Empowering Families Through Genetic Testing

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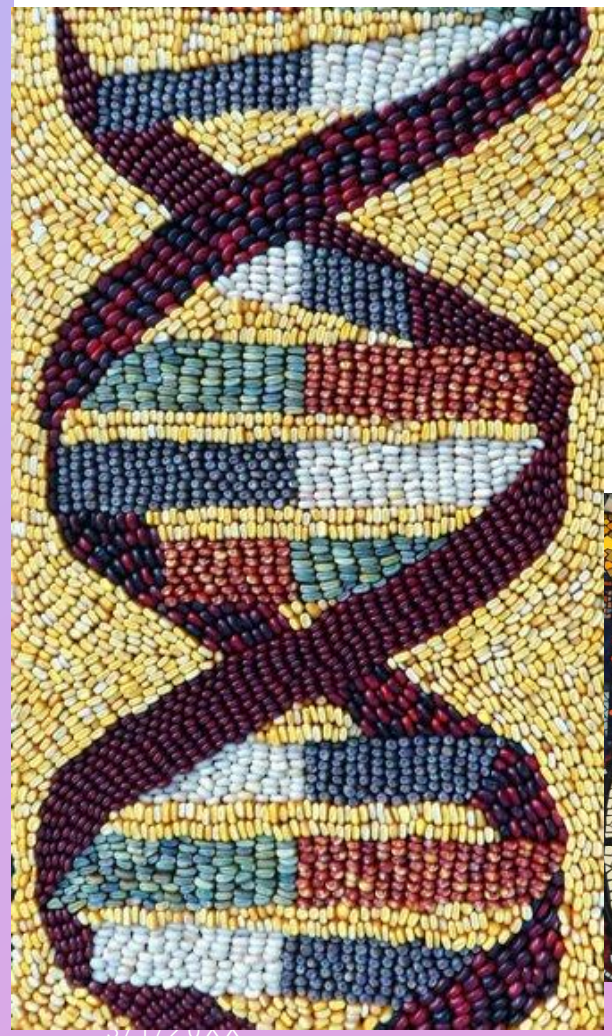
Overview

- Who needs genetic testing?
- What are we testing for?
- How is genetic testing done?
- When will results be back?
- What does it all mean?

