Knowledge, attitudes, and barriers to carrier screening for the Ashkenazi Jewish panel: a Florida experience

Education and Barriers assessment for Jewish Genetic Diseases

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Abstract The knowledge, attitudes, and barriers to Jewish genetic diseases (JGDs) and screening and their relative importance in reproductive decision-making were assessed in a population-based sample of Ashkenazi Jewish young adults in Florida. These adults attended educational screening fairs hosted by The Victor Center for the Prevention of Jewish Genetic Diseases at the University of Miami. Parametric and nonparametric tests were used as appropriate to analyze data from a single group pretest/posttest design. Four hundred twelve individuals (mean age=24.9; 54.7 % female, 45.3 % male) completed the questionnaires. Participants' level of knowledge increased from pre- to post-intervention (81.4 vs. 91.0 %; $p<0.0001$). Concern about the possibility of being a

carrier of a JGD was significantly higher after an educational session (5-point Likert scale mean difference=0.45; $p<0.0001$), as was their level of concern regarding having an affected child (mean difference=0.20; $p<0.0001$). The number of participants who agreed or strongly agreed that the test results would not have any influence on their reproductive behavior was lower after the session (17.2 vs. 20.8 %; $p<0.0001$). This study demonstrates that an educational carrier screening program increased knowledge and elucidated awareness of the attitudes and barriers toward JGDs and carrier screening.

Keywords Jewish genetic diseases · Carrier screening · Genetic education and autosomal recessive

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Introduction

As compared to other populations, several autosomal recessive diseases are more prevalent in individuals of Eastern European Jewish (Ashkenazi) descent, who comprise the majority of the North American Jewry, placing them at increased risk for having children with one of these conditions. In fact, one in five individuals of Ashkenazi ancestry are carriers of at least one of 11 heritable disorders (Klugman and Gross 2010). More recently, expanded screening for 16 of these conditions has demonstrated a carrier rate of 1 in 3.3 (Scott et al. 2010). Currently, some advocates have suggested expanding panels to include 19 disorders (Schneider et al. 2012, unpublished data). The Ashkenazi Jewish community is a prime example of the founder effect, as its members are at increased risk of inheriting rare disorders associated with alleles that have, over time, developed a higher frequency in this group as compared to the general population. Treatment is available for only some of these disorders, and with the

exception of a few, most, if not lethal in early childhood, are associated with significant morbidity including decreased quality of life, a shortened life expectancy and tremendous psychosocial and emotional burden on patients, family, and friends (Biffi and Naldini 2005; Brady 2006; Sands and Davidson 2006; Sawkar et al. 2006; Wraith 2006).

With the advent of highly sensitive and relatively inexpensive DNA-based methods for diagnosis, carrier screening, designed to identify individuals at increased risk of conceiving a child with disease, is widely available for Bloom syndrome, Canavan disease, cystic fibrosis, familial dysautonomia, Fanconi anemia type C, Gaucher disease, glycogen storage disease, maple syrup urine disease, mucopolipidosis type IV, Niemann-Pick disease type A, and Tay-Sachs disease. Additionally, carrier screening panels have very recently been expanded to include other disorders such as dihydrolipoamide dehydrogenase deficiency, hyperinsulinemia, Joubert syndrome, nemaline myopathy, spinal muscular atrophy (recommended for all populations by the American College of Medical Genetics), and Usher syndrome IF and III, and Walker Warburg syndrome, though tests for these were not yet available at the time of this study. Carrier detection has fast become an effective and widespread practice in communities pervaded by a substantial perceived burden of these diseases, as we are now able to identify healthy individuals who are carriers with no family history of a specific disorder whereas we previously would have only been able to identify carriers based on the birth of an affected child (Cao 1994; Kaback et al. 1993; Scott et al. 2010). The American College of Obstetricians and Gynecologists (ACOG) has recommended that those of Eastern European Jewish ancestry be offered the option for carrier screening for Tay-Sachs and Canavan diseases, cystic fibrosis, and familial dysautonomia (ACOG 2004). A new opinion published by ACOG in 2009 adds that “Individuals of Ashkenazi Jewish descent may inquire about the availability of carrier screening for other disorders” (ACOG 2009). As well, the American College of Medical Genetics Practice Guidelines recommend additional carrier screening for mucopolipidosis IV, Niemann-Pick disease type A, Fanconi anemia group C, Bloom syndrome, and Gaucher disease (Gross et al. 2008).

