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Summary of the experiences, knowledge, medical management, and family communication of monoallelic *MUTYH* carriers

Danielle B. McKenna[1], **Pauleen Sanchez**[1], **Jacquelyn Powers**[1], **Jamie Brower**[1], **Louise Wang**[2], **Rebecca Mueller**[1], **Heather Symecko**[1], **Jada G. Hamilton**[3,4,5], **Temima Wildman**[4], **Susan M. Domchek**[1], **Fergus J. Couch**[6], **Judy E. Garber**[7], **Kenneth Offit**[4,5], **Mark E. Robson**[4,5], **Bryson W. Katona**[2]

[1]Basser Center for BRCA, Abramson Cancer Center, University of Pennsylvania, Philadelphia, Pennsylvania, USA

[2]Division of Gastroenterology and Hepatology, Department of Medicine, University of Pennsylvania Perelman School of Medicine, Philadelphia, Pennsylvania, USA

[3]Department of Psychiatry & Behavioral Sciences, Memorial Sloan Kettering Cancer Center, New York, New York, USA

[4]Department of Medicine, Memorial Sloan Kettering Cancer Center, New York, New York, USA

[5]Weill Cornell Medical College, New York, New York, USA

[6]Department of Laboratory Medicine and Pathology, Mayo Clinic, Rochester, Minnesota, USA

[7]Department of Medical Oncology, Dana-Farber Cancer Institute, Boston, Massachusetts, USA

Correspondence Bryson W. Katona, Division of Gastroenterology and Hepatology, Department of Medicine, University of Pennsylvania Perelman School of Medicine, Philadelphia, Pennsylvania, USA. bryson.katona@pennmedicine.upenn.edu.

AUTHOR CONTRIBUTIONS

Danielle McKenna: Conceptualization; formal analysis; methodology; writing – original draft; writing – review and editing. **Pauleen Sanchez:** Data curation; visualization; writing – original draft; writing – review and editing. **Jacquelyn Powers:** Conceptualization; formal analysis; methodology; writing – original draft; writing – review and editing. **Jamie Brower:** Data curation; project administration; writing – review and editing. **Louise Wang:** Data curation; formal analysis; writing – original draft; writing – review and editing. **Rebecca Mueller:** Conceptualization; writing – review and editing. **Heather Symecko:** Project administration; writing – review and editing. **Jada Hamilton:** Methodology; project administration; writing – review and editing. **Temima Wildman:** Project administration; writing – review and editing. **Susan Domchek:** Conceptualization; investigation; project administration; supervision; writing – review and editing. **Fergus Couch:** Investigation; project administration; writing – review and editing. **Judy Garber:** Investigation; project administration; writing – review and editing. **Kenneth Offit:** Investigation; project administration; writing – review and editing. **Mark Robson:** Investigation; project administration; writing – review and editing. **Bryson Katona:** Conceptualization; formal analysis; supervision; visualization; writing – review and editing. Authors Danielle McKenna, Pauleen Sanchez, Jacqueline Powers, Jamie Brower and Bryson Katona confirm that they had full access to all the data in the study and take responsibility for the integrity of the data and the accuracy of the data analysis. All of the authors gave final approval of this version to be published and agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

CONFLICT OF INTEREST

Authors Danielle McKenna, Pauleen Sanchez, Jacquelyn Powers, Jamie Brower, Louise Wang, Rebecca Mueller, Heather Symecko, Jada Hamilton, Temima Wildman, Susan Domchek, Fergus Couch, Judy Garber, Kenneth Offit, Mark Robson, and Bryson Katona declare that they have no conflict of interest.

HUMAN STUDIES AND INFORMED CONSENT

Approval to conduct this human subject research was obtained through a centralized institutional review board (Advarra IRB). All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000. Informed consent was obtained from all patients for being included in the study.

SUPPORTING INFORMATION

Additional supporting information can be found online in the Supporting Information section at the end of this article.

