Navigating Patients to Cancer Genetics Services

Objectives

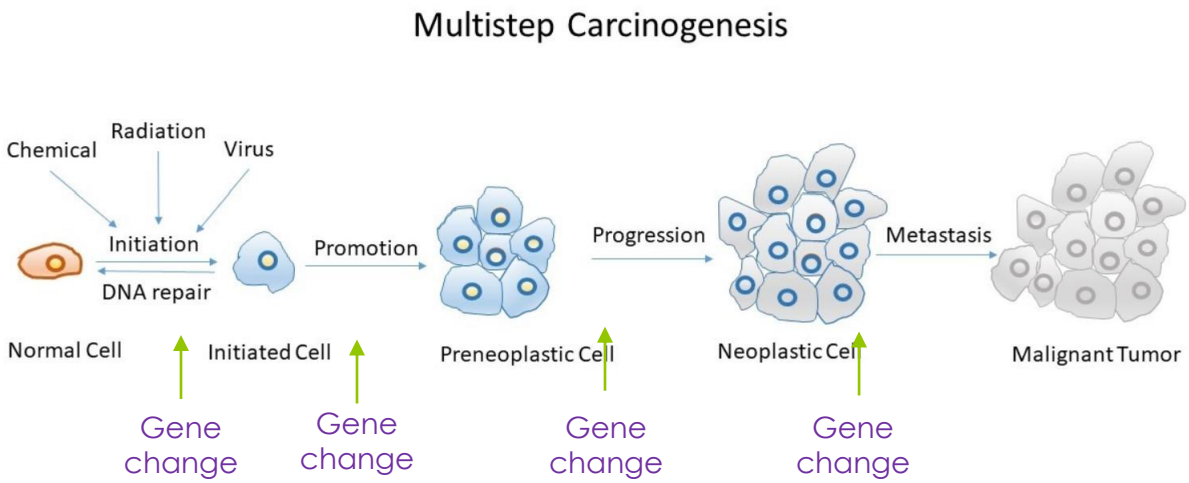
At the end of this presentation, attendees will be able to:

1. Recognize how a person's family history and personal medical history can be used to see if they are at risk of hereditary breast cancer.

- 2. Explain why it is important to identify people at risk.
- 3. Determine to how navigate those at risk to cancer genetics services.
- 4. Address patients' questions, concerns, and misconceptions about genetic counseling and possible genetic testing for cancer.

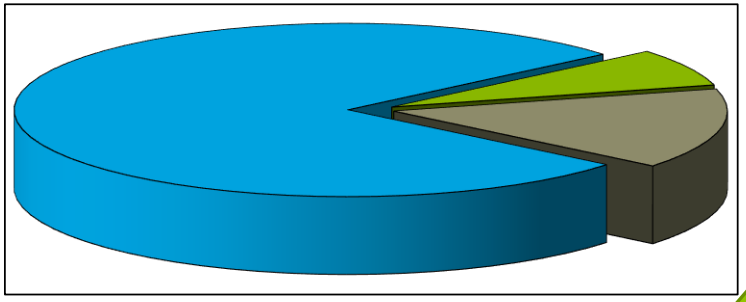
Background- Cancer Genetics

- All cancers are genetic
  - All cancers develop due to changes in our genes, that when working normally, promote normal cell growth
- **However,** only a small proportion of cancers are inherited (run in families)



What proportion of breast cancer is hereditary?

All Breast Cancers



5-10% hereditary

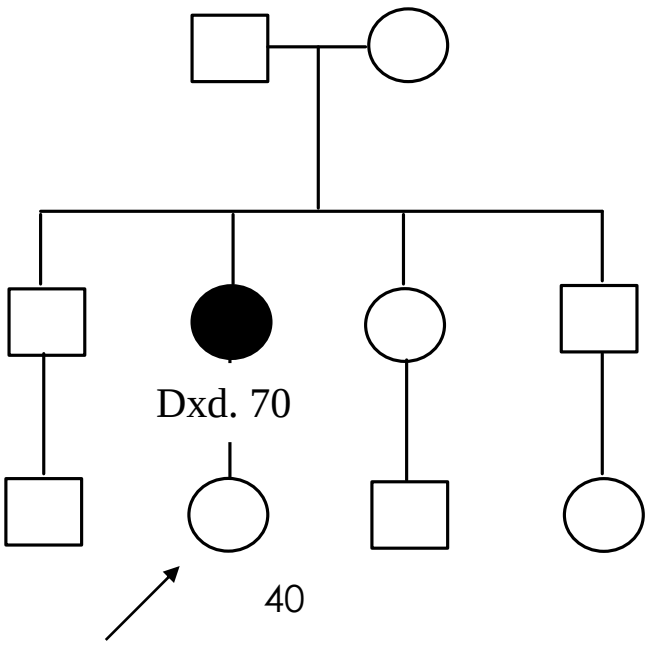
10-15% familial

75-85% sporadic

What do these look like in family histories?

