May 8th, 1:30 PM - 3:00 PM

A Case of Mistaken Identity: Biomarkers for High Risk Premalignant Breast Lesions

D. Joseph Jerry
University of Massachusetts - Amherst

Karl Simin
University of Massachusetts Medical School

Follow this and additional works at: http://escholarship.umassmed.edu/cts_retreat

Part of the Biological Factors Commons, Cancer Biology Commons, Diagnosis Commons, Neoplasms Commons, Pathological Conditions, Signs and Symptoms Commons, and the Translational Medical Research Commons

This work is licensed under a Creative Commons Attribution-Noncommercial-Share Alike 3.0 License.


This material is brought to you by eScholarship@UMMS. It has been accepted for inclusion in UMass Center for Clinical and Translational Science Research Retreat by an authorized administrator of eScholarship@UMMS. For more information, please contact Lisa.Palmer@umassmed.edu.
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
Estimated costs: $20,000 to $100,000/patient.

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

Progression to invasive cancer

50,000 new cases annually

Expect 10,000 new cases annually

Up to $1B to treat
Developing biomarkers of high risk premalignant breast lesions

Subtypes of Atypical Hyperplasias

Subtypes of lesions with malignant potential
Bioanalyzer electropherograms

Intact RNA

FFPE RNA
Log2 expression values

Normal  Ductal AH  Lobular AH

C10orf51  KIT  PROM1
GSK3B  CCL28  ANXA1  MMP16
SNORD116-21  SCARNA17  RBMS1  CNTNAP3  ACTA2  GSK3P  CRYAB  ARRD3  MAML2
NDRG2  SFRP1  ARID3A  LRIG3
NAT1  POTEGR  UGDH  EST1  SGK3  PPP53K1  L1CAM  GPRC5A
Patient Population:
Diagnosis of Breast Cancer or Benign Breast Disease

IRB-approved Registry
Patient consent
Assign study ID

Patient Registry
Survey data
Clinical data
Archived tissues

Data Manager
• Collate data
• Provides summary data
• Extract data from clinical records
• Coded data for IRB-approved projects

IRB-approved projects

Project 1
Project 2
Project 3
Project 4

Data/Tissue With Study ID
Atypical hyperplasia (UMMS 1999-2003)

2833 breast core biopsies
120 with primary diagnosis of ADH

Upstaging upon re-excision
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
Ellen Dickinson
Amy Roberts
Jeff Kane
Mary Hagen

D. Joseph Jerry, Ph.D.

Karen Dunphy
Haoheng Yan
Erick Roman
Nick Griner

University of Massachusetts - Amherst

Sallie Smith Schneider, Ph.D.
Kelly Gauger

Baystate Medical Center
Grace Makari-Judson, M.D.
Giovanna Crisi, M.D.

UMass Medical Center
Karl Simin, Ph.D.
Ashraf Khan, M.D.

Funding from
NIH, NIEHS, Avon Foundation
Rays of Hope Foundation,
Life Science Moment Fund