

Genetic Counseling and Testing for Gastric Cancer

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LEARNING OBJECTIVES

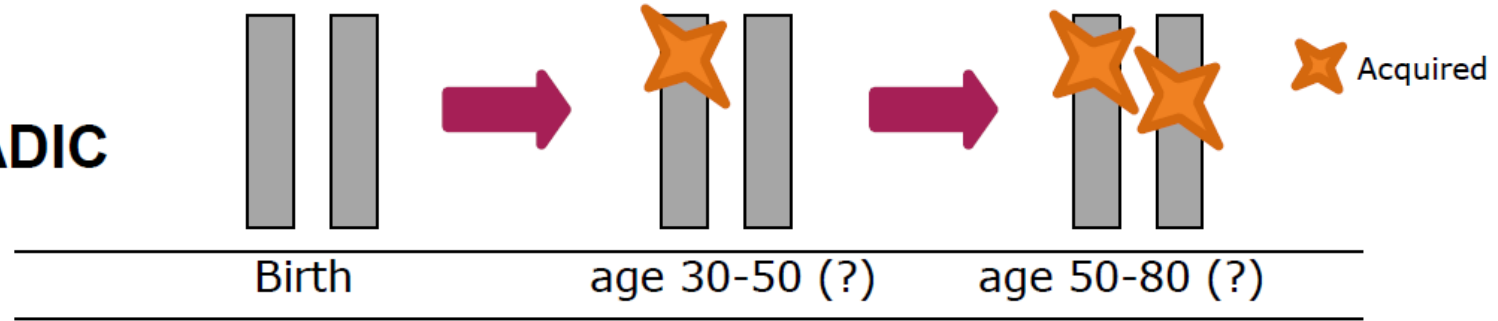
- Recognize features associated with hereditary cancer
- Understand the implications of genetic test results on medical management