The decision to undergo carrier screening to find out if one is at risk of conceiving a baby with a specific genetic disorder is especially complex: factors to be considered may include the prevalence of the disease, the carrier frequency, the severity of the disease phenotype, and the potential (or nonexistence of) therapeutic modalities, financial and emotional burdens, both present and future reproductive options and preferences, and religious beliefs. Acceptability of screening, even for lesser known disorders for which mutations were more recently discovered, is high among pregnant Ashkenazi Jewish patients, as members of this population tend to choose screening for all conditions for which heterozygote screening

is available (Klugman and Gross 2010). However, nonpregnant couples are less likely to seek screening, necessitating an explanation of both the motivations for and barriers to carrier screening (Clayton et al. 1996). From the seminal screening program for Tay-Sachs which resulted in a reduction of over 90 % of births of children with the disorder, to the highly successful Dor Yeshorim that primarily targets the Orthodox Jewish community, to secondary school-based educational initiatives in Montreal and Australia, programs designed to increase the utilization of carrier screening through improvement of genetic literacy have shown significant benefit (Bach et al. 2007; Ekstein and Katzenstein 2001; Gason et al. 2005; Kaback 2001; Mitchell et al. 1996; Rosen 2003). Yet, while much of the Jewish community is now familiar with Tay-Sachs, general knowledge is still lacking with regard to the occurrence, inheritance pattern, clinical features, and scope of other largely debilitating and potentially lethal genetic diseases. There is also a paucity of existing research to describe the social issues and ethical implications intrinsic to carrier screening and its role in reproductive decision-making.

Though screening has of late become more widely available in the USA, educational programs that encourage informed reproductive decision-making have not. The state of Florida is home to the third largest Jewish population in the USA with over 600,000 Jews (Sheskin and Dashefsky 2010). In Miami alone, there are over 60 congregations, 19 educational institutions, and 22 local Jewish agencies. This region presents an ideal opportunity to provide and evaluate outreach efforts and to elucidate attitudes toward carrier screening. In order to expand efforts to prevent serious autosomal recessive genetic diseases with a high frequency in the Ashkenazi Jewish community, The Victor Center for the Prevention of Jewish Genetic Diseases at the University of Miami was created in 2007. The overall mission of The Victor Centers is to (1) educate the Jewish community of the need to be screened for certain autosomal recessive diseases, (2) screen participants over age 18 years in order to identify healthy individuals who are carriers, and (3) counsel those at risk of having affected children and options available to them. The aim of the current study was to assess the knowledge, barriers, opinions, and predictors of interest in screening for genetic disorders prior to and after implementing an outreach and screening program for Jewish genetic diseases.

Materials and methods

Data was obtained from a single group pretest/posttest design, wherein a multisite population-based sample of self-identified Ashkenazi Jewish young adults residing in Florida were studied. The Victor Center targets adults between the ages of 18 and 44 years of age, who are more inclined to be concerned about issues relating to family planning. The study was

approved by the University of Miami IRB, and informed consent was obtained from all participants. Pregnant women/couples were not included in the study.

Screening fairs

The education and screening fairs were held by the Victor Center at several locations throughout Florida: the University of Miami Miller School of Medicine and Coral Gables campuses, the University of Florida in Gainesville, Florida State University in Tallahassee, Temple Beth Am in Kendall, the Mathew Forbes Romer Foundation event in Palm Beach County, and the March of the Living event in Miami. Participation was achieved primarily through publicity of the program to student- and community-based organizations and by referrals from local religious leaders. The screening was provided free of charge to participants after they attended an educational presentation.

Educational intervention and screening procedures

In brief, the program at each screening fair comprised the following stages: education, voluntary participation in genetic counseling for informed consent, and carrier screening. Follow-up confidential reporting of test results and reproductive counseling was provided. The educational session was delivered by the same medical geneticist, which involved an oral presentation conveying: (1) general concepts and causes of genetic disease and inheritance patterns; (2) description, clinical course and management, and prevalence of Ashkenazi Jewish genetic diseases; (3) sensitivity of screening tests and interpretation of results; (4) reproductive options; and (5) ethical considerations. The oral presentation was standardized in its content and delivery across all studies sites. Written materials were disseminated for further/future reference, and a question-and-answer period concluded the intervention.

