Referral Survey

Please complete the survey below.

Thank you!

You are being asked to participate in a research study to understand the patterns of referral for cancer genetic consultation based on family cancer history. This study is being conducted by Brandon Welch, MS, PhD, Associate Professor in Biomedical Informatics at the Medical University of South Carolina.

Understanding these patterns will allow us to evaluate the extent to which patients are being referred for cancer genetic consultation based on clinical practice guideline criteria. These findings will lead to better tools to help assist providers in evaluating patient family cancer histories according to established clinical practice guidelines.

There is a risk of loss of confidentiality as a result of participation. There will be no direct benefit to you but we hope that these findings will lead to better tools to help assist providers in evaluating patient family cancer histories according to established clinical practice guidelines.

The study should take 10 minutes. Your participation in this research study is voluntary and will not constitute any element of your job performance or become part of your record.

Your consent is implied by completing the survey. If you would like to participate in this survey, please select "I agree" to continue. If you have any questions about this research, email Dr. Welch (welchbm@musc.edu).

\bigcirc \Box	agree
\bigcirc \Box	do not agree



About you		
Are you male or female?	○ Male○ Female○ prefer not to say	
What type of healthcare professional are you?	Genetic CounselorOncologistPrimary careOB/GYNOther	
If you chose 'other', please specify		_
How many years have you been practicing as a(n) [provider_type]?		-
How many years have you been practicing as a(n) [other_provider_type]?		_

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Please consider each of the following family history cases and record whether or not the patient (proband) should be referred for genetic counseling and/or testing.

Should the patient (proband) in the following pedigree be referred for a cancer genetic consultation based on the available information?

Executive Summary

Proband is a 41 year old female with

- No reported personal history of cancer
- African American Ancestry
- No reported personal gene mutation
- No reported family gene mutation history
- Family history of Melanoma, Colorectal cancer

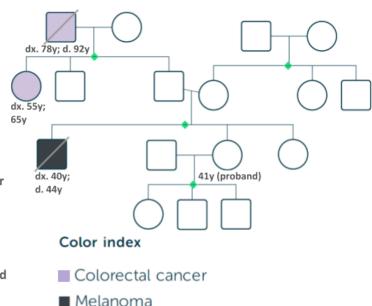
Family Cancer History

Colorectal Cancer

- Paternal Grandfather diagnosed at 78 years old
- Paternal Aunt diagnosed at 55 years old

Melanoma

Brother diagnosed at 40 years old



- YES, this patient should be referred for genetic counseling or testing
- NO, this patient should NOT be referred for genetic counseling or testing



FHx case 2/18

Should the patient (proband) in the following pedigree be referred for a cancer genetic consultation based on the available information?

Executive Summary

Proband is a 52 year old female with

- Personal history of of Cervical cancer at 49 years old
- European Ancestry
- No reported personal gene mutation
- No reported family gene mutation history
- Family history of Prostate cancer, Breast cancer

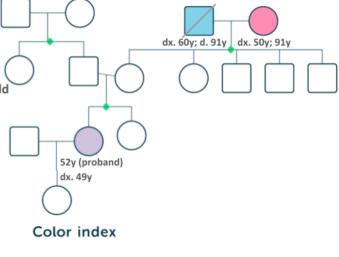
Family Cancer History

Breast Cancer

Maternal Grandmother diagnosed at 50 years old

Prostate Cancer

Maternal Grandfather diagnosed at 60 years old



Cervical cancer

Prostate cancer

Breast cancer

- counseling or testing
 NO, this patient should NOT be referred for genetic counseling or testing





FHx case 3/18

Should the patient (proband) in the following pedigree be referred for a cancer genetic consultation based on the available information?