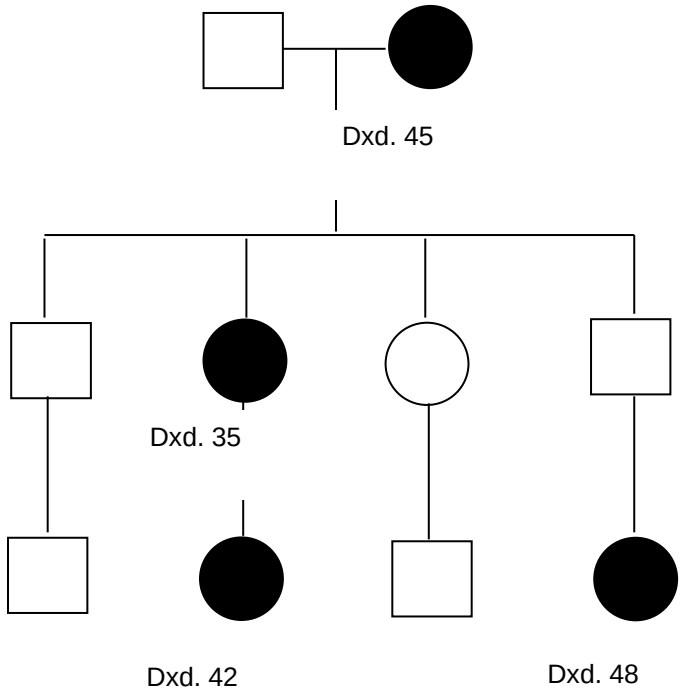
Sporadic Breast Cancer (Average Risk)

- One person in a family with breast cancer
- Typical age of onset
- Single primary tumor
- What does this history mean?
  - Relatives generally not at increased risk of cancer
  - Cause- chance or environmental factors



Medical management: population screening. Right now, genetic testing would not be recommended.

Hereditary Cancer (High Risk)

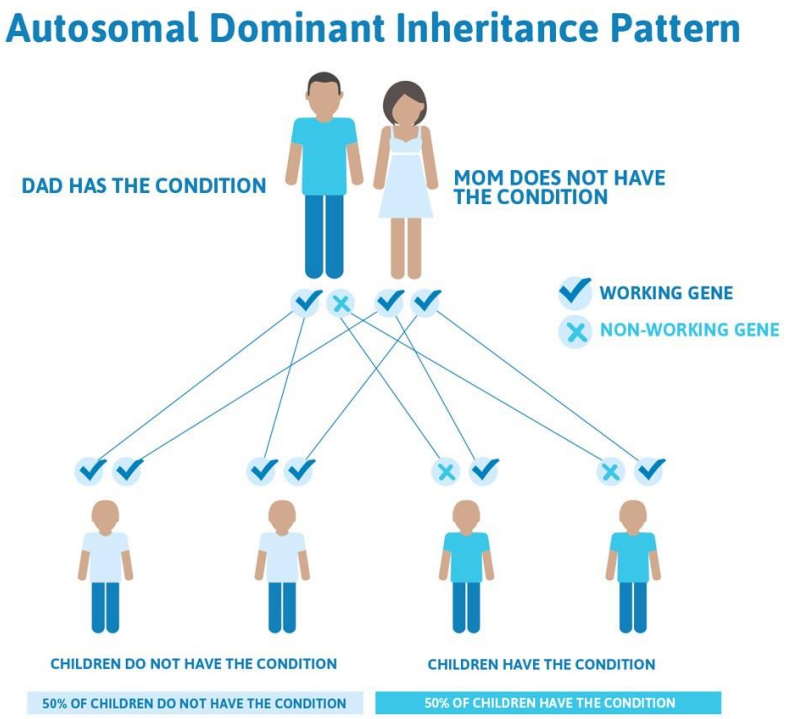


- "Red Flags"
- Two or more relatives on the **same side of the family** with the same or related cancers
- Early age of onset (<50)
- ≥2 more separate cancers in the same person (bilateral breast cancer, breast and ovarian cancer)
- Certain tumor characteristics (like triple negative BC)
- Evidence of a specific inheritance pattern (usually dominant)
- Cause- inherited change (mutation) in a single gene
- Inheritance-usually dominant with reduced penetrance.

