Genetic Counselor Utilization and Interpretation of Somatic Tumor Testing in Evaluation for Lynch Syndrome

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Lynch syndrome (LS) is a hereditary cancer predisposition syndrome characterized by increased risk for colorectal and uterine cancers. Individuals with pathogenic variants in the mismatch repair (MMR) genes (MLH1, MSH2/EPCAM, MSH6, PMS2) are diagnosed with LS and subsequently recommended to proceed with high risk screening protocols to increase prevention and early detection of LS-related cancers. Various tumor studies can help identify those at high risk for LS, but sometimes create uncertainty with discordant screening and germline results, leading to unexplained mismatch repair deficiency (UMMRD). Somatic testing of the MMR genes has created opportunities for resolving UMMRD, thus clarifying LS status and ensuring appropriate cancer surveillance. However, guidelines for such testing are currently limited. The purpose of this study was to examine current and hypothetical ordering practices of cancer genetic counselors for LS evaluation and to investigate participants’ interpretation of somatic MMR testing results. Two-hundred eligible participants were recruited through the National Society of Genetic Counselors listserv and answered questions regarding demographics, ordering practices, barriers to somatic MMR testing, theoretical patient scenarios, and need for further guidelines. Statistical analysis was done using Chi-square, Fisher exact, and Wilcoxon rank-sum tests while themes were identified from free-text responses. Most respondents did not include somatic MMR testing in the work-up for LS and did not routinely order this testing, but indicated interest in ordering this in conjunction with germline testing. The gap between preferred testing strategies and current ordering practices for somatic MMR testing may be due to reported laboratory and insurance-related barriers, particularly cost and coordination of tissue specimens. Nearly all individuals endorsed the need for additional guidelines for somatic MMR testing, which could provide support to reduce barriers, encourage insurance coverage, and allow for appropriate screening recommendations for patients and family members of those with UMMRD.

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