Septin 9 (SEPT9) methylation analysis for colorectal cancer

Record Status
This is a bibliographic record of a published health technology assessment. No evaluation of the quality of this assessment has been made for the HTA database.

Citation

Authors’ conclusions
Colorectal cancer (CRC) is one of the leading causes of cancer-related deaths in the United States. It is estimated that approximately 143,000 individuals will be diagnosed with CRC in 2012, and more than a third will die from the disease. The 5-year survival rate for patients with cancer localized to the colon or rectum is approximately 90%, but it is less than 12% for those with distant metastases. It is well-known that screening for CRC allows for the early detection and removal of cancers or precancerous polyps, resulting in improved overall survival. The U.S. Preventive Services Task Force (USPSTF) currently recommends that individuals of average risk begin screening for CRC at age 50, using colonoscopy (the current reference standard for CRC diagnosis), flexible sigmoidoscopy, and/or a high-sensitivity fecal occult blood test (FOBT). However, it is estimated that approximately half of Americans age 50 and older do not adhere to the current screening recommendations. Because of poor compliance, recent studies have focused on the development of screening tools that may be considered more acceptable to eligible individuals who are not being screened. One recently developed screening test capitalizes on the fact that epigenetic changes contribute to the pathogenesis of CRC. Epigenetic changes are independent of DNA sequence and include modifications such as DNA methylation. DNA methylation typically involves the addition of a methyl group to cytosine nucleotides located adjacent to guanosines, which are referred to as CpG dinucleotides. Methylation of CpG dinucleotides in gene promoter regions is usually associated with transcriptional silencing and an absence of gene expression. The septin 9 (SEPT9) methylation assay is based on the finding that the majority of CRCs exhibit increased methylation of the SEPT9 gene. Methylation of SEPT9, which is located on chromosome 17 at band q25 and encodes a guanosine triphosphate-binding protein that functions in cell division, is detectable in cell-free DNA circulating in the bloodstream. This allows for CRC screening using patient blood samples, a method generally considered more appealing than stool-based or invasive tests.

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