



Cancer Genetic Counseling

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Objectives

At the conclusion of this presentation, participants should be able to

- **Identify individuals at risk for hereditary cancer**
- **Understand the cancer genetic counseling process**
- **Recognize aspects of informed consent**



Genetic Counseling

- **Definition**


- Genetic counseling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease.

- **Degree**

- Master's degrees in genetic counseling are offered by programs accredited by the Accreditation Council for Genetic Counseling (ACGC)

- **Certification**

- Board certified by the American Board of Genetic Counseling (ABGC)



Why pursue genetic counseling?


- ▶ Help the patient understand the “why”
- ▶ Guide treatment
- ▶ Guide future screening and management
- ▶ Identify other relatives at risk
- ▶ Reproductive risk

An Example Session





GC Session Outline

- ▶ Contracting
 - ▶ Collecting and Interpreting History
 - ▶ Risk Assessment and Counseling
 - ▶ Genetic Testing Approach
 - ▶ *Psychosocial Counseling
 - ▶ Post-Test Counseling
- 



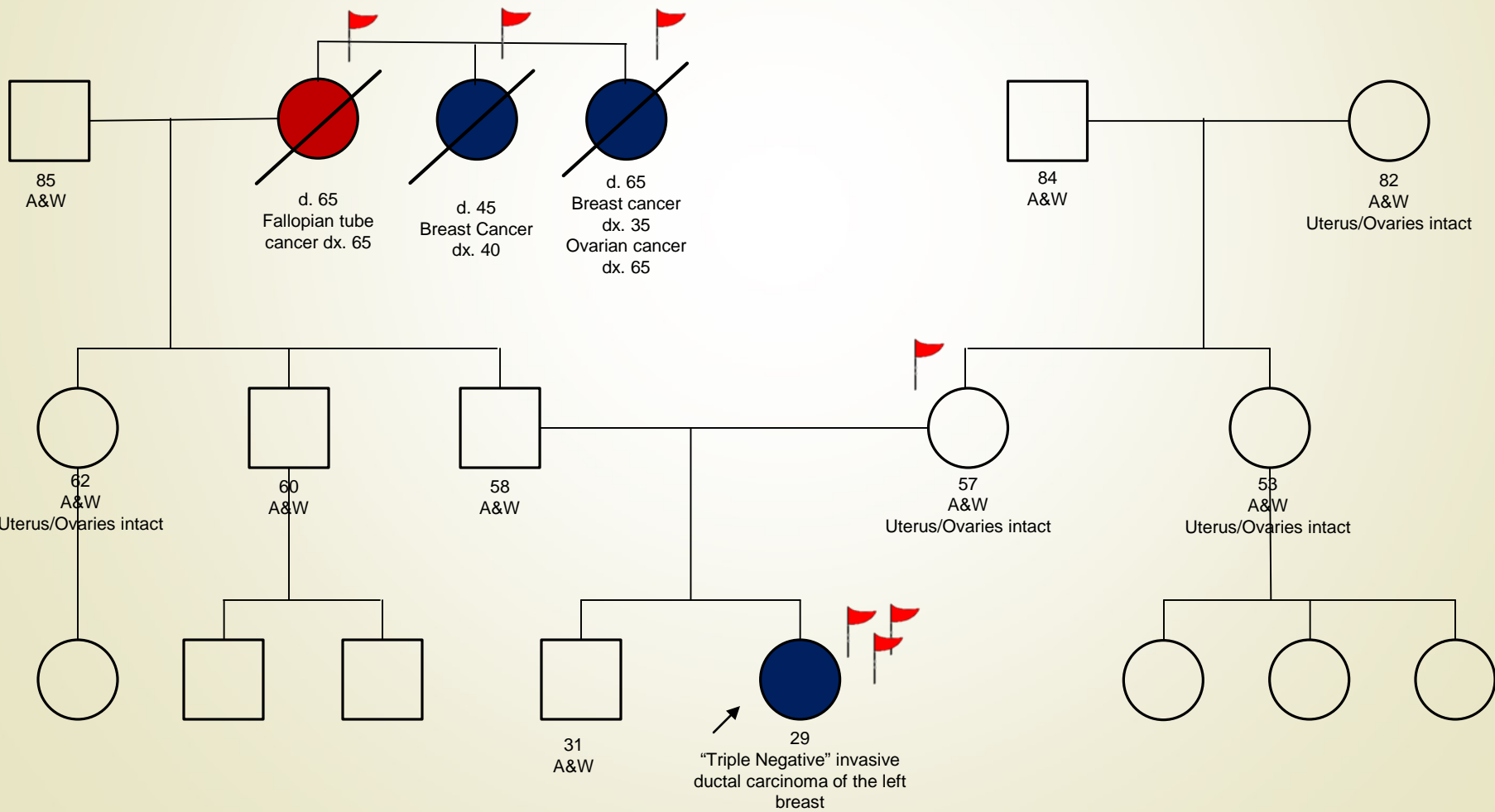
Patient A

- ▶ Case Details:
- ▶ Patient A is a 29-year old law student recently diagnosed with “triple negative” invasive ductal carcinoma of the left breast
- ▶ Referred for stat genetic counseling to discuss family history, genetic testing, and treatment options
- ▶ Scheduled for surgery in one month

Collecting a Family History: An Art and Skill

Russian

Ashkenazi Jewish



85
A&W

d. 65
Fallopian tube
cancer dx. 65

d. 45
Breast Cancer
dx. 40

d. 65
Breast cancer
dx. 35
Ovarian cancer
dx. 65

84
