

Recent advances in genetics to identify DNA changes directly linked to cancer development in CRC

Recent advances in genetic science are shifting the focus from detecting indirect signs of cancer (like blood in the stool) to identifying DNA changes directly linked to cancer development in colorectal cancer (CRC). These methods are collectively referred to as molecular or genetic biomarker tests, says Dr. Urvashi Bahadur, VP - Variant Science, Strand Life Sciences.

Genetic screening for early detection of CRC matters for India as the country faces unique challenges in detection and care. Rising incidence has been reported. CRC cases and mortality have been increasing over the past decade. There are low screening rates. Many people remain unscreened until late stages. A large proportion of CRC patients in India are under the age of 40 years, potentially pointing to lifestyle, genetic, and environmental influences. Accessibility and awareness remain barriers for many communities.

In India, CRC is growing rapidly and is now the fourth most common cancer overall, with more than 64,000 new cases and over 38,000 deaths reported in 2022. The heart of the challenge is that CRC often starts slowly and silently, with vague symptoms that are easy to overlook; this is why early detection can mean the difference between life and death.

Dr. Bahadur informs that genetic testing tools, especially when integrated with public health campaigns and awareness drives, have the potential to bridge gaps in early detection, reach more people, and reduce the burden of late-stage disease in CRC. Bringing genetic screening into broader public health efforts could also support data initiatives like the Bharat Cancer Genome Atlas and Indian Cancer Genome Atlas (ICGA), which aim to better understand genetic patterns specific to Indian populations and enable population-specific early detection strategies.

“Early detection doesn’t just save lives; it can make treatment easier and less costly. With CRC detected at a local stage, survival rates can be as high as 90%, compared to much lower survival when cancer is found late. Healthy lifestyle habits such as a balanced diet, regular exercise, maintaining a healthy weight, and avoiding tobacco and excessive alcohol complement these screening tools by lowering overall cancer risk. Genetic testing is not a replacement for medical advice, but it is rapidly becoming a powerful tool in modern cancer prevention and could transform how communities across India and the world approach CRC detection,” Dr. Bahadur adds.

Dr. Bahadur explains that advanced genetic tests include blood-based genetic tests. These tests analyze small fragments of DNA that tumours shed into the bloodstream. These include epigenetic markers, which are chemical changes in DNA that signal abnormal cell behavior of specific genes often present in CRC cells. Stool DNA test is another approach involves analyzing DNA in stool samples. Because CRC and precancerous polyps shed cells into the digestive tract, molecular tests can now detect cancer-associated genetic mutations and methylation patterns in feces. These tests have higher sensitivity than older stool blood tests and can detect cancer earlier.

Personalized screening for genetic risk profiling is now available. While most CRCs occur sporadically, a portion are influenced by inherited genetic factors. Certain individuals may carry gene variants that increase their lifetime risk of developing CRC, even if they have no symptoms. Advances in genomics are enabling the development of risk scores that combine information from many gene variants to estimate a person’s likelihood of developing CRC.

Today, genetic risk can be evaluated using targeted hereditary cancer gene panels that analyze genes known to be associated with colorectal cancer susceptibility, such as those involved in DNA repair pathways. Multigene panels analyze genes linked to hereditary colorectal cancer syndromes, including APC, MLH1, MSH2, MSH6, PMS2, MUTYH, PTEN, SMAD4, STK11, and others, which are associated with increased CRC risk are now available and can be used to guide screening and treatment.

Standard tools like colonoscopy and fecal occult blood tests (FOBTs) have long been the backbone of CRC screening. Although a colonoscopy is very accurate, it requires bowel preparation, a clinic visit, and sedation, which can be barriers for many people. Stool-based tests are easier, but they look for blood, which may not always be present in early-stage disease. Due to these limitations, participation in screening remains low even among at-risk groups in India, with colonoscopy uptake estimated below 10% in real-world settings.

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