Jewish Genetic Diseases: Knowledge of Reproductive Risk and Cancer Predisposition Among Young Adults of Ashkenazi Descent

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JEWISH GENETIC DISEASES: KNOWLEDGE OF REPRODUCTIVE RISK AND CANCER PREDISPOSITION AMONG YOUNG ADULTS OF ASHKENAZI DESCENT

by

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Bachelor of Science
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DEDICATION

This work is dedicated to the families touched by Jewish genetic diseases and BRCA1/2 gene-related mutations. Your resilience throughout the many heartaches of your medical odysseys is beyond admirable. To the innumerable medical professionals, researchers, Rabbis, Jewish organization leaders, and volunteers who have provided genetic health education to the Ashkenazi Jewish community for the many decades, thank you. May we continue to work tirelessly to raise awareness for the health and longevity of the Jewish people and future generations.
ACKNOWLEDGEMENTS

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To my parents, thank you for coming along this journey with me as I pursued my dream to become a genetic counselor. My accomplishments are possible because of a lifetime of your unconditional love and encouraging words. To Westin, Liz, and Calvin, you all light up my world. Thank you for the many laughs through the phone. I’ve held on to every one of them.

Finally, to my unwavering, supportive, and loving teammate, Ian: WE did it. We followed my dreams and we are about to accomplish a major one. Thank you for supporting me in every aspect of this project, from statistician, to chef, to dog dad, to fiancé, to friend. You wore all the hats with immense grace.
ABSTRACT

Founder mutations within the Ashkenazi Jewish (AJ) population are associated with significantly higher carrier rates for certain severe, life-limiting conditions collectively called Jewish Genetic Diseases (JGDs), and for pathogenic variants in the \textit{BRCA1} and \textit{BRCA2} (\textit{BRCA1/2}) cancer susceptibility genes. Efforts to educate AJ individuals about the implications of these founder mutations and available testing have increased the past forty years; however, studies suggest AJ individuals are not well educated on the topic (Hardy et al., 2022; Kaback, 2001; Warsch et al., 2014). Studies evaluating the reason for this gap in knowledge of young AJ adults are lacking. This study aimed to evaluate the knowledge of the young adult AJ population about their increased carrier risk for severe recessive diseases and \textit{BRCA1/2} gene-related cancer predispositions and assess factors contributing to their knowledge.

An electronic survey was distributed to Jewish and non-Jewish organizations across the United States. One hundred nineteen individuals between ages 18 and 27 (mean= 22.3; 65.5% female, 28.6% male; 5.9% gender minority) who self-identified as having at least one Ashkenazi Jewish grandparent completed the questionnaire. Demographics, personal experience with genetic testing for JGDs and \textit{BRCA1/2} mutations, and their educational experience about these topics were collected. Knowledge assessments of JGDs and \textit{BRCA1/2} mutations were also included.
Approximately 60% of participants had ‘never heard of’ or ‘did not know much about’ JGDs or BRCA1/2 gene mutations. Knowledge assessment scores for JGDs were lower compared to BRCA1/2 (69.1% vs. 78.1%) and significantly lower compared to a previous 2014 study (78.2% v. 81.4%, p value= .036). Learning most often took place at home or from parents, despite being one of the least chosen suggestions for future educational efforts. Participant preferences suggested future efforts begin as early as 11 to 14 years old in synagogues and other Jewish religious organizations.

We recommend an introductory module of Jewish genetic health concepts be incorporated into the bar/bat mitzvah curriculum by Hebrew schools and Jewish Day schools. By doing so, AJ young adults will take the first step towards achieving “AJ Genetic Health Literacy” in preparation to utilize their health literacy skills when they are of testing age (18+) to make informed, autonomous decisions about genetic testing and individualized medical management.
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LIST OF ABBREVIATIONS

ACMG ................................................................. American College of Medical Genetics and Genomics
ACOG ................................................................. American College of Obstetricians and Gynecologists
AJ .............................................................................. Ashkenazi Jewish
BRCA1/2 ........................................................................... BRCA1 gene and BRCA2 gene
ECS .................................................................................. Expanded Carrier Screening
HBOC .............................................................................. Hereditary Breast and Ovarian Cancer
JGD(s) ................................................................................ Jewish Genetic Disease(s)
LPV/PV ............................................................................ Likely Pathogenic Variant/ Pathogenic Variant
MRI ..................................................................................... Magnetic Resonance Imaging
NCCN® ........................................................................ National Comprehensive Cancer Network®
NGS .................................................................................. Next-Generation Sequencing
TTM ................................................................................ Transtheoretical Model
CHAPTER 1: BACKGROUND

1.1 Introduction to Ashkenazi Jewish Ancestry

There are approximately 15 million Jewish people around the world, with Israel and the United States being home to 85% of the total Jewish population. An estimated 7.3 million, or 39%, of the world’s population of Jews reside in the United States alone, making up approximately 2.4% of the country’s adult population (DellaPergola et al., 2020; Pew Research Center [PRC], 2021). A person’s Jewish identity often extends beyond religious beliefs and practices to include identity through culture and ancestry (Sklare, 1971). People with Jewish ancestry most often have lineage to three major groups: Ashkenazi, Sephardic, and Mizrahi. Each group has distinct origins from various places around the world with differing traditions. Between the groups, a considerable overlap of shared Jewish beliefs, practices, and customs are present (Skolnik, 2007; Skolnik & Berenbaum, 2007).

The Ashkenazi Jewish (AJ) people are known to pre-date the 14th century in Eastern and Central Europe (Waldman, 2022). The term Ashkenazi is derived from the Hebrew word ‘Ashkenaz’ meaning “Germany.” This group’s name is thought to have originated from their early inhabitation along the Rhine River valley between northern France and western Germany. Evidence suggests the group later migrated to Poland and Lithuania (Skolnik & Berenbaum, 2007). Today, 66% of Jewish adults in the United States identify as Ashkenazi, reporting to have at least one AJ parent (PRC, 2021). For genetic
testing purposes, an individual is considered to have AJ ancestry when at least one
grandparent is AJ (Gross et al., 2008; National Comprehensive Cancer Network [NCCN],
2023). The estimated number of individuals in the United States that meet this criterion
for AJ ancestry is up to 15 million people, or 4.5%, of the US population (Sheskin et al.,
2021).

Founder mutations within the AJ population have led to a significantly higher
rate of certain severe or life-limiting conditions and a predisposition to developing
certain cancers compared to rates in the general population (Behar et al., 2006).
Founder mutations refer to a specific variant in a gene that occurs more frequently in a
certain ethnic background and are a result of a population experiencing a founder event
or a bottlenecks event. A founder event refers to a small group of individuals
separating from a larger population, while a bottleneck event is a drastic reduction in a
population often associated with an environmental cause (i.e. flood, earthquake,
famine) or a human-driven cause (i.e. war, selective population control). Both types of
founder events result in a smaller gene pool for the remaining population and a higher
frequency of certain alleles or pathogenic variants (i.e. disease-causing mutations). As
reproduction continues within the smaller gene pool, the high frequency allele (i.e.
variant or mutation) continues to increase and persists for future generations, leading to
the increased prevalence of specific, rare mutations (Slatkin, 2004).

Founder events and/or bottleneck events within the AJ population are highly
suspected to have occurred through multiple expulsions and large-scale migrations over
many centuries (Bonné-Tamir et al., 1979). As a result of these happenings, the lack of
admixture from outside groups forever changed the Ashkenazi gene pool. Attempts to
determine a definitive timeline of events resulting in the founder mutations have been
made; however, it remains largely unsolved. In present day, evidence from the founder
events is observed in the high frequencies of carrier rates for many autosomal recessive
diseases, collectively known as Jewish Genetic Diseases (JGDs), and three specific
pathogenic variants in the BRCA1 and BRCA2 cancer-predisposition genes (American
College of Obstetricians and Gynecologists [ACOG] Committee on Genetics, 2017b;
NCCN, 2023).

Efforts to identify potentially at-risk individuals have increased since recognizing
the association between AJ ancestry and certain diseases. Several professional
guidelines recommend documenting a patient’s ethnic background when collecting
family history, including the National Society of Genetic Counselors (NSGC), ACOG, and
NCCN. Genetic counselors routinely ask about known AJ ancestry within a patient’s
family, with cancer genetic counselors inquiring most often of all the genetic counseling
specialties (Hubbel et al., 2022). While patients may not recognize the utility of this
question, known Ashkenazi Jewish ancestry is an important component of a provider’s
risk assessment when evaluating for the likelihood to be a carrier for a JGD or to have a
predisposition for BRCA1/2 gene-related cancers. Genetic counselors and health
professionals may also use known AJ ancestry to consider appropriate genetic testing
strategies, as discussed in the published guidelines by ACOG Committee on Genetics
(2017b) and NCCN (2023).
1.2 Jewish Genetic Diseases

1.2.1 Overview of Jewish Genetic Diseases

Individuals of Ashkenazi Jewish ancestry are more likely to be carriers of certain genetic disorders (i.e. JGDs) than the general population. In the 1970s, it was recognized that individuals of AJ ancestry were at a significantly higher risk to be carriers of Tay-Sachs disease than individuals without AJ ancestry. A blood enzyme test was then developed to identify Tay-Sachs carriers, prompting the first mass screening for a JGD in the 1970s, with the intention of identifying couples at-risk to have a child with this neurodegenerative disease (Kaback et al., 1997). These efforts reduced the number of children born with Tay-Sachs disease by more than 90% among the AJ population in North America (Kaback et al., 1993).

