

**Multi-Gene Panel Testing Detects Equal Rates  
of Pathogenic *BRCA1/2* Mutations and has a  
Higher Diagnostic Yield Compared to Limited  
*BRCA1/2* Analysis Alone in Patients at Risk for  
Hereditary Breast Cancer**

Nimmi S. Kapoor, MD,  
Lisa D. Curcio, MD, Carlee A. Blakemore, BS,  
Amy K. Bremner, MD, Rachel McFarland, BS, John G. West, MD,  
Kimberly C. Banks, MS, CGC

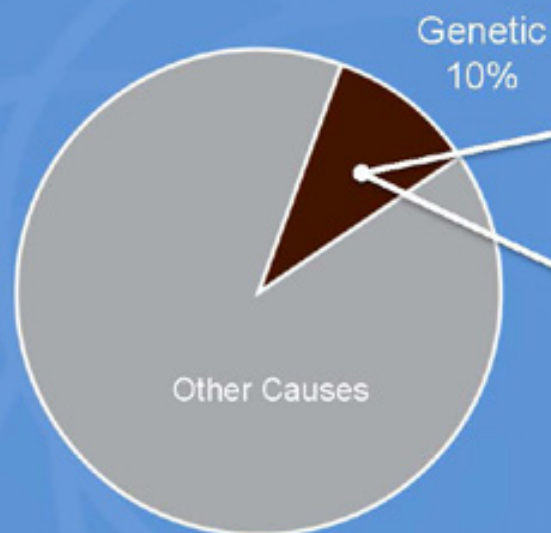
Disclosures: NSK, LDC are Consultants for Ambry Genetics, AB is a  
Consultant for Myriad Genetics, RF is and KCB was employed by  
Ambry Genetics



# Background

- The landscape of genetic testing is in rapid evolution
- Knowledge, Technology, Open Market, Celebrity Influence
- Are newer multi-gene tests as effective as single-gene tests at detecting *BRCA1/2* mutations?
- What more do we gain from expanded panels?

Identifying an inherited genetic mutation has the potential to prevent cancer and save lives



