

HOSPITAL PHYSICIAN®

GENERAL SURGERY BOARD REVIEW MANUAL

STATEMENT OF EDITORIAL PURPOSE

The *Hospital Physician General Surgery Board Review Manual* is a study guide for residents and practicing physicians preparing for board examinations in general surgery. Each manual reviews a topic essential to current practice in the specialty of general surgery.

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Inherited Cancer Syndromes

Series Editor and Contributor:

C. Neil Ellis, MD

System Chair, Division of Colon and Rectal Surgery, West Penn Allegheny Health System, Pittsburgh, PA

Contributor:

Philip A. Cole II, MD, MS

Department of Surgery, University of South Alabama, Mobile, AL

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Inherited Cancer Syndromes


Philip A. Cole II, MD, MS, and C. Neil Ellis, MD

INTRODUCTION

Efforts to improve the outcomes of patients with cancer have focused primarily on prevention and early detection. These efforts will be most beneficial for individuals who are at the highest risk. Thus, it is imperative that physicians become familiar with the tools to assess a person's genetic risk for cancer, the general benefits and limitations of genetic testing in clinical practice, and the process of making informed decisions about the use of the information obtained. This manual highlights the diagnosis, evaluation, and management of patients with the most common hereditary cancer syndromes.

HEREDITARY NONPOLYPOSIS COLON CANCER

CASE PATIENT I

 A 30-year-old woman presents to her primary care physician complaining of rectal bleeding. She is very concerned because multiple family members have had colorectal polyps and cancers. History and physical examination performed during the initial clinic visit reveal a concerning family medical history but no other abnormalities.

FAMILY CANCER HISTORY

Obtaining a detailed family medical history of cancer is an essential step in identifying patients at high risk for hereditary cancer syndromes.¹ However, many physicians do not obtain a sufficiently detailed family history of cancer. Moreover, it is not uncommon for physicians to misinterpret or fail to act on the information provided.²⁻⁷ Although patients are becoming more knowledgeable about their medical conditions, on presentation many will not know they belong to a family carrying a gene that predisposes them to cancer. Rather, it is the responsibility of physicians to identify patients at high risk for hereditary cancer syndromes.

Most hereditary cancer syndromes involve different types of cancers that affect multiple generations. In general, the risk that a patient is a member of a

high-risk hereditary cancer syndrome family increases as the number of generations and individuals with cancer increases and the age of the affected individuals decreases. One of the hallmarks of hereditary cancer is early age of onset. A history of cancers that appear in family members under the age of 50 years signals the possibility of an inherited cancer syndrome.

Gleaning the necessary information requires a structured approach to obtaining a family medical history of cancer. The most common method is to obtain the family cancer history at the time of the initial office encounter. However, patients rarely recall the detailed information necessary to adequately assess the possibility of an inherited cancer syndrome. A two-step process provides the most accurate information. During the initial office visit, the physician gathers a brief family history of cancer, explains the need to obtain more detailed information, and gives the patient a structured family cancer history questionnaire to complete after discussion with other family members. This method allows the patient the opportunity to contact family members and check medical or death records, increasing the completeness and accuracy of the information obtained. The questionnaire is subsequently returned to the physician.

The validity of this approach to collecting a family medical history of cancer and the accuracy of the information reported by patients appear good.⁸⁻¹⁴ In one study, 83% of patients correctly identified the primary cancer site in their first-degree relatives.⁹ The reporting site was accurate in 67% of second-degree relatives and 60% of third-degree relatives. The rate of false-positive results was 5%. In another study, the accuracy of cancer reports in first- and second-degree relatives was 91% and 74%, respectively.¹⁰ A mistake in identifying the presence or site of the cancer was found in only 4% of first-degree and 15% of second-degree relatives. Although a small amount of over-reporting by breast cancer patients of breast cancer in their families has been observed, the accuracy of reporting in first-degree relatives was 90%.¹² The false-negative reporting has been observed to vary by tumor site and to be greater in men, individuals of nonwhite race, and older patients. Other variables influencing false-negative reporting include time since cancer diagnosis, number of previous tumors, and type