Today, more than 50 autosomal recessive conditions are known to occur at an increased frequency in AJ individuals compared to the general population. One in every three AJ individuals is estimated to carry a genetic mutation for at least one JGD (Scott et al., 2010). Couples can utilize carrier screening to determine their carrier status to help determine their risk to have a child with a JGD. Carrier screening refers to genetic testing performed on an individual to identify the presence of a variant in a gene known to be associated with an autosomal recessive or X-linked genetic disease. It is important for individuals to be aware of their carrier status for a disease to determine the chance for a reproductive couple to have a child with a genetic disorder. In the case of autosomal recessive conditions, an affected person inherits a pathogenic variant from both their mother and father. The parents of a child with an autosomal recessive
condition are referred to as carriers of the condition and have a 25% chance of having an affected child with every pregnancy. X-linked conditions refer to the set of disorders associated with a genetic variant on the X-chromosome. The risk to have an affected child with an X-linked condition depends on which parent carries the genetic variant.

Two approaches to carrier screening have been utilized in practice: targeted and expanded. Targeted carrier screening refers to single-gene testing and ethnicity-based carrier screening. In single-gene screening, a specific gene is analyzed to look for variants known to cause a particular disease in that population. Ethnic-based carrier screening, however, analyzes a group of genes known to cause disorders that occur at higher frequencies in individuals with a particular ancestry. In contrast, expanded carrier screening (ECS) analyzes many genes for mutations, regardless of one’s ethnicity or family history, often using next-generation sequencing (NGS) technology (ACOG Committee on Genetics, 2017a/b). The development of NGS technology allows couples to accurately determine their carrier status for hundreds of genes in a timely manner (Kraft et al., 2018).

1.2.2 Professional Guidelines: Carrier Screening

In a survey of Jewish Americans, 66% reported known AJ ancestry. The same study also reported 72% of Jewish individuals marry outside of the Jewish community. As a result, it can be suspected that an increased number of individuals who do not identify as AJ will have AJ ancestry in the coming decades and may benefit from genetic screening for JGDs (PRC, 2021). Due to the estimation that 1 in every 3 AJ individuals carries a genetic mutation for at least one JGD, the American College of Medical
Genetics (ACMG) established recommendations for carrier screening in the AJ population in 2008 (Gross et al., 2008). It was recommended that all AJ individuals who are pregnant or considering having a child be offered carrier screening for four JGDs (cystic fibrosis, Canavan disease, familial dysautonomia, and Tay-Sachs disease) with the option of an additional five conditions (Fanconi anemia (Group C), Niemann-Pick (Type A), Bloom syndrome, mucolipidosis IV, and Gaucher disease) (Gross et al., 2008). While it is still known that individuals of AJ ancestry have an increased chance to be carriers of these and other genetic diseases, ACMG now recommends a pan-ethnic approach to genetic carrier screening due to the imperfect nature of patients self-identifying their ancestry (Gregg et al., 2021).

1.3 BRCA1 and BRCA2 Genes

1.3.1 Overview of BRCA1 and BRCA2 Genes

Pathogenic variants in the BRCA1 and BRCA2 (BRCA1/2) genes are associated with Hereditary Breast and Ovarian Cancer syndrome (HBOC). HBOC is characterized by an increased risk for breast (male and female), ovarian, pancreatic, and prostate cancer, as well as an increased risk for melanoma in individuals with BRCA2 pathogenic variants (Daly et al., 2021). Three founder mutations account for up to 99% of the BRCA1/2 pathogenic mutations in people with AJ background (Petruclli et al., 2022; Strueweng et al., 1997). The prevalence of the three BRCA1/2 founder mutations within the AJ population is estimated at 1 in 40, which is 10-times higher than the non-AJ population rate of 1 in 400 (DellaPergola, 2017). Because of this, the NCCN (2023) states that individuals of AJ ancestry, without additional risk factors, can consider genetic testing.
1.3.2 Professional Guidelines: Oncology Genetic Testing

It is recommended that individuals with BRCA1/2 likely pathogenic/pathogenic variants (LPV/PV) discuss increased screening and risk-reducing measures with their healthcare provider (NCCN, 2023). Females with a LPV/PV are recommended to begin screening for breast cancer at age 25 with annual breast MRIs and incorporate annual 3D mammograms into the screening regimen at age 30. They may consider reducing their risk for breast cancer by choosing a bilateral mastectomy. A bilateral salpingo-oophorectomy is recommended to reduce the risk to develop ovarian cancer. Males with a LPV/PV in BRCA1/2 are recommended to begin self-exams and annual clinical exams of the breasts at age 35. Annual mammograms for males can be considered at age 50 or 10 years prior to the earliest known male breast cancer in the family. Screening for pancreatic cancer through an MRI and/or endoscopic ultrasound is recommended only for select individuals with a family history of pancreatic cancer in a first or second-degree relative from the same side as the BRCA1/2 variant (NCCN, 2023). Regardless of whether patients choose screening or risk-reducing measures, the importance of identifying individuals at an increased risk for BRCA1/2-related cancers is the first step in early intervention and risk reduction.

1.4 Genetic Knowledge of Stakeholders

Previous generations of Jewish leaders, community members, health professionals, and researchers have dedicated time and resources to raising awareness in the Jewish community about the association between AJ ancestry, certain recessive diseases, and the BRCA1/2 cancer susceptibility genes. Studies measuring the accuracy
of educator’s and learner’s knowledge provide insight about health information known by the AJ population and the effectiveness of educational efforts. By evaluating the genetic knowledge of stakeholders, the identification of misunderstood concepts and gaps in knowledge are useful considerations during strategic planning for future educational efforts.

1.4.1 Educator Knowledge of Jewish Genetic Health

Physicians may not be consistently inquiring about ancestry and therefore may not be providing the appropriate education to AJ individuals about Jewish genetic health risks (Grinzaid et al., 2014). Similarly, one study reported the average primary care physician asked only 20% of the appropriate medical and family history questions to screen patients for inherited breast cancer syndromes (Bell et al., 2015).

Among another group of primary care providers, 54% reported not being confident about their knowledge of genetic testing and were found to have incomplete or inaccurate information regarding hereditary cancer syndromes, including inheritance and interpretation of test results (Hamilton et al., 2017). Another study suggested religious leaders who are providing education about JGDs to congregants lack accurate knowledge about JGDs themselves (Thomsen et al., 2020).

1.4.2 Jewish Population Knowledge of Jewish Genetic Health

Efforts to educate the AJ population about health risks associated with their heritage began in the 1970s with the development of screening tests for couples to determine their risk to have a child with Tay-Sachs disease (Kaback et al., 1997). In the following decades, organizations have set out to educate AJ individuals, Jewish religious
leaders, and healthcare providers about the risk for JGDs and mutations in *BRCA1/2* cancer susceptibility genes through a variety of methods. The goal of educational efforts is to encourage potentially at-risk individuals to utilize genetic services to make informed decisions about strategies to reduce the risk of undesired outcomes.

Despite efforts to raise awareness among the AJ population, several studies suggest a lack of knowledge about one’s ancestry, risk for disease, and family history. In one study, 62% of individuals who did not identify as AJ were found to have detectable AJ genetic ancestry through DNA analysis, suggesting that identifying one’s risk for disease begins with being aware of one’s correct ethnicity (Tennen et al., 2020). However, even individuals who are aware of their AJ ancestry have been shown to be unaware of their risk for disease. One study found that 42% of AJ individuals at high-risk for a *BRCA1/2* mutation did not think they were at high risk, despite 31% of participants having a mother with either breast or ovarian cancer (Wiesman et al., 2017).

