ATTITUDES TOWARD UPDATED GENETIC TESTING IN PATIENTS WITH
UNEXPLAINED MISMATCH REPAIR DEFICIENCY

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Individuals who have colorectal cancer (CRC) or endometrial cancer (EC) displaying loss of immunohistochemical (IHC) staining of one or more mismatch repair (MMR) proteins without a causative germline mutation are said to have unexplained mismatch repair deficiency (UMMRD, also known as mutation-negative Lynch syndrome). Comprehensive genetic testing that could potentially further clarify Lynch syndrome carrier status is essential to provide tailored screening guidelines to affected individuals and their family members; however, patient understanding of the potential impact of updated genetic testing for LS is unclear. This study aimed to evaluate the interest in and perceived impact of updated genetic testing among individuals with UMMRD at a tertiary academic center. A survey evaluating interest in updated genetic testing was mailed to 98 potential participants, and an electronic health record review was completed for the 31 individuals who returned the survey. Results indicate that this population is highly interested in updated genetic testing, and their perceived impact is primarily for family members to have appropriate testing and screening options. Updated risk assessment and genetic counseling, along with a discussion of the benefits and limitations of genetic testing, will allow patients with UMMRD to better understand the impact of comprehensive genetic testing for themselves and their family members.