# Assessing genetic screening barriers for hereditary breast and ovarian cancer in a high-risk Minnesota population

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Hereditary mutations in BRCA1/2, TP53, PTEN, and other genes underlie up to 10% of all breast and ovarian cancers and significantly increase carriers' lifetime risk of cancer development by up to 5 times (breast) and 20 times (ovarian) that of the general population. Multiple organizations, including the National Comprehensive Cancer Network (NCCN), American College of Obstetricians and Gynecologists (ACOG), and Society of Gynecologic Oncologists (SGO), advise that people at high risk of hereditary cancers be identified via careful collection of personal and family cancer history. It is further recommended that eligible individuals be referred to genetic counseling with a qualified provider, followed by genetic testing to identify high-risk mutations if indicated.

Correct identification of pathogenic mutations is crucial for informed patient decision-making about cancer prevention steps including increased screening, preventive chemotherapy, and risk-reducing surgery such as bilateral mastectomy or salpingo-oophorectomy. Mutation identification is also important for at-risk family members to undergo genetic evaluation if desired. Our aim was to describe patient-perceived barriers to genetic counseling and testing among people at high risk for hereditary cancers as defined by nationally accepted criteria.

## Methods

Following University of Minnesota Institutional Review Board approval, attendees of the 2019 Minnesota State Fair were asked to take a short web-based survey at the University of Minnesota's Driven to Discover (D2D) Research Building. Questions included participant demographics, experience with healthcare providers, and personal and family history of cancer. Individuals meeting criteria for cancer genetic counseling (GC) referral were identified by branching logic and prompted to answer additional questions about their history of GC referral and uptake. Demographic and clinical measures were summarized using mean (SD), range, frequencies, and percentages, and compared between relevant groups using t- and Chi-square tests. Statistical significance was set at $p<0.05$.

## Results

Some 1,594 fairgoers participated, and 673 (42%) met GC referral criteria. Of this group, 184 (27%) knew their high-risk (HR) status. Participants were more likely to know their status if they identified a provider as "their doctor" (83.6% vs 75.4%, p=.023) or had discussed family history with their provider (94.5% vs 82.4%, p<.001). All 91 HR participants (13.6%) referred to GC underwent genetic testing, but only 49 (52%) completed the GC referral. The most common reasons HR participants cited for not receiving GC referral were a doctor not telling them they were at high risk (294 HR participants, 43.7%) or a doctor not taking a family history (56 HR participants, 8.3%).

## Conclusions

In this large survey study, most participants at high risk for hereditary cancers were unaware of their status and eligibility for cancer genetic screening. Contrary to published evidence that physicians and genetic counselors consider patients generally uninterested in genetic screening, our findings highlight the key role physicians play in ensuring high-risk patients have appropriate access to genetic services including counseling, testing, and treatment. Improved family history collection and patient awareness of hereditary cancers, in addition to accessible physician education on genetic referral guidelines in locations where genetic counselors are available, are all viable strategies to aid in the identification and referral of high-risk individuals. **MM**

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