

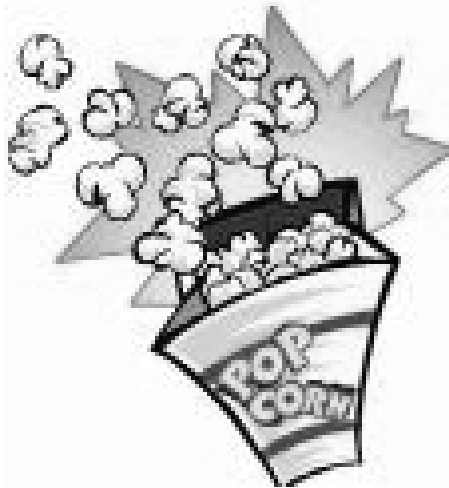
Genetic Risk Assessment and Genetic Counseling Research

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Outline for today

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

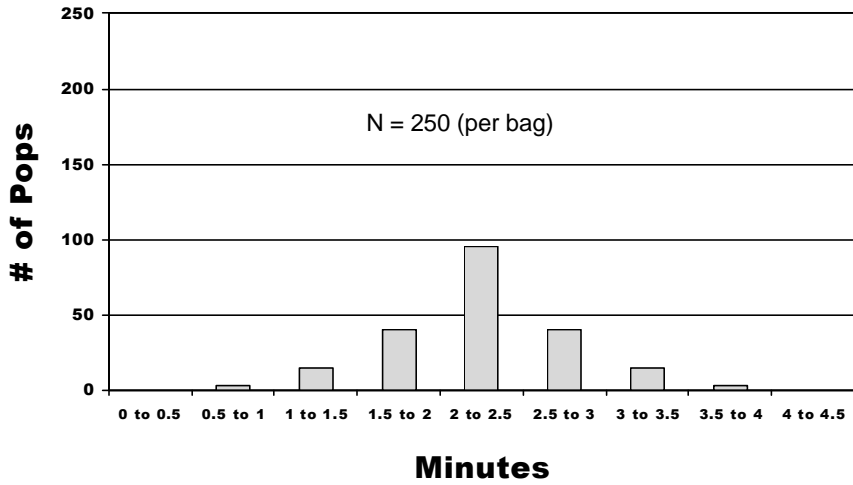
Of Risk and Popcorn



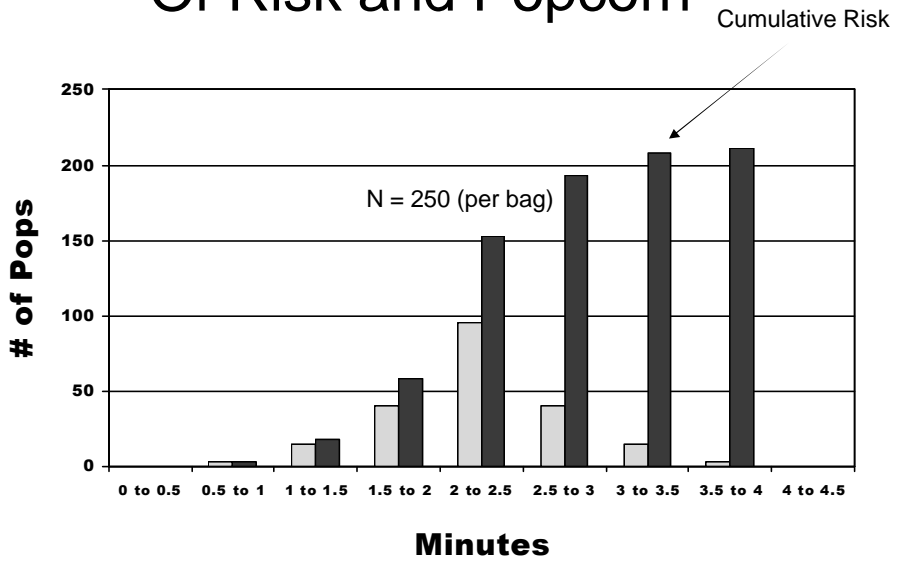
Of Risk and Popcorn



