

## Division of Laboratory Medicine

### Cellular Pathology

# Histopathology - Mismatch Repair (MMR) testing

## Clinical Information

Identification of MMR can identify mistakes in the genes responsible for the mismatch repair system. This system is needed for ensuring that as cells grow and divide there are no errors in the process. If this does not work properly, errors (genetic mutations) can accumulate. This can lead to various health issues, including certain types of cancer.

**Mismatch Repair System:** The mismatch repair system includes several genes. These include MLH1, MSH2, MSH6, PMS2, and EPCAM. These genes contain the information to make (encode) proteins that find and fix mistakes in the DNA. This process stops errors (genetic mutations) from being passed down to new cells.

Lynch syndrome is a condition associated with MMR gene defects. Lynch syndrome is sometimes called hereditary nonpolyposis colorectal cancer, or HNPCC. People with Lynch syndrome have a higher risk for some types of cancer. These include colorectal cancer, endometrial cancer, ovarian cancer, and others.

How MMR Genetic Testing Works:

- The test involves analysing a person's DNA to look for mutations or changes in one or more of the MMR genes.
- This can be done using blood or tissue samples, such as biopsies.
- The test can help identify inherited mutations or somatic mutations (mutations that occur during a person's lifetime) associated with cancer.

### 1. Results of the Test:

- **Positive Test:** Indicates a mutation in one of the MMR genes, suggesting a potential increased risk for Lynch syndrome or other related cancers.
- **Negative Test:** No mutation is found, but it doesn't entirely rule out the presence of a risk (especially if there are other suspicious clinical signs).
- **Variants of Uncertain Significance (VUS):** Sometimes, genetic testing may reveal changes in a gene that are not yet well understood, and further studies may be needed.

### 2. Benefits of MMR Genetic Testing:

- **Helps assess the risk of developing cancer,** particularly for individuals with a family history of cancer.
- **Early Detection and Prevention:** For individuals with Lynch syndrome, regular screenings (such as colonoscopies) can be done to catch cancers early or even prevent them.
- **Guiding Treatment Choices:** Certain cancers with defective MMR (known as microsatellite instability, or MSI) may respond better to specific treatments, such as immunotherapy.

**(Last updated August 2025)**