Executive Summary

Proband is a 58 year old female with

- No reported personal history of cancer
- European Ancestry
- No reported personal gene mutation
- No reported family gene mutation history
- Family history of Hemangioblastoma, Breast cancer

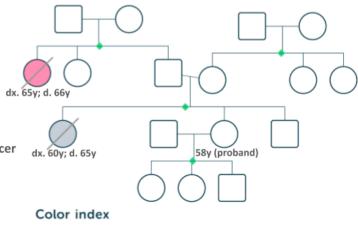
Family Cancer History

Breast Cancer

Paternal Aunt diagnosed at 65 years old

Hemangioblastoma

Sister diagnosed at 60 years old



Breast cancer

Hemangioblastoma

- YES, this patient should be referred for genetic counseling or testing
- NO, this patient should NOT be referred for genetic counseling or testing

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FHx case 4/18

Should the patient (proband) in the following pedigree be referred for a cancer genetic consultation based on the available information?

Executive Summary

Proband is a 59 year old female with

- Personal history of of Squamous cell lung cancer at 43 years old
- European Ancestry
- No reported personal gene mutation
- No reported family gene mutation history
- Family history of Ewing's sarcoma, Breast cancer

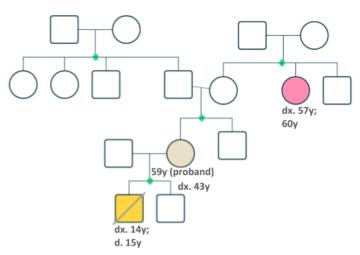
Family Cancer History

Breast Cancer

Maternal Aunt diagnosed at 57 years old

Ewing's Sarcoma

Son diagnosed at 14 years old



Color index

Squamous cell lung cancer

Breast cancer

Ewing's sarcoma

- YES, this patient should be referred for genetic counseling or testing
- NO, this patient should NOT be referred for genetic counseling or testing



FHx case 5/18

Should the patient (proband) in the following pedigree be referred for a cancer genetic consultation based on the available information?

Executive Summary

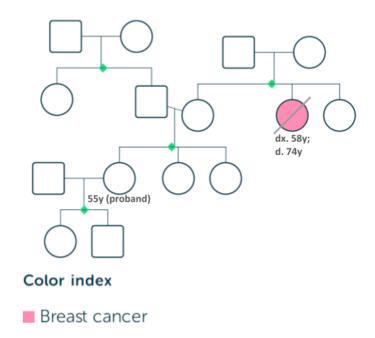
Proband is a 55 year old female with

- No reported personal history of cancer
- Ashkenazi Jewish Ancestry
- No reported personal gene mutation
- No reported family gene mutation history
- Family history of Breast cancer

Family Cancer History

Breast Cancer

- Maternal Aunt diagnosed at 58 years old
 - ER negative
 - PR negative
 - HER2 negative
 - Unilateral



- YES, this patient should be referred for genetic counseling or testing
- NO, this patient should NOT be referred for genetic counseling or testing

FHx case 6/18

Should the patient (proband) in the following pedigree be referred for a cancer genetic consultation based on the available information?

Executive Summary

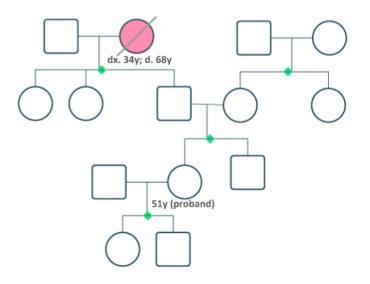
Proband is a 51 year old female with

- No reported personal history of cancer
- Ashkenazi Jewish Ancestry
- No reported personal gene mutation
- No reported family gene mutation history
- Family history of Breast cancer

Family Cancer History

Breast Cancer

- Paternal Grandmother diagnosed at 34 years old
 - o Bilateral



Color index

Breast cancer

- YES, this patient should be referred for genetic counseling or testing
- NO, this patient should NOT be referred for genetic counseling or testing



FHx case 7/18

Should the patient (proband) in the following pedigree be referred for a cancer genetic consultation based on the available information?