Abstract

Germline genetic testing for inherited cancer risk is increasingly being performed with multigene panel testing with *MUTYH* often included on colorectal cancer- and polyposis-focused panels, as well as on broader pan-cancer panels. With up to 1%–2% of the general population being monoallelic *MUTYH* carriers, pathogenic/likely pathogenic (P/LP) variants in *MUTYH* are one of the most common findings on multigene cancer panels. However, little is known about patient experience and understanding of monoallelic *MUTYH* P/LP variants, nor whether such findings influence medical management recommendations and familial communication, which this study aims to better understand. Monoallelic P/LP *MUTYH* carriers were recruited from the Prospective Registry of Multiplex Testing (PROMPT) and completed a cross-sectional self-report survey on sociodemographic characteristics, medical and family history, experiences with *MUTYH* genetic testing, genetics and *MUTYH* knowledge, perceived cancer risk, and familial communication. Of 115 eligible PROMPT participants, 49 (43%) completed the survey who were primarily female (94%), white (96%), had a history of cancer (61%), and a median age of 51.4 years. Most participants (61%) reported satisfaction with how their healthcare provider managed their genetic test result and care, and 65% of survey participants reported their provider recommended colonoscopy based on their genetic test results. Participants' responses also reflected variable levels of knowledge regarding cancer risks and screening recommendations for *MUTYH* carriers. The majority (98%) of participants shared their genetic test results with at least some of their relatives; however, only 13% of eligible relatives reportedly underwent cascade testing. Taken together, this study provides needed insight into the overall experiences of monoallelic *MUTYH* carriers and highlights numerous areas for improvement in clinician education, communication, and management of these individuals.

Keywords

cascade testing; genetic testing; knowledge; *MUTYH*; polyposis; risk management

1 | INTRODUCTION

Currently, the majority of patients who present for cancer risk evaluation will be offered the option of multigene panel testing to simultaneously assess for hereditary risk of multiple cancer types. *MUTYH* is a gene that is commonly included on cancer-focused multigene panels, including panels focused on colorectal cancer (CRC) and polyposis as well as pan-cancer panels. *MUTYH*-associated polyposis (MAP) results from biallelic inactivating pathogenic variants (PV) in the *MUTYH* gene and is characterized by increased risk for adenomatous colonic polyposis, CRC, and possibly other malignancies (Al-Tassan et al., 2002; Vogt et al., 2009). In contrast, individuals who have a monoallelic PV in *MUTYH* do not have MAP, and there are conflicting views as to whether these individuals carry substantially increased CRC risk (Ma et al., 2014; Win et al., 2014). Up to 1–2% of the general population are monoallelic *MUTYH* carriers, making this one of the most common findings on multigene panel testing (Yurgelun et al., 2017). The National Comprehensive Cancer Network (NCCN) provides the only recommendation to date to address monoallelic *MUTYH*-associated risk, recommending earlier and more frequent screening only for

carriers with a family history of CRC in a first degree relative (NCCN, 2021). However, for carriers lacking family history of CRC, current NCCN guidelines do not recommend more aggressive CRC screening as it is unknown if earlier and increased screening is justified (Katona et al., 2018). Furthermore, cascade testing for *MUTYH* carriers is unique for cancer genetics for various reasons including autosomal recessive inheritance, limited actionable clinical recommendations for carriers, and the importance to consider screening the other biological parent (OBP) to assess reproductive risks for existing or future offspring for MAP.

One potential downside of multigene panel testing is the inclusion of many low-to-moderate risk genes on panels, such as *MUTYH*, where there is less established data on cancer risk estimates and less clarity on risk management strategies (Bradbury et al., 2015; Rainville & Rana, 2014). Misinterpretation of genetic results can ultimately have significant consequences, including incorrect diagnoses, unnecessary treatments and interventions, increased psychosocial stress on patients and their families, and sometimes missed diagnoses (Bensend et al., 2014; Bonadies et al., 2014; Brierley et al., 2010; Brierley et al., 2012; Helm et al., 2018; Mahon, 2017; Mahon, 2019; Pasic et al., 2013; Riley et al., 2015). Even with correct interpretation of genetic test results, the implications of these results can still be misconstrued due to misunderstanding of or lack of familiarity with genetic concepts, misreading of the genetic report, and/or poor communication (Donohue et al., 2021). Given the uncertainty regarding medical management recommendations for monoallelic *MUTYH* carriers, this study aims to survey a cohort of *MUTYH* monoallelic carriers to assess how this monoallelic genetic variant is interpreted, assess whether *MUTYH* carrier status contributes to changes in medical management recommendations, and explore familial cascade testing in *MUTYH* carriers. To our knowledge, this is the first study to survey monoallelic *MUTYH* carriers on the aforementioned topics.