Of Risk and Popcorn



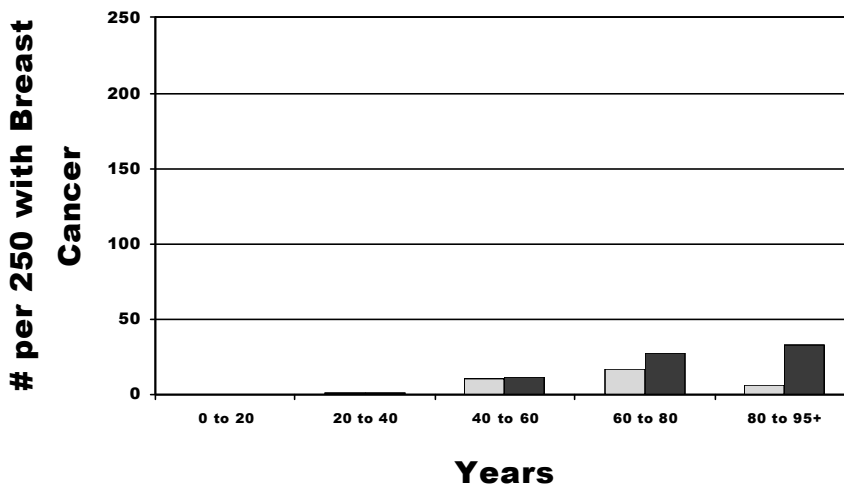
Of Risk and Popcorn



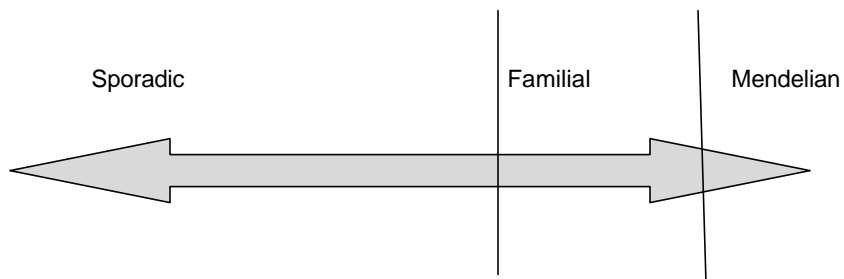
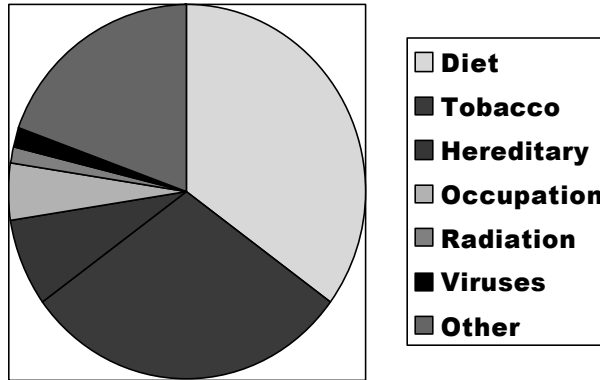
What determines if a kernel pops?

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

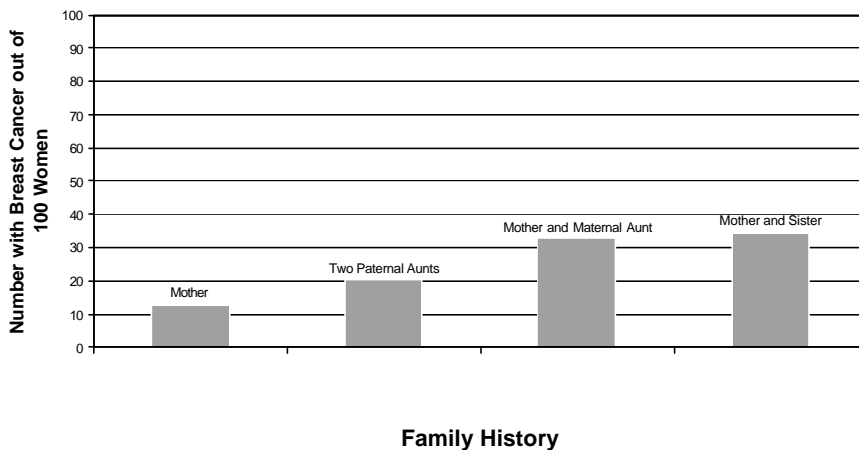
Breast Cancer Risk



What affects cancer risk?