Some Examples of Hereditary Breast Cancer Syndromes

Gene	Syndrome
BRCA1 & BRCA	Hereditary breast ovarian cancer syndrome (HBOC): Breast, ovarian, male BC, prostate cancers, and pancreatic cancer, melanoma
p53	Li-Fraumeni syndrome: Breast, sarcoma, lung, leukemia, colon, adrenal cortical tumor
PTEN	Cowden syndrome: Breast, thyroid, endometrial cancers; less often brain, renal cancers. Skin manifestations, thyroid abnormalities, polyps, macrocephaly
CDH1	Hereditary diffuse gastric cancer syndrome: Lobular breast cancer and diffuse gastric cancer.

Hereditary Cancer Inheritance



- Autosomal dominant means first degree relatives have a 1 in 2 chance of inheriting the cancer-predisposing mutation
- When a mutation is identified in a family (ideally in a person with cancer first), testing is then available for other relatives.
  - Usually once they are 18 or older with a few exceptions.
- BUT, not everyone with a mutation develops cancer (reduced penetrance)

Cascade Testing

- Once a mutation has been found in a family, it is important to share that information with other relatives so they can decide whether to have genetic counseling, with or without genetic testing
- Family member with the mutation is usually responsible for telling relatives
  - Genetic counselors can help by providing letters and resources
  - Support organizations also have resources for cascade testing
  - https://www.cdc.gov/genomics/disease/cascade_testing/cascade_hboc.htm#print
  - https://www.curetoday.com/view/saving-lives-with-a-cascade

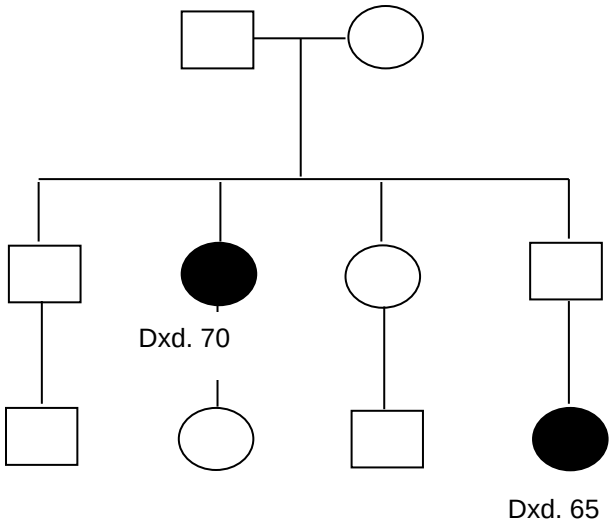
Why would a person want to know about hereditary cancer risk?

- Medical care is different than in sporadic cases.
- Depending on the gene involved:
  - Breast cancer screening usually starts at an earlier age and often includes both MRI and mammography.
  - Screening for cancers in other organs may be recommended.
  - Preventative or risk-reducing steps may be available.

Taking these steps can help identify cancers at an earlier, more treatable stage reduce the risk of cancer, and save lives.

Not all family histories mean high risk- Familial Cancer (Moderate Risk)

- Two close relatives with a specific type of cancer like breast cancer
- Typical age of onset
- Single primary tumor
- No apparent pattern of inheritance.
- What does it mean?
  - Other relatives at moderately increased risk of cancer
  - Cause- usually multifactorial (combination of genes and environment)