Genetics of cancer

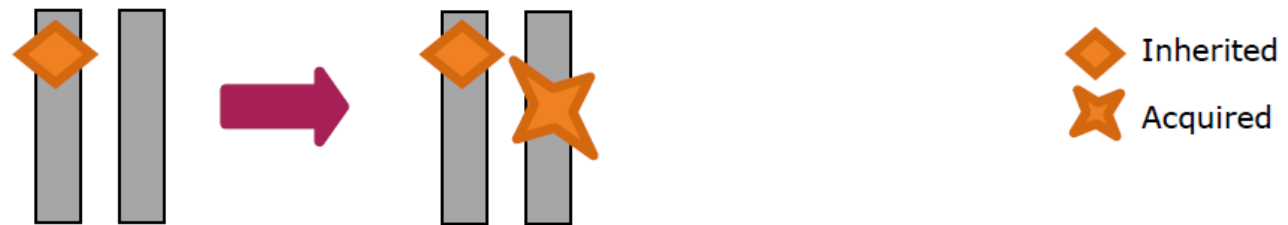
90%

SPORADIC

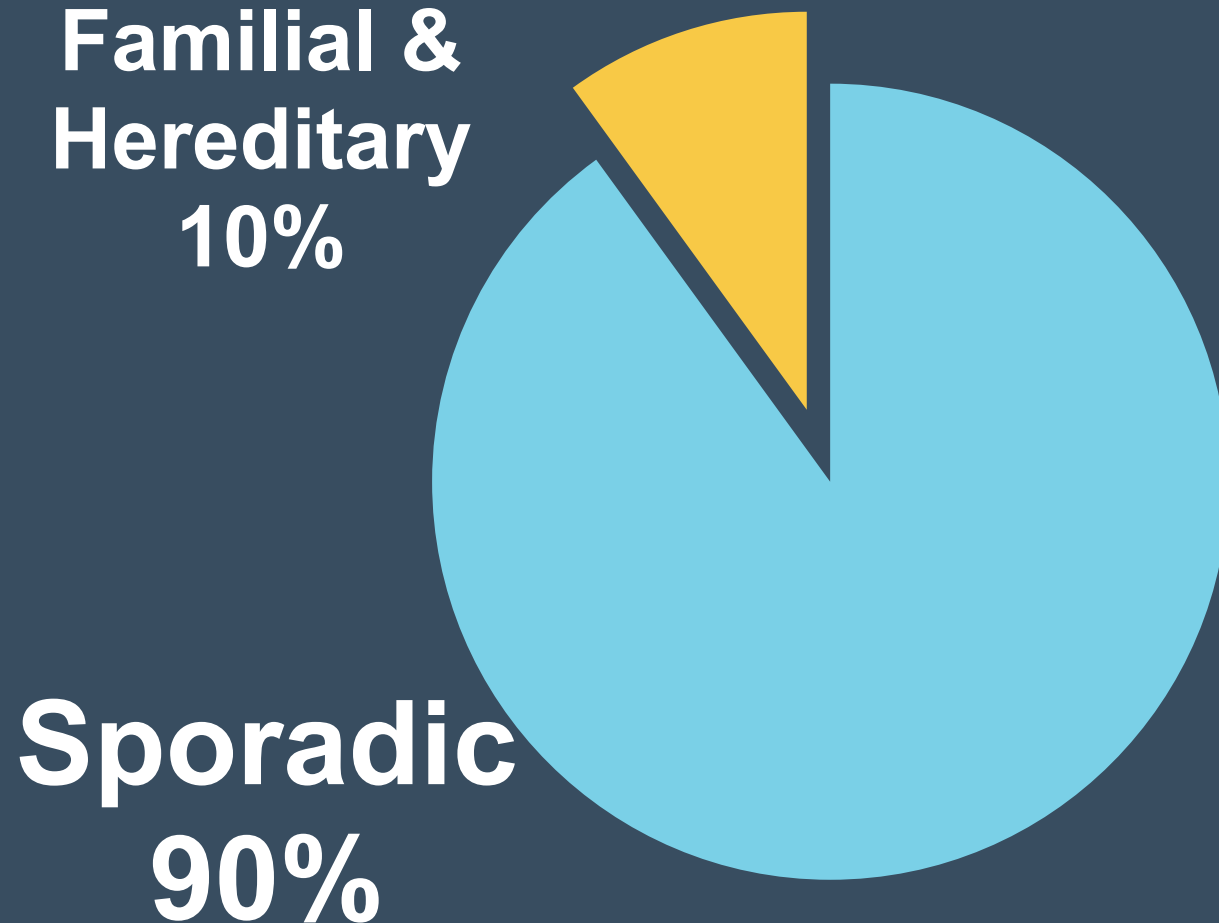


10%

HEREDITARY



Cancer Patterns in Families



Genetic testing may help to identify those at risk for **FAMILIAL** or **HEREDITARY** cancer

Cancer Patterns in Families



Gastric cancer:

- 1-3% CDH1
- Other genes:
 - Lynch syndrome
 - Juvenile Polyposis
 - Peutz-Jeghers syndrome
 - Familial Polyposis (FAP)
 - ?CTNNA1

Features of Hereditary Cancer

- Multiple cases of the same cancer in a family
 - ≥ 2 cases of diffuse stomach cancer in first- or second-degree relatives, where at least one was diagnosed $< 50y$
 - ≥ 3 or more cases of diffuse stomach cancer in first- or second-degree relatives, regardless of the age at diagnosis
[25% to 40% will have a CDH1 gene mutation]
- Diagnosis of gastric cancer prior to age 40

Features of Hereditary Cancer

- Cancer diagnosed prior to age 50
 - Breast (you, 1st, or 2nd degree relative)
 - Colorectal or Endometrial (you or 1st-degree relative)
- Any ONE of the following
 - Ovarian cancer (you, 1st, or 2nd degree relative)
 - Pancreatic adenocarcinoma (you or 1st-degree relative)
 - Metastatic Prostate cancer (you or 1st-degree relative)
 - Personal history of >10 colorectal polyps

Features of Hereditary Cancer

- Combination of cancers in a family:
 - ≥ 3 or more cases of breast, ovarian, pancreatic, or prostate cancers
 - ≥ 3 or more cases of colorectal, endometrial, gastric, ovarian, pancreas, glioblastoma, and small intestine cancers
 - ≥ 2 or more cases of lobular breast cancer and/or diffuse gastric cancer

