

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 people², 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

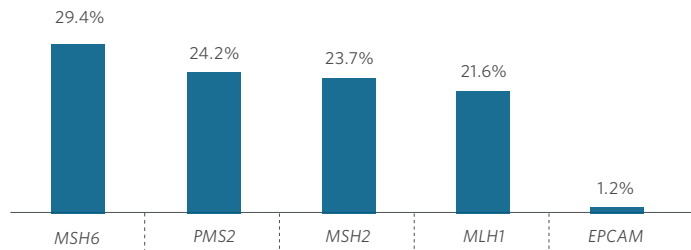
POINTS FOR YOUR PRACTICE

- 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

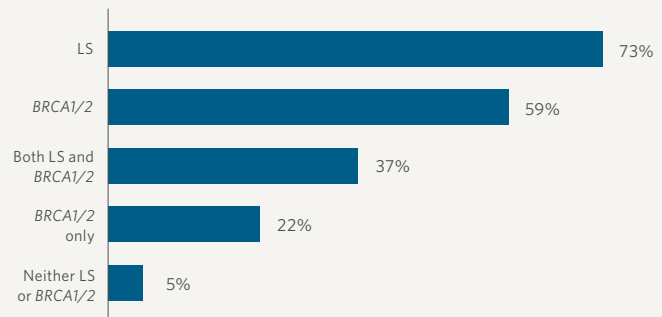
SIGNIFICANT FINDINGS

- *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

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:



Learn more about our research [here](#).

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