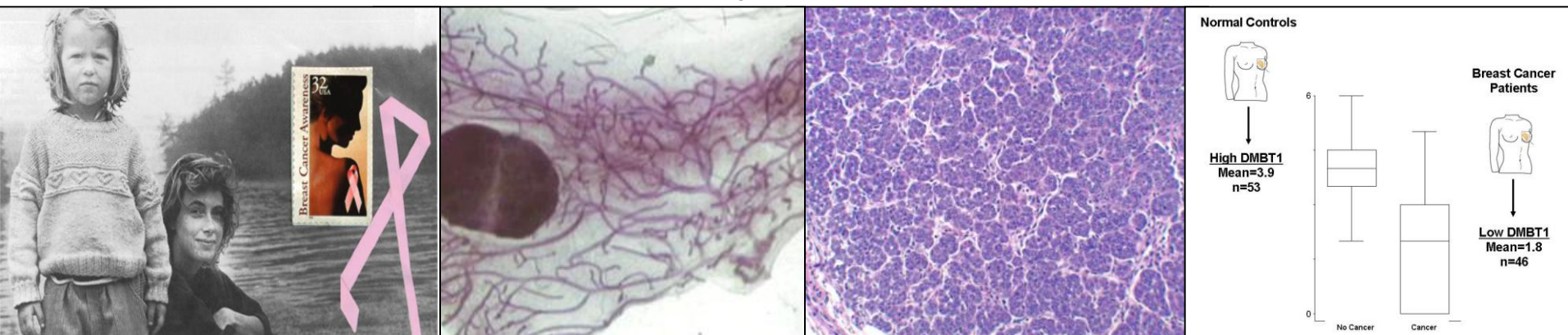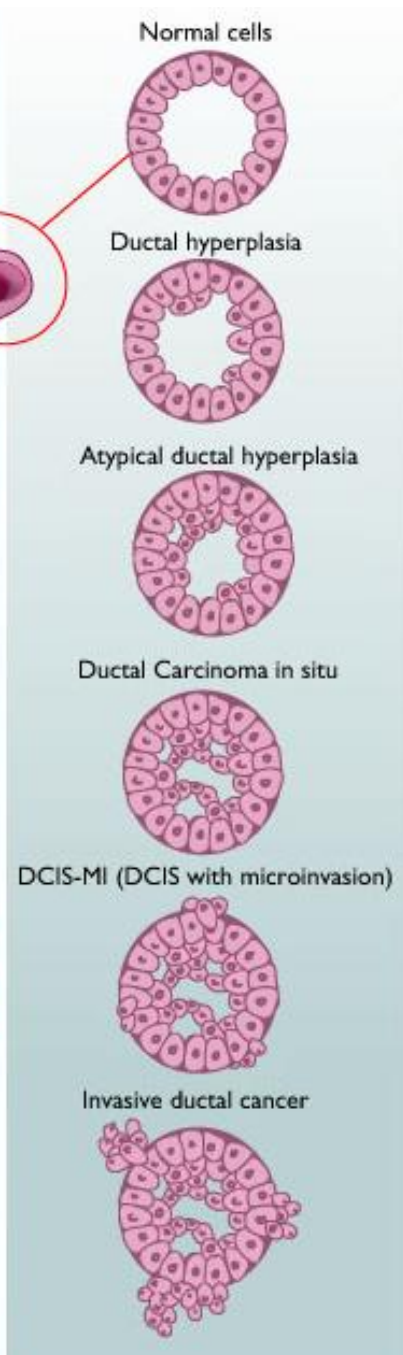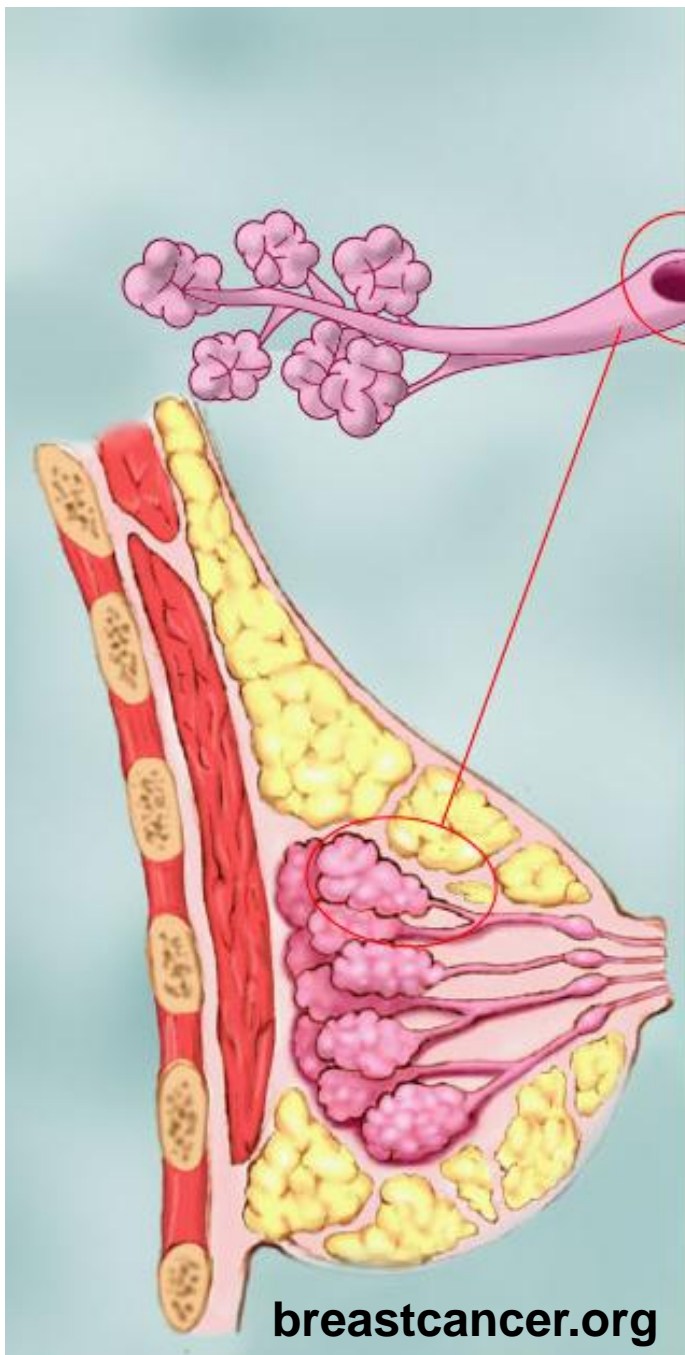# *A case of mistaken identity --- Biomarkers for high risk premalignant breast lesions*

