PASS IT ON

For Individuals and Families With a History of Cancer

Newsletter of the Cancer Genetic Counseling Service

OUR FAMILY TREE HAS GROWN!

WELCOME THE NEWEST MEMBER OF THE CANCER GENETIC COUNSELING TEAM

ASHLEY COSENZA, MS

Ashley Cosenza joined our team as a genetic counselor with the Cancer Genetic Counseling Service at the Karmanos Cancer Institute in June 2021. Ashley received her Bachelor of Science in Honors Biology with a concentration in Applied Genetics from the University of Detroit Mercy in 2016 and her Master of Science in Genetic Counseling from Wayne State University in 2021. Ashley previously worked as a genetic counseling assistant for Progenity, Inc. She is excited to provide genetic counseling as part of the Karmanos team and to contribute to outreach and clinical research related to improving access to cancer genetic counseling services.

Ashley will be servicing downtown Detroit, McLaren Macomb, McLaren Bay City, and McLaren Flint.

IN THIS ISSUE

BARBARA ANN Karmanos CANCER INSTITUTE Wayne State University

LITERATURE REVIEW

SCIENTIFIC HIGHLIGHTS IN GENETICS

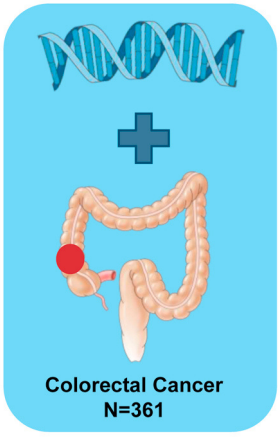
In this edition of the Cancer Genetic Counseling Service newsletter, we highlight a new article released from the Journal of Clinical Gastroenterology and Hepatology in April 2021. The article is titled "Germline Cancer Susceptibility Gene Testing in Unselected Patients With Colorectal Adenocarcinoma: A Multicenter Prospective Study" by Uson et al. This study was a collaboration between researchers and clinicians at the Mayo Clinic and Invitae Laboratories. See below for more details!

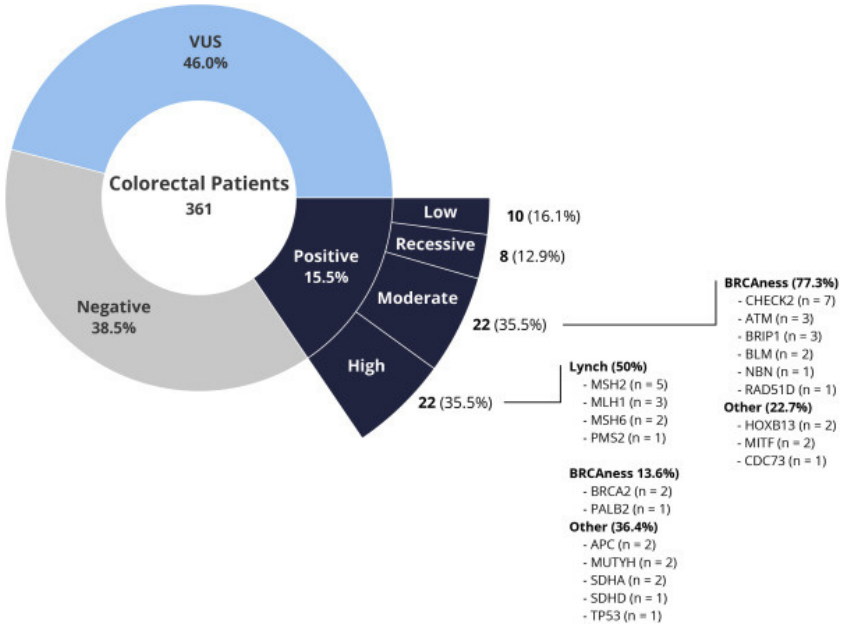
ARTICLE TAKEAWAYS

Background: Hereditary factors play a role in the development of colorectal cancer. Identification of such genetic mutations that can run in families can have implications on treatment and cancer prevention. This study aimed to determine how often these genetic mutations were found in colorectal cancer patients using a universal genetic testing approach (every patient was tested with the same large genetic panel). Researchers also examined how often family members pursued testing when a genetic mutation was found.

