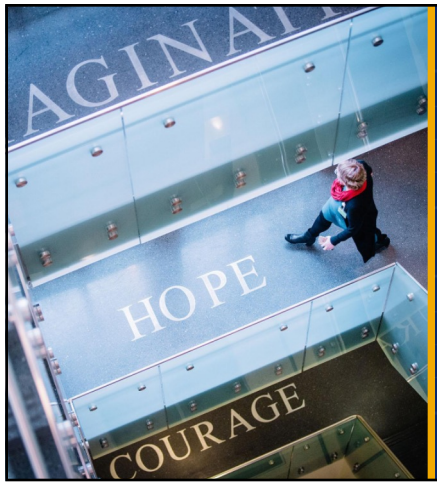

NAVIGATING RISK: GENETIC COUNSELING & HIGH-RISK SCREENING

Brittany Adams, MMSc, CGC
Licensed, Certified Genetic Counselor
Winship Cancer Institute at Emory Johns Creek Hospital

Breast Cancer Awareness Month Symposium, 2025
October 11, 2025

EMORY WINSHIP CANCER INSTITUTE
NCI Designated Comprehensive Cancer Center

1

CONFLICTS OF INTEREST

None

2

LEARNING OBJECTIVES

Define the role of genetic counseling in the evaluation and management of patients at risk for hereditary cancer syndromes

Identify appropriate candidates for genetic testing based on personal and family history criteria

Describe recommended high-risk screening strategies and how they integrate into patient care following genetic test results

3

THE ROLE OF A GENETIC COUNSELOR

Genetic counselors

- Master's degree in genetic counseling
- Certified by the American Board of Genetic Counseling (ABGC) or the American Board of Medical Genetics (ABMG)

Other healthcare providers with specialized training in genetics

- MD geneticists
- Genetic nurse clinicians
- Other genetic sub-specialists

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WHAT HAPPENS DURING A GENETIC COUNSELING VISIT

An initial genetic counseling visit includes:

- Review of personal and family medical histories
- Assessment of risk for hereditary cancer
- Discussion of cancer biology and genetics
- Discussion of genetic testing options and/or referrals for additional evaluation if appropriate
- Discussion of implications of testing for the patient and their family
- Review insurance concerns

The initial genetic counseling typically lasts 1 to 1½ hours

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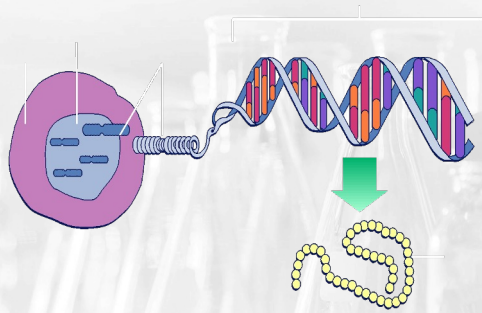
DNA, GENES, AND GENETIC TESTING



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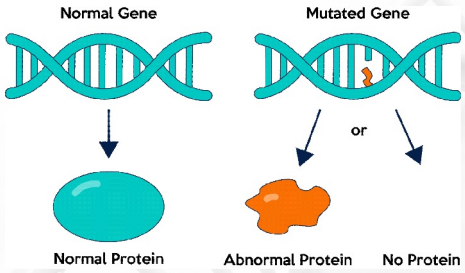
DNA, GENES, AND GENETIC TESTING



- A type of medical test that identifies changes in chromosomes, genes, or proteins.
- The results of a genetic test:
  - can confirm or rule out a suspected genetic condition
  - or help determine a person's chance of developing or passing on a genetic disorder

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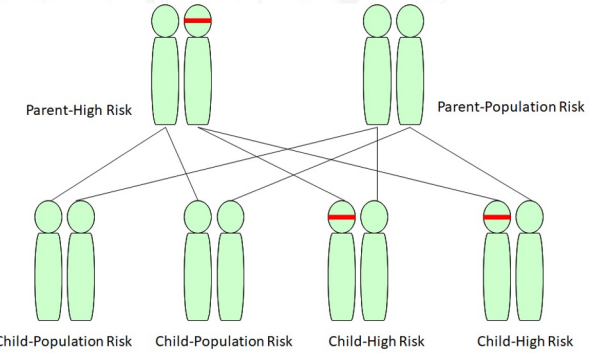
AUTOSOMAL DOMINANT INHERITANCE