**D. Joseph Jerry, UMass Amherst and PVLSI**

**Karl Simin, UMass Medical**

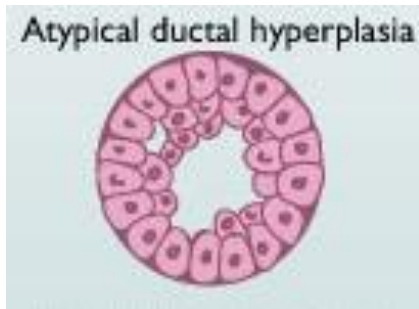*May 8th, 2013*

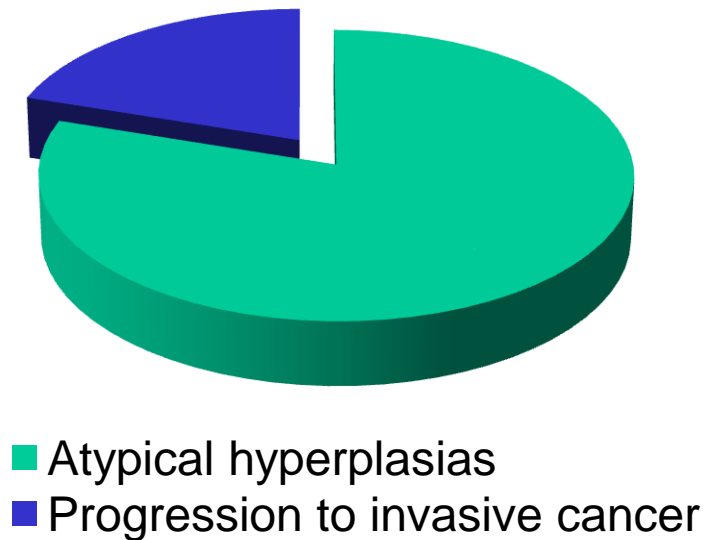A Baystate Medical Center ■ UMass Amherst Research Partnership

