Pinpointing the prevalence of mismatch repair (MMR)-deficient rectal adenocarcinomas

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A widely-circulated clinical trial published earlier this year in the *New England Journal of Medicine* (NEJM) described a new treatment that eradicated cancer in all 12 of its patients with locally advanced mismatch repair (MMR)-deficient rectal adenocarcinomas. Clinicians say the finding will spark a paradigm shift in the treatment of MMR-deficient rectal cancer, but there remains uncertainty about how many patients have that specific condition.

Researchers at the Brigham have analyzed a collection of 16,083 colorectal adenocarcinoma biopsies to determine how many patients would benefit from the promising new treatment. In a letter published in *NEJM*, they write that whereas the earlier clinical trial estimated that 5-10% of rectal cancer patients have the MMR-deficient subtype, their data suggest that 2.65% do.

They also observed that while younger patients appear to be at higher risk of developing MMR-deficient adenocarcinoma—a previously described phenomenon—an unexpectedly high number of older patients also had the condition, highlighting the importance of screening at all ages. The letter suggests that analysis of biopsy material is sufficient for MMR testing in 99.9% of cases.

"We have leveraged the volume of the largest gastrointestinal pathology group in the U.S. to study the prevalence of mismatch repair deficiency in colorectal adenocarcinoma," said lead author David J. Papke, Jr., MD, Ph.D., of the Pathology Department. "We used our study cohort to determine the prevalence of mismatch repair deficiency in rectal adenocarcinoma, thereby defining the population likely to benefit from this novel therapeutic approach."


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