Executive Summary

Proband is a 60 year old female with

- Personal history of of Breast cancer at 56 years old
- African American Ancestry
- No reported personal gene mutation
- No reported family gene mutation history
- Family history of Brain cancer, Bone cancer, Breast cancer

Family Cancer History

Breast Cancer

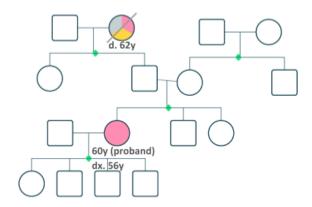
Paternal Grandmother diagnosed at 56 years old

Bone Cancer

• Paternal Grandmother diagnosed at 62 years old

Brain Cancer

Paternal Grandmother diagnosed at 62 years old



Color index

Breast cancer

Bone cancer

Brain cancer

- YES, this patient should be referred for genetic counseling or testing
- NO, this patient should NOT be referred for genetic counseling or testing



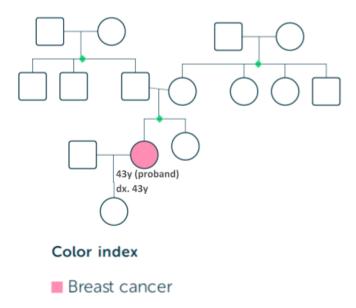
FHx case 8/18

Should the patient (proband) in the following pedigree be referred for a cancer genetic consultation based on the available information?

Executive Summary

Proband is a 43 year old female with

- Personal history of of Breast cancer at 43 years old
- European Ancestry
- No reported personal gene mutation
- No reported family gene mutation history
- No Family history of cancer



- YES, this patient should be referred for genetic counseling or testing
- NO, this patient should NOT be referred for genetic counseling or testing



FHx case 9/18

Should the patient (proband) in the following pedigree be referred for a cancer genetic consultation based on the available information?

Executive Summary

Proband is a 60 year old female with

- No reported personal history of cancer
- European Ancestry
- No reported personal gene mutation
- No reported family gene mutation history
- Family history of Breast cancer, Pancreatic cancer

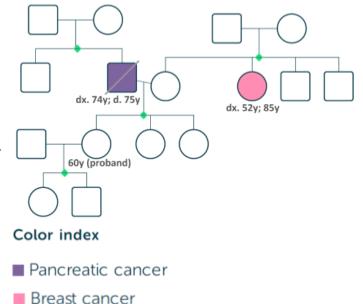
Family Cancer History

Breast Cancer

Maternal Aunt diagnosed at 52 years old

Pancreatic Cancer

Father diagnosed at 74 years old



- YES, this patient should be referred for genetic counseling or testing
- NO, this patient should NOT be referred for genetic counseling or testing



FHx case 10/18

Should the patient (proband) in the following pedigree be referred for a cancer genetic consultation based on the available information?

Executive Summary

Proband is a 60 year old female with

- Personal history of Medullary thyroid cancer at 48 years old, Breast cancer at 57 years old
- European Ancestry
- No reported personal gene mutation
- No reported family gene mutation history
- Family history of Medullary thyroid cancer,
 Stomach cancer

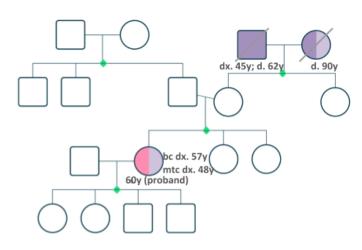
Family Cancer History

Medullary thyroid cancer

Maternal Grandmother diagnosed at 45 years old

Stomach cancer

- Maternal Grandmother diagnosed at 49 years old
- Maternal Grandfather diagnosed at 60 years old



- Medullary thyroid cancer
- Breast cancer
- Stomach cancer
 - YES, this patient should be referred for genetic counseling or testing
 - NO, this patient should NOT be referred for genetic counseling or testing



FHx case 11/18

Should the patient (proband) in the following pedigree be referred for a cancer genetic consultation based on the available information?