Normal cells

Ductal hyperplasia

Atypical ductal hyperplasia

Ductal Carcinoma in situ

DCIS-MI (DCIS with microinvasion)

Invasive ductal cancer

**breastcancer.org**

*Estimated costs: $20,000 to $100,000/patient.*

Campbell JD, Ramsey SD. Pharmacoeconomics. 2009;27(3):199-209. PMID: 19354340

Atypical ductal hyperplasia

**Expect 10,000 new cases annually**

Invasive ductal cancer
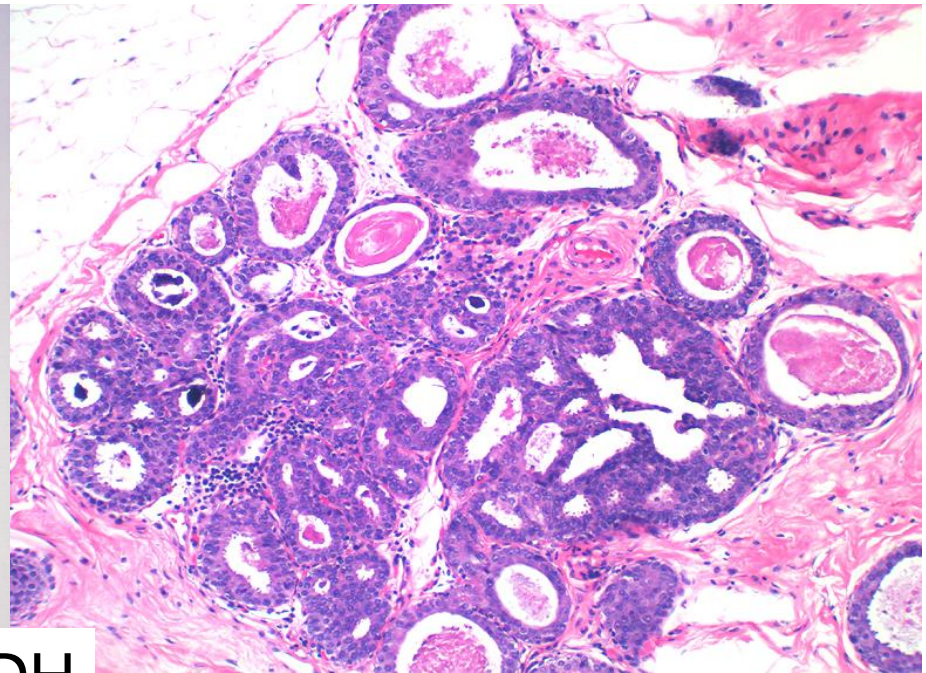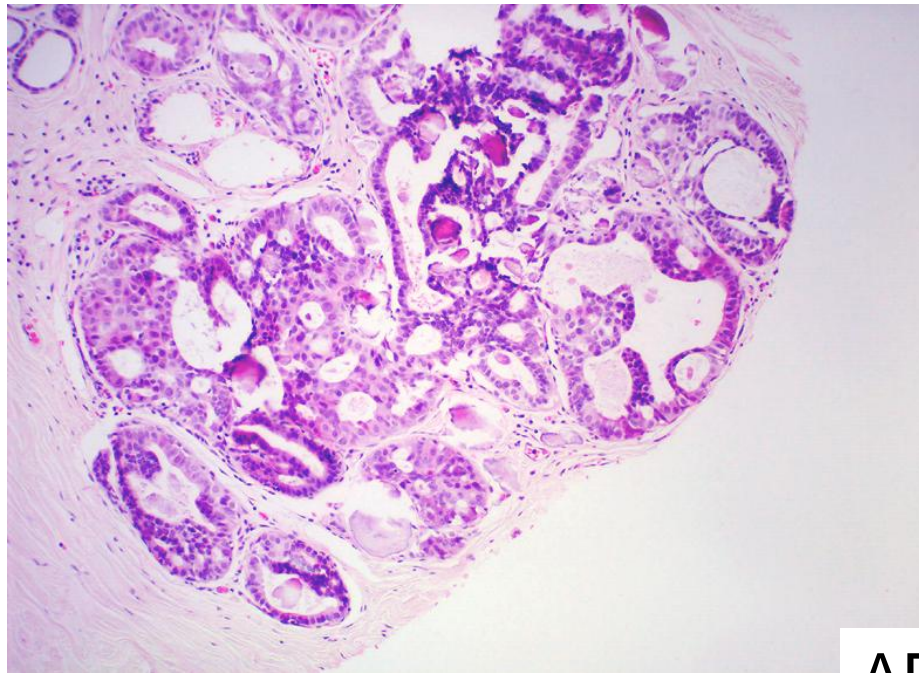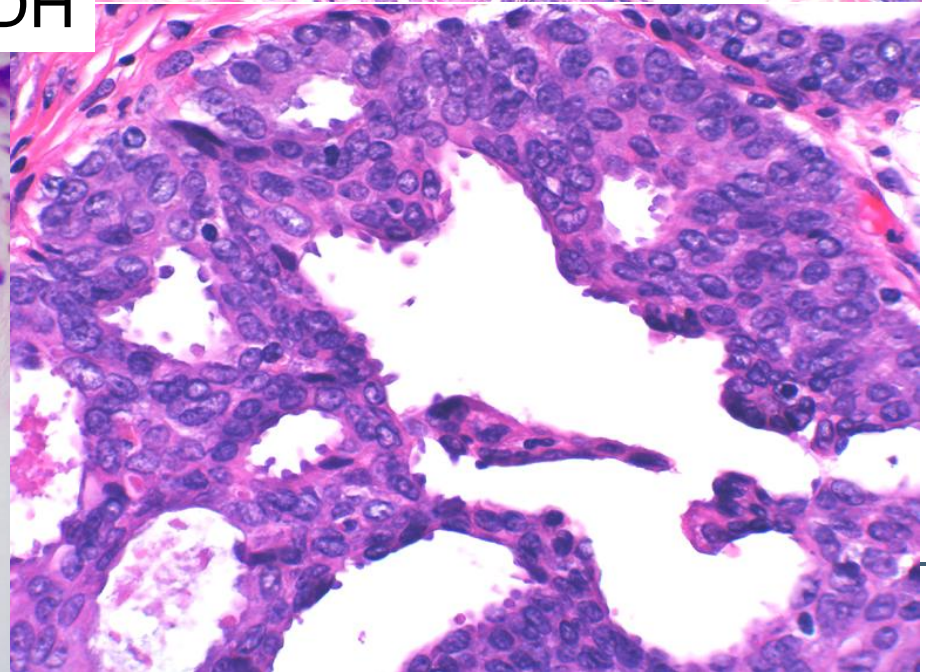
**50,000 new cases annually**

