Ambry Genetics®
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# Identifying At-Risk Patients with CancerNext

## CASE EXAMPLE

## Who is the Patient?

- 33 year old female
- No personal history of cancer
- No reported history of colorectal polyps
- No previous genetic testing

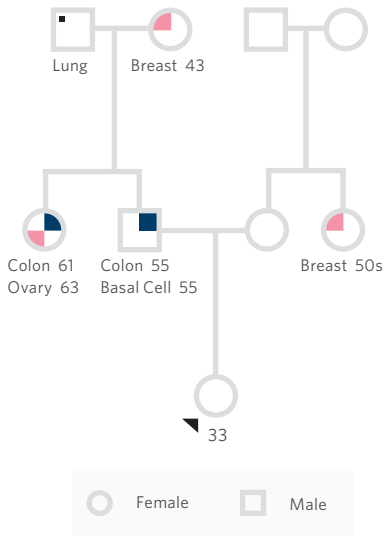
## What is the Family History?

### PATERNAL FAMILY HISTORY

- Father with colorectal cancer diagnosed at age 55 and basal cell skin cancer diagnosed at age 55
- Aunt with colorectal cancer diagnosed at age 61 and ovarian at age 63
- Grandmother with breast cancer at age 43
- Grandfather with lung cancer at an unknown age

### MATERNAL FAMILY HISTORY

- Aunt with breast cancer in her 50s




## What Happened with Genetic Testing?

Provider ordered **Ambry's CancerNext, a 34-gene hereditary cancer panel**

### Genetic Testing Criteria:

- Patient meets NCCN® genetic testing criteria for *BRCA1/2*[1]
- Patient does not meet testing guidelines for Lynch or other colorectal cancer testing.[2]

### Genetic Test Results:

- Positive finding on CancerNext
- *MLH1* pathogenic (disease-causing) variant
- Consistent with a diagnosis of Lynch syndrome

## HOW DID GENETIC TESTING IMPACT THE PATIENT AND FAMILY?

### INCREASED LIFETIME CANCER RISKS
(see reverse for details)

- Colorectal
- Uterine
- Ovarian
- Other

### PERSONALIZED SCREENING AND PREVENTION OPTIONS

- Recommend colonoscopies every 1-2 years
- Consider TAH/BSO (post-childbearing)
- Possible EGD every 3-5 years

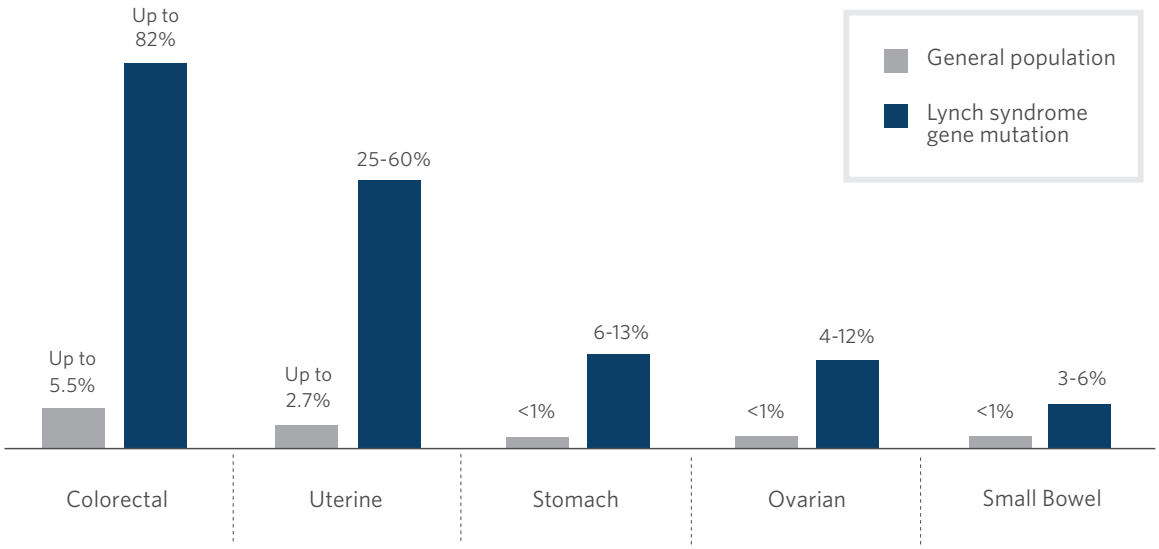
### IMPACT FOR THE PATIENT'S FAMILY

- Siblings and children have a 50% of having Lynch syndrome and should be tested for the familial mutation
- Parents can be tested to determine which side of the family is at-risk, then other relatives, such as aunts/uncles and cousins, can be tested for the familial mutation

## What is Lynch Syndrome?

Lynch syndrome is the most common hereditary form of colorectal cancer and it affects about 1 in 440 individuals in the U.S. It is caused by a defect in *MLH1*, *MSH2*, *MSH6*, *PMS2* or *EPCAM*. A mutation in one of these genes makes it difficult for the body to fix errors that occur in the DNA. This causes a significantly increased risk for multiple types of cancer, including colorectal, uterine, ovarian, stomach, small bowel, hepatobiliary tract, upper urinary tract, brain, pancreatic, and sebaceous cancers. Cancer risks may vary by gene.

## Lynch Syndrome Cancer Risks




**Lower risks suggested for MSH6 and PMS2 mutation carriers.*[1]

## POINTS FOR YOUR PRACTICE

This patient met criteria for **only** *BRCA1/2* testing. By ordering **CancerNext**, rather than only *BRCA1/2* testing, the healthcare provider was able to determine a diagnosis of Lynch syndrome that otherwise could have been missed.

Ambry's study of >34,000 individuals revealed that 22% of patients identified to have Lynch syndrome only met NCCN® guidelines for *BRCA1/2* genetic testing. Therefore, CancerNext may identify unexpected hereditary cancer syndromes.[3]

Identifying if a patient has Lynch syndrome allows you to guide informed, personalized healthcare decisions for that patient and his/her family members.

## References


1. National Comprehensive Cancer Network®. NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®). Genetic/Familial High-Risk Assessment: Breast and Ovarian. Version 2.2017
2. National Comprehensive Cancer Network®. NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®). Genetic/Familial High-Risk Assessment: Colorectal. Version 2.2016.
3. Espenschied C, *et al.* J Clin Oncol. April 2017.