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