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The Genetic Counselor

An Important Surgical Ally in the Optimal Care of the Cancer Patient

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Key Points
- Genetic counselors are healthcare providers specialized in the risk assessment and genetic evaluation of the cancer patient.
- Genetic counseling and genetic testing may lead to surgical-based implications for the cancer patient.
- The genetic counselor and surgeon may work together in multiple settings to enhance patient care.

With increased knowledge and greater availability of genetic testing for hereditary cancer predisposition syndromes, the role of the surgeon has further expanded. Often, surgeons are the initial treatment providers to patients recently diagnosed with cancer. This position provides the opportunity and responsibility to look beyond a patient's current disease treatment plan and consider possible future risks to patients and possible risks to their family members. Genetic counselors, equipped with specialized knowledge and skills in risk assessment and management of patients with hereditary cancer predisposition syndromes and their at-risk family members, are important allies to surgeons managing individuals and families afflicted with a hereditary cancer syndrome. Genetic counseling has been incorporated into published practice guidelines [1–4] and is now a standard recommendation for patients at increased risk for a hereditary cancer predisposition syndrome. A partnership between surgeons and genetic counselors provides a unique opportunity to identify, assess, and manage at-risk patients and their

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family members in an optimal manner. This article discusses in detail the role of a genetic counselor, multiple settings for genetic counseling, and opportunities for coordination of care between a surgeon and genetic counselor. Although focused on patients with various forms of hereditary colorectal cancer (CRC), other hereditary cancer syndromes are discussed briefly and readers are referred to primary source documents for greater detail. Table 1 provides basic details about a few hereditary cancer predisposition syndromes. Surgeons are urged to recognize the importance of identifying and collaborating with a local genetic counselor because this relationship sets the stage for optimal care of cancer patients.

DEFINITION OF GENETIC COUNSELING AND GENETIC COUNSELORS

In 2005, the National Society of Genetic Counselors defined genetic counseling as the "process of helping people understand and adapt to the medical, psychological, and familial implications of genetic contributions to disease. This process integrates: (1) the interpretation of family and medical histories to

Table 1
Hereditary cancer predisposition syndromes—basic details

Syndrome name	Associated genes	Mode of inheritance	Associated tumors	References
Lynch syndrome/ HNPCC (hereditary non-polyposis colorectal cancer)	*MLH1* *MSH2* *MSH6* *PMS2* *EPCAM*	Autosomal dominant	Colorectal, endometrial, gastric, ovarian, urothelial, small bowel, biliary tract, pancreas, brain, sebaceous gland neoplasm	[32,36,43,44]
Familial adenomatous polyposis (FAP) & attenuated FAP	*APC*	Autosomal dominant	Colorectal, gastric, small bowel, thyroid, pancreas, hepatoblastoma	[43,45,46]
MYH-associated polyposis (MAP)	*MYH*	Autosomal recessive	Colorectal, gastric, small bowel	[45,47,48]
Hereditary breast and ovarian cancer syndrome	*BRCA1* *BRCA2*	Autosomal dominant	Breast, ovarian, prostate, pancreas	[7,43,49,50]
Hereditary diffuse gastric cancer (HDGC) syndrome	*CDH1*	Autosomal dominant	Diffuse gastric, lobular breast, signet ring cell colorectal	[13]
Multiple endocrine neoplasia (MEN)-2A, MEN-2B, and familial medullary thyroid carcinoma (FMTC)	*RET*	Autosomal dominant	Medullary thyroid, pheochromocytoma (only in MEN-2A and MEN-2B)	[43,51,52]