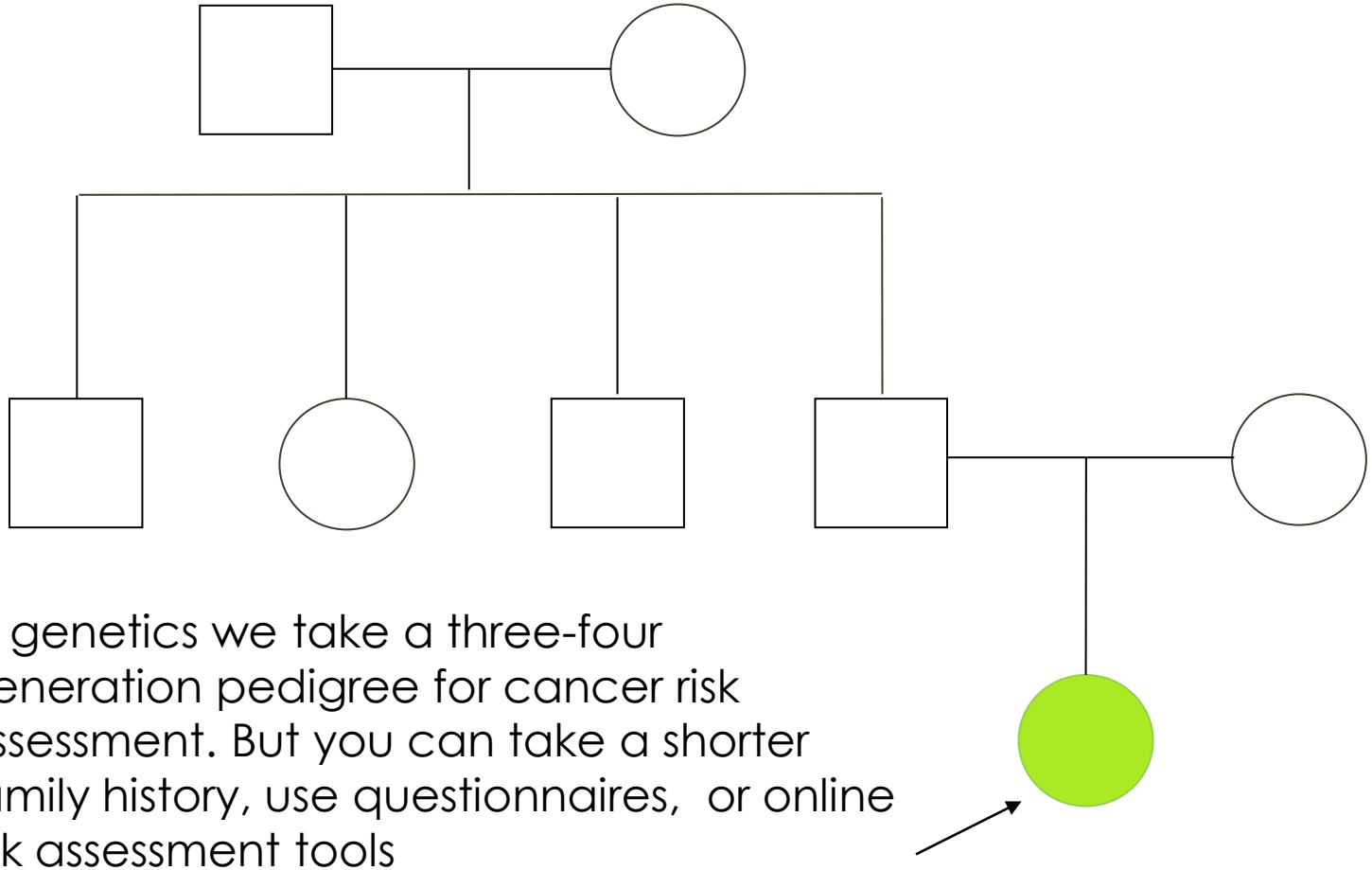
What do we do? Increased screening, generally starting at standard age of onset. Discussion of preventative steps balancing risks of cancer versus risk of intervention. Genetic testing less likely to help.

Key Points

- All cancers are genetic but most are not inherited.
- Risk factors in a person or their family that increase the chance of hereditary breast cancer are
  - Early age of onset (<50)
  - Bilateral or ≥2 different primary cancers in a person
  - More affected relatives than expected based on chance
  - Certain tumor characteristics
- People at risk of hereditary cancer may benefit from increased screening starting at an earlier age than usual, discussions about ways to prevent cancer, and genetic counseling with or without genetic testing.
  - Includes relatives of people with cancer

Navigating Patients to Genetic Counseling

Step 1: Identify Patients at Risk through Personal and Family History Information



In genetics we take a three-four generation pedigree for cancer risk assessment. But you can take a shorter family history, use questionnaires, or online risk assessment tools

