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Recent progress in Lynch syndrome and other familial colorectal cancer syndromes

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Abstract

The current understanding of familial colorectal cancer was limited to descriptions of affected pedigrees until the early 1990s. A series of landscape-altering discoveries revealed that there were distinct forms of familial cancer, and most were related to genes previously not known to be involved in human disease. This review largely focuses on advances in our understanding of Lynch syndrome because of the unique relationship of this disease to defective DNA mismatch repair and the clinical implications this has for diagnostics, prevention, and therapy. Recent advances have occurred in our understanding of the epidemiology of this disease, and the advent of broad genetic panels has altered the approach to germline and somatic diagnoses for all of the familial colorectal cancer syndromes. Important advances have been made toward a more complete mechanistic understanding of the pathogenesis of neoplasia in the setting of Lynch syndrome, and these advances have important implications for prevention. Finally, paradigm-shifting approaches to treatment of Lynch-syndrome and related tumors have occurred through the development of immune checkpoint therapies for hypermutated cancers. CA Cancer J Clin 2018;68:217-231. © 2018 American Cancer Society.

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