Executive Summary

Proband is a 49 year old female with

- Personal history of of Cervical cancer at 35 years old
- European Ancestry
- No reported personal gene mutation
- No reported family gene mutation history
- Family history of Breast cancer, Osteosarcoma, Colon cancer

Family Cancer History

Breast Cancer

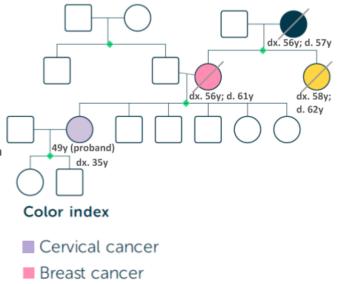
Mother diagnosed at 56 years old

Colon Cancer

• Maternal Grandmother diagnosed at 56 years old

Osteosarcoma

Maternal Aunt diagnosed at 58 years old



- YES, this patient should be referred for genetic counseling or testing
- NO, this patient should NOT be referred for genetic counseling or testing

Colon cancer

Osteosarcoma



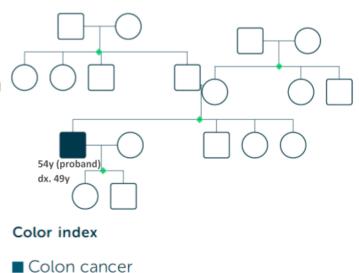
FHx case 12/18

Should the patient (proband) in the following pedigree be referred for a cancer genetic consultation based on the available information?

Executive Summary

Proband is a 54 year old female with

- Personal history of of Colon cancer at 49 years old
- African American Ancestry
- No reported personal gene mutation
- No reported family gene mutation history
- No Family history of cancer



- YES, this patient should be referred for genetic counseling or testing
- NO, this patient should NOT be referred for genetic counseling or testing



FHx case 13/18

Should the patient (proband) in the following pedigree be referred for a cancer genetic consultation based on the available information?

Executive Summary

Proband is a 49 year old female with

- Personal history of Non-Hodgkin's lymphoma at 48 years old
- European Ancestry
- No reported personal gene mutation
- No reported family gene mutation history
- Family history of Ovarian cancer, Small cell lung cancer, Hodgkin's lymphoma, Breast cancer,
 Pancreatic cancer

Family Cancer History

Breast Cancer

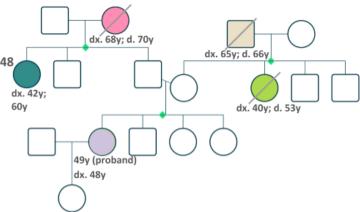
- Paternal Grandmother diagnosed at 68 years old Hodgkin's lymphoma
 - Maternal Aunt diagnosed at 40 years old

Ovarian Cancer

Paternal Aunt diagnosed at 42 years old

Small cell lung cancer

Maternal Grandfather diagnosed at 65 years old



- Non-Hodgkin's lymphoma
- Breast cancer
- Small cell lung cancer
- Hodgkin's lymphoma
- Ovarian cancer
 - YES, this patient should be referred for genetic counseling or testing
 - NO, this patient should NOT be referred for genetic counseling or testing



FHx case 14/18

Should the patient (proband) in the following pedigree be referred for a cancer genetic consultation based on the available information?

Executive Summary

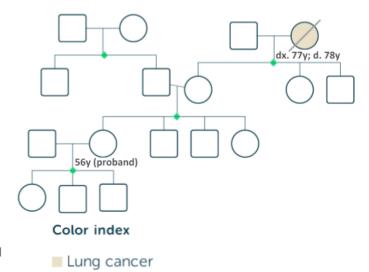
Proband is a 56 year old female with

- No reported personal history of cancer
- European Ancestry
- No reported personal gene mutation
- No reported family gene mutation history
- Family history of Lung cancer

Family Cancer History

Lung Cancer

Maternal Grandmother diagnosed at 77 years old



- YES, this patient should be referred for genetic counseling or testing
- NO, this patient should NOT be referred for genetic counseling or testing



FHx case 15/18

Should the patient (proband) in the following pedigree be referred for a cancer genetic consultation based on the available information?