2 | METHODS

2.1 | Participants and procedures

Individuals with a pathogenic or likely pathogenic (P/LP) *MUTYH* variant were recruited from the Prospective Registry of Multiplex Testing (PROMPT). PROMPT is an internet-based, patient-directed ascertainment study for those who completed multiplex panel testing for cancer susceptibility, and is a partnership between Memorial Sloan Kettering Cancer Center, University of Pennsylvania, Mayo Clinic, and Dana Farber Cancer Institute. The overall objective of this registry is to ascertain families who underwent multigene panel testing to allow penetrance calculations for mutations in less characterized genes (Balmaña et al., 2016; Brower et al., 2019; Symecko et al., 2018) as well as to promote other research among these variant carriers. Since September 2014, healthcare providers and commercial laboratories have provided PROMPT information to eligible participants and ordering providers with test results. The PROMPT registry self-enrolls those with P/LP variants and variants of unknown significance (VUS) in cancer susceptibility genes, where registrants provide personal and familial cancer history and genetic testing information, as well as consent to be contacted about additional sub-studies. Genetic testing reports are not required for PROMPT enrollment; however, participants are encouraged to upload reports. PROMPT participants can self-report variant reclassifications on annual PROMPT follow-up

surveys; however, none of the participants in this study reported a reclassification of their *MUTYH* variant prior to completing this survey. Additionally, all reported *MUTYH* variants were confirmed as pathogenic/likely pathogenic in ClinVar prior to the time of survey completion.

From September 2014 to January 2021, 7100 participants enrolled in PROMPT. Of this cohort, eligible participants were invited via email to participate in this PROMPT sub-study. Eligible individuals were identified to be those with a monoallelic P/LP *MUTYH* variant, and individuals were excluded if they did not have a P/LP *MUTYH* variant, were a biallelic *MUTYH* carrier, or if they were monoallelic *MUTYH* carriers but had another P/LP variant in a different CRC risk gene (4 with *MSH2* and 1 with *MSH6*). Participants who consented completed a cross-sectional self-report survey on the following topics: sociodemographic characteristics, medical and family history, experiences with *MUTYH* genetic testing, genetics and *MUTYH* knowledge, perceived cancer risk, and familial communication.

2.2 | Measures

Surveys were completed between April 2020 and January 2021, with the following sections (see Supplemental Material for full survey):

2.2.1 | Demographics—We assessed age, biological sex, race, ethnicity, marital status, and education.

2.2.2 | Personal and family history—We assessed personal and family cancer history (age of diagnosis, type of cancer) and personal surgical history.

2.2.3 | Screening history—Twenty investigator-designed items assessed gastrointestinal cancer screening such as colonoscopy and endoscopy including “yes” or “no,” age at initiation, as well as reported outcome(s) and provider recommendations.

2.2.4 | Genetics knowledge—Genetic knowledge was assessed with 14 items regarding basic genetics concepts selected or modified from the University of North Carolina Genomic Knowledge Scale (Langer et al., 2017). *MUTYH* knowledge was assessed with six investigator-designed items regarding cancer risks and inheritance of *MUTYH* variants using “true,” “false,” and “do not know.” Participant understanding of their *MUTYH* result was assessed using one Likert-scale item (1= “Not at all confident” to 5 = “Very confident”).

