The presentation will begin shortly

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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® 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 longitudinally to assess whether they have the patience to engage with an interrogative search for new variants for POI
Collaborators

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Thank You

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