*BRCA1/2* accounts for part of this hereditary risk

Numerous other genes account for the rest

Breast Cancer  
Etiology

# Limited Tests vs Multi-Gene Panel Tests



## Limited-Gene

Tests for *BRCA1/2* mutations



## Multi-Gene\*

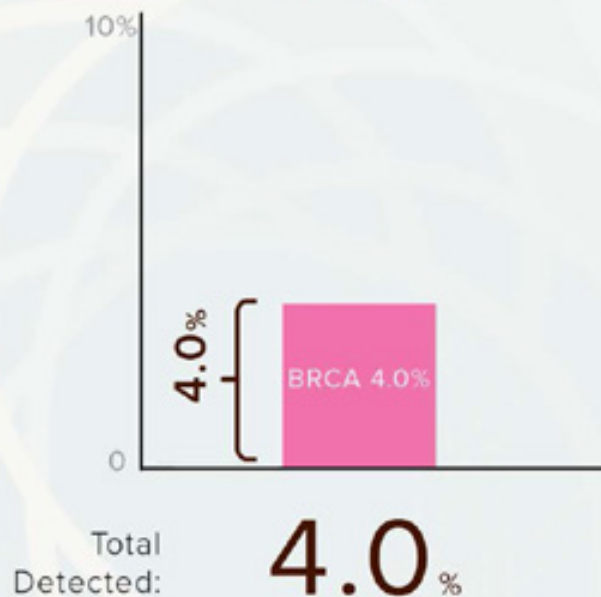
Tests up to 43 genetic mutations



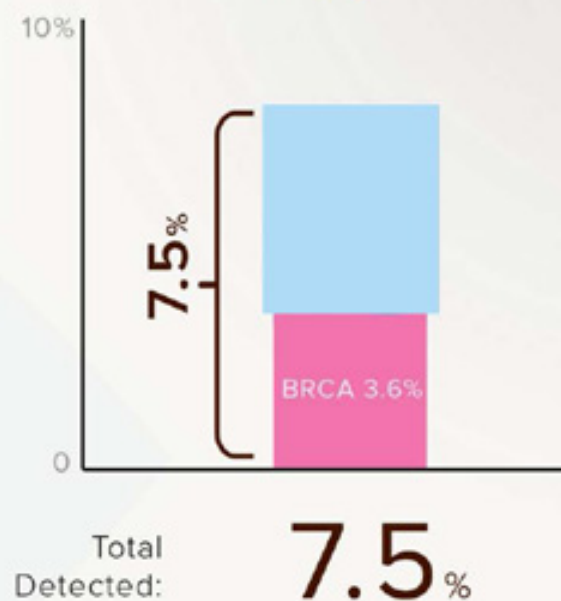
# 966 patients

National guidelines were used to determine eligibility for testing based on personal and family history

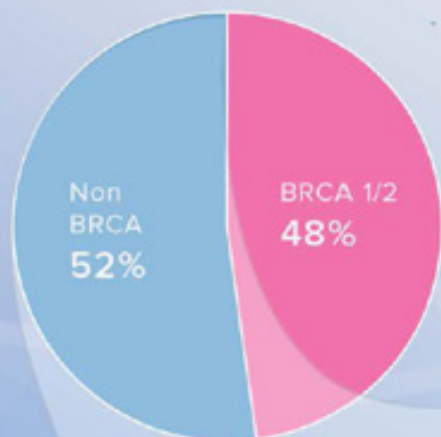
Limited Group  
n=629



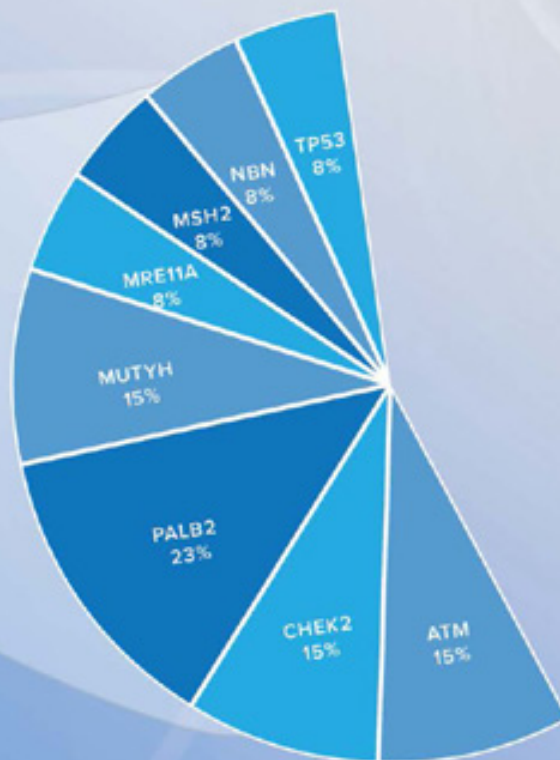
Panel Group  
n=337



The most common non-BRCA mutations identified were in *PALB2*, *CHEK2*, and *ATM*



NON-BRCA1/2  
MUTATIONS



Multi-Gene Panel  
Testing  
DOUBLES  
Detection Rate

# KNOW YOUR RISK

## Benefits of multi-gene testing

Effectively evaluates *BRCA1/2* AND other breast cancer-related mutations.

Empowers women with more information about their risk.

Provides clinical information to enhance our knowledge of breast cancer biology.

Doctors are the gatekeepers to genetic testing.  
We have an obligation to help patients understand genetic risk and how testing can guide health care decisions.

# Thank You!

To be Presented at the 2015 Annual Meeting of the American Society of Breast Surgeons by  
**Nimmi Kapoor, MD**

Cancer Research Collaboration

Shuwei Li

Luke Gale

Anne Lee

Andrew Ausanka-Cruess

Paul Colacchio

Holly Laduca

Nimmi S. Kapoor, MD,  
Lisa D. Curcio, MD, FACS,  
Carlee A. Blakemore, BS, Amy  
K. Bremner, MD, Rachel  
McFarland, BS, John G. West,  
MD, FACS, Kimberly C. Banks,  
MS, CGC, MBA