2.2.5 | Genetic counseling and genetic testing—Personal experiences with genetic counseling and testing were assessed with 19 selected and modified items from a published, novel measure of the impact of multigene panel testing (Lumish et al., 2017). Satisfaction and confidence in the healthcare provider most responsible for providing information regarding the *MUTYH* result was assessed using two Likert-scale items (1= “Not at all confident” to 5 = “Very confident”).

2.2.6 | Risk perception—Four modified items from the National Cancer Institute Health Information National Trends Survey (HINTS, 2019) assessed perceived cancer risks

("Please rate how likely you are to develop [breast, ovarian…] cancer; "Very likely" to "Very unlikely") and beliefs regarding influencing factors (genetic test result, family history, lifestyle; "Not at all" to "A lot").

2.2.7 | Familial communication—Eleven investigator-designed items assessed whether participants shared their *MUTYH* results with family and other biological parent (OBP, if applicable), who they shared with, if their family member pursued genetic testing, and reasons why this information was not shared with family and/or the OBP.

Only select data from the survey administered to *MUTYH* carriers are herein reported.

2.3 | Data analysis

We presented descriptive statistics as medians and interquartile ranges (IQRs) or as percentages. We compared characteristics between *MUTYH* carriers who completed the survey versus individuals who did not. Continuous and categorical variables were compared using Wilcoxon rank-sum and chi-squared tests (Fisher's exact test for samples <5), respectively. All statistics and calculations were performed using Stata Version 16.0 (StataCorp, College Station, TX).

3 | RESULTS

3.1 | Survey participants

Of 7100 participants in PROMPT, 6976 were excluded who did not have a P/LP *MUTYH* variant, and 9 additional individuals were excluded because these participants carried biallelic *MUTYH* P/LP variants ($N = 4$) or another CRC risk gene ($N = 5$) (Figure 1). In total, 115 *MUTYH* monoallelic carriers were invited via email to participate in this PROMPT survey sub-study, of which 49 (43%) individuals enrolled and complete the survey.

3.2 | Demographics

Of the 115 invited *MUTYH* monoallelic P/LP variant carriers in PROMPT, the majority were white, non-Hispanic, and female, with a median age of 49 (IQR 39–61) (Table 1). Of these carriers, 72 (63%) had a history of any type of cancer while 9 (8%) had a personal history of CRC. A family history of CRC in any relative was reported by 45 (39%), and of these 15 (13%) reported having a first degree relative with CRC. In addition, of these *MUTYH* carriers, 26 (23%) also had a non-CRC-associated P/LP variant. Comparing *MUTYH* monoallelic P/LP variant carriers who completed the survey to those who did not complete the survey, there were no statistically significant differences between the two groups (Table 1).

Of the 49 individuals who participated in the *MUTYH*-specific survey, the majority were also white, non-Hispanic, and female, with a median age of 51.4 (IQR 39–61) (Table 1). Of these carriers, 30 (61%) had a history of any type of cancer while 3 (6%) had a personal history of CRC. A family history of CRC in any relative was reported by 18 (37%), and of these 6 (12%) reported having a first degree relative with CRC. In addition, of these

MUTYH carriers, 12 (24%) also had a non-CRC-associated P/LP variant. While we did not have education data on the *MUTYH* carriers who did not complete the survey, 42 of the 49 *MUTYH* survey participants (86%) reported having at least a college education.

3.3 | Genetic results disclosure

The 49 *MUTYH* survey participants reported receiving information regarding their *MUTYH* genetic test result from a genetic counselor (32, 65%), advanced practice professional (nurse practitioner, physician assistant) (5, 10%), oncologist (3, 6%), other healthcare professional (3, 6%), gastroenterologist (2, 4%), surgeon (2, 4%), primary care physician (1, 2%), or gynecologist (1, 2%) (Table S1). Thirty (61%) participants expressed satisfaction with their healthcare provider's knowledge and management of their *MUTYH* monoallelic variant. Of the 32 participants (65%) who received their genetic information from a genetic counselor, 69% were satisfied with their healthcare provider's knowledge and management (Table S1). Satisfaction rates were highest for gastroenterologists (100%) and surgeons (100%); however, sample size for these providers was small. The 19 participants who were not satisfied received their information from a genetic counselor (10, 53%), oncologist (3, 17%), advanced practice professional (2, 12%), primary care physician (1, 6%), gynecologist (1, 6%), or other healthcare professional (2, 12%).