Prepare Yourself
Know what questions to ask

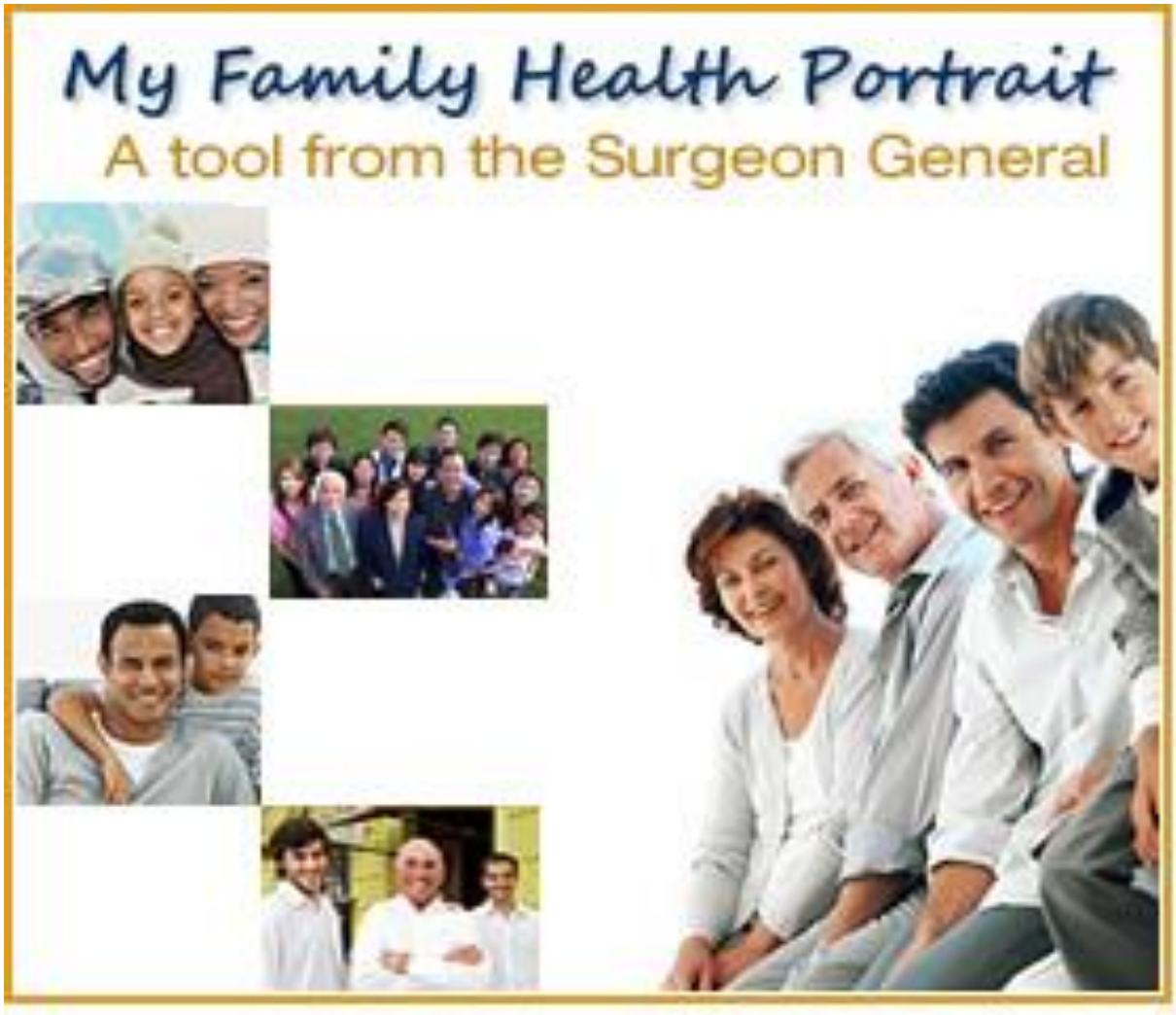
Questions to Ask About All Relatives	Questions to Ask About Relatives Who Have Had Cancer
•Age •Personal history of benign or malignant tumors? •Major illnesses •Hospitalizations •Surgeries (including risk-reducing ones) •Biopsy history •Reproductive history* *Especially important for women at increased risk of breast, ovarian, or endometrial cancer.	•Organ in which tumor developed •**Age** at time of diagnosis •Number of tumors* •Pathology, stage, and grade of malignant tumor •Pathology of benign tumors •Treatment regimen (surgery, chemotherapy, radiation) *Primary or recurrence?