At the conclusion of the educational session, attendees were offered the opportunity to, free-of-charge, be tested at that time or schedule a genetic screening appointment at their convenience. Genetic counseling was provided and written informed consent obtained at the same visit at which blood was drawn so as to ensure that the decision to obtain screening had been carefully evaluated. Blood samples were sent to the Human Genetics Laboratory at Jacobi Medical Center in the Bronx, NY, which offers enzymatic diagnostic screening for Tay-Sachs disease and DNA panels targeted to the mutations most commonly found in persons of Ashkenazi Jewish descent. The screening panel included nine disorders: Bloom syndrome, Canavan disease, cystic fibrosis, familial dysautonomia, Fanconi anemia type C, Gaucher disease, mucopolidosis type IV, Niemann-Pick disease type A, and Tay-Sachs mutations and Hex A enzyme. Patients with a positive screening result were contacted by phone by the genetic counselor, and a

follow-up appointment with a geneticist was offered at that time. Those screening negative for all disorders received a letter by mail explaining the results including the lab reports, with contact information for the Victor Center should they wish to also schedule a follow-up appointment.

Assessments

The evaluation process included the use of several tools developed to assess level of pre- and post-intervention knowledge, attitude, feelings, concerns, and reasons for screening choice, as well as a measure of participant satisfaction. All screening fair attendees, whether ultimately receiving carrier screening or not, were provided a folder containing all study materials and asked to complete these questionnaires. Prior to the start of the educational session, individuals were informed of the purpose of the research and asked to then complete the consent form, and the pre-questionnaire should they agree to participate; to ensure that any changes in the abovementioned dimensions were likely a result of the educational intervention, participants were asked to complete the post-questionnaire at the close of the session. The self-administered, coded questionnaires used were designed specifically for the purposes of the Victor Center's studies, loosely based upon constructs adapted from similar previously published scales (Barlow-Stewart et al. 2003; Bekker et al. 1994). The questionnaire consisted of a series of demographic questions, scales designed to assess knowledge, perceived risk, barriers, consequences of test results, significance, and discrimination (the pre/post component), and a separate fair evaluation and participant feedback section to be completed with the post-questionnaire.

Analyses

The goal of the analysis phase of this study was to conduct a process, outcome, and impact type evaluation of the qualitative and quantitative data gleaned from the fairs in order to assess the fidelity and feasibility of the project, its influence on participants' knowledge and self-efficacy, and its ability to assess the attitudes of the participants.

SAS statistical software (version 9.2, SAS Institute, Cary, NC) was used for data management and all analyses and statistical tests. Descriptive statistics, with respect to age, gender, religious affiliation, and education, were generated for the entire sample in order to determine the efficiency of the program characterize the population in attendance and provide a context within which to interpret the outcome analyses. Responses to the pre- and post-questionnaires were transformed as follows. Percentages of responses for items concerning previous screening, affected/carrier family members, and education about Jewish genetic diseases were calculated. Items included in the domains of concern, attitudes, and knowledge were analyzed as continuous variables.

Knowledge responses were coded as “0,” incorrect or “1,” correct and summed for a maximum possible score of seven. Missing values were handled with listwise deletion ($N=2$). Each variable was checked for normality, and parametric tests and their nonparametric analogs were used accordingly. Changes in the knowledge score from pre- to post-education were assessed with Student's t test for paired samples. The impact of categorical variables such as gender and religious affiliation on knowledge score prior to the educational session was evaluated with one-way ANOVA. The Wilcoxon signed-rank test for continuous dependent variables was employed to investigate the impact of the intervention program on subjects' knowledge, attitudes, and concern. The Wilcoxon-Mann-Whitney test was used to evaluate differences in level of concern, perceived risk, barriers, consequences, and discrimination by site, gender, and religious affiliation, and the Kruskal-Wallis test, followed by Dunn's test for multiple comparisons for significant results was used to investigate differences in participant responses by various categories (e.g., parental religious affiliation, level of concern, or perceived risk). Statistical significance was assessed according to a preset two-sided alpha level of 0.05. Responses and suggestions gleaned from the fair evaluation and participant feedback form were examined qualitatively by content analysis in order to appraise the fair from subjects' perspectives and to aid in guiding any revisions to or improvement of the program.