3.4 | Medical management

Thirty-nine participants (80%) reported having had a prior colonoscopy (Table 2). By self-report, 32 participants (65%) were recommended to get a colonoscopy based upon their genetic test result, and of those, 15 (47%) were under the age of 50, which at the time of survey administration would have constituted early initiation of CRC screening, and 12 (38%) did not report a personal history of colon polyps. Of note, in 2016 the NCCN made a recommendation for earlier colonoscopy initiation for *MUTYH* heterozygotes that was subsequently reversed in 2017; 12 (24%) of survey participants and 8 (25%) of those recommended to get a colonoscopy based on their genetic testing results had their testing in 2016 or earlier. Additionally of those who were recommended to undergo colonoscopy based on their genetic test result, 20 (63%) had no personal or family history of CRC.

3.5 | *MUTYH* knowledge

Most participants answered questions correctly that addressed that P/LP *MUTYH* variants are associated with an increased risk of colon cancer and colon polyps, but not lung cancer (Table 3, questions 1 and 2). Furthermore, 80% of participants recognized that monoallelic and biallelic carriers have different cancer risks (Table 3, question 3). However, less than half of survey participants correctly answered that *MUTYH* homozygous carriers, but not *MUTYH* heterozygous carriers, have polyposis (Table 3, questions 4 and 5). Furthermore, there was uncertainty about screening recommendations for monoallelic *MUTYH* carriers, with only 47% correctly answering a question regarding colon cancer screening recommendations for *MUTYH* monoallelic carriers (Table 3, question 6).

3.6 | Familial communication

Of survey participants, 98% reported sharing their genetic test results with at least one relative (Table 4). Of these, 67% reported sharing their genetic test results with their "entire family." Of the 183 reportedly disclosed relatives, only 23 (13%) of these relatives were reported to have had their own genetic testing. Of the 31 participants reporting biological children, 61% reportedly shared their genetic test results with the OBP. Out of the 19 disclosed OBPs, only 4 were reported to have pursued genetic testing, with all of these individuals reporting lack of insurance coverage for their testing.

4 | DISCUSSION

MUTYH is often included on cancer-focused multigene panels and given that up to 1–2% of the general population carries a monoallelic P/LP *MUTYH* variant, it is inevitable that *MUTYH* monoallelic carriers will be commonly identified (Yurgelun et al., 2017). However, despite the high number of these carriers, very limited research exists on the experiences of monoallelic *MUTYH* carriers identified during multigene panel testing. To fill this gap, we surveyed *MUTYH* monoallelic carriers enrolled in PROMPT on their satisfaction with provider communication of their genetic results, reported medical management changes, family communication of results, and cascade practices. To our knowledge, this is the first survey of its kind for the *MUTYH* carrier population.

The majority of *MUTYH* carriers reported satisfaction with how their healthcare provider managed their genetic test result and care. Most of the results were disclosed by genetic counselors and given the small number of other providers who disclosed results it is not possible to make meaningful conclusions about patient satisfaction based on which healthcare provider disclosed results. However, of more concern is that 39% of patients were not satisfied with the provider who disclosed their genetic test results. Possible explanations for this dissatisfaction could include lack of knowledge by providers or other factors affecting dissatisfaction that were not assessed by this survey. Furthermore, a majority of the patients who were dissatisfied had a history of breast or gynecologic cancer (10 of 19, 53%). In such a scenario, the discovery of the monoallelic *MUTYH* mutation would be incidental and would not provide any insight into the individual's cancer. Therefore, it is certainly plausible that an incidental genetic testing result could produce dissatisfaction among these patients.

