

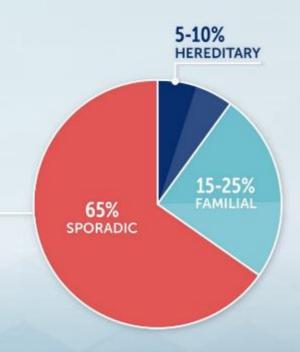
# Ruth Paul Hereditary Cancer Genetics & Prevention Service

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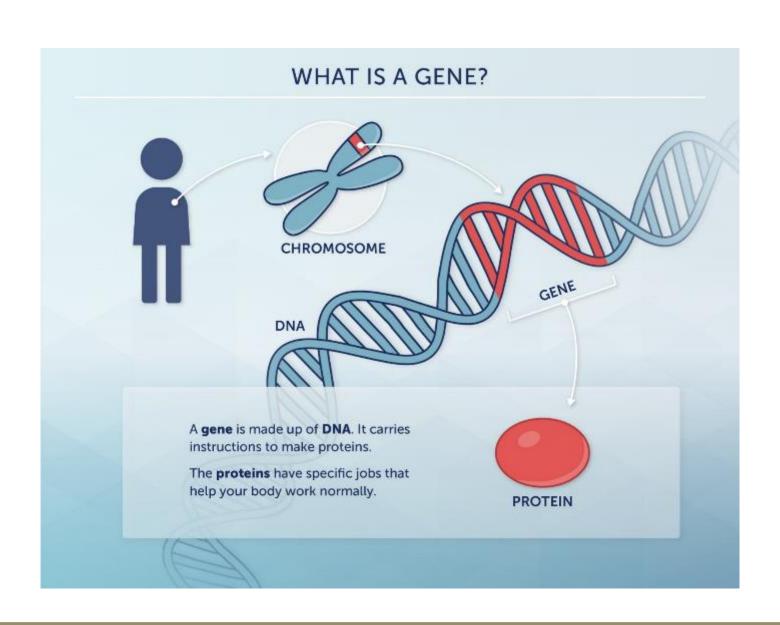
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## CANCER CAN BE HEREDITARY, FAMILIAL, OR SPORADIC

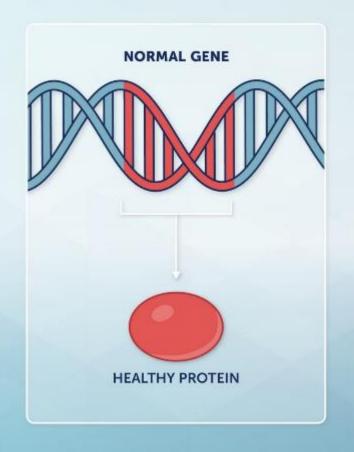


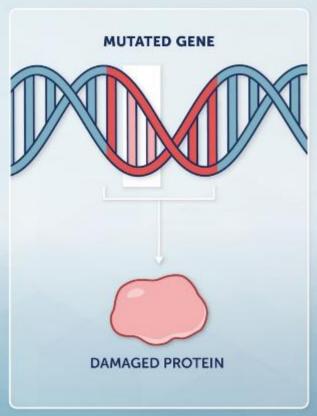


Understanding which category your cancer falls into will help guide the management of your risk better.



## WHAT HAPPENS WHEN THERE IS A GENETIC MUTATION?

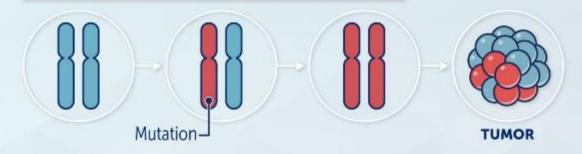




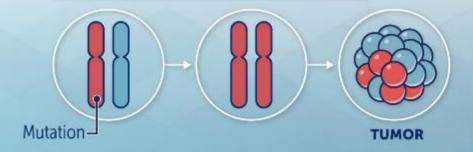
# **HOW ARE GENES INHERITED?** MOTHER **FATHER** Gene inherited from mother-Gene inherited from father

#### THE TWO-HIT HYPOTHESIS

#### INDIVIDUAL WHO DOES NOT CARRY A MUTATION



#### **INDIVIDUAL WHO CARRIES A MUTATION**



### FLAGS FOR GENETIC TESTING

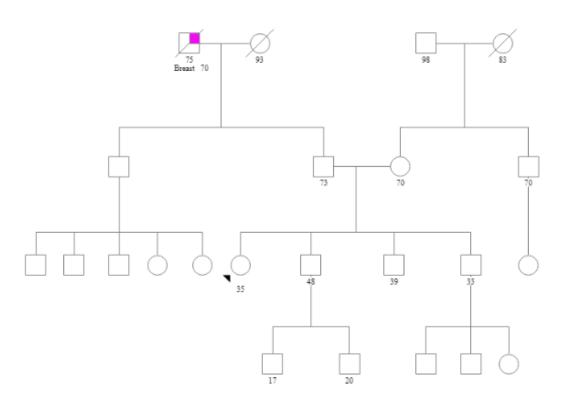
What are the common traits associated with hereditary cancer?