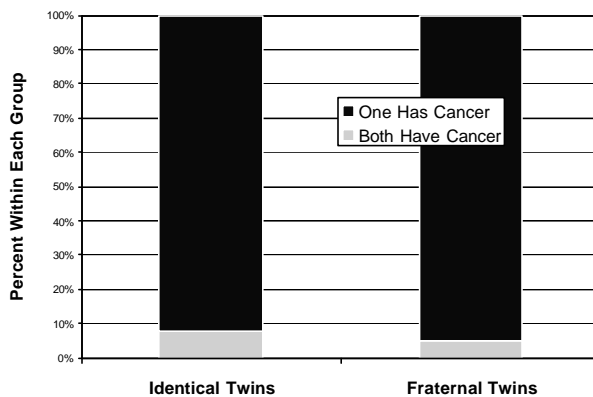
Family History and Chance of Breast Cancer



Claus, 1990

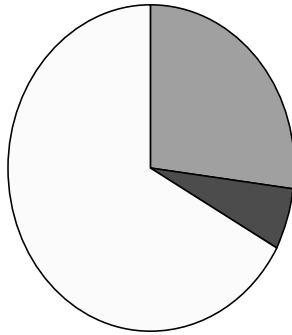
Twin Studies

Breast Cancer in Twins



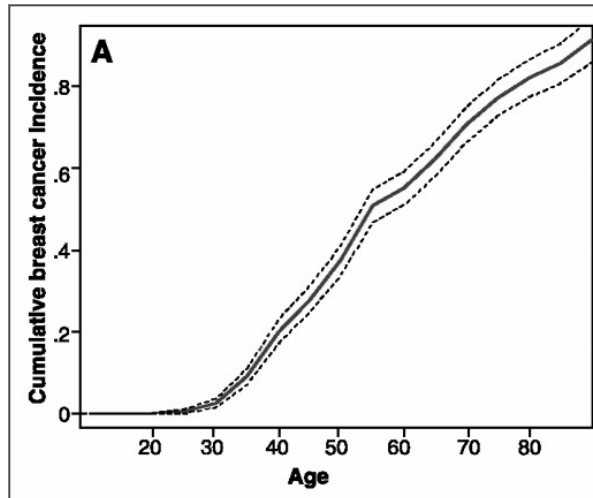
N Engl J Med 2000;343:78-85

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

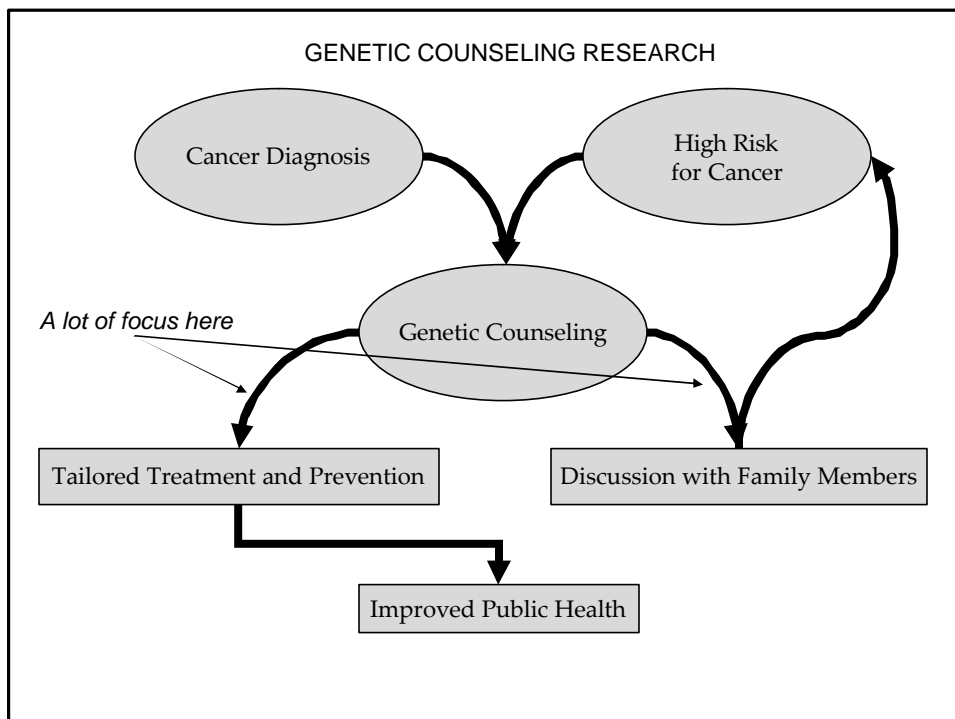
- Primary prevention
 - Medication (e.g., tamoxifen, sulindac)
 - Risk-reducing surgery
 - Lifestyle (e.g., exercise)
 - Diet (coffee??)
- Secondary prevention
 - Timing (age of initiation, interval)
 - Method (MRI)
 - Whether or not to screen at all