The survey data also revealed that a high percentage of monoallelic *MUTYH* carriers reported that their health care providers recommended enhanced colonoscopy screening based upon their genetic test result. Undoubtedly, it is challenging to deduce whether such recommendations resulted from genetic testing results alone, a combination of genetic testing results along with personal and/or family history, or personal and/or family history alone. Furthermore, NCCN recommended more aggressive colonoscopies for all *MUTYH* heterozygotes in their 2016 guidelines, which was then reversed the following year; however, this guideline change may have shifted provider and patient preferences about colonoscopy use in *MUTYH* heterozygotes. Of most concern is the potential for unnecessary colonoscopy screening of *MUTYH* heterozygotes based on the genetic test results alone; however, at this time we do not have sufficient data to allow determination

of whether unnecessary colonoscopy screening was performed in survey participants. Additional research is needed to determine whether healthcare providers are making medical management recommendations based on the presence of a monoallelic P/LP variant in *MUTYH*.

Furthermore, it is possible that monoallelic *MUTYH* carriers may be recommended to undergo colonoscopy based on misinterpretation of genetic test results among medical providers, potentially mistaking *MUTYH*-associated polyposis due to biallelic *MUTYH* P/LP variants for a monoallelic *MUTYH* P/LP variant. Other studies have shown that genetic test results can be mismanaged due to the complex nature of results, constantly evolving practice changes, and lack of formal genetics training among healthcare providers (Bensend et al., 2014; Eccles et al., 2015; Farmer et al., 2019). The potential for mismanagement and harm is a serious one that must be taken into consideration given the genetic counselor workforce is limited and alternative care delivery models are increasingly being implemented. While CRC screening itself has not been associated with adverse psychological harm (Kirkøen et al., 2016), colonoscopies are an invasive procedure that do pose some risk. However, the risks of an unnecessary colonoscopy are far less than those associated with other risk-reducing procedures such as mastectomies and oophorectomies.

Most participants also shared their genetic test results with at least some of their relatives, and it is notable that familial communication should be so prevalent regarding carrier status of an adult-onset cancer predisposition syndrome. However, given that participants were recruited through PROMPT, a patient-initiated research registry, there could be selection bias as participants may be especially motivated about their genetic testing results at baseline. Despite disclosure of these results to relatives, few relatives were reported to have followed through with cascade testing which is consistent with the experience reported in the case of cascade testing for carrier status in other common recessive syndromes such as cystic fibrosis (Gorrie et al., 2018). It is unknown if relatives failed to pursue cascade testing because they were not interested or because they lacked the access or information to find a local genetics provider to order the testing. In an effort to increase cascade testing, genetics providers may wish to follow-up with their proband by providing additional resources for family members on how to proceed with genetic testing if they are interested. Furthermore, for *MUTYH* carriers, testing the OBP of their children should always be considered, especially if their children are too young to consider cascade testing. Although our numbers are small, more than half of participants shared results with the OBP but a very small percentage of OBPs actually pursued testing and for those who did pursue testing, it was never reported to be covered by insurance. These data certainly raises the question of whether insurance carriers should be paying for carrier testing of the OBP once one parent is determined to be a *MUTYH* carrier.

4.1 | Study limitations

This study has several limitations including that participants were ascertained through a patient-initiated research registry that necessitated access to a computer or smart phone with internet capabilities, which could result in ascertainment bias given these patients are highly motivated to participate in research. Furthermore, given the study design, there is

also potential for recall bias among survey participants. The majority of the cohort were white females, and therefore this work does not provide insights among more diverse populations. Additionally, out of the 115 individuals who were eligible, there were only 49 participants who completed the survey, and this small sample size limited our ability to make meaningful statistical comparisons between survey responses. The survey also did not capture family history of polyps or advanced adenomas, nor did it capture when participants had polyps detected, which are factors that could contribute to increased colonoscopy screening recommendations.

4.2 | Practice implications

The potential for mismanagement of patients with monoallelic P/LP variants in *MUTYH* is a concerning possibility that warrants further investigation. Providers may wish to refer *MUTYH* carriers to specialists who are familiar with the current management recommendations. Furthermore, genetics providers might want to pay special attention to or offer additional resources to family members and the OBP of patients with monoallelic P/LP variants in *MUTYH* to assess reproductive risks for existing or future offspring.