Taking a family history can…

- Help you and the patient recognize patterns of cancer in the family
- Help you understand their experience with cancer and what it means to them
- Help you identify the patient's motivations or concerns about learning about cancer risk
- Help you identify family dynamics

Preparing the Patient In Advance Can Help

- Explain what you are doing and why
- Explain what type of information you are looking for
- When possible, notify patients ahead of time that you will be requesting family history information so they can prepare
  - Can provide family history questionnaires
- Ask patients what questions or concerns they have before you get started



https://phgkb.cdc.gov/FHH/html/index.html

Consider Risk Assessment Tools

- May have limited time to take a family history
- Tools available on line to help identify patients at risk of hereditary cancer
- https://migrc.org/cancer-risk/
  - Based on recent risk guidelines like those from the National Comprehensive Cancer Network, www.nccn.org
- https://www.cdc.gov/cancer/breast/young_women/bringyourbrave/health_care_provider_education/risk-assessment-management-strategies/risk-assessment.htm

Step 2. Assessing Risk-Knowing When to Refer

Family history approach

- **Look for red flags (signs)**
- Relative with mutation in a cancer predisposition gene
- Early age of onset
- Bilateral or multifocal cancers in an individual
- Several family members affected with the same or related cancer
- Other non-cancer signs or symptoms of an inherited syndrome

Personal history factors that prompt referral

- Diagnosis of ovarian cancer at any age
- Women with metastatic breast cancer (for cancer therapy decisions)
- Diagnosis of pancreatic cancer
- Women with breast cancer or men with high grade prostate cancer (Gleason ≥7) and Ashkenazi Jewish ancestry
- Males with breast cancer or metastatic prostate cancer
- Tumor test result that shows a PV in a gene associated with a cancer syndrome or microsatellite instability

What if you identify a need for referral?

