Colorectal cancer (CRC) is one of the most common cancers in industrialized countries and affects men and women almost equally (1). The prognosis for a patient largely depends on the stage of the tumor at the time of diagnosis with the 5-year survival over 90% in cases where the cancer has not spread to the outer wall of the colon, but only 5% for stages where the cancer has spread outside the colon (2). CRC is a complex disease where environmental factors such as diet and lifestyle play an important role (3). Besides these factors, it has been shown that CRC occurs more frequently in certain families and a number of syndromes mainly with Mendelian dominant inheritance, which predispose to CRC development, have been described. The most common syndromes include hereditary non-polyposis CRC (HNPCC), familial adenomatous polyposis (FAP) and MUTYH associated polyposis.
CRC develops in a multistep manner over 10-15 years and the tumorigenic process covers a wide range of both premalignant and malignant lesions, such as hyperplastic polyps and adenomas. These lesions are well characterized both morphologically and genetically and can be easily detected and removed by colonoscopy. Even though CRC develops during a long period, carcinomas are usually recognised at advanced stages of tumor development and surgical resection can be performed in only about two thirds of patients (4). This situation is alarming because CRC is one of the most curable tumors when diagnosed early. Therefore, the identification of mutated gene carriers and people at risk is of major clinical importance.