5 | CONCLUSION

Monoallelic P/LP variants in *MUTYH* are a common finding on multigene panel testing for cancer predisposition, and at present, *MUTYH* carriers are frequently identified but represent an understudied cohort. The presented data provide needed insight into the overall experiences of *MUTYH* carriers and may contribute to improving care of these individuals in clinical practice. Further research is needed to determine whether *MUTYH* carriers are being over-screened based on their genetic testing results.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

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DATA AVAILABILITY STATEMENT

The data that support the findings of this study are available on request from the corresponding author. The data are not publicly available due to privacy or ethical restrictions.

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What is known about this topic

Although P/LP variants in *MUTYH* are one of the most common findings on multigene cancer panels (up to 1–2% of the general population), little is known about patient experience and understanding of monoallelic *MUTYH* P/LP variants.

What this paper adds to the topic

This paper provides insight into the experiences, knowledge, medical management, and familial communication of monoallelic *MUTYH* carriers.

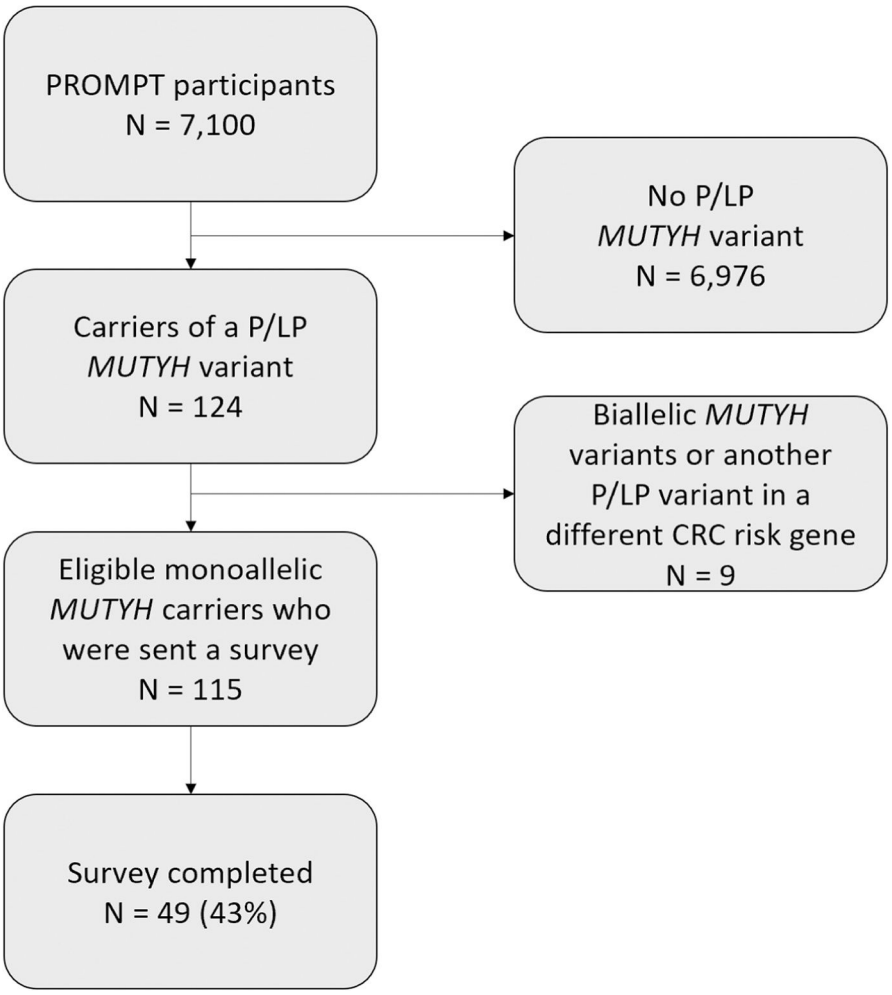

FIGURE 1.
Flow chart of PROMPT participants who completed the study survey

TABLE 1

Demographics of monoallelic *MUTYH* carriers

	***MUTYH* carriers (*N* = 115)**	***MUTYH* carriers who did not participate in the survey (*N* = 66)**	***MUTYH* carriers who participated in the survey (*N* = 49)**	***p* value (comparing those who participated and those who did not)**
Female Sex	105 (91%)	59 (89%)	46 (94%)	0.40
Age, median [IQR], years	49 [39–61]	48 [37–61]	51.4 [39–61]	0.93
Race				
White	77 (67%)	30 (45%)	47 (96%)	1.00 *
Asian	3 (3%)	1 (2%)	2 (4%)	
Unknown	35 (30%)	35 (53%)	0 (0%)	
Ethnicity				
Hispanic/Latino	4 (3%)	1 (2%)	3 (6%)	0.31 *
Non-Hispanic/Latino	74 (64%)	28 (42%)	46 (94%)	
Unknown	37 (32%)	37 (56%)	0 (0%)	
Have another known P/LP variant	26 (23%)	14 (21%)	12 (24%)	0.68
Personal History				
Cancer (any type)	72 (63%)	42 (64%)	30 (61%)	0.79
Colorectal cancer (CRC)	9 (8%)	6 (9%)	3 (6%)	0.73
