

### **New Insights into Lynch Syndrome: A Narrative Review**

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### **Abstract**

Lynch syndrome, characterized by DNA mismatch repair deficiency, represents a significant paradigm among cancer predisposition syndromes and is notably associated with heightened susceptibility to various cancers, particularly colorectal and endometrial malignancies. The primary aim of this research paper is to scrutinize specific associations and delve into the underlying molecular mechanisms of Lynch syndrome. Genetic alterations in MMR genes, including MLH1, MSH2, MSH6, PMS2, and EPCAM, compromise DNA repair mechanisms, predisposing affected individuals to a spectrum of malignancies. This paper comprehensively investigates current screening methodologies and preventive measures tailored for individuals identified or at risk of Lynch syndrome. The integration of advanced sequencing technologies and refined bioinformatics tools has significantly improved mutation detection accuracy, facilitating precise identification of mutation carriers and their at-risk relatives. Moreover, this review emphasizes the evolving diagnostic landscape, which have revolutionized the identification of potential mutation carriers. The structured diagnostic algorithm, incorporating clinical criteria, tumor testing, and genetic analysis, plays a pivotal role in systematically identifying and managing individuals with Lynch syndrome. While the well-established association of Lynch syndrome with colorectal and endometrial cancers is recognized, emerging evidence suggests an increased risk for other types of malignancies. A crucial aspect of this literature review is to extensively analyze the less commonly acknowledged correlation between Lynch syndrome and prostate or testicular malignancies. Understanding these correlations holds significant importance in guiding tailored screening protocols and preventive strategies for individuals carrying Lynch syndrome-associated genetic mutations. The comprehensive assessment of this diverse spectrum of cancers underscores the necessity for tailored surveillance strategies and multidisciplinary approaches to effectively manage and mitigate risks in individuals harboring Lynch syndrome-associated genetic alterations.

**Key words:** Lynch syndrome, MMR, germline testing, carcinogenesis, colorectal cancer