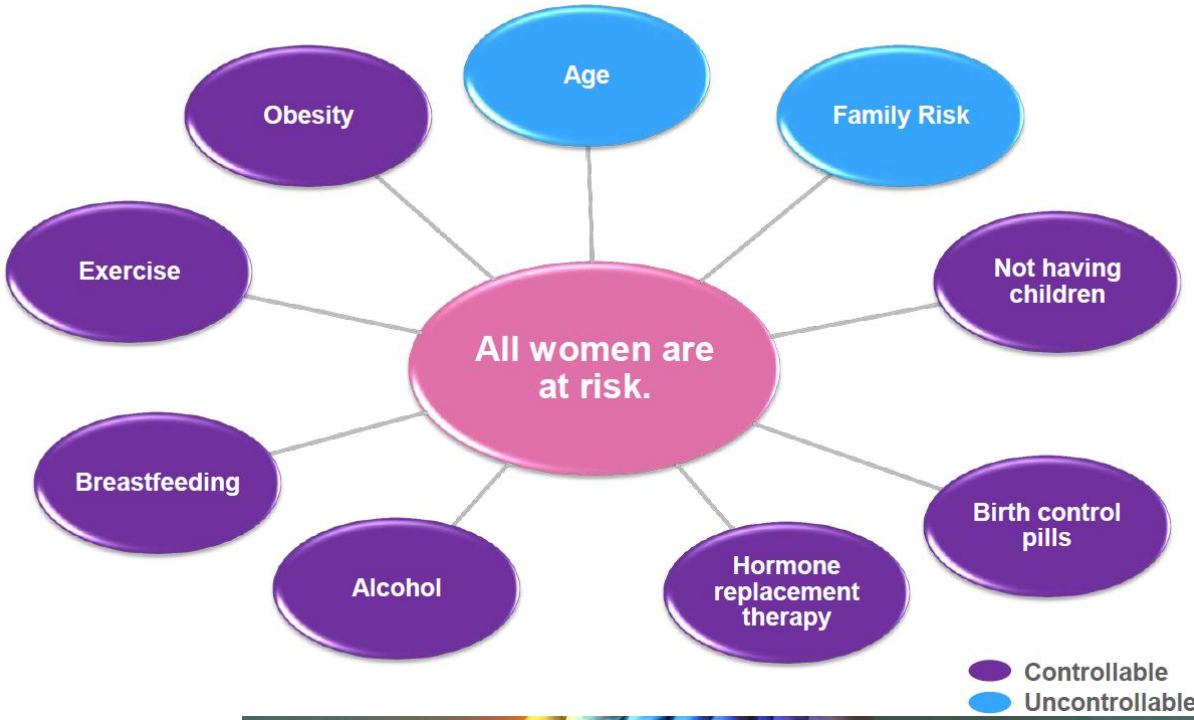


Introduction

Approximately 5 to 10% of breast cancers are attributed to inherited factors.

We are all a mixture of genes and our environment.

All of us have something genetic!



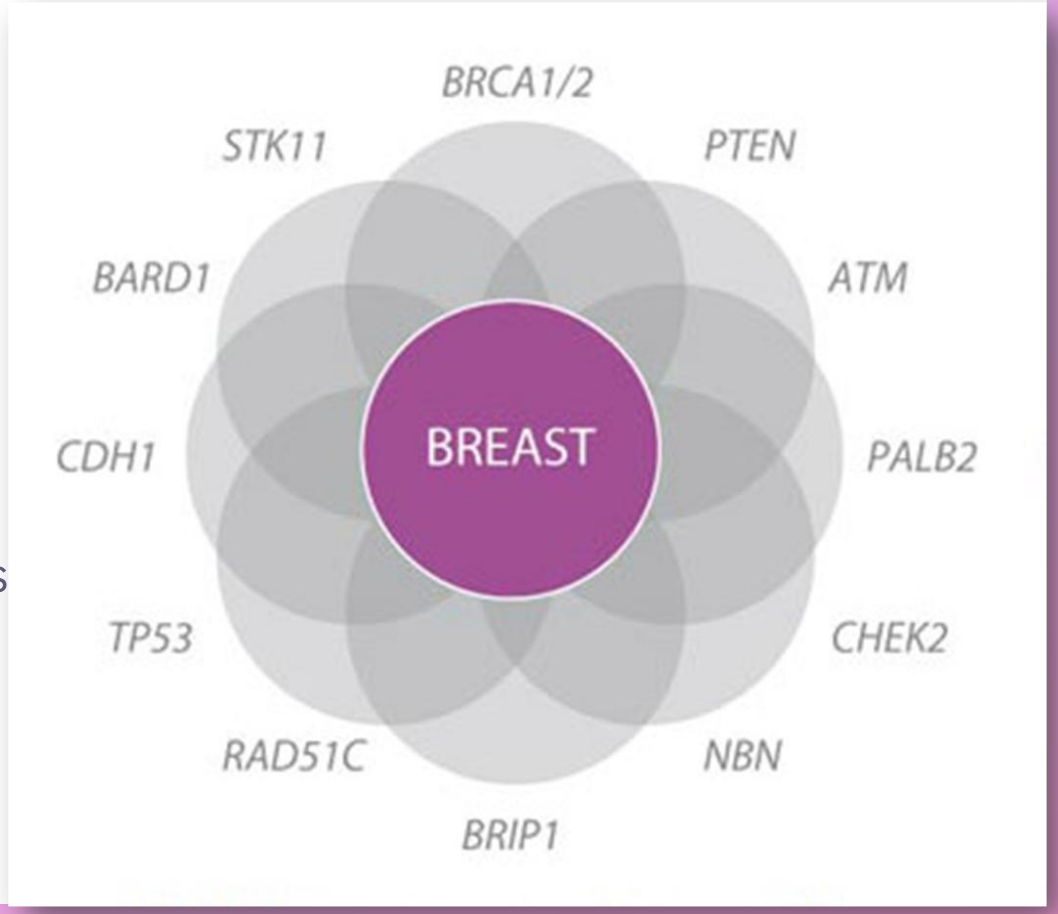
Who Should Be Tested?