1. Find a genetic counselor
2. Prepare your patient
3. Prepare yourself

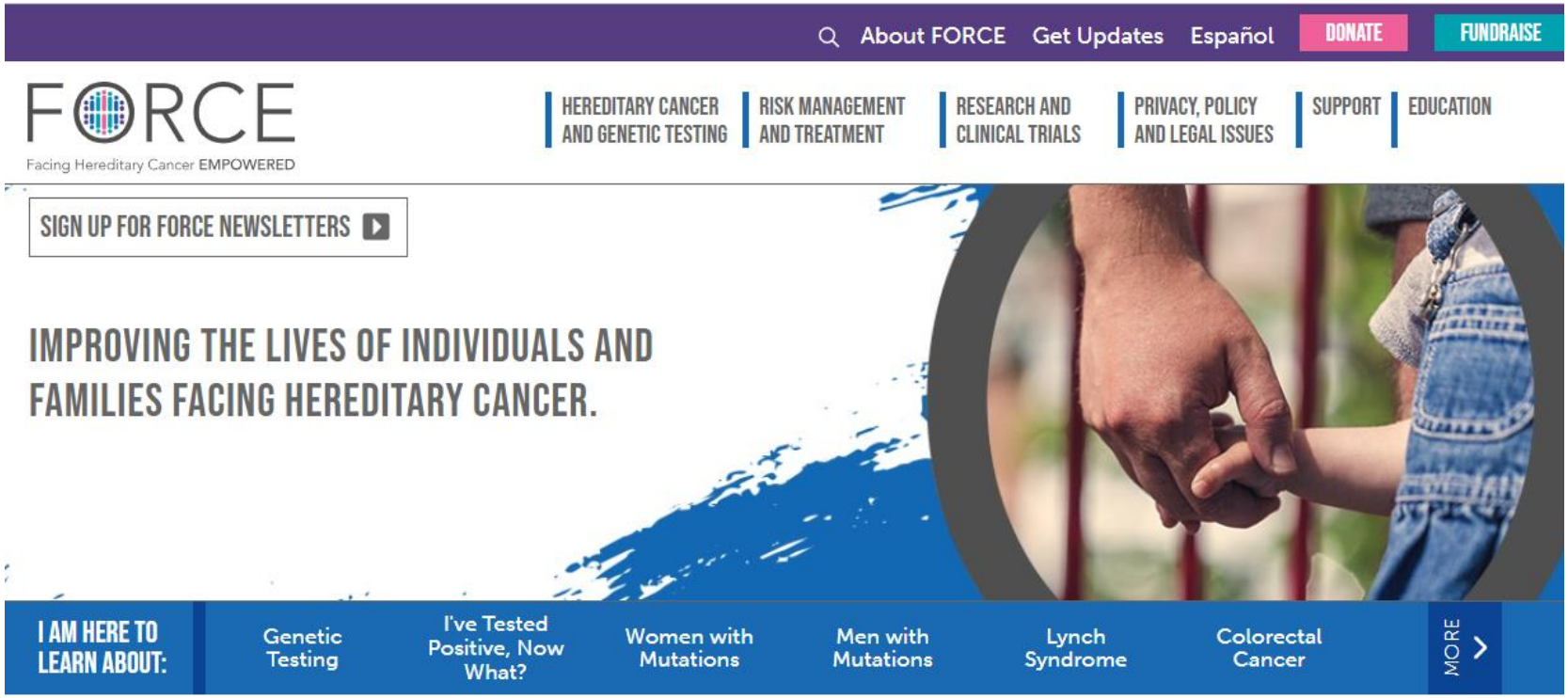
3. Finding a Genetic Counselor

- National Society of Genetic Counselors Find a Counselor search engine
  - http://www.nsgc.org/page/find-a-gc-search
- Medline Plus
  - https://medlineplus.gov/genetics/understanding/consult/findingprofessional/
- Find a Genetics Clinic https://clinics.acmg.net/
- Find a Genetics Clinic in Texas
- https://www.tsgc.org/genetics-clinics-in-texas

4. Preparing your Patient

- What is genetic counseling? Why is it important?
  - https://www.facingourrisk.org/info/hereditary-cancer-and-genetic-testing/how-to-get-testing/genetic-counseling
- What will happen?
  - http://www.cancer.net/navigating-cancer-care/cancer-basics/genetics/what-expect-when-meeting-genetic-counselor
- Do I have to have a genetic test?
- Should I bring someone with me?

Resources- Facing our Risk of Cancer Empowered



Resources

- Centers for Disease Control and Prevention- Bring Your Brave Campaign https://www.cdc.gov/cancer/breast/young_women/bringyourbrave/index.htm
  - *Provides information about breast cancer to women younger than age 45 by sharing real stories about young women whose lives have been affected by breast cancer.*

5. Preparing Yourself

- What does the genetic counselor need from you?
- If your patient has testing, will you get the results? When?
- How can you work with the genetic counselor to best care for your patient?

Resource from MDHHS

*** Do you have questions about hereditary cancer? ***
Call our HOTLINE at 866-852-1247, 9am-4pm Monday thru Friday.

Below are no-cost resources for clinical use and professional development in cancer genomics:

Risk Assessment

- Guide to Hereditary Cancer Indications for Referral - online interactive tool
- 4 Steps to Making Appropriate Referrals for Cancer Genetic Risk Assessment and Counseling
- Cancer-Specific Risk Factors for Referral for Hereditary Cancer Risk Assessment and Counseling
- NIH Breast Cancer Risk Assessment Tool
- USPSTF Recommendations for BRCA-Related Cancer
- EGAPP Guidelines for colorectal cancer patients
- National Comprehensive Cancer Network Guidelines

Genetic Counseling, Testing and Referral

- Video: Genetic Testing for Inherited Risk for Cancer - An overview of understanding inherited risk for cancer and why genetic te help in determining if someone might be at risk
- Infographic: Cancer Genetic Counseling: What to Know - A guide for patients addressing frequently asked questions and comm about cancer genetic counseling and testing
- Directory of Cancer Genetics Clinics in Michigan
- NSGC Tool: Find a Genetic Counselor
- Genetic Counseling and Testing Guidelines - a summary of national recommendations related to clinical practice in cancer gen
- Position Paper on Genetic Counseling and Testing from MCGA

Health Plan Policy

- Information for Insurers and Health Plan Administrators - a guide for navigating cancer genomics policies

Cancer Genomics Education

- Video: Genetics 101 - an introduction to cancer genomics, hereditary breast and ovarian cancer (HBOC) and Lynch syndrome

https://www.michigan.gov/mdhhs/0,5885,7-339-73971_4911_4916_47257_68337_94208_94214---,00.html

Summary

- All cancers are genetic but just a small percent are inherited
- Identifying individuals at risk for inherited breast cancer is important, though, because it changes medical management and can save lives
- Identifying patients at risk involves collecting family history information and identifying red flags. Several tools and guidelines are available to help.
- Providers like you are key to helping at risk individuals get access!