Genome Medicine

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Predictors of patient uptake of colorectal cancer gene environment risk assessment

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Abstract

Background: In an ongoing clinical trial, the genetic and environmental risk assessment (GERA) blood test offers subjects information about personal colorectal cancer risk through measurement of two novel low-to-moderate risk factors. We sought to examine predictors of uptake of the GERA blood test among participants randomized to the Intervention arm.

Methods: Primary care patients aged 50 to 74 years eligible for colorectal cancer screening are randomized to receive a mailed stool blood test kit to complete at home (Control) or to the control condition plus an in-office blood test called GERA that includes assessment of red blood cell folate and DNA-testing for two *MTHFR* (methylenetetrahydrofolate reductase) single nucleotide polymorphisms (SNPs) (Intervention). For the present study, baseline survey data are examined in participants randomized to the Intervention.

Results: The first 351 intervention participants (161 African American/190 white) were identified. Overall, 249 (70.9%) completed GERA testing. Predictors of GERA uptake included race (African American race, odds ratio (OR) 0.51 (0.29 to 0.87)), and being more knowledgeable about GERA and colorectal cancer screening (OR 1.09 (1.01 to 1.18)). Being married (OR 1.81 (1.09 to 3.00)) was also significant in the multivariable model.

Conclusions: Participant uptake of GERA testing was high. GERA uptake varied, however, according to socio-demographic background and knowledge.

Background

Colorectal cancer (CRC) is the third most common cancer of US men and women, with approximately 150,000 new diagnoses in 2012 [1]. While a small number of CRCs are known to be caused by mutations in high penetrance cancer genes such as those associated with familial adenomatous polyposis or Lynch syndrome, most cases of CRC appear to be sporadic, and likely arise from risks associated with both low penetrance genes and environmental risks such as dietary or toxin exposures. Colonoscopy screening in adults is proven to lower the risk of developing CRC and is endorsed by the US Preventive Services Task Force [2]. Despite recent increases in general US population screening, improvements are still needed. Screening rates among underserved populations continue to lag behind those of white Americans [3]. Improving CRC screening rates remains a national health care goal [2,3].

Experts have hypothesized that providing personalized genetic susceptibility feedback may serve as an important link between public health goals and individual motivation to engage in healthy behaviors such as cancer screening [4,5]. A number of SNPs associated with generally modest (5 to 20%) increases in cancer risk have been identified, and several studies have to date examined the impact of genetic susceptibility feedback on health behaviors either through hypothetical scenarios or through offering single-gene or so-called multiplex genetic testing [6-9]. In adult populations both at increased risk for cancer and unselected for cancer risk, interest in genetic susceptibility feedback is generally high, and experts have supported a potential for large impact on prevention behaviors [4,5]. A recent meta-analysis of the impact of genetic susceptibility feedback on smoking and physical activity outcomes, however, suggested limited effectiveness [10].

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