A&W

82
A&W
Uterus/Ovaries intact

62
A&W
Uterus/Ovaries intact

60
A&W

58
A&W

57
A&W
Uterus/Ovaries intact

53
A&W
Uterus/Ovaries intact

31
A&W

29
"Triple Negative" invasive
ductal carcinoma of the left
breast

Key Family History Information

- ▶ Who in the family has cancer?
 - ▶ How old are they now?
 - ▶ How old were they when they were diagnosed?
 - ▶ What type of cancer?
 - ▶ Primary tumor vs. metastatic disease
- ▶ Who in the family does not have cancer?
 - ▶ How old are they or how old were they when they passed away?
 - ▶ Other non-cancerous features
- ▶ Does the information seem accurate?



Hereditary Cancer Syndromes



- ▶ Several relatives with the same or related cancers
 - ▶ Breast, ovarian, pancreas, prostate,
 - ▶ Colon, uterus, small intestine, kidney
- ▶ Younger age of onset than typical
- ▶ Autosomal dominant pattern of cancer
 - ▶ Is more than one generation affected?
 - ▶ How many men vs. women?
- ▶ Presence of rare cancers
 - ▶ Ovarian cancer
 - ▶ Pheochromocytoma
 - ▶ Male breast cancer
 - ▶ Teenagers with colon cancer



Hereditary Cancer Syndromes



- ▶ Multifocal or bilateral cancer
 - ▶ Right sided colon and rectal
 - ▶ Bilateral breast cancer
- ▶ Multiple primary cancers
 - ▶ Breast and ovarian cancer
 - ▶ Parathyroid tumors and pancreatic cancer
- ▶ Non-malignant features
 - ▶ Lumps, bumps, and spots
 - ▶ Hearing loss
 - ▶ A large head size
- ▶ Absence of environmental risk factors
 - ▶ Mesothelioma without asbestos exposure



Rule of Thumb



- ▶ 3 individuals with similar or related cancers
- ▶ 2 affected generations
- ▶ 1 person diagnosed at an unusually young age (<age 50 for adult-onset cancers)