- Breast cancer diagnosed < age 50 years old
- Multiple primary cancers
- Family history of cancer

What are we testing for in Genetics?

Cancer Susceptibility Genes

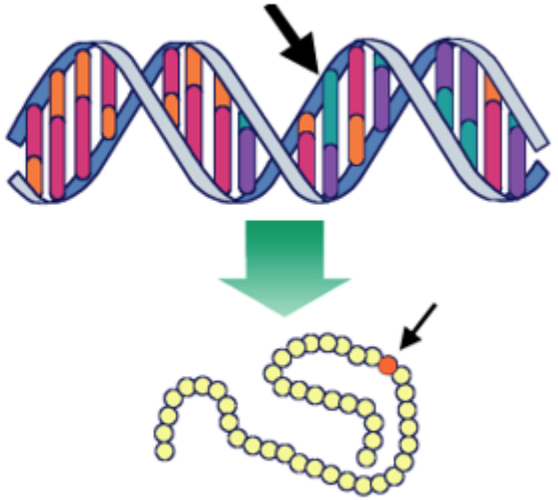
- *BRCA1* and *BRCA2*
- Lower Penetrance Breast Cancer Genes
  - *CHEK2*
  - *ATM*
  - *PALB2*
  - *NBN*
- Other Hereditary Cancer Predisposition Genes
  - *TP53*
  - *PTEN*
  - *STK11*
- Undiscovered Genes



How is genetic testing preformed?

Functional protein

Nonfunctional or missing protein



Functional protein

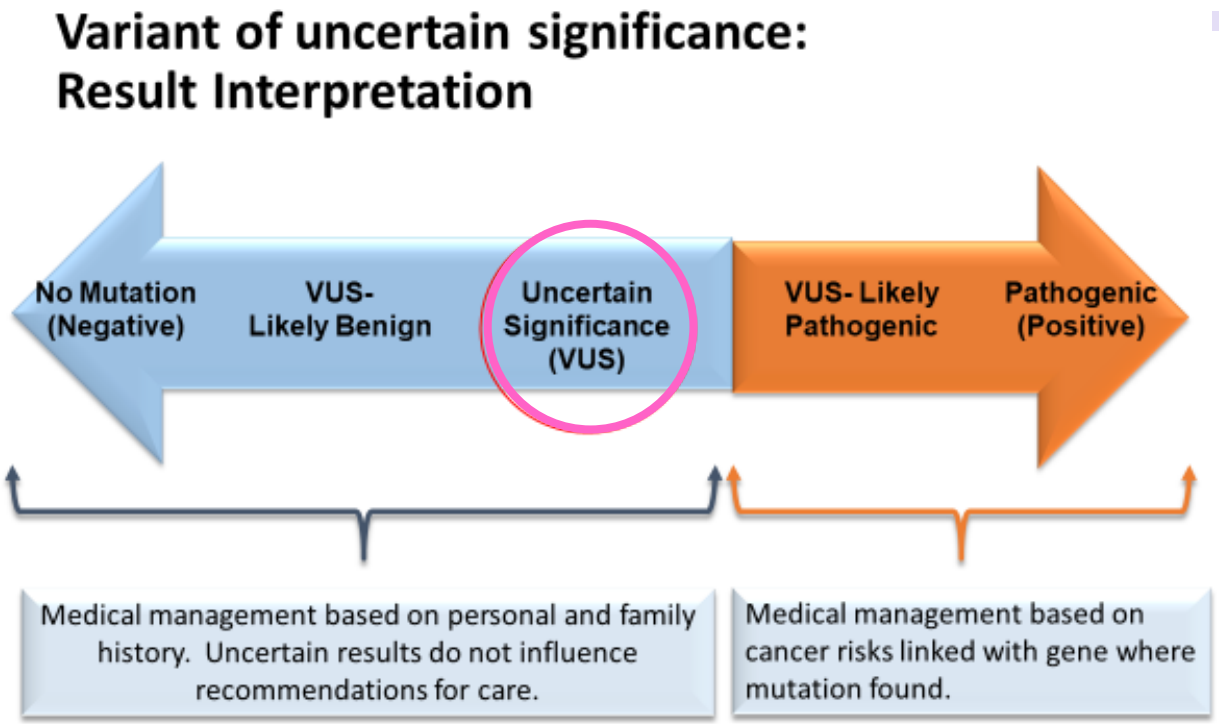
Insurance Concerns

- Coverage?? Yes- for guidelines-based diagnostic genetic testing at an in-network laboratory.
  +/- Genetic screening = testing unaffected individuals
- Insurance Discrimination??
  - GINA
    - **'Genetic Information Nondiscrimination Act'**
    - NOT for long term care, disability or life insurance

When do test results come back?