More generally, studies have shown AJ individuals are simply unfamiliar with JGD and *BRCA1/2* concepts. A study surveying young adults with AJ ancestry revealed that of the 412 individuals surveyed, only 13.9% reported having ever been educated about JGDs (Warsch et al., 2014). Additionally, a more recent study reported that 54% of participants said they had never heard of or knew little about *BRCA1/2* (Hardy et al., 2022). At this point in time, it is unclear what is driving the gap in knowledge of AJ individuals regarding their risk for disease.
1.5 Rationale

With one in three AJ individuals at risk to be a carrier of a JGD and one in forty at risk for a BRCA1/2 gene mutation, it is vital to educate and raise awareness about a person’s background risk to promote the utilization of genetic counseling and genetic testing (DellaPergola, 2017; Gross et al., 2008). A couple that is found to be carriers of the same JGD can utilize alternative reproductive techniques to increase the chance to have a healthy child without the JGD. Such techniques include prenatal diagnosis, in vitro fertilization (IVF) with preimplantation genetic testing (PGT), using an egg or sperm donor that has tested negative as a carrier for the JGD, or adoption. Additionally, individuals with a BRCA1/2 pathogenic variant can be offered additional cancer screening to identify the disease at an early stage when it is more treatable. People with a BRCA1/2 gene mutation have the option to reduce their risk for breast and ovarian cancer through prophylactic surgeries (NCCN, 2023). Additionally, BRCA1/2 carriers may choose pharmacological prevention to reduce the risk of breast cancer (Nazarali et al., 2014). It is also useful to know one’s BRCA1/2 mutation status to aid oncologists in identifying the most appropriate treatment plan if a cancer is diagnosed. Alternative reproductive technologies can be used to increase the chance to have a child without the BRCA1/2 mutation. While the risk for disease is well documented, there is limited data about the level of knowledge individuals of AJ ancestry have regarding their potential reproductive risk for JGDs and the increased chance to have mutations in BRCA1/2 cancer susceptibility genes. Additionally, no research, to our knowledge, has surveyed the young adult population about when and where the education, if any,
occurs. Research also has not assessed the knowledge and awareness of participants for both reproductive risks for JGDs and BRCA1/2 cancer susceptibility genes within the same questionnaire. This study will allow the knowledge and awareness of each risk category to be compared. Results of this study may help to identify current gaps in knowledge and inform future educational efforts.

With the majority of Jewish Americans reporting Ashkenazi Jewish ancestry and 72% recently electing to marry outside of the Jewish community, it should be known that children resulting from these interfaith marriages have an increased chance to be a carrier for a JGD and/or a cancer predisposition syndrome due to AJ ancestry compared to those without (PRC, 2021). It is imperative to anticipate the lack of awareness of genetic disease risk among interfaith couples in the coming decades and continue to study the impact and accuracy of educational efforts. In doing so, future efforts can be recognized and implemented.

1.6 Purpose

Young adults with Ashkenazi Jewish ancestry who are educated about their genetic disease risks can better utilize screening and diagnostic tools to make health decisions. We predicted young adults of AJ ancestry with a family and/or personal history of JGDs or BRCA1/2 mutations have more knowledge regarding genetic health risks than those without such a history. Additionally, we predicted that individuals primarily learn about genetic risks within their household. We hypothesized that the denomination of Judaism will have little influence on the knowledge level of participants. We predicted that young adults of AJ ancestry are familiar with their
increased carrier frequency for autosomal recessive JGDs and are less aware of their risk for mutations in the \textit{BRCA1/2} cancer susceptibility genes; however, we anticipated knowledge overall to be limited.

The aim of this study was to evaluate the knowledge of young adults with AJ ancestry about their reproductive risk for genetic diseases and cancer predisposition. We aimed to assess factors that contribute to increased awareness about one’s chance for health risks associated with AJ ancestry by collecting demographic information, personal and family history, perception of health risk, and prior education on Jewish genetic diseases. Finally, we hoped to identify useful strategies to better educate individuals with AJ ancestry about their reproductive risks and potential cancer predisposition. The primary objectives of the study were to:

1. Determine the knowledge base of young adults with AJ ancestry about JGDs and \textit{BRCA1/2} mutations:
   a. Compare study group’s knowledge of JGDs to previous study (Warsch et al., 2014);
   b. Compare study group’s knowledge of \textit{BRCA1/2} mutations to previous study (Hardy et al., 2022);
   c. Identify factors that may influence knowledge; and
   d. Compare knowledge of autosomal recessive conditions to knowledge of autosomal dominant cancer predisposition;

2. Identify where and when AJ individuals learn about genetic diseases related to their ancestry; and
3. Develop recommendations to help educate the AJ population regarding genetic diseases and cancer predisposition.
CHAPTER 2:

JEWFISH GENETIC DISEASES: KNOWLEDGE OF REPRODUCTIVE RISK AND CANCER PREDISPOSITION AMONG YOUNG ADULTS OF ASHKENAZI DESCENT

1 Granger, H.K., Edwards, J.G., Grinzaid, K., Schneider, A., Say, C. To be submitted to Journal of Community Genetics
2.1 ABSTRACT

Founder mutations within the Ashkenazi Jewish (AJ) population are associated with significantly higher carrier rates for certain severe, life-limiting conditions collectively called Jewish Genetic Diseases (JGDs), and for pathogenic variants in the BRCA1 and BRCA2 (BRCA1/2) cancer susceptibility genes. Efforts to educate AJ individuals about the implications of these founder mutations and available testing have increased the past forty years; however, studies suggest AJ individuals are not well educated on the topic (Hardy et al., 2022; Kaback, 2001; Warsch et al., 2014). Studies evaluating the reason for this gap in knowledge of young AJ adults are lacking. This study aimed to evaluate the knowledge of the young adult AJ population about their increased carrier risk for severe recessive diseases and BRCA1/2 gene-related cancer predispositions and assess factors contributing to their knowledge.

An electronic survey was distributed to Jewish and non-Jewish organizations across the United States. One hundred nineteen individuals between ages 18 and 27 (mean= 22.3; 65.5% female, 28.6% male; 5.9% gender minority) who self-identified as having at least one Ashkenazi Jewish grandparent completed the questionnaire. Demographics, personal experience with genetic testing for JGDs and BRCA1/2 mutations, and their educational experience about these topics were collected. Knowledge assessments of JGDs and BRCA1/2 mutations were also included.

Approximately 60% of participants had ‘never heard of’ or ‘did not know much about’ JGDs or BRCA1/2 gene mutations. Knowledge assessment scores for JGDs were lower compared to BRCA1/2 (69.1% vs. 78.1%) and significantly lower compared to a
previous 2014 study (78.2% v. 81.4%, p value= .036). Learning most often took place at home or from parents, despite being one of the least chosen suggestions for future educational efforts. Participant preferences suggested future efforts begin as early as 11 to 14 years old in synagogues and other Jewish religious organizations.

We recommend an introductory module of Jewish genetic health concepts be incorporated into the bar/bat mitzvah curriculum by Hebrew schools and Jewish Day schools. By doing so, AJ young adults will take the first step towards achieving “AJ Genetic Health Literacy” in preparation to utilize their health literacy skills when they are of testing age (18+) to make informed, autonomous decisions about genetic testing and individualized medical management.

2.2 INTRODUCTION

Individuals with Ashkenazi (i.e. Central/ Eastern European) Jewish (AJ) ancestry have an increased chance to be carriers of severe, life-limiting Jewish genetic diseases (JGDs) and mutations in BRCA1/2 cancer susceptibility genes than non-AJ individuals. In the 1970s, efforts to educate and screen the AJ community for JGDs began and proved successful when the number of children born with autosomal recessive Tay-Sachs disease in the AJ population decreased by 90% (Kaback et al., 1993). Today, more than fifty autosomal recessive and X-linked genetic diseases are categorized as JGDs. Estimates predict one in three AJ individuals are carriers for at least one of these conditions and one in forty individuals have a BRCA1/2 cancer susceptibility mutation, which is ten-times that of a non-AJ individual (DellaPergola, 2017; Scott et al., 2010). The increased frequency of the genetic variants found in the AJ community (i.e. founder
mutations) ultimately result from the lack of admixture with other groups of people, likely due to expulsions and large-scale migrations over many centuries (Slatkin, 2004).

Despite efforts to raise awareness within the AJ population, several studies suggest a lack of awareness about one’s risk for disease, family history, and general knowledge of JGDs and BRCA1/2 gene mutations. One study found that 42% of AJ individuals at high-risk for a BRCA1/2 mutation did not think they were at high risk, despite 31% of participants having a mother with either breast or ovarian cancer (Wiesman et al., 2017). Regarding a lack of JGD knowledge, a study surveying 412 young AJ adults revealed only 13.9% reported ever having been educated about JGDs (Warsch et al., 2014). Similarly, another study surveying AJ individuals reported that 54% of participants said they had never heard of or knew little about BRCA1/2 genes. (Hardy et al., 2022). It is unclear what factors are driving the gap in knowledge of AJ individuals regarding their chance to be a carrier of a JGD or have a BRCA1/2 gene mutation. Furthermore, data evaluating the knowledge of young AJ adults is lacking.

This study explored the current knowledge of young adults with AJ ancestry regarding their reproductive risk for genetic diseases and cancer predisposition. By surveying individuals from different parts of the country, we aimed to assess factors that contribute to increased awareness of health risks associated with AJ ancestry by collecting demographic information, personal and family history, perception of health risk, and familiarity with the topic prior to the survey. Finally, results from this study may help identify updated strategies and build recommendations to better educate
individuals with AJ ancestry about their reproductive risks and potential cancer predisposition.

2.3 METHODS

2.3.1 Survey Design

A survey focused on assessing the knowledge of young adults regarding JGDs and mutations in the BRCA1/2 cancer susceptibility genes was constructed using a combination of new questions and questions from similar knowledge assessments (Hardy et al., 2018; Hardy et al., 2022; Warsch et al., 2014). The survey was designed and administered online through Qualtrics.com and included demographic information, Likert-scale items, multiple choice questions, and open-ended questions divided into five categories: demographic questions, JGD Awareness, JGD Knowledge, BRCA1/2 Awareness, and BRCA1/2 Knowledge.

