Genetic Risk Assessment and Genetic Counseling Research

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Outline for today

• Of Risk and Popcorn
• Cancer Risk Factors
• Genetic Risk Assessment
• Implications for Cancer Control
• Genetic Counseling Research
Of Risk and Popcorn

Of Risk and Popcorn

![Graph](chart.png)
Of Risk and Popcorn

N = 250 (per bag)

Of Risk and Popcorn

Cumulative Risk

N = 250 (per bag)
What determines if a kernel pops?

- **External factors**
  - How long it's in the microwave
  - The type of microwave
  - Oil/seasonings
- **Internal factors**
  - Genes (genetically-modified corn??)
  - Soil/nutrients

Breast Cancer Risk

![Breast Cancer Risk Chart](chart.png)
What affects cancer risk?

- Diet
- Tobacco
- Hereditary
- Occupation
- Radiation
- Viruses
- Other
Family History and Chance of Breast Cancer

Claus, 1990

Twin Studies

Breast Cancer in Twins

Causes of Breast Cancer

- Genetics
- Shared Environment
- Unique Environment

BRCA1 and BRCA2

Science 2003;302(5645):643-6
More likely to be hereditary if…

- Earlier-than-expected age of onset
- Multiple primary cancers
- Rare cancers
- Inheritance pattern
- Syndrome (malignant and non-malignant characteristics)
- No clear environmental association

Peutz-Jeghers Syndrome
Cancer Control Implications

- Primary prevention
  - Medication (e.g., tamoxifen, sulindac)
  - Risk-reducing surgery
  - Lifestyle (e.g., exercise)
  - Diet (coffee??)

- Secondary prevention
  - Timing (age of initiation, interval)
  - Method (MRI)
  - Whether or not to screen at all

Cancer Control Implications

- Tertiary prevention
  - Chemotherapy
  - Extent of surgery
  - Radiation
Why genetic counseling?

- Risk communication is complicated
- Diagnosis/risk assessment often precedes effective intervention
  - Right not to know
  - Psychosocial cost/benefit
- Limitations of testing (VUS, false negative)
- Expensive
- Discrimination

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**GENETIC COUNSELING RESEARCH**

- Cancer Diagnosis
- High Risk for Cancer
- Genetic Counseling
- Tailored Treatment and Prevention
- Discussion with Family Members
- Improved Public Health

*A lot of focus here*
Maternal/Paternal History

Figure 1. Pie Chart Showing Side of Family from which Breast Cancer Was Reported.

Figure 2. Path diagram of final modeled relationship (standardized estimates) between spiritual coping and breast cancer risk perceptions on the remaining sample of 450 participants. Note: Unstandardized estimates were not significant ($P > 0.05$). *The unstandardized values for these paths were fixed to 1 to identify the model.* In LISREL, the exogenous observed variable race was modeled as a single indicator latent construct with no measurement error and path coefficient fixed to 1.
Summary

• About 5-10% cancers hereditary
• Prevention/management implications
• DNA testing available but
  – Imperfect, expensive, complicated
• Identification and access are important challenges