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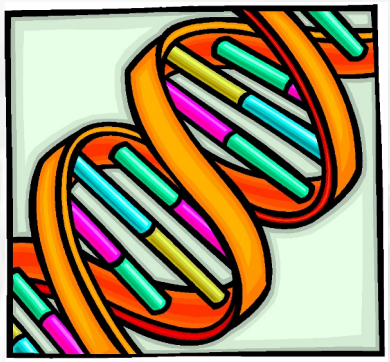
ALL CANCER IS GENETIC, BUT NOT ALL CANCER IS HEREDITARY



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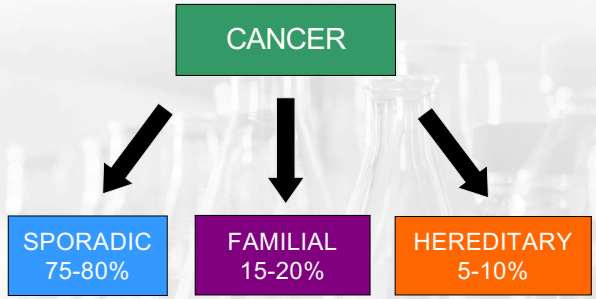
WHEN IS CANCER HEREDITARY?



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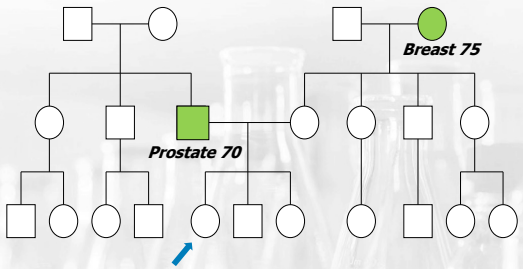

SPORADIC CANCER

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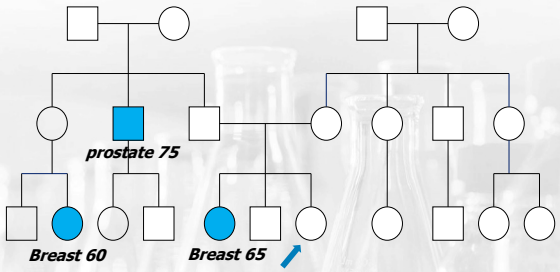

FAMILIAL CANCER

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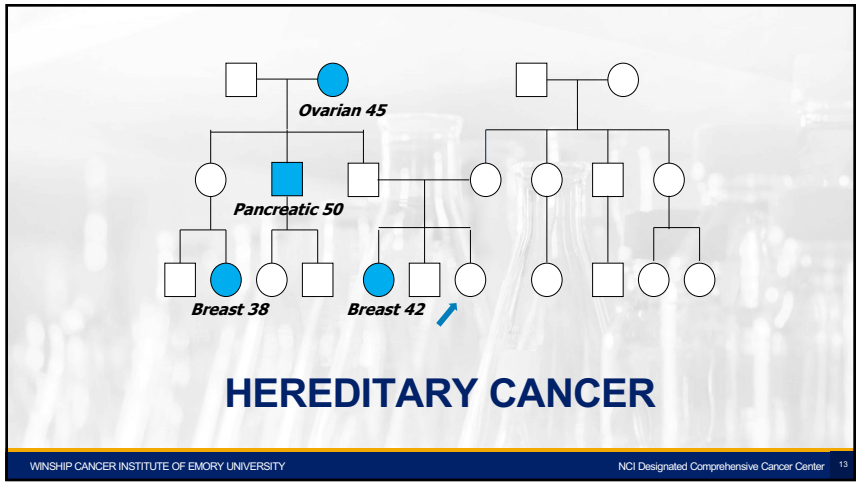

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RISK FACTORS FOR HEREDITARY BREAST CANCER

- **Early onset cancer diagnosis**
  - Diagnosed at age 50 or younger
- **Multiple cancer primaries in the same individual**
  - Bilateral breast cancer, breast and ovarian cancer
- **Same or related types of cancer in 2+ close on the same side of the family**
  - Parent, sibling, child, aunt/uncle
  - Breast/ovarian/pancreatic/prostate cancer can be "related"
- **Multiple generations of cancer within a family**
  - Grandparents, children, and their grandchildren with cancer
- **Rare types of cancer**
  - Male breast cancer, ovarian cancer, pancreatic cancer
- **Ashkenazi Jewish ancestry**
- **Anyone who has a close family member who meets the above criteria**

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NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®)

Genetic/Familial High-Risk Assessment:

Breast, Ovarian, Pancreatic, and Prostate

Testing is clinically indicated in the following scenarios:

- See General Testing Criteria on CRIT-1.
- Personal history of breast cancer with specific features:
  - ≤50 y
  - Any age:
    - ◊ Treatment indications
      - To aid in systemic treatment decisions using PARP inhibitors for breast cancer in the metastatic setting[k,l] (See NCCN Guidelines for Breast Cancer)
      - To aid in adjuvant treatment decisions with olaparib for high-risk,[m] HER2-negative breast cancer[l]
    - ◊ Pathology/histology
      - Triple-negative breast cancer
      - Multiple primary breast cancers (synchronous or metachronous)[k]
      - Lobular breast cancer with personal or family history of diffuse gastric cancer (See NCCN Guidelines for Genetic/Familial High-Risk Assessment: Colorectal, Endometrial, and Gastric)
    - ◊ Male breast cancer
    - ◊ Ancestry: Ashkenazi Jewish
  - Any age (continued):
    - ◊ Family history[o]
      - ≥1 close blood relative[p] with ANY:
        - breast cancer at age ≤50 y
        - male breast cancer
        - ovarian cancer
        - pancreatic cancer
        - prostate cancer with metastatic,[q] or high- or very-high-risk group (Initial Risk Stratification and Staging Workup in NCCN Guidelines for Prostate Cancer)
      - ≥3 diagnoses of breast and/or prostate cancer (any grade) on the same side of the family including the patient with breast cancer
- Family history criteria: unaffected; or affected but does not meet above criteria
  - Individual with a first- or second-degree blood relative meeting any of the criteria listed above (except unaffected individuals whose relatives meet criteria only for systemic therapy decision-making).[r]
  - Individuals who have a probability >5% of a *BRCA1/2* P/LP variant based on prior probability models (eg, Tyrer-Cuzick, BRCAPro, CanRisk).[s]

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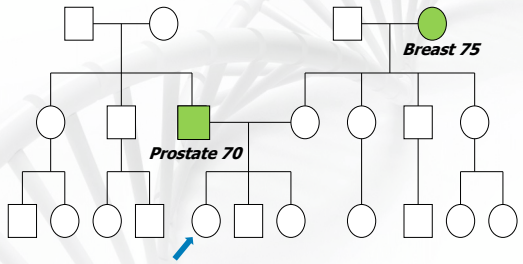
DOES THIS FAMILY MEET GENETIC TESTING CRITERIA?



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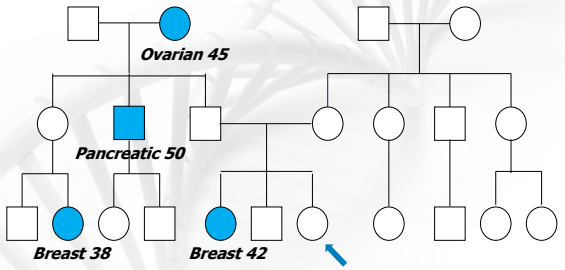
DOES THIS FAMILY MEET GENETIC TESTING CRITERIA?



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Breast/Ovarian cancer risk genes

	GENE (S)	ASSOCIATED CANCERS
High-risk genes 4-5 fold cancer risk	*BRCA1 / BRCA2*	Breast, ovarian, pancreas, prostate, fallopian
	TP53	Breast, sarcoma, brain, adrenocortical, and more
	PTEN	Breast, thyroid, uterine, and non-cancer findings
	CDH1	Breast and stomach
	STK11	Breast, colon, stomach, specific polyps, and more
Moderate-risk genes 2-3 fold cancer risk	*ATM*	Breast, ovarian, prostate and pancreatic
	CHEK2	Breast, prostate, thyroid, and uterine
	PALB2	Breast, ovarian, and pancreatic
Newer genes 1-2 fold cancer risk	*BARD1*	Breast and ovarian
	BRIP1	Breast and ovarian
	FANCC	Breast
	NBN	Breast and ovarian
	RAD51C, RAD51D	Breast and ovarian

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HIGH RISK SCREENING VS MODERATE RISK SCREENING

BRCA1/2
Lifetime risk of breast cancer: >60%

Age 18: Breast awareness

Age 25: Clinical breast exam, every 6-12 months

Age 25-29: Annual breast MRI

Age 30-75: Annual mammogram and breast MRI

Optional:

- Bilateral mastectomy- reduce risk of breast cancer by ≥90%

ATM
Lifetime risk of breast cancer: 20-40%

Age 30-35: Consider annual breast MRI

Age 40-75: Annual mammogram and breast MRI

*begin at age 40 or 10 years before the earliest breast cancer diagnosis

Risk reducing mastectomy not recommended

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HIGH RISK, BUT NEGATIVE GENETIC TESTING RESULT

Patient has a lifetime risk ≥20% as defined by models that include a comprehensive family history

Models are used to help assess someone's risk for breast cancer or a *BRCA* mutation

Model Examples:

- BRCAPRO, **Tyrer Cuzick**, BOADICEA/CanRisk, Breast Cancer Surveillance Consortium (BCSC) Invasive Breast Cancer Risk Calculator

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TYRER CUZICK/IBIS MODEL

More comprehensive for breast cancer risk

Uses personal information and family history information to provide a lifetime risk of breast cancer:

- Age
- Height and weight
- Breast density
- Age at first menstrual period
- Age at birth of first child
- Age at menopause
- Personal history of hormone use
- Family history of breast and ovarian cancer

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HIGH RISK BREAST CANCER SCREENING: ≥20% LIFETIME RISK

To begin when patient is identified as high risk:

- Initiate annual mammographic screening at age 40, or 10 years prior to the earliest breast cancer diagnosis in a relative—whichever comes first—but not before age 30
- Initiate annual breast MRI screening at age 40, or 10 years prior to the earliest breast cancer diagnosis in a relative—whichever comes first—but not before age 25.
- Consider risk reduction strategies
- Breast awareness

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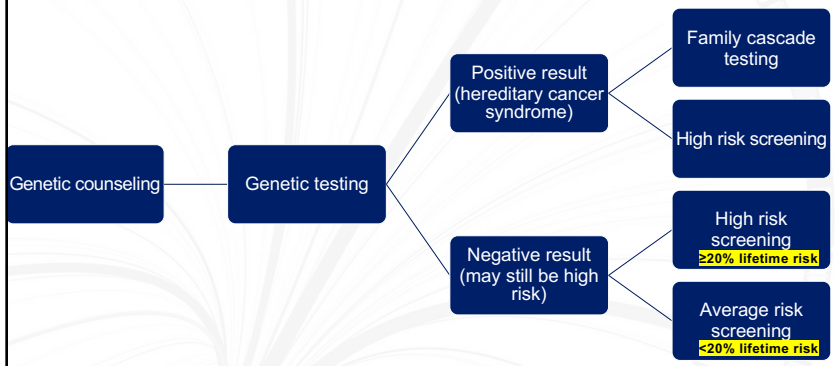
GENETIC COUNSELING CARE PATHWAY



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WINSHIP GENETIC COUNSELING PROGRAM

Genetic Counselors

- **Christine Stanislaw, MS, CGC** – Clifton
- **Suzy Cahn, MMSc, CGC** – Clifton, Decatur
- **Claire Kelso, MS, CGC** – Clifton
- **Fabienne Ehivet, MS, CGC** – St. Joseph's
- **Rachel Cohn-Lynch, MS, CGC** – Midtown
- **Tatiana Garrison Johnson, MMSc, CGC** – Midtown
- **Brittany Adams, MMSc, CGC** – Johns Creek

Appointments

- 404.778.1900
- Internal referral in EPIC: Add order – select Genetics – Cancer Genetics
- Telemedicine and in-person available

Website

- https://winshipcancer.emory.edu/genetic-counseling.html

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SOURCES

National Comprehensive Cancer Network. (2025). *Breast cancer screening and diagnosis (Version 1.2025)*. NCCN Clinical Practice Guidelines in Oncology. https://www.nccn.org/guidelines/guidelines-detail?category=1&id=1410

National Comprehensive Cancer Network. (2025). *Genetic/familial high-risk assessment: Breast, ovarian, pancreatic, and prostate (Version 1.2026)*. NCCN Clinical Practice Guidelines in Oncology. https://www.nccn.org/guidelines/guidelines-detail?category=2&id=1545

Tyrer J, Duffy SW, Cuzick J. A breast cancer prediction model incorporating familial and personal risk factors. *Stat Med.* 2004;23(7):1111–1130. doi:10.1002/sim.1668

IBIS Breast Cancer Risk Evaluation Tool. Available at: https://www.ems-trials.org/riskevaluator/

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Thank you!
Do you have any questions?

Brittany.a.adams@emoryhealthcare.org

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