**Up to $1B to treat**

■ Atypical hyperplasias
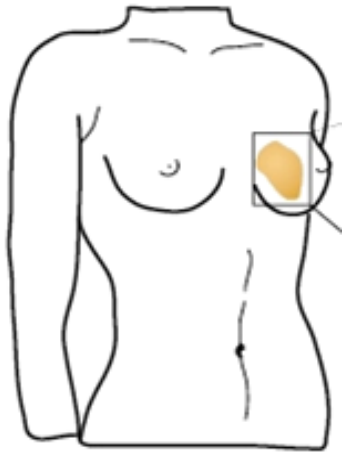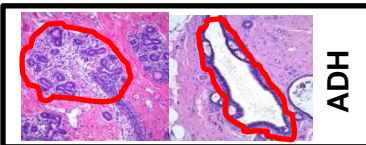■ Progression to invasive cancer

ADH

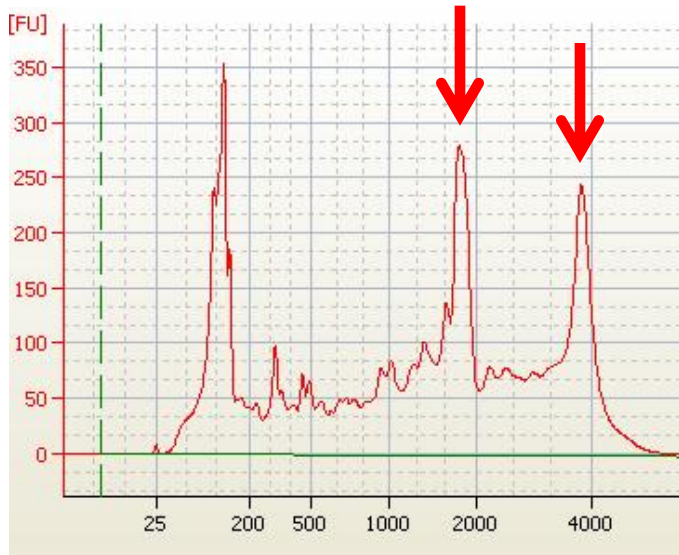# Developing biomarkers of high risk premalignant breast lesions



Micro-dissect
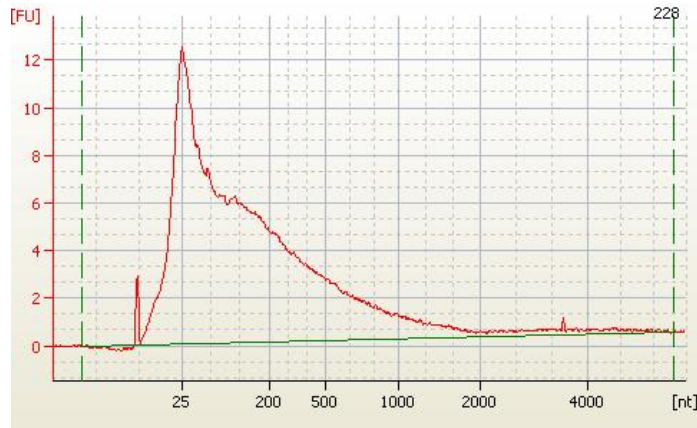
**Subtypes of Atypical Hyperplasias**
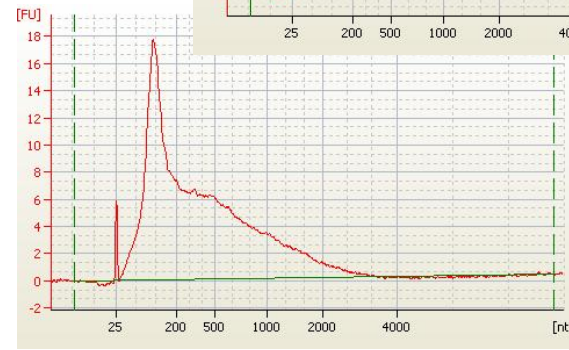
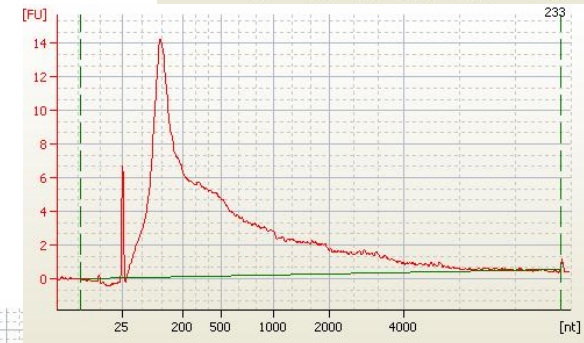LN

ADH

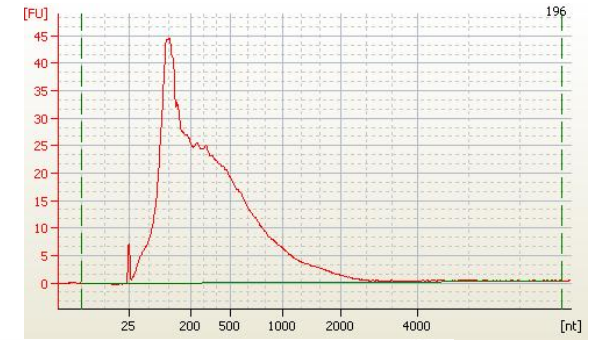**Subtypes of lesions with malignant potential**

