

# GENETIC TESTING FOR COLORECTAL CANCER

COLORECTAL CANCER IS THE THIRD MOST COMMON CANCER IN the world and ranks the top in Singapore. The local numbers have been escalating at an average annual rate of approximately 2.6 percent. This disease is also the second highest cause of cancer-related deaths, especially for Chinese males.

The chances of getting colorectal cancer increases with age and more than 75 percent of the cases are diagnosed after 50 years old. Affluent lifestyle, obesity, smoking, dietary changes and environmental pollution are some of the contributing factors. Genetic inheritance also plays an important role in the development of colon cancer. While we can control our diet and attempt to manipulate the external factors, our genetic make-up is determined at birth.

Colonoscopy and x-ray examinations are some of the available tests for diagnosing colorectal cancer. However these screening tests can only detect the presence of polyps or cancer, but does not predict one's risk of hereditary cancers.

Genetic testing has been advocated by international bodies for high-risk patients with strong family history of colorectal cancer. Ministry of Health Singapore recommends individuals or families suspected to have hereditary cancer syndromes to go for genetic risk evaluation and testing. This enables the identification of an entire family at risk of future cancers. It also allow individuals to better manage their lifestyle and frees the non-carriers from continuous anxiety.

The accuracy of genetic testing depends on many factors. It must be interpreted in the context of the person's medical history, family history and the methods used in the test. Targeted testing of a strongly suspected genetic syndrome is very accurate in determining the presence of the disease when a positive test is obtained.

Up to 30 percent of all colorectal cancers exhibit some degrees of familial inheritance. At least five to eight percent of these are inherited in an autosomal dominant fashion. The children of affected individuals carrying the mutated gene(s) have a 50 percent chance of inheriting the defective gene. Patients carrying these genes have extremely high risks of developing cancers of multiple organs, including the colon, rectum, uterus, ovaries, stomach, small intestine, liver, kidneys and the brain. Their estimated lifetime risk of colorectal cancers is 80 percent and the lifetime risk of uterine and ovarian cancer is 30 percent.

An individual should consider genetic testing if:

- There is a strong family history of three or more colorectal cancers affecting two or more generations, especially if one of the affected is a first degree relative



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(such as a parent or sibling)

- There is a personal or family history of colorectal cancer diagnosed before 50 years old
- You were found to have multiple polyps in the colon, rectum or the rest of the gastrointestinal tract
- You are from a family known to have hereditary conditions such as:
  - a) Familial Adenomatous Polyposis (FAP)
  - b) Lynch Syndrome (LS)
  - c) Pueritz-Jegher's Syndrome (PJS)
  - d) Cowden's Syndrome (CS)
- Your managing physician believes there is a possible hereditary component to your colorectal cancer

A specialist will give a detailed clinical consultation by assessing the person's medical history and family history. Blood samples will be sent to a specialized laboratory for analysis. Further testing may be necessary after the initial screening tests to confirm the diagnosis and very often, testing other affected and unaffected family members can help to further confirm these results.

Capstone Colorectal Surgery Centre is the only specialist colorectal clinic that provides comprehensive genetic counseling and genetic testing. The close collaboration with major research institutes allows them to research on the best methods for genetic diagnosis. This novel integrative approach is unique in South East Asia and takes colorectal cancer care to a new, personalized level. ■