Preface

The majority of colorectal cancers arise from sporadic polyps which are likely secondary to a combination of underlying acquired genetic mutations potentiated by environmental exposures. However, approximately 5% of people who develop colorectal cancer will have an underlying hereditary colon cancer syndrome that is associated with an increased polyp burden. Classification of a patient into a specific polyposis syndrome is based on both clinical features and when possible genetic test results, yet determining the precise diagnosis is no small task, given the complexity of the histological variations and clinical features that may overlap between these syndromes. Similarly, the risks for extracolonic polyps or cancers as well as the risk for the development of colorectal cancer necessitate an extensive knowledge of how best to diagnose and manage polyp and cancer screening and surveillance for these patients.

This book outlines the known hereditary polyposis syndromes including familial adenomatous polyposis (FAP) syndrome, attenuated FAP, MUTYH-associated polyposis, juvenile polyposis syndrome, Peutz–Jeghers syndrome, hereditary mixed polyposis syndrome, inflammatory cap polyposis, PTEN hamartoma syndrome, serrated polyposis syndrome, polymerase proofreading-associated polyposis, and several other newly identified polyposis syndromes of hereditary colorectal cancer. Though not known to be hereditary, Cronkhite–Canada syndrome is also included as an intestinal polyposis condition that is associated with an increased risk for colorectal cancer. The first chapter of the book is an overview of these intestinal polyposis syndromes, demonstrating the degree of overlap among polyp histologies and cancer risks while highlighting the diversity of these conditions. Following this overview, a chapter is dedicated to each of the known polyposis syndromes and outlines the clinical features that are associated with the polyps, the histologic features of these polyps, the risk for colorectal and extraintestinal malignancies, the known molecular genetic mechanisms that lead to the development of the polyps and likely the associated malignancies, cancer surveillance and screening recommendations, and, when available, chemopreventive therapies.

The future for those who have an intestinal polyposis syndrome and those who care for them will evolve as these intestinal polyposis syndromes are more
meticulously categorized. Ultimately, the molecular etiologies and even the definition of how few polyps constitute a polyposis syndrome will be expanded as whole genome and other “omics” technologies are applied to all patients who have developed polyps and/or colorectal cancer. In fact, during the development of this book, four new germline genetic mutations were found to cause intestinal polyposis.

Colorectal cancer screening has proved invaluable, and, in fact, the incidence of colorectal cancer in the general population has decreased with the use of colonoscopy and polypectomy. This is particularly useful in some of the polyposis syndromes, but, unfortunately, for patients with polyps that cannot be managed endoscopically, the need for surgery continues to be a necessity. The long-term hope for patients with polyposis syndromes may be met by further development of chemopreventive agents directed at genetically relevant targets, more tailored screening and surveillance programs, and possibly methodologies for gene editing or correction of genetic defects that may ultimately help these patients avoid the need for invasive surgeries. In the meantime, our intention is to provide a clear, comprehensive guide for recognition and management of individuals with polyposis syndromes.

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