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Colon cancer screening practices after genetic counseling and testing for hereditary nonpolyposis colorectal cancer.

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PURPOSE: Hereditary nonpolyposis colorectal cancer (HNPCC) is the most common hereditary form of colon cancer. Cancer screening recommendations differ between individuals identified to carry an HNPCC mutation and those who do not carry a known family mutation. We assessed the impact of genetic counseling and testing (GCT) on the use of endoscopic screening procedures and adherence to recommended endoscopic screening guidelines in 56 asymptomatic at risk individuals from families known to carry an HNPCC mutation. **PATIENTS AND METHODS:** We analyzed data on colonoscopy and flexible sigmoidoscopy screenings collected before GCT and 6 months and 12 months post-GCT on 17 mutation-positive and 39 true mutation-negative individuals. Main outcome measures were use of endoscopic screening and adherence to recommended guidelines for the relevant mutation status. Mutation status, age, sex, employment, and income were analyzed as predictor variables. **RESULTS:** Among mutation-negative individuals, use of colonoscopy and flexible sigmoidoscopy decreased significantly between pre- and post-GCT ($P < .00001$ and $P < .0003$, respectively). Among mutation-positive individuals, a nonsignificant increase ($P = .24$) in use was noted. Age was also associated with use of endoscopic screening after GCT ($P = .03$). Mutation status (odds ratio [OR], 7.5; $P = .02$) and employment (OR, 8.6; $P = .025$) were associated with nonadherence to endoscopic screening guidelines. More mutation-negative individuals strictly adhered to guidelines than did mutation-positive individuals (87% v 65%). **CONCLUSION:** Genetic counseling and testing for HNPCC significantly influences the use of colonic endoscopy and adherence to recommendations for colon cancer screening.