Inherited Cancer Syndromes
Inherited Cancer Syndromes
Current Clinical Management
The goal in preparation of this text was to provide a source of information about the diagnosis, evaluation, and management of inherited cancer syndromes. Two decades ago, these syndromes were thought to be extremely rare, if they existed at all. Over the last 10 years, it has become apparent that 20% of patients with breast, colorectal, or gynecologic cancer will have a family history suggestive of one of these syndromes, and 5% will have the condition. The ramifications of having one of these diseases are significant for both the patient and their family, with a high risk of developing a malignancy in many organs at an early age.

Clinicians should be prepared to recognize and manage these diseases. This is not as simple as it sounds. While data derived from the technological advances in genetics can provide important information about the prognosis and possible manifestations of the disease, only in special circumstances can it be used to confirm or exclude the diagnosis. Instead, the diagnosis is made from the clinical information obtained by examination of a patient’s family medical history. Despite the best efforts of the patient and their clinician the pedigree developed frequently has omissions and inaccuracies.

Another issue is the lack of consensus regarding the diagnostic criteria for these conditions. All of the currently accepted guidelines have been criticized, leaving the clinician confused as to who should be diagnosed with one of these syndromes. Avoidance of false-positive and -negative diagnosis is of particular importance given the implications of these diagnoses for patients and their families.

After the diagnosis is suspected or confirmed, the management of these diseases is also controversial. Treatment options can include aggressive cancer screening, preventive medications, and/or prophylactic surgery. There are few randomized, controlled trials showing the superiority of one strategy over another for patients with an inherited predisposition for cancer. Management of these patients is therefore based on clinical judgment, taking into account the clinical manifestations of these syndromes and the risks and limitations of the various therapeutic options.
It is hoped that this text will provide a source of information that is useful to the clinician in the recognition and management of these syndromes, and in the prevention of the suboptimal outcomes frequently associated with these diseases.

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