Identifying Patients with Colorectal Cancer

CASE EXAMPLE

Who is the Patient?

- 48 year old male patient
- Colorectal cancer diagnosed at age 47
- Ashkenazi Jewish descent
- History of mixed polyps in last colonoscopy

What is the Family History?

- Sister with adenomatous polyps at 38

PATERNAL FAMILY HISTORY

- Father with colorectal cancer at 49 and history of polyps
- Uncle with colorectal cancer at age 52
- Grandmother with colon cancer at age 52
- Grandfather with prostate cancer at 50

What Happened with Genetic Testing?

1. Initial provider ordered Lynch Syndrome genetic testing only which was negative.
2. A second healthcare provider ordered Ambry’s ColoNext, a 17 gene hereditary colorectal cancer panel.

Genetic Testing Criteria:
- Patient meets NCCN® genetic testing criteria for Lynch Syndrome testing

Genetic Test Results:
- Positive finding on ColoNext
- GREM1 pathogenic (disease-causing) variant
- Consistent with a diagnosis of hereditary colorectal cancer
HOW DID GENETIC TESTING IMPACT THE PATIENT AND FAMILY?

INCREASED LIFETIME CANCER RISKS
(see reverse for details)
- Colorectal

PERSONALIZED SCREENING AND PREVENTION OPTIONS
- Begin colonoscopy screening age 25-30

IMPACT FOR THE PATIENT’S FAMILY
- Siblings and children have a 50% chance of having the same GREM1 mutation and should be tested for the familial mutation
- Need to determine which side of the family is at-risk, so other relatives, such as aunts/uncles and cousins, can be tested for the familial mutation

What is GREM1?

GREM1 is a gene that helps protect our cells from turning cancerous. Germline (hereditary) mutations in this gene have been associated with an increased lifetime risk for colorectal cancer and polyps. Research is ongoing to further our understanding of the cancer risks for individuals with a GREM1 mutation.

POINTS FOR YOUR PRACTICE

- Negative results from germline Lynch Syndrome genetic testing does not completely rule out hereditary cancer for a patient with colorectal cancer
- Comprehensive multigene panel testing that includes Lynch syndrome genes as well as other colorectal cancer genes such as GREM1, POLD1, and POLE should be considered for a patient with personal and/or family history of colorectal cancer and/or polyposis
- Identifying if a patient has hereditary colorectal cancer allows informed, personalized healthcare decisions for that patient and his or her family members.

References