



# Genetic Counseling for Hereditary Colorectal Cancer Syndromes: Empowering Patients through Early Detection and Prevention

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## INTRODUCTION

Colorectal Cancer (CRC) is one of the most prevalent cancers worldwide, with a significant portion attributed to inherited genetic mutations. Among the various forms of CRC, hereditary colorectal cancer syndromes contribute to roughly 5-10% of all cases. Genetic counseling plays a pivotal role in identifying individuals at increased risk for these syndromes, providing personalized risk assessments, and guiding preventive strategies. This article explores the importance of genetic counseling in managing hereditary colorectal cancer syndromes, its benefits, and the approaches used to support affected individuals and their families.

## DESCRIPTION

### Understanding hereditary colorectal cancer syndromes

Hereditary colorectal cancer syndromes are caused by mutations in specific genes, which significantly increase an individual's risk of developing colorectal cancer and, in many cases, other types of cancers. The most common hereditary syndromes associated with CRC include Lynch syndrome (also known as hereditary nonpolyposis colorectal cancer), Familial Adenomatous Polyposis (FAP), MUTYH-associated polyposis, and Peutz-Jeghers syndrome. Each syndrome is characterized by distinct genetic mutations, cancer risks, and clinical manifestations, necessitating tailored counseling and surveillance approaches.

**Lynch syndrome:** Lynch syndrome is the most common hereditary CRC syndrome, accounting for approximately 3-5% of all CRC cases. It results from Mutations in Mismatch Repair (MMR) genes (*MLH1*, *MSH2*, *MSH6*, *PMS2*, and *EPCAM*), which lead to DNA replication errors and increased cancer risk. Individuals with Lynch syndrome are at a higher risk of CRC, as well as endometrial, ovarian, stomach, and other cancers.

**Familial Adenomatous Polyposis (FAP):** FAP is characterized by hundreds to thousands of colorectal polyps and is primarily

caused by mutations in the *APC* gene. Left untreated, these polyps will almost inevitably progress to cancer. Attenuated FAP, a milder form, presents with fewer polyps but still carries a significant cancer risk.

**MUTYH-Associated Polyposis (MAP):** Caused by mutations in the *MUTYH* gene, MAP is an autosomal recessive condition that leads to the development of multiple adenomas in the colon and an elevated risk for CRC.

**Peutz-Jeghers Syndrome (PJS):** PJS, associated with mutations in the *STK11* gene, causes distinctive pigmented spots on the skin and mucous membranes along with hamartomatous polyps in the gastrointestinal tract. Individuals with PJS face an increased risk of several cancers, including CRC, breast, pancreatic, and ovarian cancers.

### The role of genetic counseling in hereditary CRC syndromes

Genetic counseling is essential for individuals at risk of hereditary CRC syndromes, providing education on their condition, assessing risk for family members, and guiding clinical decision-making. Genetic counseling typically involves gathering a detailed family history, assessing individual risk based on genetic and clinical factors, discussing the benefits and limitations of genetic testing, and interpreting test results.

**Risk assessment and family history analysis:** Counselors collect a detailed family medical history, paying close attention to patterns of CRC and related cancers across generations. Identifying cases of early-onset cancer, multiple cancers in a single individual, or clustering of cancers in close relatives can indicate a hereditary cancer syndrome. Family history analysis allows counselors to determine if genetic testing is appropriate and to select the specific tests that may be most informative.

**Genetic testing and interpretation:** Once a potential hereditary syndrome is suspected, genetic testing is recommended to confirm or rule out the presence of specific gene mutations. Genetic counselors are critical in interpreting these test results,

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which can be complex. For instance, a positive result may indicate a pathogenic mutation linked to an increased cancer risk, while a negative result may suggest that no known mutations were detected. In cases of variants of uncertain significance, counselors provide context, explaining that while a mutation is present, its impact on cancer risk is not fully understood.

**Education and informed decision-making:** Genetic counseling sessions empower patients and their families with the knowledge needed to make informed choices about their healthcare. Patients are educated about the inheritance patterns, risks, and prevention strategies associated with their syndrome. For example, individuals with Lynch syndrome may benefit from regular colonoscopies starting at an early age, typically every one to two years, to catch potential cancers early. Counseling also addresses preventive measures, including the option of risk-reducing surgeries or medications.

**Psychosocial support:** Genetic counseling offers a supportive environment where patients can explore the emotional and psychological impacts of a hereditary cancer syndrome diagnosis. Concerns about cancer risks, the possibility of passing the syndrome on to future generations, and the psychological burden of regular surveillance are common topics. Genetic counselors provide a safe space for individuals and families to discuss these challenges, often connecting them to additional resources such as support groups or mental health professionals.

### Prevention and surveillance strategies

Through genetic counseling, patients are introduced to surveillance and preventive strategies tailored to their specific syndrome and individual risk profile. For instance:

**Lynch syndrome:** Frequent colonoscopies and screenings for other cancers associated with Lynch syndrome are advised. Prophylactic surgeries, such as hysterectomy and oophorectomy, may be recommended for women with a high risk of gynecologic cancers.

**FAP and MAP:** Individuals with FAP or MAP benefit from early and frequent colonoscopic surveillance, with a possibility of colectomy if polyp burden becomes high.

**Peutz-jeghers syndrome:** Patients undergo regular endoscopic screenings and imaging for cancers in the gastrointestinal tract and other organs.

## CONCLUSION

Genetic counseling for hereditary colorectal cancer syndromes is a critical component of modern cancer care, offering at-risk individuals the opportunity to understand their genetic predisposition and take proactive steps toward cancer prevention and early detection. By combining education, risk assessment, and psychosocial support, genetic counseling empowers patients to make informed healthcare decisions, ultimately reducing the impact of these hereditary cancer syndromes on individuals and families.