Executive Summary

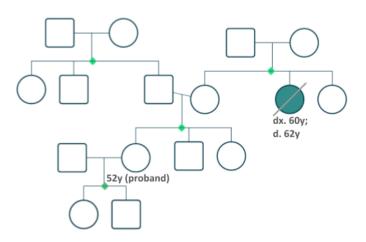
Proband is a 52 year old female with

- No reported personal history of cancer
- European Ancestry
- No reported personal gene mutation
- No reported family gene mutation history
- Family history of Ovarian cancer

Family Cancer History

Ovarian Cancer

Maternal Aunt diagnosed at 60 years old



Color index

Ovarian cancer

- YES, this patient should be referred for genetic counseling or testing
- NO, this patient should NOT be referred for genetic counseling or testing



FHx case 16/18

Should the patient (proband) in the following pedigree be referred for a cancer genetic consultation based on the available information?

Executive Summary

Proband is a 55 year old female with

- Personal history of of Squamous cell skin cancer at 51 years old
- European Ancestry
- No reported personal gene mutation
- No reported family gene mutation history
- Family history of Liver cancer, Chronic myelogenous leukemia (CML), Breast cancer

dx. 60y; d. 62y d. 65y 55y (proband) dx. 51y

Family Cancer History

Breast Cancer

Maternal Aunt diagnosed at 60 years old

Chronic myelogenous leukemia (CML)

Maternal Grandfather diagnosed at 60 years old

Liver Cancer

Maternal Aunt diagnosed at 65 years old

- Squamous cell skin cancer
- Chronic myelogenous leukemia (CML)
- Breast cancer
- Liver cancer
 - YES, this patient should be referred for genetic counseling or testing
 - NO, this patient should NOT be referred for genetic counseling or testing



FHx case 17/18

Should the patient (proband) in the following pedigree be referred for a cancer genetic consultation based on the available information?

Executive Summary

Proband is a 42 year old female with

- No reported personal history of cancer
- European Ancestry
- No reported personal gene mutation
- No reported family gene mutation history
- Family history of Prostate cancer, Breast cancer

dx. 49y; d. 88y dx. 42y; 76y dx. 58y; 69y cer 42y (proband)

Family Cancer History

Breast Cancer

- Paternal Grandmother diagnosed at 49 years old
- Paternal Aunt diagnosed at 42 years old

Prostate Cancer

- Father diagnosed at 58 years old
 - Gleason score: 8

- Prostate cancer
- Breast cancer
 - YES, this patient should be referred for genetic counseling or testing
 - NO, this patient should NOT be referred for genetic counseling or testing



FHx case 18/18

Should the patient (proband) in the following pedigree be referred for a cancer genetic consultation based on the available information?

Executive Summary

Proband is a 50 year old female with

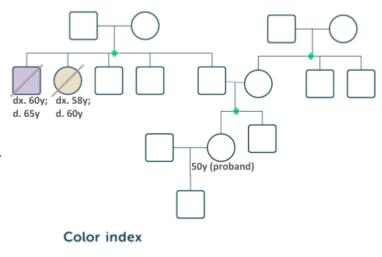
- No reported personal history of cancer
- European Ancestry
- No reported personal gene mutation
- No reported family gene mutation history
- Family history of Lung cancer, Bladder cancer

Family Cancer History

Bladder Cancer

 Paternal Uncle diagnosed at 60 years old Lung Cancer

Paternal Aunt diagnosed at 58 years old



Bladder cancer

Lung cancer

- YES, this patient should be referred for genetic counseling or testing
- NO, this patient should NOT be referred for genetic counseling or testing



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