As part of the ‘JGD Awareness’ and ‘BRCA1/2 Awareness’ questions, participants were asked about their familiarity with the topics. Those who selected an answer other than ‘I have never heard of’ for both JGDs and BRCA1/2 genes were prompted to select the age group in which they learned about the topics and choose the place(s) where they initially learned about these conditions. Respondents were able to choose more than one answer when identifying the place(s) of learning.

The three questions of the survey asked all participants about recommendations for future education efforts. Participants were asked to choose one age group from a list of options that they felt were the most appropriate for AJ individuals to receive education about Jewish genetic health. The other two questions asked participants to
identify a place AJ individuals may want to learn about their genetic health and ideas about how to reach unaware AJ individuals about this education. The following examples were provided following each question in an effort to further clarify the question: “i.e. a presentation at a synagogue, public online modules, etc.” and “i.e social media, flyer in a community building, etc.”

The final question of the survey allowed participants to provide an email address if they wished to participate in the raffle for the sole purpose of the principal investigator contacting the participant if they were a raffle winner. Age, targeted personal and family history, Jewish heritage, and email address were the only identifying information collected on the survey.

2.3.2 Participant Recruitment

Approval for this study was obtained from the University of South Carolina’s Institutional Review Board (Pro00121753). Eligible participants included individuals between ages 18 and 27 who reported Ashkenazi Jewish ancestry, which was defined as having at least one Ashkenazi Jewish grandparent. To increase participation, an incentive was provided in the form of an opportunity to enter a raffle and win one of five $100 gift cards by completing the questionnaire. The funding for the project was provided on behalf of the Columbia Jewish Federation (CJF) in the amount of $500. CJF was not further involved in the planning or conducting of the project beyond their donation.

Participation in the study was voluntary, as addressed in the introduction letter to the participants prior to beginning the survey (Appendix A). Participants were also
made aware in the introduction letter about the option to enter a personal email address following the completion of the survey in order to enter a raffle to win one of the $100 gift cards, and were informed that their email address would only be used for purposes pertaining to the raffle. Reading the introduction letter and choosing to continue to the questionnaire served as the participant’s consent.

Individuals in leadership roles at Jewish organizations or in Jewish communities across the United States that interact with this age group (i.e. synagogues, Hillels, Chabads on Campus, Jewish Community Centers, non-religious campus organizations, etc.) were contacted by email or phone and encouraged to share the electronic survey amongst their community via a link or QR code. Pre-written verbiage for a newsletter announcement and social media post were included in a supplied Microsoft Office Word document along with a recruitment flyer (Figure 2.1) to print or post on social media and instructions to share on social media. The recruitment flyer was developed using Canva.com.

Responses were collected from October 2022 to March 2023. Of the 563 responses, 119 participants met inclusion criteria, completed the survey in its entirety, and were included in data analysis. The remaining 444 unanalyzed responses consisted of 80 participants that did not meet inclusion criteria, 70 responses that did not complete 100% of the survey, 44 duplicate responses, and 249 responses that appeared to be artificial robot responses. Internal Qualtrics measurements were used to differentiate artificial responses from human responses.
2.3.3 Data Analysis

Descriptive statistical analysis of the quantitative data was completed using Microsoft Office Excel software, while quantitative hypothesis testing was completed using R software. In most analyses, a chi-squared test of Independence was used to identify associations between categorical variables. The two independent-samples t-test was used to calculate the p value when comparing participants’ ages from the survey responses collected for this project and those from previous studies (Hardy et al., 2022; Warsch et al., 2014). Fisher’s exact test was applied when the estimated value of a cell was five or less and was therefore used when calculating the p value for gender and
denomination of Judaism between study participants and previous participants of the Warsch et al. (2014) study. When analyzing free-response questions, a constant comparison approach was used to assign each response a category. Responses were coded and thematic frequencies were reported. Figures and tables were constructed using Microsoft Office’s Word and Excel software programs.

2.4 RESULTS

2.4.1 Demographic Information

The age, gender identity, highest completed level of education, and state of residence were obtained for all 119 participants (Table 2.1). The average age of the cohort was 22.3 years ($SD = 2.7$) with the majority identifying as female (65.5%) and having completed between 12 and 16 years of education, at minimum. Florida (N=16), California (N=14), and New York (N=10) were the most reported states of residence out of the 23 states represented in the cohort, although most respondents were from the southern region of the United States. About 92% (N=109) of individuals identified themselves as AJ, while the remaining 8.2% identified as AJ with hesitancy by choosing ‘yes, I think so’ rather than a definitive ‘yes.’ The Reform and Conservative denominations of Judaism were represented by most participants, while Orthodox and Reconstructionist Jews were represented the least. To gain insight about communication practices regarding genetic health education within this targeted population, participants were asked to identify the primary source that shared the survey with them. Hillel organizations, social media, and personal referral totaled 78.2% of all distribution sources.
<table>
<thead>
<tr>
<th>Variable</th>
<th>Frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Age (mean, SD)</strong></td>
<td>22.3 (2.71)</td>
</tr>
<tr>
<td>18</td>
<td>12 (10.1)</td>
</tr>
<tr>
<td>19</td>
<td>9 (7.6)</td>
</tr>
<tr>
<td>20</td>
<td>14 (11.8)</td>
</tr>
<tr>
<td>21</td>
<td>14 (11.8)</td>
</tr>
<tr>
<td>22</td>
<td>21 (17.6)</td>
</tr>
<tr>
<td>23</td>
<td>9 (7.6)</td>
</tr>
<tr>
<td>24</td>
<td>9 (7.6)</td>
</tr>
<tr>
<td>25</td>
<td>13 (10.9)</td>
</tr>
<tr>
<td>26</td>
<td>9 (7.6)</td>
</tr>
<tr>
<td>27</td>
<td>9 (7.6)</td>
</tr>
<tr>
<td><strong>Gender</strong></td>
<td></td>
</tr>
<tr>
<td>Female</td>
<td>78 (65.5)</td>
</tr>
<tr>
<td>Male</td>
<td>34 (28.6)</td>
</tr>
<tr>
<td>Gender Minority</td>
<td>7 (5.9)</td>
</tr>
<tr>
<td><strong>Education</strong></td>
<td></td>
</tr>
<tr>
<td>High School</td>
<td>57 (47.9)</td>
</tr>
<tr>
<td>Associate’s Degree</td>
<td>8 (6.7)</td>
</tr>
<tr>
<td>Bachelor’s Degree</td>
<td>45 (37.8)</td>
</tr>
<tr>
<td>Graduate/ Professional Degree</td>
<td>8 (6.7)</td>
</tr>
<tr>
<td>Medical School</td>
<td>1 (0.8)</td>
</tr>
<tr>
<td><strong>Region</strong></td>
<td></td>
</tr>
<tr>
<td>Northeast</td>
<td>21 (17.6)</td>
</tr>
<tr>
<td>Midwest</td>
<td>14 (11.8)</td>
</tr>
<tr>
<td>South</td>
<td>58 (48.7)</td>
</tr>
<tr>
<td>West</td>
<td>25 (21.0)</td>
</tr>
<tr>
<td>International</td>
<td>1 (0.8)</td>
</tr>
<tr>
<td><strong>Ashkenazi Jewish</strong></td>
<td></td>
</tr>
<tr>
<td>Yes</td>
<td>109 (91.6)</td>
</tr>
<tr>
<td>Yes, I think so</td>
<td>10 (8.4)</td>
</tr>
<tr>
<td><strong>Affiliated Denomination of Judaism</strong></td>
<td></td>
</tr>
<tr>
<td>Reform</td>
<td>66 (55.5)</td>
</tr>
<tr>
<td>Conservative</td>
<td>30 (25.2)</td>
</tr>
<tr>
<td>Orthodox</td>
<td>6 (5.0)</td>
</tr>
<tr>
<td>Other/ Unaffiliated</td>
<td>14 (11.8)</td>
</tr>
<tr>
<td>Reconstructionist</td>
<td>3 (2.5)</td>
</tr>
<tr>
<td><strong>Survey Source</strong></td>
<td></td>
</tr>
<tr>
<td>Chabad on Campus</td>
<td>5 (4.2)</td>
</tr>
<tr>
<td>Hillel</td>
<td>44 (37.0)</td>
</tr>
</tbody>
</table>

Table 2.1 Participant Demographics (N=119)
2.4.2 Outcomes of JGD Knowledge Assessment

Knowledge of JGDs were measured by asking participants nine questions related to general information about JGDs, including mode of inheritance, carrier risk among AJ individuals compared to the general population, and defining characteristics of a carrier. Questions 1 through Question 7 were adopted from the Warsch et al. (2014) study. Participants scored an average 69.1% (mean= 6.2, SD=1.7) on the JGD Knowledge Assessment (Table 2.2). Questions pertaining to the spectrum of JGD severity and an individual’s potential carrier status for a JGD without a family history or AJ ancestry were the highest scoring questions, with all three scoring above 80%. Of the 119 participants, only 22 (19.1%) correctly identified the chance for a person with AJ background to be a carrier for at least one JGD. Zero (0) participants answered all nine questions correctly.