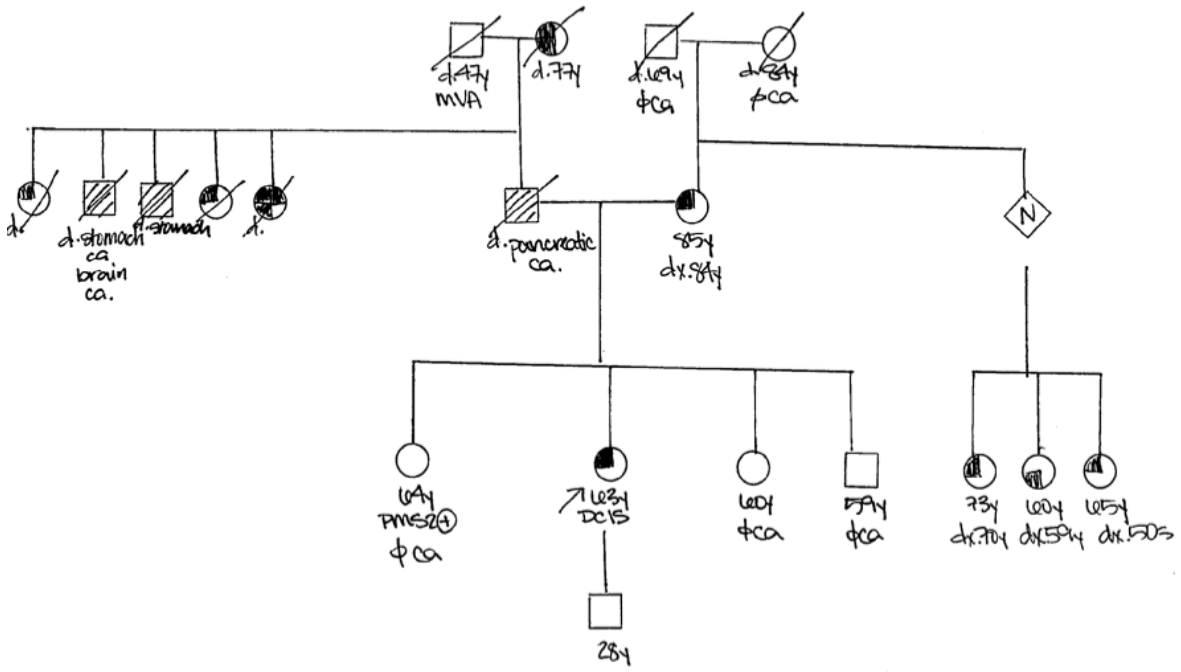
- Typically 3 to 4 weeks,
- Some cancer genetic testing is done *"STAT"* which returns in as little as 5 days.
- RNA analysis takes longer.

What does it all mean?



- 3 possible outcomes:
  - **+ Positive** "Pathogenic variant" identified
  - **-- Negative** "No variants"
  - **? Uncertain** results → *"Variant of Uncertain Significance"* meaning there is insufficient data.

Cancer Genetics and the Family



- For a positive result, it is important to share this information with family members.
- Family variant testing letter.

Very empowering!