Results

Participant characteristics

A total of 412 individuals (mean age=24.9; 54.7 % female, 45.3 % male) completed questionnaires at the beginning and end of the educational program (Table 1). The rate of uptake of carrier screening was nearly 100 %, as only two of the 412 individuals opted not to have the screening done (one was Sephardic and the second one had fear to blood drawn). Almost one third (27.5 %) of participants were unsure if they had a family member affected by a Jewish genetic disease, while more than twice as many (60.7 %) were unsure if any family members were carriers of a Jewish genetic disease. Only 13.9 % reported having ever been educated about Jewish genetic diseases prior to attending the fair.

Knowledge assessment

Participants performed well on the knowledge assessment (Fig. 1). There was a significant increase in the participants' level of knowledge from pre- to post-intervention (81.4 vs. 91.0 %; $p<0.0001$). Pre-intervention knowledge scores were marginally statistically associated ($p=0.05$) with mother's religious affiliation only, with the highest scores among those with

Table 1 Characteristics of overall study sample ($N=412$)

Demographic	<i>N</i> (%)
Site	
UMMSM	34 (8.2)
UM—Coral Gables	41 (9.9)
UF	127 (30.8)
FSU	45 (10.9)
Temple Beth Am	89 (21.6)
Romer	23 (5.6)
March of the Living	53 (12.9)
Age, years	24.9±4.7
Education ^a	
High school	9 (2.3)
Associate's degree	10 (2.6)
Bachelor's degree ^b	190 (48.7)
Graduate/professional Degree ^b	127 (32.6)
Medical school	54 (13.8)
Gender ^a	
Male	178 (45.3)
Female	215 (54.7)
Religious affiliation	
Reform	145 (40.6)
Conservative	169 (47.3)
Orthodox	43 (12.0)
Other/unaffiliated	55 (13.3)
Mother's religion	
Reform	162 (45.5)
Conservative	166 (46.6)
Orthodox	28 (7.9)
Other/unaffiliated	56 (13.6)
Father's religion	
Reform	150 (43.7)
Conservative	160 (46.6)
Orthodox	33 (9.6)
Other/unaffiliated	69 (16.7)
Individuals with family members known to be:	
Affected by a JGD	3 (0.7)
Carriers of a JGD	5 (1.2)
Previously educated about JGDs	57 (13.9)

^a N is less than 412 due to incomplete data for these questions only

^b In progress or completed

mothers practicing Reform (mean=83 %) and Conservative Judaism (mean=81 %) and the lowest among those with Orthodox mothers (mean=75 %).

Level of concern, perceived risk, and barriers

Specific items included in the pre- and posttests section for these domains are presented in Table 2. The percent of

Fig. 1 Knowledge Assessment. Asterisk percent responding correctly on the pretest and posttest are significantly different (McNemar's test, $p < 0.0001$). 1 Jewish genetic disorders are only found in the Ashkenazi (Central/Eastern European lineage) Jewish population, true/false. 2 If both parents are carriers of a Jewish genetic disorder, all of their children will be affected, true/false 3 All Jewish genetic disorders are fatal with no

known treatment or cure, true/false. 4 Children of interfaith couples are not at risk of Jewish genetic disorders, true/false. 5 If a family has no known history of Jewish genetic disorders, future generations will not be at risk, true/false. 6 Carriers of an autosomal recessive disorder have no symptoms, true/false. 7 If the carrier test is negative, you have no risk of having a child with any of these conditions, true/false

participants reporting a high or very high level of concern or perceived risk prior to and after the educational session is shown in Fig. 2 and did not differ by gender or religious

Table 2 Level of concern, perceived risk, and barriers

Specific items included in pre- and post-test questionnaires^a

What is your level of concern regarding the possibility of being a carrier of the genetic disorders listed above?
What is your level of concern regarding having a child who is affected?
How do you perceive your risk of being a carrier compared to that of a non-Jewish individual?
Please rate, on a scale of 1 to 5, the extent to which you agree or disagree with the following statements:
Offering testing for Jewish genetic diseases will cause anxiety in couples planning a pregnancy.
I want to avoid the birth of a child with a Jewish genetic disease.
There are reproductive options for couples that are found to be carriers of Jewish genetic diseases.
It is important to share my genetic test results with my family.
It is important to share my genetic test results with my partner.
The test results will not have any influence on my reproductive behavior.
I am afraid of discrimination of carriers of Jewish genetic diseases by society.
I am afraid of discrimination of carriers of Jewish genetic diseases by insurance companies.

affiliation. Individuals' concern about the possibility of being a carrier of a Jewish genetic disease was significantly higher after education than before (5-point Likert scale mean difference=0.45; $p < 0.0001$), as was their level of concern regarding having a child who is affected (mean difference=0.20; $p < 0.0001$).

