Hypertension BMI Triglycerides Low HDL Cholesterol High LDL Cholesterol Lipoprotein (a)
- Alzheimer's disease Inflammatory Bowel Disease (IBD) Prostate cancer Ancestry

#### Polygenic Risk Map – Cardiovascular Disease

Order Code:87101

\$ 199

• CAD • Atrial fibrillation • Type 2 Diabetes • Hypertension • BMI • Triglycerides • Low HDL Cholesterol • High LDL Cholesterol • Lipoprotein (a) • Ancestry

#### Polygenic Risk Map – Female Essential

Order Code:87102

\$ 199

Breast cancer • Osteoporosis • Early Menopause • Ancestry

Polygenic Risk Map – Prostate Cancer

Order Code:87105

\$ 199

Prostate cancer • Ancestry

Polygenic Risk Map – Alzheimer's Disease

Order Code:87106

\$ 199

Alzheimer's disease
 Ancestry

Polygenic Risk Map – Inflammatory Bowel Disease (IBD)

Order Code:87107

\$ 199

POLYGENIC RISK SCORE EXPLAINED

A polygenic risk score (PRS) measures the component of disease risk from many genetic variants spread throughout the genome. Lifetime risk of CAD is calculated by comparing

the tested individual's PRS to a reference population. PRS above the percentile is considered high because it confers a greater

than 45% lifetime risk, which is three times the risk of disease compared to the remainder of the population. The chart shows how PRS translates to lifetime risk of coronary artery

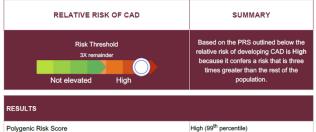
disease. This CAD PRS comprises 1,926,521

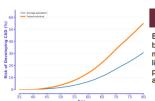
• Inflammatory Bowel Disease (IBD) • Ancestry

The \$199 panels can be combined: \$199 for the first and \$49 for each additional.

## Sample CAD Report

#### CAD RISK REPORT





Everyone's CAD risk increases with age. The blue line on the chart shows this increase for a males with an average PRS. The PRS (orange line) of the tested individual shows that this person's risk rises more quickly than the average (blue line).

times greater than the rest of the population.

High (99<sup>th</sup> percentile)

CAD RISK OVER TIME

# RECOMMENDATIONS

The suggested next step is a conversation with a physician to discuss assessing the LDL cholesterol levels and 10 year absolute risk of cardiovascular disease\*. Additional non-genetic risk factors will also affect your lifetime risk. There are behavioral and dietary approaches to lowering risk, including following a healthy lifestyle and regular exercise.

CAD POLYGENIC RISK SCORE REPORT

\*Information on national heart disease guidelines can be found on the AHA website here AHA Guidelines on management of blood lipids (2018) Circ 139:e1144-e1161\_

## Specimen Requirements

Buccal Swab

# For more information, contact your **Area Sales Manager** or Customer Care at **877.425.1252** or **customercare@bostonheartdx.com**

#### References

1. Mujwara D, Henno G, Vernon ST, et al. Integrating a Polygenic Risk Score for Coronary Artery Disease as a Risk-Enhancing Factor in the Pooled Cohort Equation: A Cost-Effectiveness Analysis Study. J Am Heart Assoc. 2022;11(12):e025236

877.425.1252 bostonheartdiagnostics.com