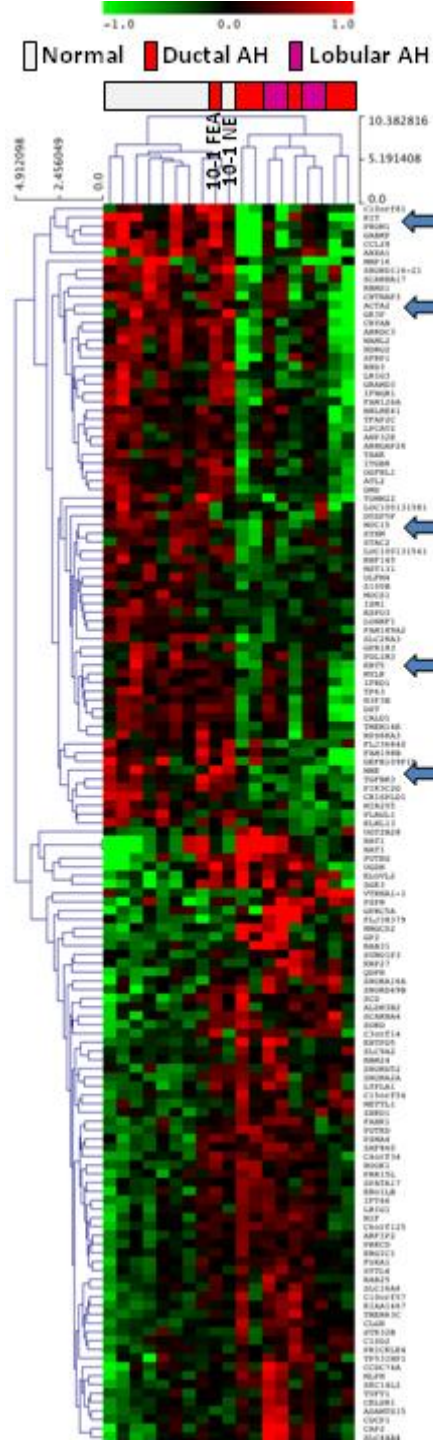# Bioanalyzer electropherograms



**Intact RNA**

**FFPE RNA**

# Atypical hyperplasia (UMMS 1999-2003)

Upstaging upon re-excision

2833 breast core biopsies
120 with primary diagnosis of ADH

# Summary

- Value of diagnostic test
  - Large interobserver variability in diagnosing premalignant lesions (Jain et al., 2011. PMID 21532546)
  - Identify subgroups to benefit from preventive therapies.
  - Identify molecular pathways to provide appropriate preventive treatments
- Technical challenges
  - Minute amounts of tissue
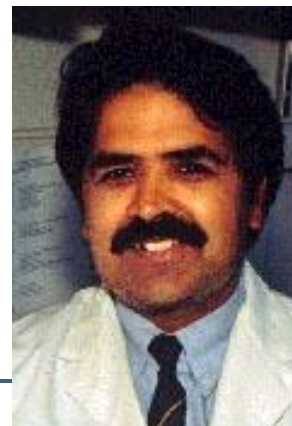  - Fragmentation of RNA in FFPE tissues
  - Amplification and labeling for robust detection

## University of Massachusetts - Amherst

### D. Joseph Jerry, Ph.D.

Ellen Dickinson
Amy Roberts
Jeff Kane
Mary Hagen

Karen Dunphy
Haoheng Yan
Erick Roman
Nick Griner

## UMass Medical Center

**Karl Simin, Ph.D.**

**Ashraf Khan, M.D.**

### Sallie Smith Schneider, Ph.D.

Kelly Gauger

## Baystate Medical Center

**Grace Makari-Judson, M.D.**

**Giovanna Crisi, M.D.**