The percentage of participants who agreed or strongly agreed that the test results would not have any influence on their reproductive behavior was lower after the session (posttest 17.2 % vs. pretest 20.8 %; $p < 0.0001$), as was the percentage that agreed or strongly agreed that they were afraid of discrimination of carriers by insurance companies (posttest 35.0 % vs. pretest 40.9 %; $p < 0.0001$). On the other hand, after the educational session, a greater percentage of participants agreed or strongly agreed that offering screening would cause anxiety in couples planning a pregnancy (posttest 31.1 % vs. pretest 19.9 %; $p < 0.0001$) that they wanted to avoid the birth of a child with a Jewish genetic disease (posttest 90.4 % vs. pretest 86.1 %; $p < 0.0001$) that there are reproductive options for couples that are found to be carriers (posttest 91.5 % vs. pretest 70.4 %; $p < 0.0001$), and that they are afraid of discrimination of carriers by society (posttest 14.5 % vs. pretest 11.6 %; $p < 0.0001$). These results are presented in Fig. 3. Women felt more strongly than men that they wanted to avoid the birth of a child with a Jewish genetic disease ($z = -2.08$; $p = 0.04$), while men felt more strongly than

Fig. 2 Levels of concern and perceived risk reported by participants

women that the test results would not have any influence on their reproductive behavior ($z=2.01$; $p=0.04$). Those

individuals who reported a high or very high perceived risk of being a carrier compared to non-Jewish persons were more

Fig. 3 Assessment of Attitudes and Barriers. 1 Offering screening for Jewish genetic diseases will cause anxiety in couples planning a pregnancy. 2 I want to avoid the birth of a child with a Jewish genetic disease. 3 There are reproductive options for couples that are found to be carriers of Jewish genetic diseases. 4 The test results will not have any influence on my reproductive behavior. 5 I am afraid of discrimination of carriers of Jewish genetic diseases by society. 6 I am afraid of discrimination of carriers of Jewish genetic diseases by insurance companies

concerned with avoiding the birth of a child with a Jewish genetic disease (Kruskal-Wallis $p=0.0008$; Dunn's $p<0.05$) and were more afraid of discrimination of carriers by insurance companies (Kruskal-Wallis $p=0.03$; Dunn's $p<0.05$).

Attitudes toward carrier screening and evaluation of the screening fair

Attitudes toward carrier screening and participants' evaluation of the screening fair were gleaned from the separate fair evaluation and participant feedback section completed with post-questionnaire. Nearly all participants felt that it is important for someone to know their carrier status (97.1 %; $N=400$), to be tested even if you do not have a family history (98.1 %; $N=404$), and to share this information with one's family (90.8 %; $N=374$) and significant other (98.8 %; $N=407$). About 39 % ($N=158$) said they would have participated in the screening fair if it were not free, stating that they would have paid a mean value of \$120 (range \$10–5,000) for the screening and education. The most common reasons for obtaining carrier screening were “to guide future reproductive decisions,” “parents' influence,” and “I just want to know.”

Discussion

Our study assessed awareness of and attitudes toward inherited disorders that are more prevalent among young adults of Ashkenazi Jewish descent and elucidated perceptions of carrier screening for these disorders. To our knowledge, this study is the first of its kind in Florida, a region home to one of the country's largest Jewish populations, and is novel in its assessment of multiple constructs of concern, perceived risk, and barriers in a preconception population of young adults. Our results are in line with other attitudinal studies in this area, demonstrating that educational carrier screening programs increase knowledge, while also producing a mild increase in level of concern (Hegwer et al. 2006), and consistent with similar research on the favorability of carrier screening, the practice is well received by the majority of individuals, with nearly all participants reporting that it is important for someone to know their carrier status, to be tested even without a family history, and to share this information with one's family and significant other (Archibald et al. 2012).