	Lifetime risk for Gastric Cancer	Gastric Screening	Other considerations
Hereditary Diffuse Gastric Cancer (CDH1)	56%-83% females 67%-70% males	Gastrectomy at 18 - 40. Upper endoscopy every 6-12 months with random biopsies	Prophylactic gastrectomy, ideally 3rd decade of life. Breast cancer, lobular-type (39%-52%) – annual breast MRI and mammogram starting at age 30. Option for preventative mastectomies.
Lynch syndrome (MLH1, MSH2, MSH6, PMS2, EPCAM)	5% - 7% (MLH1) 0.2% - 9% (MSH2) ≤1% - 7.9% (MSH6)	Upper endoscopy starting age 40, every 3-5y	Colorectal cancer, also endometrial, ovarian, pancreas, biliary tract, glioblastoma, and renal pelvis (kidney) cancer. 1 in 280 people ? Immunotherapy
Peutz-Jeghers syndrome (STK11)	29%	Upper endoscopy starting in late teens, every 2-3y	Breast cancer (32%-54%), also colorectal, small intestine, pancreas, reproductive organs, and lung cancer. Blue pigmentation on mouth, lips, face, genitalia, and fingers. Polyps of the intestines.
Juvenile Polyposis syndrome (BMPR1A, SMAD4)	21%	Upper endoscopy starting at age 15, every 1-3y	Numerous polyps of the intestines , often diagnosed in teenage years, and may require surgery to remove the colon. Possible risk for recurrent nosebleeds.
Familial Adenomatous polyposis (APC)	0.5% - 1.3%	Upper endoscopy starting at age 20-25, every 1-4y	Numerous polyps of the intestines , often diagnosed in teenage years, and often requiring surgery to remove the colon. Risk for duodenal cancer, and thyroid cancer. Rare to consider gastrectomy.
Li-Fraumeni syndrome (TP53)	Unknown	Upper endoscopy starting at age 25, every 2-5y	Rare syndrome. Very high risk for early-onset cancers diagnosed prior to age 50. Most often breast, glioblastoma, sarcomas.

Features of Hereditary Cancer

HEREDITARY CANCER RISK ASSESSMENT QUESTIONNAIRE

Please complete this form to be reviewed by your healthcare provider at your visit

YES	NO	
		Do you have any of the following in you or your relatives? (circle relative, if applicable)
<input type="checkbox"/>	<input type="checkbox"/>	Metastatic Prostate cancer (brother, father, son, or myself - if male)
<input type="checkbox"/>	<input type="checkbox"/>	Pancreatic adenocarcinoma (myself, son, daughter, mother, father, sister, brother)
<input type="checkbox"/>	<input type="checkbox"/>	Ovarian, Fallopian tube, or Primary Peritoneal cancer (myself, mother, sister, daughter, aunt, or grandmother)
<input type="checkbox"/>	<input type="checkbox"/>	Breast cancer diagnosed <i>before age 50</i> (myself, mother, sister, daughter, aunt, niece, or grandmother)
<input type="checkbox"/>	<input type="checkbox"/>	Breast cancer in a MALE (brother, father, son, uncle, nephew, grandfather, or myself - if male)
<input type="checkbox"/>	<input type="checkbox"/>	Colon or Rectal cancer diagnosed <i>before age 50</i> in you, son, daughter, mother, father, sister, or brother
<input type="checkbox"/>	<input type="checkbox"/>	Endometrial (uterine) cancer diagnosed <i>before age 50</i> in you, your mother, daughter, or sister
YES	NO	Do you have 3 or more relatives on the <u>same side</u> of the family (related to each other by blood) with: Colon, Rectal, Endometrial (uterine), Gastric, and/or Pancreatic cancers ?
<input type="checkbox"/>	<input type="checkbox"/>	
YES	NO	Do you have 3 or more diagnoses on the <u>same side</u> of the family (related to each other by blood) with: Breast cancer, Pancreatic cancer, and/or metastatic (stage 4) Prostate cancers ?
<input type="checkbox"/>	<input type="checkbox"/>	

How Do I Get Tested?

- Ask your family about history of cancers
- Talk to your healthcare provider
- See a cancer genetic counselor
 - Testing is available for more than just CDH1
 - Experience with rare cancer syndromes
 - www.aboutgeneticcounselors.org





Every life deserves world class care.