Table 2.2 JGD Knowledge Assessment Scores

<table>
<thead>
<tr>
<th>Questions</th>
<th>Correct Responses</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Jewish genetic disorders are only found in the Ashkenazi (Central/Eastern European lineage) Jewish population (Agree/Disagree)</td>
<td>89 (74.8)</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Overall Scores (N=119)</th>
<th>mean (SD)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Composite Knowledge Score</td>
<td>6.2 (1.7)</td>
</tr>
<tr>
<td>Average % Correct</td>
<td>69.1%</td>
</tr>
</tbody>
</table>
2. If both parents are carriers of a Jewish genetic disorder, all of their children will be affected (Agree/ Disagree)  91 (76.5)

3. All Jewish genetic disorders are fatal with no known treatment or cure (Agree/ Disagree)  106 (88.1)

4. Children of interfaith couples are not at risk of Jewish genetic disorders (Agree/ Disagree)  109 (91.6)

5. If a family has no known history of Jewish genetic disorders, future generations will not be at risk (Agree/ Disagree)  104 (87.4)

6. Carriers of an autosomal recessive disorders typically have no symptoms (Agree/ Disagree)  67 (56.3)

7. If the carrier test is negative, you have no risk of having a child with any of these conditions (Agree/ Disagree)  85 (71.4)

8. Which of the following puts a child at risk to inherit a recessive genetic disease?
   a) Only one partner is a carrier  67 (56.3)
   b) Both partners are carriers, but for different genetic diseases
   c) Both partners are carriers for the same genetic disease
   d) I am not sure

9. Approximately how many American Jews of Ashkenazi descent are a carrier for at least one JGD?
   a) 1 in 4  22 (19.1)
   b) 1 in 10
   c) 1 in 25
   d) 1 in 100

All 9 Questions Correct  0 (0)

*Correct answers are in bold.
Q1–Q7 from Warsch et al., 2014

JGD knowledge scores from the young AJ adults (N=412) surveyed by Warsch et al. in 2014 scored significantly better overall than the individuals in our study (81.4% vs. 78.2%; p value = .036). Participants in the current study scored significantly lower on Question 2 (76.5% vs. 91.3%; p value= <.001) and Question 3 (89.1% vs. 94.9%; p value=.022) in comparison to the 2014 cohort. On Figure 2.2, Questions 6 is noticeably the
lowest scoring question in both groups, with approximately 56% of participants answering correctly.

![Figure 2.2 Comparing JGD Knowledge: 2014 to Current Day](image)

* p value <.001
** p value <.05

When asked about the perceived chance to be a carrier of a JGD, participants in this study reported a significantly higher risk than previously reported by a similar age group in 2014 (Table 2.3). In contrast, our participants felt the availability of reproductive options for JGD carriers to be significantly less than the Warsch et al. (2014) cohort (52.9% vs. 70.4%; p value= <.001).
2.4.3 Awareness and Predictors of JGD Knowledge

The degree of JGD familiarity was assessed on a scale ranging from ‘I have never heard of JGDs’ to ‘I know a great deal about these diseases.’ Nearly 20% of participants reported they had never heard of JGDs prior to the survey, while the same number of individuals reported knowing a ‘fair amount’ about JGDs. Prior familiarity with JGDs was a significant predictor in JGD knowledge scores ($p$ value = .045). Table 2.4 displays the average knowledge scores for the varying levels of JGD familiarity, with the lowest familiarity level corresponding with the lowest average knowledge score (mean=5.9; $SD=1.7$). The study also assessed if known carrier screening of relatives impacted knowledge scores, however this did not hold true ($p$ value = .203). The majority of

<table>
<thead>
<tr>
<th>Question</th>
<th>Granger et al., 2023 ($N=119$)</th>
<th>Warsch et al., 2014 ($N= 412$)</th>
<th>$p$ value</th>
</tr>
</thead>
<tbody>
<tr>
<td>How do you perceive your chance of being a carrier of a genetic disease compared to that of a non-Jewish individual?</td>
<td>44 (37.0)</td>
<td>111 (26.9)</td>
<td>.034**</td>
</tr>
<tr>
<td>‘High’ or ‘Very High’</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>‘Agree’ or ‘Somewhat Agree’</td>
<td>63 (52.9)</td>
<td>290 (70.4)</td>
<td>&lt;.001*</td>
</tr>
</tbody>
</table>

* $p$ value <.001
** $p$ value <.05

2.4.3.1 Comparing JGD Risk Perceptions: 2014 to Current Day

<table>
<thead>
<tr>
<th>Question</th>
<th>Granger et al., 2023 ($N=119$)</th>
<th>Warsch et al., 2014 ($N= 412$)</th>
<th>$p$ value</th>
</tr>
</thead>
<tbody>
<tr>
<td>How do you perceive your chance of being a carrier of a genetic disease compared to that of a non-Jewish individual?</td>
<td>44 (37.0)</td>
<td>111 (26.9)</td>
<td>.034**</td>
</tr>
<tr>
<td>‘High’ or ‘Very High’</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

* $p$ value <.001
** $p$ value <.05

2.4.3.2 There are reproductive options for couples found to be carriers for the same Jewish genetic disease. (Agree or Somewhat Agree)

<table>
<thead>
<tr>
<th>Question</th>
<th>Granger et al., 2023 ($N=119$)</th>
<th>Warsch et al., 2014 ($N= 412$)</th>
<th>$p$ value</th>
</tr>
</thead>
<tbody>
<tr>
<td>There are reproductive options for couples found to be carriers for the same Jewish genetic disease. (Agree or Somewhat Agree)</td>
<td>63 (52.9)</td>
<td>290 (70.4)</td>
<td>&lt;.001*</td>
</tr>
</tbody>
</table>

* $p$ value <.001
** $p$ value <.05
participants reported being well-informed of their relatives’ having completed carrier screening or not (‘yes’=46.2%; ‘no’= 19.3), although more than a third of participants were unsure. Education level was determined to be a significant predictor of JGD knowledge scores (p value=.031), as was the primary survey source (p value=.014).

Table 2.4 Familiarity with JGDs and Family Genetic Health as Predictors of Knowledge

<table>
<thead>
<tr>
<th>Variable</th>
<th>Frequency N (%)</th>
<th>mean (SD)</th>
<th>p value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Familiarity with JGDs</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>I have never heard of Jewish Genetic Diseases (JGDs).</td>
<td>24 (20.2)</td>
<td>5.9 (1.7)</td>
<td>0.045**</td>
</tr>
<tr>
<td>I have heard of JGDs, but I do not know much about them.</td>
<td>66 (55.5)</td>
<td>6.1 (1.8)</td>
<td>0.058</td>
</tr>
<tr>
<td>I know a fair amount about these diseases.</td>
<td>24 (20.2)</td>
<td>6.8 (1.4)</td>
<td></td>
</tr>
<tr>
<td>I know a great deal about these diseases.</td>
<td>5 (4.2)</td>
<td>7.2 (0.8)</td>
<td></td>
</tr>
<tr>
<td>Familiarity with association between AJ ancestry and genetic diseases</td>
<td></td>
<td></td>
<td>0.203</td>
</tr>
<tr>
<td>I have never heard of this association.</td>
<td>24 (20.2)</td>
<td>5.9 (1.8)</td>
<td></td>
</tr>
<tr>
<td>I have heard of this association, but I do not know much about them.</td>
<td>45 (37.8)</td>
<td>5.8 (2.0)</td>
<td></td>
</tr>
<tr>
<td>I know a fair amount about this association.</td>
<td>41 (34.5)</td>
<td>6.8 (1.2)</td>
<td></td>
</tr>
<tr>
<td>I know a great deal about this association.</td>
<td>9 (7.6)</td>
<td>6.8 (1.0)</td>
<td></td>
</tr>
<tr>
<td>Carrier Screening of Relatives and/or Self</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Yes</td>
<td>23 (46.2)</td>
<td>6.7 (1.3)</td>
<td></td>
</tr>
<tr>
<td>No</td>
<td>55 (19.3)</td>
<td>6.3 (1.6)</td>
<td></td>
</tr>
<tr>
<td>Unsure</td>
<td>41 (34.5)</td>
<td>5.9 (1.9)</td>
<td></td>
</tr>
</tbody>
</table>

*p value <.001  **p value <.05

In an effort to hypothesize causes for the discrepancy in JGD knowledge between the current study and the Warsch et al. (2014) study, the demographic variables were compared (Table 2.5). Significant demographic differences exist between the two groups, with p<.001 for age, gender, education, and religious denomination. It should be noted that the Warsch et al. (2014) study asked participants to report their highest level
of education, whether in progress or completed. Participants that completed our survey reported on their highest level of completed education only.