Primary prevention such as that offered by the Victor Center aims to identify carriers and high risk couples before the birth of affected offspring in order to provide them with genetic counseling and educate them about reproductive options for their future family planning. Less than 14 % of participants reported having ever been educated about Jewish genetic diseases prior to attending the screening fairs. Though attendees did in fact perform well on the pre-

intervention knowledge assessment, we found a statistically significant increase in the participants' level of knowledge from pre- to post-intervention (81.4 vs. 91.0 %; $p<0.0001$), and while the Genetic Information Nondiscrimination Act of 2008 prohibits the use of genetic information, including carrier testing, by health insurers or employers, 41 % of individuals agreed or strongly agreed that they were afraid of discrimination of carriers by insurance companies. This number was significantly lower after the educational session, and therefore, may be related to education on the difference between what it means to be a carrier versus having the disease, as well as on current health policy, further indicating the need for more interventions of this type within the Florida Ashkenazi Jewish community. Education is paramount to increasing rates of screening and ultimately preventing these diseases. In Montreal, for instance, programs to educate, screen, and counsel senior high school students at high risk for Tay-Sachs disease and β -thalassemia have existed for over 30 years. Two decades following the programs' inception, virtually, all carriers identified still remember their status have had their partner tested if his or her status was unknown and opted for reproductive counseling and/or fetal diagnosis. The incidence of both diseases declined by a remarkable 90–95 % (Mitchell et al. 1996). In Melbourne, Australia, a program offering education, screening, and counseling to Jewish secondary school students found too that education plays a critical role in enabling informed decisions about screening and in mitigating possible related psychosocial harms (Gason et al. 2005).

The utilization of population screening programs is based on the health belief model. When infused with the principles of the theory of reasoned action, which focuses on personal attitudes and subjective norms as the major predictors of behavioral intention, justifies our study of psychological constructs including perception of significance, consequences of test results, barriers, and discrimination as well as predictors of carrier screening acceptability and utilization (Ajzen and Fishbein 1975; Becker 1974). We found that the concern about the possibility of being a carrier of a Jewish genetic disease was in fact significantly higher after education than before, as was the level of concern regarding having a child who is affected. Perhaps, even more importantly, we anticipate that the participants' newfound knowledge will translate into more informed and meaningful decision-making with regard to their reproductive future. Indeed, in this study, we saw a statistically significant increase after the educational intervention in both the percentage of participants that felt their test results would influence their reproductive behavior and in the percentage that felt there are reproductive options for couples that are found to be carriers. Special care should therefore be taken to provide at risk individuals or couples with preconception counseling to ensure that they are truly informed. This is especially important with carrier screening continuing to

become available for a wider range of diseases, as a recent study in Australia found that increasing the number of conditions in a screening program was related to a decrease in knowledge and an increase in predicted negative feelings if found to be a carrier (Ioannou et al. 2010). Appropriate counseling will not only avail individuals of their reproductive options but also permit an understanding of residual risk and the opportunity to impress upon individuals who are identified as carriers of a gene mutation that while they are healthy and not at risk for developing the disease, there is risk of passing the gene mutation to their offspring. Opening the lines of communication is critical to reducing the burden of these diseases. While over 90 % felt it to be important to share one's carrier status with family, nearly two thirds of participants were unsure if any family members were carriers of a Jewish genetic disease, with one third unsure if they had an affected family member. This outreach program was ultimately designed to start a dialogue among young adults and their families about these conditions and the consequences associated with having an affected child.

Though we acknowledge the methodological limitations to the single group pretest/posttest design, this type of study remains commonly used in naturalistic settings. Moreover, it is not plausible to employ a control group for this research given the ethical and practical concerns in denying participation in the educational intervention to those individuals interested in undergoing genetic testing. While our study population was comprised of a convenience sample rather than a probability sample, participants were not preselected and were likely representative of the Jewish young adult population in Florida; however, the extent to which selection biases such as volunteerism impact our results cannot be discerned given that we are unable to assess those who chose not to participate in the screening fairs.

Our study contributes to the field of applied genetics and further characterizes the attitudes, opinions, and predictors of interest in autosomal recessive disorders that affect the Ashkenazi Jewish population, as well as implications for health improvement and disease prevention. This model can be used in any community with a high risk of specific autosomal recessive disorder. However, further research is still needed in order to fully understand the psychosocial implications of carrier screening and of knowing one's carrier status.

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Compliance and ethics guidelines The experiments comply with the current laws of the country in which they were performed.

Conflict of interest The authors declare no conflicts of interest.

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