Ambry Genetics

RESEARCH FOR YOUR PRACTICE

# Lynch Syndrome: It's more common than you think

## >1/4 WITH LYNCH SYNDROME ARE MISSED BY CURRENT GENETIC TESTING GUIDELINES

Ambry Genetics and The Ohio State University research of nearly 35,000 patients, published in the Journal of Clinical Oncology, expands how we think about genetic testing strategies, lifetime cancer risks, and medical management for people with Lynch syndrome.[1]

## WHY THIS MATTERS TO YOU

This study found that 27.3% of people with Lynch syndrome are missed by current genetic testing guidelines. Additionally, >25% of Lynch syndrome gene mutation carriers presented with breast or ovarian cancer as their first primary cancer.[1] As a result we recommend ordering a larger panel, such as CancerNext, to identify patients with mutations in *BRCA1/2*, in addition to Lynch syndrome.

## BACKGROUND

Lynch syndrome (LS) is estimated to occur in 1 in 440 people[2], but recent research suggests this should be as high as 1 in 279 – making LS the most common hereditary cancer syndrome.[3]

Genetic testing for LS is increasingly performed using multigene panels; results can impact cancer risk assessment and direct medical management, often following published guidelines.[4]

Many current LS guidelines are based on studies biased toward colorectal cancer and/or endometrial cancer, and many did not analyze all LS genes.[5-10] Basing LS genetic testing on a patient meeting Amsterdam criteria or Bethesda guidelines may miss as many as 72% and 27% of those with LS, respectively.[8] Screening for LS with microsatellite instability (MSI) and/or immunohistochemistry (IHC) for the mismatch repair (MMR) proteins may miss 13-23%.[8]

**34,890**

People Tested for Lynch Syndrome

## POINTS FOR YOUR PRACTICE

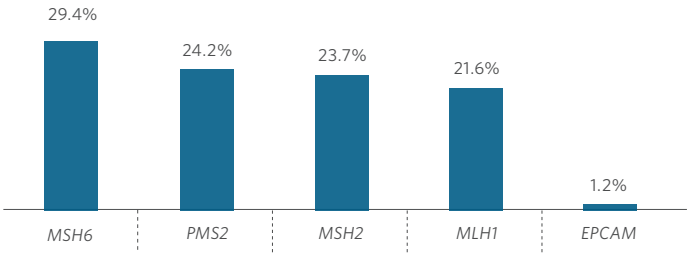
- This study reports on the largest group of individuals tested for and diagnosed with LS through multigene panel testing
- Current genetic testing guidelines may miss more than 1 in 4 people with LS
- Some women with LS may have a hereditary breast and ovarian cancer (HBOC) clinical presentation

## SIGNIFICANT FINDINGS

- *MSH6* and *PMS2* mutations were most commonly seen
- *MSH6* and *PMS2* mutation carriers were significantly more likely to just have breast cancer and only meet *BRCA1/2* NCCN® testing criteria
- Overall, 27.3% of people did not meet any current criteria for LS testing

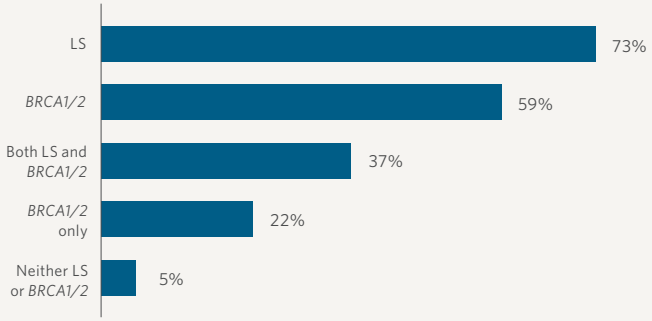
Learn more about our research here.

### Overall MMR and *EPCAM* Gene Distribution (n=579)




### How Current Genetic Testing Guidelines Stacked Up[4,11]

PATIENTS WHO MET NCCN® GENETIC TESTING GUIDELINES FOR:

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