- ▶ When in doubt, refer out



Cancer Risk Assessment



- ▶ **High Risk:** strong evidence for having an inherited predisposition, pattern of cancer is highly suggestive of a particular syndrome, meets clinical diagnostic criteria or testing criteria
 - ▶ Patient A:
 - ▶ Very clear family history suggestive of an inherited predisposition
 - ▶ Patient meets criteria for testing
 - ▶ Based on age of onset we did a panel including BRCA1, BRCA2, and TP53
 - ▶ Pathogenic variant in BRCA1
 - ▶ Scheduled consult with surgeon regarding next steps

- 
- 
- ▶ **Moderate Risk/Possible Genetic Syndrome:** some features suggestive of an inherited predisposition, but may not meet criteria for a specific syndrome
 - ▶ *Should we do testing?*
 - ▶ *If so, who in the family is the best person to test?*
 - ▶ *What genes should we test?*
 - ▶ *If testing is negative: discussion includes why this is uninformative*
 - ▶ *What is the patient's personal risk for cancer?*



▶ **Low Risk/Likely Sporadic Cancers:**

- ▶ Few first- or second-degree relatives with cancer
- ▶ Cancers that are not usually associated with a hereditary syndrome
- ▶ Cancers that have occurred at typical ages
- ▶ Cancers that commonly occur in the general population
- ▶ No unusual tumors or features

- 
- 
- **Risk Models:** a tool to help predict the lifetime risk of cancer or the likelihood of having a genetic change (mutation) related to a hereditary cancer syndrome
 - Helps guide management
 - Helps understand risk
 - Important to understand limitations



Risk Model Examples

- ▶ CancerGene (BRCAPro, MMRpro, PancPRO, MelaPRO)
 - ▶ Mutation probabilities based on family history
- ▶ Myriad Tables
 - ▶ Mutation probabilities based on lab data and family history
- ▶ BOADICEA
 - ▶ Family history
- ▶ Tyrer-Cuzick (IBIS)
 - ▶ Family history
 - ▶ Hormone history
 - ▶ Ashkenazi Jewish ancestry
- ▶ Claus
 - ▶ Family history of breast cancer
- ▶ Gail
 - ▶ Hormone History
 - ▶ Breast cancer in 1st degree relatives
 - ▶ Biopsy history
 - ▶ Race

Who should have genetic testing?


- ▶ Key points:
 - ▶ Test the family member most likely to test positive
 - ▶ Dependent on family structure and communication
 - ▶ Counseling and testing unaffected individuals is an extensive conversation
 - ▶ Patients are lost to follow-up
 - ▶ Chance a patient may not receive a risk assessment

Genetic Testing Considerations


- Testing should be considered in appropriate high-risk individuals where it will impact the medical management of the tested individuals and/or their at-risk family members. It should be performed in a setting in which it can be adequately interpreted.¹
- The probability of pathogenic/likely pathogenic variant detection associated with these criteria will vary based on family structure. Individuals with unknown or limited family history/structure, such as fewer than 2 female first- or second-degree relatives having lived beyond age 45 in either lineage, may have an underestimated probability of familial pathogenic/likely pathogenic variant detection. The estimated likelihood of pathogenic/likely pathogenic variant detection may be very low in families with a large number of unaffected female relatives.


Genetic Testing Approach

- If more than one family member is affected with cancers highly associated with a particular inherited cancer susceptibility syndrome, consider testing first a family member with youngest age at diagnosis, bilateral disease, multiple primary cancers, or other cancers associated with the syndrome, or most closely related to the proband/patient. If there are no living family members with cancer that is a cardinal feature of the syndrome in question, consider testing first- or second-degree family members affected with other cancers thought to be related to the gene in question (eg, prostate or pancreas with *BRCA1/2*).
- Testing for unaffected family members when no affected member is available should be considered. Significant limitations of interpreting test results should be discussed.
- If no pathogenic/likely pathogenic variant is found, consider referral for expert genetics evaluation if not yet performed; testing for other hereditary cancer syndromes may be appropriate. For additional information on other genetic pathogenic/likely pathogenic variants associated with breast/ovarian cancer risk for which genetic testing is clinically available, see [GENE-1](#).