Impact of Genetic Test Results on Family Management

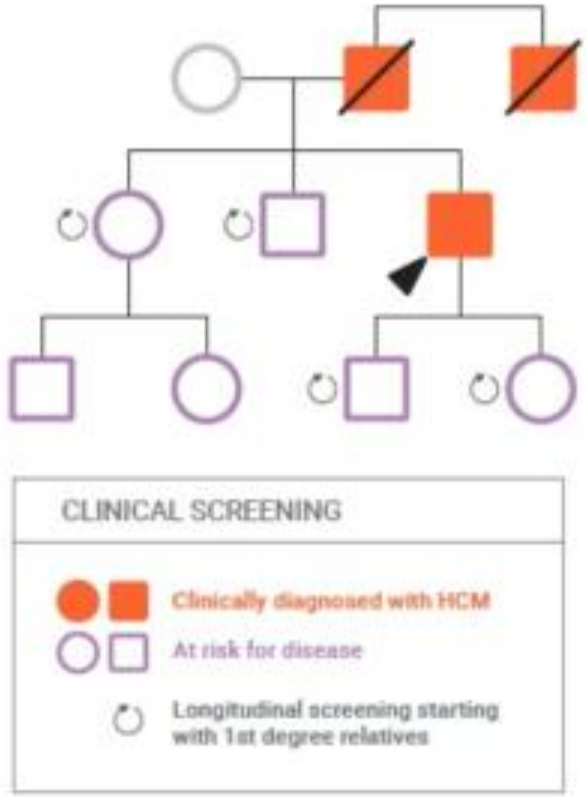
Scenario 1 : Negative

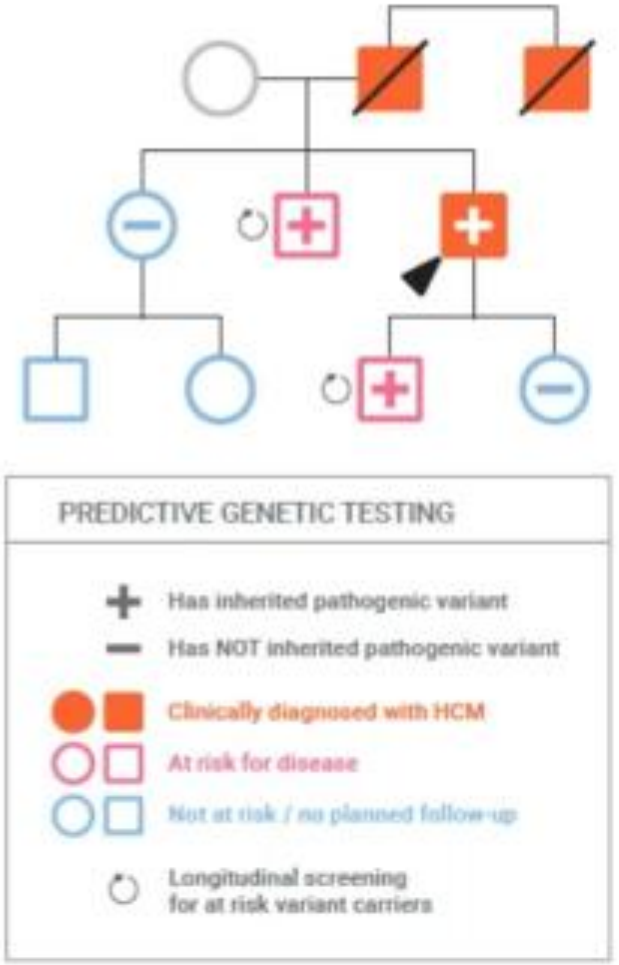
No pathogenic variant identified

Scenario 2: Positive

Pathogenic variant identified

What about Uncertain data?





Kontorovich AR et al. ACC Heart Fail. 2023 Feb;11(2):133–142.

Cancer Genetic Resources

- **NSGC: www.nsgc.org**
- **FORCE:**

www.facingourrisk.org/

Commission on Cancer®

A QUALITY PROGRAM of the AMERICAN COLLEGE OF SURGEONS

COC Standard 7.3

*Cancer risk assessment, genetic counseling, and testing services are provided to patients either on-site or by referral, by a qualified genetics professional.**

Summary: 5 things done at a Cancer Genetics Visit

1. Perform hereditary cancer risk assessment and counseling.
2. Coordinate genetic testing and provide information to help you understand treatment options and manage your health.
3. Recognize familial cancer risks and assist in notifying/testing for other relatives.
4. Interpret genomic test results.
5. Work with our cancer care team to provide interdisciplinary care for persons and families affected by breast cancer.

Thank you!

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