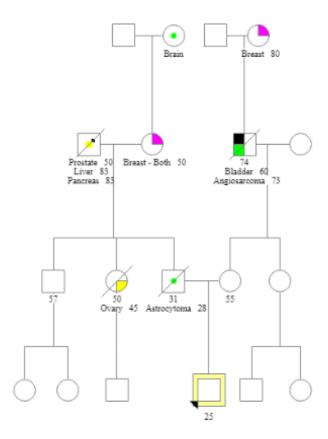
CANCER AT AN EARLY AGE
At an age younger than average

2 CERTAIN RARE CANCERS
Such as male breast cancer, sarcoma etc.

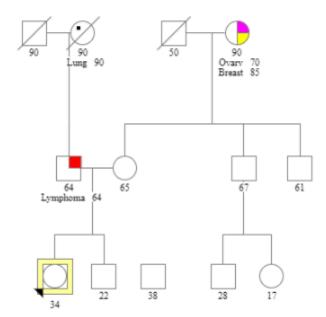
MULTIPLE CANCERS

Multiple individuals within the family may have cancer. Or one individual may have multiple cancers









Paternal Ancestry Italian, Other:

Maternal Ancestry Italian, Other:

Ashkenazi Jewish Pedigree YES

## Many testing options available

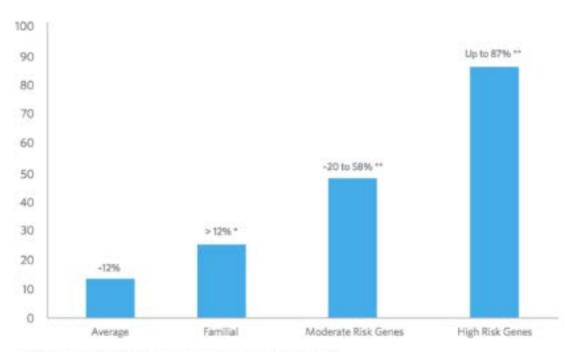
Women of Ashkenazi Jewish ancestry and a family member diagnosed with breast or ovarian cancer are covered for testing under the Affordable Care Act with no cost sharing.

Most women have no out of pocket expense

Insurance coverage is actually very good for family histories of breast and ovarian cancer

We have resources to help determine what the best test for you is

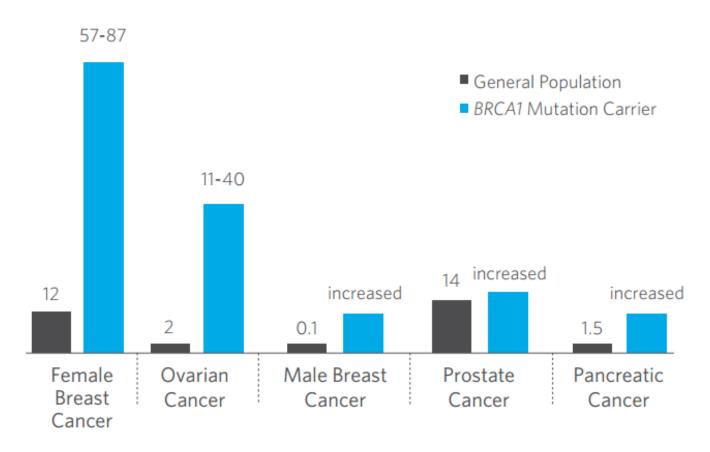
# Not all genes are created equal: High vs. Moderate Penetrant Genes



<sup>\*</sup> Specific level of risk depends on the family history of breast cancer

<sup>\*\*</sup> Specific level of risk depends on the gene involved

#### BRCA1 MUTATION LIFETIME CANCER RISKS (%)\*



## Just because your results are negative....

Doesn't mean you don't have an increased risk for developing breast or other cancers!

Personalized models and family history help us target options

ID:

Woman's age is 35 years.

Age at menarche was 13 years.

Age at first birth was 27 years.

Person is premenopausal.

Height is 5 ft 3 in.

Weight is 10 st 5 lb.

Woman has never used HRT.

Risk after 10 years is 2.4%.

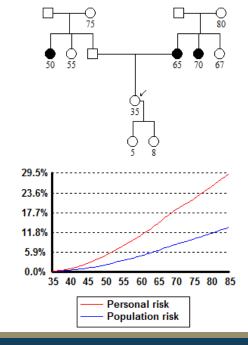
10 year population risk is 1%.

Lifetime risk is 29.1%.

Lifetime population risk is 13.2%.

Probability of a BRCA1 gene is 0.21%.

Probability of a BRCA2 gene is 0.66%.



## GINA

#### Genetic Infomation Non-Discrimination Act



GINA protects most patients from discrimination with health insurance or an employer. Active duty military personnel are an exception.



However, it does not protect a patient from discrimination with life insurance or disability.

## Common Misconceptions

- Paternal family history of cancer doesn't matter
  - Mutations can be inherited from the mother OR the father
- There is nothing to do but wait and see
  - We have ways of preventing cancer, or at the very least, detecting it an early, treatable stage
- Testing is expensive
  - Most people pay \$0 out of pocket.
- •I can lose my health insurance
  - GINA is an excellent protection for health insurance

## How to find a genetic counselor

https://www.FindAGeneticCounselor.com/

Please don't hesitate to contact me if I can be of help!