Cancer Control Implications

- Tertiary prevention
 - Chemotherapy
 - Extent of surgery
 - Radiation

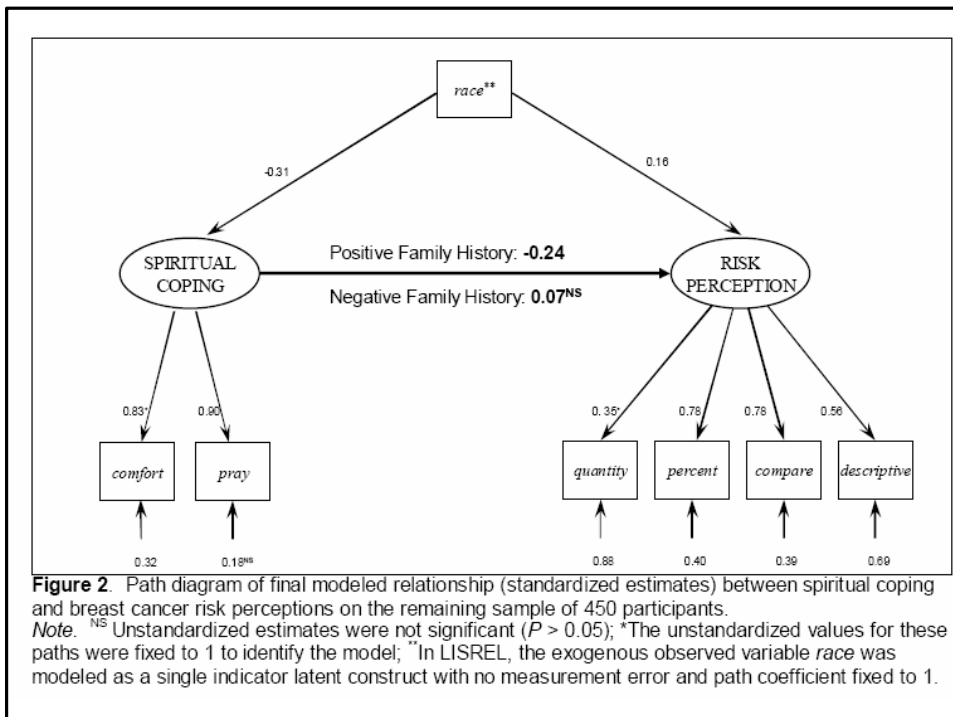
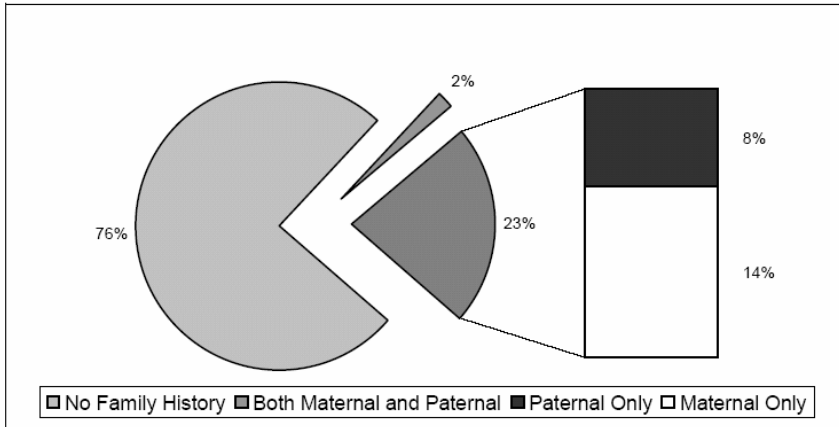
Why genetic counseling?

- 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



Maternal/Paternal History

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



Summary

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