

# BEHAVIORAL RESEARCH

CANCER CONTROL AND POPULATION SCIENCES

# The presentation will begin shortly

\* This presentation is being recorded. You may disconnect at any time if you do not wish to be recorded.



National Institutes of Health



## BEHAVIORAL RESEARCH

CANCER CONTROL AND POPULATION SCIENCES

# Decision-Making Steering Committee Speaker Series

Barbara Biesecker, Ph.D., M.S.,  
C.G.C.

August 5, 2014



National Institutes of Health





---

*Most Challenging issues in  
CA Prevention in the Genome Era  
Decision Making Steering Committee*

---

Barbara Bowles Biesecker, PhD, MS

# Identifying genetic risk for CA

Primarily based on hereditary CA risk due to single gene variants

Family history and characteristics of CA used to identify higher risk patients/clients

Used for recurrence risk, risks to relatives and increasingly, treatment/risk management

Comprises a relatively small subset of CA cause

# What does the genomics era look like?

With the explosion of genome technologies more tests are being offered

- Prognostic testing (Oncotype Dx™, Mammaprint™)
- Risk assessment (single gene, panel, sequencing)

Tests are migrating from hereditary cancer clinics and into primary care

# What are the testing options?

Past decisions involve risk assessing risk  
(family hx, *BRCA1/2* testing)

What's new is the scope and types of choices-  
more of them and less targeted

BreastNext (Ambry) 17 genes

Br/Ovarian CA (GeneDx) 21 genes

BROCA (Univ Wash) 26 genes

# Current challenges CA prevention

New genome technologies offer more options but hard to assess their usefulness

Tests not distinctly preference-based v. recommended

CA predisposition among IF recommendations

Choices based on insurance reimbursement

Disparities in cancer genetics services!

# Genetic counselors as choice architects

Most decisions are preference-based

Decisions should be informed

Most of choices patients/clients face are unfamiliar

Shared decision-making may be the best approach

Responsibility on genetic counselors to ensure that the way the choice is presented optimizes patient preference



# Decisions within uncertainty

Current state of testing panels or sequence result includes a significant degree of uncertainty

While there may be benefits, there is also a great deal of information that cannot be interpreted

There insufficient evidence to guide clinical practice in how to engage in shared decision making within significant uncertainty



# Uncertainty in genomic sequencing Information

How clients perceive uncertainty is likely to predict decisions:

- to learn sequence results
- to act on the information

Practitioners who consent clients face the challenges of conveying uncertainties to ensure informed choice and mitigate unrealistic expectations of the information



# Sequencing research

Use of genome sequencing is widespread

Investigators seek to identify elusive variants that contribute to diagnosed conditions

As well, pursuit of variants that contribute to common conditions is underway

More than one variant is likely contributing and the interpretation is more complex

# Multiple participant decisions

Decision to participate in sequencing studies

Intention to receive various types of results  
(hypothetical)

Decision to receive various types of results:  
    Medically actionable (recommendation?)  
    Health related but non-actionable  
    Carrier status

Decisions to use or act on the results

# Social and behavioral studies

ClinSeq<sup>®</sup> longitudinal cohort study that includes return of results

- Perceptions of uncertainty
- Intervention study comparing web-based platform to a genetic counselor
- Returning uncertain variants related to hypertrophic cardiomyopathy

Randomized controlled trial of consent to undergo genome sequencing (NICHD)

# Genome sequencing consent study

Data from the RCT may inform development of NIH CC consents for genome sequencing

Assesses perceptions of uncertainty among other outcomes

Need to follow participants for a longitudoinally to assess whether they have the patience to engage with an interrogative search for new variants for POI

# Collaborators

## ClinSeq®

Les Biesecker, MD

Katie Lewis, MS

Gillian Hooker, PhD, ScM

William Klein, PhD

Paul Han, MD, MPH, MS

## Consent

Larry Nelson, MD

Amber Cooper, MD

Holly Peay, MS

# BEHAVIORAL RESEARCH

CANCER CONTROL AND POPULATION SCIENCES

# Thank You

Questions/Comments, contact:

[NCI.BRPwebinars@icfi.com](mailto:NCI.BRPwebinars@icfi.com)

301-407-6608



National Institutes of Health