Methods: A prospective multisite study of germline genetic testing using a more than 80-gene panel among colorectal cancer patients (not selected for age or family history) receiving care at Mayo Clinic Cancer Centers between April 1, 2018 and March 31, 2020.

Results: Of 361 patients, the median age was 57 years, 43.5% were female, 82% were white, and 38.2% had stage IV disease. Genetic mutations were found in 56 (15.5%) patients, including 44 mutations in moderate- and high-penetrance cancer predisposition genes. Thirty-four (9.4%) patients had clinically actionable findings that would not have been detected by practice guideline criteria or a colon cancer specific gene panel alone. Eleven percent of patients had changes in their treatment based on genetic findings. Further testing among family members once a mutation was identified was low (16%).





Conclusions: Universal multigene panel testing in individuals with colorectal cancer was associated with a modest, but significant, detection of heritable mutations over guideline-based testing. **One in 6 patients with colorectal cancer was found to carry a genetic mutation.** One in 10 patients had changes in their management based on test results. Uptake of genetic testing in family members once a mutation was identified was low, which is a concerning observation that warrants further study.

WHAT IS PALB2?

PALB2 is a tumor suppressor gene that works to prevent the development of cancer in our bodies.

WHAT ARE THE RISKS?

People who have a mutation in their PALB2 gene are at an increased risk to develop breast, ovarian, and pancreatic cancer. Risks are higher for those with a family history of cancer.

	LIFETIME RISK FOR INDIVIDUALS WITH A PALB2 MUTATION	LIFETIME RISK FOR THE GENERAL POPULATION
BREAST CANCER (FEMALE)	40% - 60%	10% - 12%
OVARIAN CANCER	3% - 5%	1%
BREAST CANCER (MALE)	~1%	0.1%
PANCREATIC CANCER	5% - 10%	1%

WHAT CAN I DO IF I HAVE THIS MUTATION?

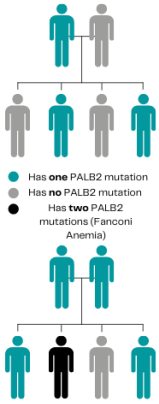
Enhanced cancer screening or risk reducing surgeries are options for individuals who have a PALB2 mutation.

- Women are recommended to begin yearly mammogram screening at age 30. They may also consider yearly breast MRIs in addition to mammograms, alternating each every 6 months. Some women with a PALB2 mutation also consider undergoing a risk-reducing mastectomy (surgical removal of the breasts).
- Women with a PALB2 mutation and family history of ovarian cancer may also consider screening such as transvaginal ultrasound and CA-125 tests. While not standardly recommended at this time, some women opt to have their ovaries surgically removed for personal reasons.
- Men may consider clinical or self breast exams every 6-12 months.
- Individuals with a family history of pancreatic cancer are recommended to begin yearly abdominal MRI and/or endoscopic ultrasounds at age 50 or 10 years younger than the earliest age of pancreatic cancer diagnosis in the family.

HOW CAN THIS AFFECT MY FAMILY?

Children and siblings of individuals with a PALB2 mutation each have a 50% chance of also having inherited the mutation. When a mutation is identified in the family, it is strongly recommended that such information be shared with relatives so that they can make informed decisions about their medical care.

When two individuals with a PALB2 mutation have children, there is a 25% (or 1/4) chance each parent will pass on their mutation. People who inherit two PALB2 mutations have a medical condition called Fanconi Anemia (FA). This condition is associated with various developmental abnormalities, organ defects, early-onset bone marrow failure, and an increased risk for cancer.



Infographic created by Morgan Devlin

REMINDER: We offer annual virtual genetic counseling appointments for any individual who has a positive genetic testing result. These appointments give us the chance to update you regarding any new information learned about that gene.

Screening recommendations or known cancer risks may change over time. In addition, it gives us the opportunity to update your family history, review the importance of testing your relatives for your cancer gene mutation, and help facilitate such testing for your family members.

You can call us at 313-576-8748 to schedule your annual Genetics follow-up appointment!

Currently, all genetic counseling sessions are being conducted over the telephone or via our secure video platform.

If you have questions about upcoming genetic counseling visits or scheduling, please email us at genetics@karmanos.org or call 313-576-8748.

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