



## Colorectal Cancer and Microsatellite Instability Testing (MSI)

### What is MSI testing?

MSI testing is done in a lab on a sample of colorectal tumour tissue that was removed during surgery, to determine whether **microsatellite instability** is present. A blood sample is also needed in order to compare to the tumour tissue results.

**Microsatellites** are repeated sequences of DNA (our genetic code, found on our chromosomes). Every person's DNA contains microsatellites; they are both common and normal. Each microsatellite has a specific number of times that it should repeat.

Microsatellite instability refers to abnormally long or short microsatellites in a sample of DNA. If the number of repeats is increased or decreased, the region of DNA is considered unstable. Microsatellite instability can be seen in both hereditary and non-hereditary forms of colorectal cancer.

### Why is MSI testing done?

MSI testing may be used as a screening test for Lynch syndrome (named after Dr Henry Lynch). **Lynch syndrome** (HNPCC) is the most common form of hereditary colorectal cancer, and accounts for up to 5% of all colorectal cancers. It is caused by an inherited mutation (mistake) in one of several genes. Being born with a Lynch syndrome gene mutation increases a person's risk for colorectal cancer and some other cancers.

When a person is diagnosed with colorectal cancer (especially before age 50), MSI testing can help to predict whether that cancer is related to Lynch syndrome.

### What do MSI results mean?

MSI results are reported as High (Unstable) or Stable. **Most results are MSI-Stable.**

An **MSI-High** result for a colorectal tumour suggests that further tests could be done. This result is a clue that a person *may* have been born with a Lynch syndrome gene mutation. A referral to the Hereditary Cancer Program is suggested. It is important to know that not all MSI-High cancers are hereditary.

An **MSI-Stable** result suggests that further testing is not needed. This result is unlikely to be seen in a person with a Lynch syndrome gene mutation.

This information was prepared by the BC Cancer Agency's Hereditary Cancer Program for the GI Tumour Group as a resource for patients and their physicians. April 2008