Proactive Health
INTEGRATED PRS™
BREAST CANCER

TEST CODE: PR42009

Overview

The MyOme Proactive Health Plus Integrated Polygenic Risk Score™ (iPRS™) Breast Cancer test uses a Blended Genome-Exome (BGE) backbone built from whole exome sequencing and low coverage whole-genome sequencing to estimate risk of developing breast cancer.

Clinical Use

This test is a comprehensive risk assessment tool (not a diagnostic test) intended for women who have not previously been diagnosed with breast cancer and who do not have a clinically relevant variant in a breast cancer predisposition gene. This tool provides a 5-year and remaining lifetime risk of developing breast cancer and may assist with the development of a personalized treatment and management strategy in conjunction with standard clinical assessment.

Method

Genomic DNA is obtained from submitted samples and sequenced using Illumina technology. A PCR-free whole-genome library is constructed and a sub-aliquot is taken through PCR amplification and exome selection. The blended genome and exome libraries are sequenced to generate 2x150 bp paired-end reads resulting in low-coverage whole-genome and higher coverage exome data. Reads are aligned to human genome reference assembly GRCh38. Genotype likelihoods are estimated for bases covered by at least one sequencing read. Genotypes at additional sites are imputed based on a genotype reference panel. A PRS is calculated for each of 5 continental ancestries– African, Admixed American, East Asian, South Asian, and European–and standardized and weighted to produce a cross-ancestry PRS (caPRS). The caPRS is integrated with an individual's clinical risk based on the Tyrer-Cuzick (TC) model to estimate a 5-year and remaining lifetime risk of developing breast cancer.[1,2]

Sample Types

- Blood (2 EDTA tubes)
- Saliva (2 tubes)
- Buccal (2 swabs)

Turn Around Time

- From initial sample received, approximately 6 to 8 weeks
- For previously processed sample, approximately 24 to 48 hours

Included

- Cohesive report with 5-year and remaining lifetime risk of developing breast cancer that includes actionable recommendations

1. Tshiaba P et al. Integration of a cross-ancestry polygenic model with clinical risk factors improves breast cancer stratification. JCO Precis Oncol. 2023. doi: 10.1200/PO.22.00447. **2**. Tyrer J et al. A breast cancer prediction model incorporating familial and personal risk factors. Stat Med. 2004. doi: 10.1002/sim.1668.