Our Story (with a detective hunt)

- Lisa is studying how people think about health risks
  - But having to make them up to control their experimental properties
  - Studying them among people at population risk, not high risk
  - Genetic testing is interesting because people are perfectly well, yet may have a highly elevated risk they cannot see or modify
  - Today, we will share what we have learned in 16 years of asking questions about whether and why genetic testing motivates improvements in prevention behaviors

Melanoma Is Environmental AND Genetic

Melanoma: A cancer of melanocytes (skin cells that make brown pigment and produce a tan when damaged)

Cause: A combination of an individual’s genetic makeup and their environmental exposures to ultraviolet light (both UVA and UVB)

Melanoma Is a Deadly Cancer If Caught Late

Prevention & Early Detection Can Save Lives:

- Sunscreen
- Protective clothing
- Avoidance of UVR
- Self Skin-exams
- Provider Skin-exams

The survival rate for melanoma is 88% if treated early, and as low as 9% if it is detected late.

Family History of Melanoma:
- Shared family traits: Common, lower risk genes for UVR vulnerability include those for red hair, light skin/eyes

Hereditary Melanoma:
- Single gene (e.g., p16), high penetrance, not visible on the outside, very high risk
What is the percent chance that a person with red hair will get a melanoma in their lifetime?

What is the percent chance that a person with a p16 mutation will get a melanoma in their lifetime?

About 4% Lifetime Risk!

About 76% Lifetime Risk!

About 76% Lifetime Risk!

About 4% Lifetime Risk!

How likely do you think these patients are to follow our recommendations for using sunscreen?

1. Never
2. Rarely
3. Occasionally
4. Half the time
5. Fairly regularly
6. Almost all of the time
7. All of the time

Our 1st melanoma genetic test reporting study

- Provided clinical genetic test results to prior research participants, N=60
  - 1/3 had prior melanoma diagnosis
  - 2/3 unaffected (no prior diagnosis)
- Why focus on unaffected family members?

Does genetic testing provide benefits?

- In our 1st test-reporting study
  - unaffected carriers reported improvements in...
    - Thoroughness of skin self-examinations
    - Adherence to annual TBSEs
    - Daily routine sun-protection
    - Use of protective clothing
    - Reduced # of sunburns
  - These gains sustained at 2-year follow-up
- No increases in anxiety, depression, or worry
- Multiple informational & behavioral benefits
  - Understanding of risk
  - Motivation to practice photoprotection & screening

Aspinwall et al., 2008, 2013, Cancer Epidemiology, Biomarkers and Prevention
Aspinwall et al., 2014, Genetics in Medicine
A et al., 2013, Psycho-Oncology
Does genetic testing really provide benefits?

- These results cannot distinguish effects of test reporting from those of genetic education & counseling re risk management
- Need to show impact of genetic test reporting > standard recommendations based on family history

The BRIGHT Project
Behavior, Risk Information, Genealogy & Health Trial

- All study participants are unaffected members of high-risk families
  - At least three 1st- or 2nd-degree relatives with melanoma
  - They do not yet have a personal diagnosis of melanoma
- Design
  - Prospective nonexperimental control group
  - Individualized cancer risk assessment with or without genetic testing (no-test controls counseled based on family history)

The Utah BRIGHT Project
Behavior, Risk Information, Genealogy & Health Trial

Hereditary Melanoma Population

- p16 Family
  - Genetic Testing Available?
    - YES
    - NO
- p16
  - POSITIVE
  - NEGATIVE
- Non-p16 Family

Counseled Risk

- 70X
- 2X
- 30-70X

ASSESSMENTS: Baseline, post-counseling, 1 month, 1 year

Illness Coherence
"I have a clear picture or understanding of melanoma."

Understanding of melanoma risk (PAGIS)
"I feel certain that I understand the meaning of having this melanoma risk."
"I understand how I came to have this melanoma risk."

Comparative lifetime risk
"Compared to other people of your age, gender, and skin color, what is the likelihood that you will develop melanoma in your lifetime?"
Urgency & priority of managing risk

“Dealing with my risk of getting melanoma is a priority to me.”
“My risk of getting melanoma is something I need to take action on.”

Motivation to reduce sun exposure

“I am more motivated to reduce my sun exposure.”
“I protect myself more from the sun.”

Carriers increased significantly over time, but noncarriers returned to baseline. No change among no-test controls. (Taber et al., 2020, Annals of Beh Med)

Groups significantly different (F(2, 108) = 4.06, p < .02.
Carriers > no-test controls & noncarriers, p < .03. 4 items, alpha = .87

UVR dosimetry

- Similar to wristwatch
- Worn for 3 27-day periods
- Daily UVR dose @ 10-sec intervals
- Computed standard erythemal dose (SED; J/m²)
  - For individuals with fair skin, SED of 2 = slight reddening of skin

Stump et al. (2019), Genetics in Medicine

Reflectance spectroscopy measures skin color at typically exposed sites

Stump et al. (2019), Genetics in Medicine

Melanin Index Scores via reflectance

Controlling for age, skin type, seasonality, sex, propensity scores

Dorsal wrist

Facial composite

Stump et al. (2019), Genetics in Medicine
Understanding the impact of genetic testing on the management of familial cancer risk

Familial cancer risk

Genetic testing

- Greater understanding of risk & prioritization of its management
- Greater acceptance of behavioral management recommendations & motivation to perform them

Goal of personalized medicine: Improve cancer outcomes through sustained changes in prevention & early detection behaviors

Pre-existing distress
Beliefs about risk & its management
Can healthy behaviors alter genetic risk?