Polyps	DNC	DNC	25 (51%)	-
Breast/gynecologic cancer	53 (46%)	31 (47%)	22 (45%)	0.85
Family History of CRC				
Any relative	45 (39%)	27 (41%)	18 (37%)	0.65
FDRs	15 (13%)	9 (14%)	6 (12%)	0.83

Abbreviation: DNC, data not collected; FDR, first-degree relative.

* *p* values are only based on comparison of known values.

TABLE 2

Medical management of monoallelic *MUTYH* carriers

Medical management	***N* = 49**
Prior colonoscopy	39 (80%)
Recommended colonoscopy based on genetic testing result	32 (65%)
<50 years old	15 (47%)
No personal nor family history of CRC (FDR, SDR, TDR)	20 (63%)
No personal history of CRC	29 (91%)
No personal history of CRC or FDR with CRC	27 (84%)
Family history of CRC	10 (31%)
≥1 FDR with CRC	5 (16%)

TABLE 3

MUTYH knowledge among survey participants

	True	False	Do Not Know
1. *MUTYH* is a gene that is linked to an increased risk for colon (large intestine) cancer and colon polyps	44 (90%)	0 (0%)	5 (10%)
2. *MUTYH* is a gene that is linked to an increased risk for lung cancer	0 (0%)	33 (67%)	16 (33%)
3. People who have two *MUTYH* gene mutations have different cancer risks than people who have one *MUTYH* gene mutation	39 (80%)	1 (2%)	9 (18%)
4. People who inherit one *MUTYH* gene mutation (from either mother or father) have a condition known as “polyposis”	3 (6%)	24 (49%)	22 (45%)
5. People who inherit two *MUTYH* gene mutations (one from mother and one from father) have a condition known as “polyposis”	17 (35%)	9 (18%)	23 (47%)
6. An adult with one *MUTYH* gene mutation should receive colon cancer screening (for example, colonoscopy) every 1–2 years	11 (22%)	23 (47%)	15 (31%)

Note: Gray boxes denotes correct answer.

TABLE 4

Familial communication of genetic results.

Familial Communication	***N* = 49**
Participants who shared their genetic testing results	
Shared with at least 1 relative	48 (98%)
Shared with their “entire family”	33 (67%)
Number of relatives genetic testing results disclosed to *	183
Number of relatives reported to be tested	23 (13%)
Participants with biological children	31 (63%)
Shared genetic testing results with the other biological parent (OBP)	19 (61%)
OBPs who pursued testing	4 (21%)
OBP testing covered by insurance	0 (0%)

Note:

* No denominator for total amount of relatives.