Components of Informed
Consent and Pretest
Education in Clinical Cancer
Genetics





Traditional Pretest Counseling for Susceptibility Testing

1. Information on the specific genetic mutation(s) or genomic variant(s) being tested, including whether the range of risk associated with the variant will affect medical care
2. Implications of positive, negative, or uncertain result
3. Possibility that test will not be informative



Traditional Pretest Counseling for Susceptibility Testing

4. Risk that children and/or other family members may have inherited genetic condition
5. Fees involved in testing and counseling; for Direct-to-Consumer (DTC) testing, whether counselor is employed by testing company
6. Psychological implications of test results (benefits and risks)




Traditional Pretest Counseling for Susceptibility Testing

7. Risks and protections against genetic discrimination by employers and insurers

8. Confidentiality issues; including DTC testing companies and policies related to privacy and data security

9. Possible use of DNA samples for future research



Traditional Pretest Counseling for Susceptibility Testing

10. Options and limitations of medical surveillance and strategies for prevention after genetic or genomic testing

11. Importance of sharing genetic and genomic test results with at-risk relatives so they may benefit from this information

12. Plans for disclosing test results and providing follow-up



Genetic Information Nondiscrimination Act

GINA & Health Insurance

- Illegal for health insurers to request, require, or use genetic information to make decisions about:
 - Your eligibility for health insurance
 - Your health insurance premium, contribution amounts, or coverage terms
- Illegal for your health insurer to:
 - Consider family history or a genetic test result a pre-existing condition
 - Ask or require that you have a genetic test
 - Use any genetic information they do have to discriminate against you, even if they did not mean to collect it



GINA & Employment

- Illegal for employers to use your genetic information in the following ways:
 - To make decisions about hiring, firing, promotion, pay, privileges or terms
 - To limit, segregate, classify, or otherwise mistreat an employee
- Illegal for an employer to request, require, or purchase the genetic information of a potential or current employee, or his or her family members.



GINA & Caveats

- ▶ Tricare and the VA
- ▶ Indian Health Services
- ▶ Federal Employee Health Benefits Plans
- ▶ Life Insurance
- ▶ Long-Term Care Insurance
- ▶ Disability Insurance

Cancer development

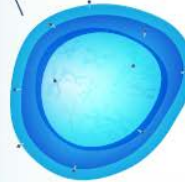
Germline DNA



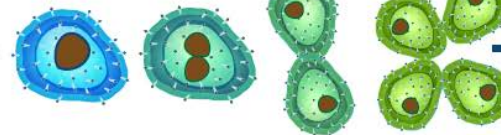
Tumor DNA



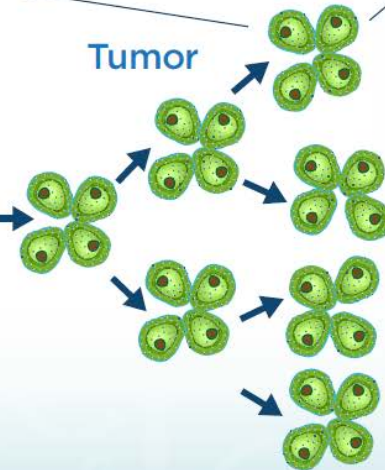
Normal cell



DNA mutation(s)



Tumor



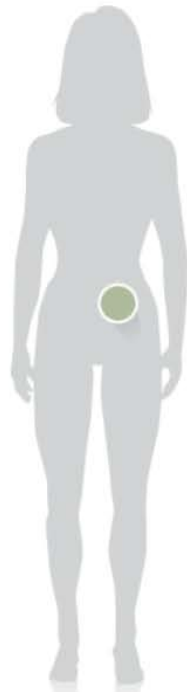


Somatic mutation

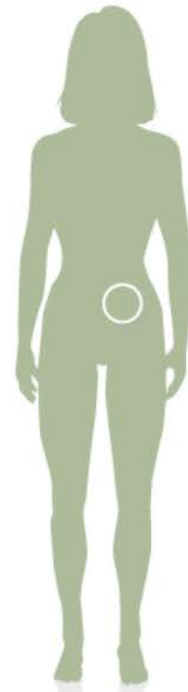
Germline mutation

 BRCA mutated cells

 BRCA intact cells



 { **Tumor
BRCA
Positive** } 





Pretest Education for Somatic Mutation Profiling with Potential for Incidental Germline Findings