**Table 2.5 Descriptive Statistics of JGD Knowledge Studies**

<table>
<thead>
<tr>
<th>Variables</th>
<th>Granger et al., 2023 (N=119)</th>
<th>Warsch et al., 2014 (N=412)</th>
<th>p value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Age (mean, SD)</td>
<td>22.3 (2.7)</td>
<td>24.9 (4.7)</td>
<td>&lt;.001*</td>
</tr>
<tr>
<td>Gender</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Female</td>
<td>78 (65.5)</td>
<td>215 (54.7)</td>
<td>&lt;.001*</td>
</tr>
<tr>
<td>Male</td>
<td>34 (28.6)</td>
<td>178 (45.3)</td>
<td></td>
</tr>
<tr>
<td>Gender Minority</td>
<td>7 (5.9)</td>
<td>--</td>
<td></td>
</tr>
<tr>
<td>Education</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>High School</td>
<td>57 (47.9)</td>
<td>9 (2.3)</td>
<td></td>
</tr>
<tr>
<td>Associate’s Degree</td>
<td>8 (6.7)</td>
<td>10 (2.6)</td>
<td>&lt;.001*</td>
</tr>
<tr>
<td>Bachelor’s Degree</td>
<td>45 (37.8)</td>
<td>190 (48.7)a</td>
<td>&lt;.001*</td>
</tr>
<tr>
<td>Graduate/ Professional Degree</td>
<td>8 (6.7)</td>
<td>127 (32.6)a</td>
<td></td>
</tr>
<tr>
<td>Medical School</td>
<td>1 (0.8)</td>
<td>54 (13.8)</td>
<td></td>
</tr>
<tr>
<td>Religious Denomination</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Reform</td>
<td>66 (55.5)</td>
<td>145 (40.6)</td>
<td>&lt;.001*</td>
</tr>
<tr>
<td>Conservative</td>
<td>30 (25.2)</td>
<td>169 (47.3)</td>
<td></td>
</tr>
<tr>
<td>Orthodox</td>
<td>6 (5.0)</td>
<td>43 (12.0)</td>
<td></td>
</tr>
<tr>
<td>Other/ Unaffiliated</td>
<td>14 (11.8)</td>
<td>55 (13.3)</td>
<td></td>
</tr>
<tr>
<td>Reconstructionist</td>
<td>3 (2.5)</td>
<td>--</td>
<td></td>
</tr>
<tr>
<td>Individuals with a family member known to be:</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Affected by a JGD</td>
<td>1 (0.8)</td>
<td>3 (0.7)</td>
<td></td>
</tr>
<tr>
<td>Carrier(s) of a JGD</td>
<td>5 (4.2)</td>
<td>5 (1.2)</td>
<td></td>
</tr>
</tbody>
</table>

* p value < .001
** p value < .05

*a Degree in progress or completed
b p value cannot be calculated due to ‘Affected by a JGD’ N=1 for Granger et al., 2023
2.4.4 Outcomes of BRCA1/2 Knowledge Assessment

BRCA1/2 knowledge was assessed through a series of nine questions about the mode of inheritance related to the genes, likelihood to be a carrier depending on AJ ancestry or family history, and understanding of genetic test result types. Questions 2, 4, 5, 6, and 7 were adopted from a previous study completed by Hardy et al. (2022). An average score of 78.1% (mean= 7.3; $SD=1.3$) was obtained on the BRCA1/2 Knowledge Assessment (Table 2.6) was correctly answered. Five of the nine questions had an average score of 90% or above. Question 6 and Question 9 asked about the inheritance pattern and carrier frequency of BRCA1/2 gene mutations. Participants scored the lowest on these questions with only 20.2% and 45.4% correctly answering the question. Unlike JGD knowledges scores, 6 participants (5%) answered all nine questions correctly.

Table 2.6 BRCA1/ BRCA2 Knowledge Assessment Scores

<table>
<thead>
<tr>
<th>Overall Scores (N=119)</th>
<th>mean (SD)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Composite Knowledge Score</td>
<td>7.3 (1.3)</td>
</tr>
<tr>
<td>Average % Correct</td>
<td>78.1%</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Question</th>
<th>Correct Response</th>
<th>N (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Pathogenic variants in BRCA1/2 increase the chance for both men and women to develop cancer. (Agree/ Disagree)</td>
<td>106 (89.1)</td>
<td></td>
</tr>
<tr>
<td>2. If a female tests negative for mutations in BRCA1 and BRCA2, then she definitely will not get breast or ovarian cancer. (Agree/ Disagree)</td>
<td>110 (92.4)</td>
<td></td>
</tr>
<tr>
<td>3. If a male or female tests positive for mutations in BRCA1 or BRCA2, then they are both at risk to develop breast cancer. (Agree/ Disagree)</td>
<td>108 (90.8)</td>
<td></td>
</tr>
<tr>
<td>4. A mother can pass down a BRCA1 or BRCA2 mutation to her sons. (Agree/ Disagree)</td>
<td>110 (92.4)</td>
<td></td>
</tr>
<tr>
<td>5. A father can pass down a BRCA1 or BRCA2 mutation to his daughters. (Agree/ Disagree)</td>
<td>107 (89.9)</td>
<td></td>
</tr>
</tbody>
</table>
6. If an individual is a carrier of a BRCA1 or BRCA2 mutation, each of his or her children has a 1 in 4 chance of also having the mutation. (Agree/ Disagree)  
24 (20.2)

7. If there is no history of breast or ovarian cancer in my family, I am definitely not at risk for carrying a BRCA1 or BRCA2 mutation. (Agree/ Disagree)  
108 (90.8)

8. Individuals with AJ ancestry may want to consider genetic testing for BRCA1 and BRCA2, even if they have no personal or family history of cancer. (Agree/ Disagree)  
109 (91.6)

9. Approximately how many American Jews of Ashkenazi descent have a genetic mutation in BRCA1/2 cancer susceptibility genes?
   a) 1 in 10
   b) 1 in 40
   c) 1 in 100
   d) 1 in 400  
54 (45.4)

All 9 Questions Correct  
6 (5.0)

Q2, Q4, Q5, Q6, Q7= Hardy et al., 2022 Questions  
*Correct answers are in bold.

Participants in our study scored lower on the BRCA1/2 knowledge scores compared to the Hardy et al. (2022) participants (p value=.074), with an average percent correct of 77.1% (mean=3.9; SD=.87) compared to 80.0% (mean=4.0; SD=.69). Significant differences in percent correct between the two cohorts were observed for Question 1, Question 2, (92.2% vs. 96.7%; p<.001) and Question 5 (Table 2.6, Question 7; 90.4% vs. 93.7%; p value=.011), as seen in Figure 2.3.
When AJ adults ages 18-27 from the Hardy et al. (2022) study were analyzed and compared to participants in this study of the same age group, knowledge scores were relatively similar (78.5% vs. 77.1%; p value=.511). Figure 2.4 depicts participants in the Hardy et al. (2022) study that scored significantly higher on Question 1 (Table 2.6, Question 2) and Question 5 (Table 2.6, Question 7), just as the ‘All Ages’ scored. Question 4 was the lowest scoring question of both cohorts, although the current study’s participants scored higher than the Hardy et al. (2022) age-adjusted group (20.9% vs. 14.2%; p value=.234).
2.4.5 Awareness and Predictors of BRCA1/2 Knowledge

Among respondents of our survey, prior familiarity with BRCA1/2 genes was a significant predictor of BRCA1/2 knowledge (p value=.032; Table 2.7). The 30% of participants who had never heard of BRCA1/2 genes prior to the survey scored lower than those who reported a higher degree of familiarity. Approximately 60% of respondents reported a family history of cancers known to be associated with BRCA1/2 gene mutations; however, the majority of respondents did not report known BRCA1/2 genetic testing for themselves or their family (‘no’ and ‘unsure’= 75.6%). Of those who reported known BRCA1/2 genetic testing within their family, 39.1% (N=9) recalled the involvement of a genetic counselor (Table 2.7).
Table 2.7 Familiarity with BRCA1/2 ‘Cancer Susceptibility Genes’ and Family Genetic Health as Predictors of Knowledge

<table>
<thead>
<tr>
<th>Variable</th>
<th>Frequency</th>
<th>mean (SD)</th>
<th>p value</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Familiarity with BRCA1/2 Genes</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>I have never heard of these genes</td>
<td>34 (28.6)</td>
<td>6.8 (1.3)</td>
<td></td>
</tr>
<tr>
<td>I have heard of these genes, but I do not</td>
<td>37 (31.1)</td>
<td>6.8 (1.3)</td>
<td>.032**</td>
</tr>
<tr>
<td>know much about them.</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>I know a fair amount about these genes.</td>
<td>37 (31.1)</td>
<td>7.3 (1.1)</td>
<td></td>
</tr>
<tr>
<td>I know a great deal about these genes.</td>
<td>11 (9.2)</td>
<td>7.6 (1.0)</td>
<td></td>
</tr>
<tr>
<td><strong>Relatives with Breast, Ovarian, Prostate, or</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Pancreatic Cancer</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Yes</td>
<td>71 (59.7)</td>
<td>7.0 (1.3)</td>
<td>.951</td>
</tr>
<tr>
<td>No</td>
<td>41 (34.5)</td>
<td>7.0 (1.3)</td>
<td></td>
</tr>
<tr>
<td>Unsure</td>
<td>7 (5.9)</td>
<td>7.0 (1.2)</td>
<td></td>
</tr>
<tr>
<td><strong>Relatives/ Self tested for BRCA1/2</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Yes</td>
<td>29 (24.4)</td>
<td>7.2 (1.3)</td>
<td>.170</td>
</tr>
<tr>
<td>No</td>
<td>40 (33.6)</td>
<td>7.0 (1.3)</td>
<td></td>
</tr>
<tr>
<td>Unsure</td>
<td>50 (42.0)</td>
<td>6.9 (1.2)</td>
<td></td>
</tr>
<tr>
<td><strong>Relatives/ Self- BRCA1/2 Testing Results</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Positive (i.e. pathogenic variant identified)</td>
<td>6 (20.7)</td>
<td>7.5 (0.5)</td>
<td>.202</td>
</tr>
<tr>
<td>Negative</td>
<td>19 (65.5)</td>
<td>7.4 (1.0)</td>
<td></td>
</tr>
<tr>
<td>Unsure</td>
<td>4 (13.8)</td>
<td>6.0 (2.8)</td>
<td></td>
</tr>
<tr>
<td><strong>Genetic Counselor Involved in BRCA1/2</strong></td>
<td>(N=23)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Testing</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Yes</td>
<td>9 (39.1)</td>
<td>7.7 (0.5)</td>
<td>.159</td>
</tr>
<tr>
<td>No</td>
<td>6 (26.1)</td>
<td>7.5 (0.8)</td>
<td></td>
</tr>
<tr>
<td>Unsure</td>
<td>8 (34.8)</td>
<td>6.9 (2.0)</td>
<td></td>
</tr>
</tbody>
</table>

** p value <.05

\( ^{a} \) Six participants with reported personal/ family history of BRCA1/2 testing were not appropriately presented this question due to technical error; \( N=23 \).

The cohort surveyed by Hardy et al. (2022) \( N=331 \) was significantly different \( (p<.001) \) from our participants in nearly all demographic categories, including age, gender, region of the country, education, family history of cancer, and a personal or family history of BRCA1/2 genetic testing. The majority of the Hardy et al. (2022)
participants were female (60.1% vs. 39.9% male) with at least a college degree (88.5%) from the northeast region of the United States (47.7%) with an average age of 29.9 (SD=5.8). Prior familiarity with BRCA1/2 was also significantly different between the two cohorts, with 15.7% of the Hardy et al. (2022) group reporting having ‘never heard of BRCA1/2 genes’ compared to 28.6% reported by our participants (p value=.003).

After adjusting the Hardy et al. (2022) cohort to fit within the age-related inclusion criteria of our study (ages 18-27), the age, gender, region of the country, and education from the 108 participants remained significantly different in comparison to our participants (Table 2.8). Although gender appears to be significantly different, the structure of the Hardy et al. (2022) survey question was bivariant while the survey question in this study was multivariant with the option to write in their gender identity. Therefore, the significance of gender between these groups should be interpreted with caution.

Table 2.8 shows that the age-adjusted Hardy et al. (2022) (N=108) cohort consisted of majority female (61.1% vs. 38.9%) with a minimum education of a college degree (74.1%) residing in the northeast region of the U.S. (44.4%). Awareness of BRCA1/2 testing among relatives was similar between the groups, despite nearly all of the Hardy et al. (2022) group reporting a family history of cancers associated with BRCA1/2 gene mutations (98.1%). Regarding familiarity with BRCA1/2 genes, our participants were nearly 1.5 times more likely to have ‘never heard of BRCA1/2 genes’ prior to the survey. In contrast, the age-adjusted Hardy et al. (2022) group was almost twice as likely to report knowing ‘a great deal’ about BRCA1/2 genes.
Table 2.8 Descriptive Statistics of Participants BRCA1/2 ‘Cancer Susceptibility Genes’ Knowledge Studies, Ages 18-27

<table>
<thead>
<tr>
<th>Variables</th>
<th>Granger et al., 2023 (N=119)</th>
<th>Hardy et al., 2022 (N=108)</th>
<th>p value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Age (18-27)</td>
<td>22.3 (2.7)</td>
<td>23.8 (2.5)</td>
<td>&lt;.001*</td>
</tr>
<tr>
<td>Gender</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Female</td>
<td>78 (62.1)</td>
<td>66 (61.1)</td>
<td></td>
</tr>
<tr>
<td>Male</td>
<td>34 (28.7)</td>
<td>42 (38.9)</td>
<td>.016**</td>
</tr>
<tr>
<td>Gender Minority</td>
<td>7 (6.1)</td>
<td>--</td>
<td></td>
</tr>
<tr>
<td>Region</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Northeast</td>
<td>21 (17.6)</td>
<td>68 (44.4)</td>
<td>&lt;.001*</td>
</tr>
<tr>
<td>Midwest</td>
<td>14 (11.8)</td>
<td>14 (7.4)</td>
<td></td>
</tr>
<tr>
<td>South</td>
<td>58 (48.7)</td>
<td>43 (39.8)</td>
<td>&lt;.001*</td>
</tr>
<tr>
<td>West</td>
<td>25 (21.0)</td>
<td>6 (5.6)</td>
<td></td>
</tr>
<tr>
<td>International</td>
<td>1 (0.8)</td>
<td>1 (0.4)</td>
<td></td>
</tr>
<tr>
<td>Education</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>&lt; College graduate</td>
<td>65 (54.6)</td>
<td>26 (24.1)</td>
<td>&lt;.001*</td>
</tr>
<tr>
<td>College graduate +</td>
<td>54 (45.4)</td>
<td>80 (74.1)</td>
<td></td>
</tr>
<tr>
<td>Relatives with Breast, Ovarian, Prostate, or Pancreatic Cancer</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Yes</td>
<td>71 (59.7)</td>
<td>106 (98.1)</td>
<td>.065</td>
</tr>
<tr>
<td>No/ Unsure</td>
<td>48 (40.3)</td>
<td>2 (1.9)</td>
<td></td>
</tr>
<tr>
<td>Relatives tested for BRCA1/BRCA2</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Yes</td>
<td>29 (24.4)</td>
<td>26 (24.1)</td>
<td>.169</td>
</tr>
<tr>
<td>No</td>
<td>40 (33.6)</td>
<td>25 (23.1)</td>
<td></td>
</tr>
<tr>
<td>Unsure</td>
<td>50 (42.0)</td>
<td>57 (52.8)</td>
<td></td>
</tr>
<tr>
<td>Familiarity with BRCA1 and BRCA2</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Never heard of them</td>
<td>34 (28.6)</td>
<td>20 (18.5)</td>
<td></td>
</tr>
<tr>
<td>Did not know much about them</td>
<td>37 (31.1)</td>
<td>43 (39.8)</td>
<td>.241</td>
</tr>
<tr>
<td>New a fair amount about them</td>
<td>37 (31.1)</td>
<td>27 (25.0)</td>
<td></td>
</tr>
<tr>
<td>Knew a great deal about them</td>
<td>11 (9.2)</td>
<td>18 (16.7)</td>
<td></td>
</tr>
</tbody>
</table>

* p value <.001
** p value <.05
2.4.6 The ‘When’ and ‘Where’ of Jewish Genetic Health Education

Figure 2.5 demonstrates that most individuals initially learned about JGDs between ages 11 and 14 (26.1%), which is earlier than the ages 15-17 for BRCA1/2 education (28.6%). Of the participants educated about JGDs and BRCA1/2 genes, the majority of all learning was reported to have occurred before age 22. More respondents were reported to have never been educated about BRCA1/2 genes (28.6%) compared to JGDs (20.2%). The participants recommended ages 15-17 as the most appropriate age group to learn about AJ genetic health.

**Figure 2.5 Age of Jewish Genetic Health Education vs. Personal Recommendation**

Participants reported receiving both JGD and BRCA1/2 cancer susceptibility gene education at home or from their parents most often, followed by friends and family outside of the home (Figure 2.6). Similar frequencies were reported for both ‘formal
education’ and ‘social media, internet, entertainment, advertisement’ for JGD learning 
(N=15 vs. N=14). While zero participants reported receiving their education about JGDs 
or BRCA1/2 genes in synagogue (referenced here as both a place of worship and as a 
Jewish institution) or by a rabbi, it was recommended most often as the setting AJ 
individuals may want to learn about Jewish genetic health (N=42), with Hebrew school 
mentioned a total of seven times. Jewish organizations, including Hillel International, 
Chabad on Campus, Birthright Israel, and youth groups, were the second-most 
recommended educational setting to teach about Jewish genetic health. Seven 
participants suggested collaborating with Jewish celebrities and social media influencers 
to reach a larger, potentially unaware audience, along with targeted advertisements on 
social media platforms. The remaining 30 participants in the ‘Social Media, Internet, 
Advertisement, Public Online Modules’ consisted of recommendations for educational 
online modules.
Figure 2.6 Place of Jewish Genetic Health Education vs. Personal Recommendation
2.5 DISCUSSION

This was one of the first studies to survey young adults with AJ ancestry about their familiarity, knowledge, and education experience regarding reproductive risks for recessive genetic diseases and BRCA1/2 cancer predispositions. Furthermore, our study explored factors that influence knowledge for the purpose of developing recommendations to update educational strategies for the AJ community. While genetic education and screening recommendations in the AJ community began on a large-scale decades ago, research on the success of educational efforts for both JGDs and BRCA1/2 cancer predisposition genes across the United States is lacking (Kaback, 2001). Additionally, little quantitative research on the current state of knowledge and familiarity with JGDs within the young adult AJ population has been conducted in nearly a decade (Warsch et al., 2014). The main findings of our research highlighted familiarity with JGDs and BRCA1/2 genes as a significant predictor for knowledge and revealed gaps in young adult AJ knowledge for both genetic health categories. We propose a model to achieve “Ashkenazi Jewish Genetic Health Literacy,” with education at the center, to better equip an AJ individual with the tools to choose healthy behaviors (i.e. identify an individualized management plan) regarding genetic health.