1. Discussion of possibility of discovering information relevant to inherited risk and range of possible germline risks that may be identified; explain if there will be mandatory search for secondary findings and provide option to decline
2. Describe criteria that will be used to identify returnable germline variants



Pretest Education for Somatic Mutation Profiling with Potential for Incidental Germline Findings

3. Emphasize the purpose of testing is NOT to identify germline risk; dedicated testing directed by personal or family history is available
4. Discuss how incidental findings may be relevant to family members
5. Consider identifying a surrogate who could receive incidental results on behalf of patient in the event patient has died or is unable to receive results




Post-test Counseling






Result Disclosure and Interpretation

- ▶ Negative, Positive, Variant of Uncertain Significance (VUS)
 - ▶ Clarify the result in terms of personal and family history
 - ▶ True negative vs. uninformative negative
- 



Cancer Risk Assessment

- ▶ Based on genetic test result, risk assessment models, or empiric data
 - ▶ Include basic risk assessments for family members when available and applicable
- 




Cancer Screening Recommendations

- ▶ Will be addressed regardless of result
 - ▶ Individuals with negative test result but increased cancer risk will receive individual screening recommendations
- ▶ Discuss general guidelines (ACS, NCCN, USPSTF) for the early detection of cancer and preventative measures



Appropriate Referrals

- ▶ Long term follow up programs
 - ▶ Clinicians/specialty clinics for subsequent medical management
- 




Resources



- ▶ Pre-test and post-test genetic counseling medical record documentation provided to patient
 - ▶ Family and physician letters
- ▶ Specialized resources:
 - ▶ Provide template or custom letter to family to explain testing results and implications to other family members
 - ▶ Psychosocial support
 - ▶ Facing Our Risk of Cancer Empowered (FORCE)
 - ▶ Bright Pink
 - ▶ Alive and Kickin'
 - ▶ Children's Tumor Foundation



Additional Testing Options

- ▶ Other hereditary cancer syndromes indicated by personal or family history
 - ▶ Clinical Trials and future discoveries/developments in the field of cancer genetics
- 

Find a genetic counselor

➤ Findageneticcounselor.com



The screenshot shows the top section of the website. On the left is an illustration of a woman with dark hair, wearing a white lab coat, holding a clipboard and a pen. To the right of the illustration, the text reads "Brought to you by National Society of Genetic Counselors" with the organization's logo. Below this is the main heading "Genetic Counselors" in large blue font, followed by the subtitle "Personalized Care For Your Genetic Health" in orange. A red navigation bar contains the text "BACK TO ABOUTGENETICCOUNSELORS.COM".

Find a Genetic Counselor

This directory has been developed to assist physicians, patients and genetic counselors in accessing genetic counseling services.

DISCLAIMER
SEARCH TIPS
MEET BY PHONE
MEET IN PERSON
ABOUT GENETIC COUNSELORS
PATIENT RESOURCE SITE
MEMBER DIRECTORY

FIND A GENETIC COUNSELOR

The Find a Genetic Counselor directory offers access to over 3,300 genetic counselors (US and Canada). Check with your insurance company to verify coverage of genetic counseling, testing and authorized providers. For more information, visit AboutGeneticCounselors.com.

To start your search, first tell us **how you would prefer to meet with a genetic counselor:**



In Person



By Phone

Additional searches:

- If you are a student, healthcare provider or other individual interested in speaking with a genetic counselor, [click here](#).
- NSGC members are offered an expanded directory that contains additional information for use in searching for colleagues. Access the [NSGC Member Directory](#).



QUESTIONS?