As stated by the National Human Genome Research Institute, genomic health literacy is essential to improving decision-making capability and choosing healthy behaviors (Hurle et al., 2013). Health literacy is most often defined as “the degree to which individuals have the capacity to obtain, process, and understand basic health information and services needed to make appropriate health decisions” (Ratzan &
Parker, 2000). Our data showed a lack of familiarity, a high perceived risk, and inaccurate knowledge among AJ young adults, all of which play a role in the development of health literacy (Miyoshi & Watanabe, 2023).

Familiarity with JGDs and BRCA1/2 cancer susceptibility genes was a significant predictor of the respective knowledge assessments and indicates a stepwise process for learning. The perceived risk to be a carrier of a JGD was significantly higher in our cohort compared to the Warsch et al. (2014) cohort. Other studies have found similarly high-risk perceptions among young adults with a family history of BRCA1/2 mutations (Mellon et al., 2009; Patenaude et al., 2013). Risk perception is formed from a variety of sources but most often from parents, relatives, and healthcare providers (Werner-Lin, 2008; Wiesman et al., 2010). Research suggests personal relevance and perceived risk is a key influence in a person’s decision to proceed with genetic screening, further emphasizing the importance of educating the AJ community about the relevance of carrier screening and BRCA1/2 testing and/or screening options due to their AJ ancestry (Archibald & McClaren, 2012).

Participants were less knowledgeable about JGDs than BRCA1/2 cancer susceptibility genes and scored significantly lower on the JGD knowledge assessment compared to a young adult AJ cohort from nine years prior (Warsch et al., 2014). Historical context may play an important role in the significant knowledge difference between the 2014 cohort and the current 2023 cohort. In 2017, ACOG released updated guidelines endorsing pan-ethnic, expanded carrier screening as one of many acceptable approaches to carrier screening (ACOG Committee on Genetics, 2017a/b). In 2021,
ACMG published updated guidelines recommending a pan-ethnic approach to carrier screening and suggested a screening panel consisting of 113 conditions (Gregg et al., 2021). While the clinical utility of pan-ethnic carrier screening is widely recognized and supported, it is important to consider the possible implications of shifting away from an ethnic-based approach and its impact on genetic health education. It can be speculated that offering a standard carrier screening panel to all patients, regardless of ethnicity, places less importance on identifying a patient’s ancestry, including AJ. As providers inquire less about ancestry for testing strategy purposes, opportunities to educate individuals about the potential for reproductive risk for disease and cancer predisposition may be missed.

Our study found formal education to be a significant predictor of JGD knowledge, and formal education was significantly lower among study participants compared to the Warsch et al. (2014) cohort. It is likely that the level of completed formal education is a contributing factor to overall JGD knowledge. This finding, along with the participant’s reported preference for formal education as a setting for discussing potential genetic-related risks, provides support for utilizing the formal education setting as a place to implement JGD education. Although genetic concepts are typically part of high school curriculum, teaching AJ individuals about genetic concepts as it relates to AJ ancestry prior to high school is likely to benefit their long-term health behaviors.

Genetics is a complex topic with ever-evolving information that requires constant learning from the most well-versed genetic experts. For AJ individuals, the
complex information comes with medical decisions and an additional emotional component with the potential to impact not only themselves, but their future and family. Familiarity with reproductive risks to be a carrier and *BRCA1/2* cancer susceptibility genes begins the process of AJ Genetic Health Literacy. For this reason, it is important to begin familiarizing the AJ population about Jewish genetic health topics from a young age and over time to allow for additional and necessary processing, learning, and decision-making.

The earlier AJ individuals are exposed to genetic concepts related to their ancestry, the more time and opportunity a person has to improve their “AJ Genetic Health Literacy.” Elements from the Health Belief Model and the Transtheoretical Model (TTM), also called the Stages of Change Model, were used in combination with the results from our study to propose the “Stages of Achieving AJ Genetic Health Literacy” model (Figure 2.7) (Prochaska & DiClemente, 1983; Rosenstock, 1960). The model serves as a helpful framework for understanding the process of developing “AJ Genetic Health Literacy” and healthy genetic behaviors. Stakeholders invested in AJ community education would benefit from utilizing this framework when developing long-term and sustainable strategies to elicit a community-movement towards healthy genetic behaviors (i.e. sharing carrier results with relatives, following NCCN cancer screening guidelines, consideration of reproductive options for carrier couples, etc.).

**Figure 2.7 Stages of Achieving AJ Genetic Health Literacy**
The goal of the educational efforts established using this model is for AJ individuals to achieve “Ashkenazi Jewish Genetic Health Literacy” prior to being of testing age (18+). We define “Ashkenazi Jewish Genetic Health Literacy” as the degree to which individuals with one or more grandparent(s) of Ashkenazi Jewish ancestry have the competence to seek out, process, and understand basic genetic health information and services necessary to make appropriate, autonomous health decisions regarding the inherited risk to be a carrier of Jewish genetic diseases and BRCA1/2 cancer susceptibility mutations.

As identified by our study, familiarity is the initial steppingstone towards knowledge. Following familiarity, an individual’s interpretation of their risk to be a carrier of a JGD or have a BRCA1/2 mutation needs to be identified as personally relevant enough to take action towards seeking out additional information. The ‘Educational Curiosity’ step involves the individual having a desire to learn the information, whether through a medical provider, social media, local presentation, etc.

It is important for educators to be well-informed themselves and ensure accurate, useful educational materials and resources are easily accessible for all age groups in order for “AJ Genetic Health Literacy” to be achieved. Educational efforts should not cease after health literacy has been achieved, as genetic information and technology are ever-evolving.

Following ‘Educational Curiosity’, a working knowledge of the genetic information will allow individuals to consider additional implications of genetic testing
without pressure to make an immediate decision due to approaching reproductive age or cancer screening age.

By beginning the process towards “AJ Genetic Health Literacy” between 11-14 years old, as suggested by many of our participants, it is plausible to equip AJ individuals by age 18 with the necessary skills to utilize their health literacy to make informed, autonomous decisions about genetic testing and medical management. The 11-14 year old age group is an important time for many Jewish adolescents as they prepare to become adults in the eyes of Jewish law through the celebrations of ‘bar mitzvah’ at age 13 for boys and ‘bat mitzvah’ at age 12 for girls. Hebrew schools and Jewish Day schools begin preparing Jewish adolescents for the ceremony nearly two years prior, around age 11. The extensive preparation includes a formal education component with a primary focus of studying the Torah and the Hebrew language.

It is our recommendation that an introductory module about Jewish genetic health concepts be incorporated into the bar/bat mitzvah curriculum by Hebrew schools and Jewish Day schools. By doing so, young AJ adults will take the first step towards achieving “AJ Genetic Health Literacy” and implementing healthy behaviors. Individuals with an understanding of their genetic tie to Judaism may also feel a deeper connection to continue practicing Jewish traditions beyond their bar/bat mitzvah, adding additional value of our recommendation to the Jewish community.

The slow, intentional transition from familiarizing, processing, learning, decision-making to managing one’s health will aid Jewish genetic health educators in identifying continuous, long-term, and sustainable efforts completed over time.
instance education sessions are helpful to bring awareness to the Jewish community about their potential to be a carrier of a JGD or a BRCA1/2 mutation, the method should not be relied upon as complete education on the complex and detailed topics. Eliminating the standard, single-instance education sessions currently being utilized is also likely to lessen the burden placed on the small percentage of Jewish genetic health educators as they begin to share the responsibility of educational efforts with other stakeholders.

Organizations dedicated to providing Jewish genetic health education currently offer accurate information through a variety of resource materials, tool kits, and presentations for all ages and assist in the coordination of free, local screening events. The study showed these resources are likely underutilized, yet participants identified Jewish and non-profit organizations and similar resources as recommendations for future educational efforts. It is our recommendation that organizations utilize Figure 2.7 to review and modernize their current resources and identify updated educational outreach strategies for all age groups within the AJ community and for stakeholders.

Young adults of AJ ancestry are in need of updated strategies to improve their health literacy regarding their reproductive risk for JGDs and risk for cancer predisposition. In order for the AJ community to have the opportunity to achieve “AJ Genetic Health Literacy,” collaborative educational efforts between healthcare providers, religious leaders, schools, various organizations, and family members are a vital component. The implementation of an introductory module to AJ genetic health concepts by Hebrew schools and Jewish Day schools into the bar/bat mitzvah curriculum
will allow for education, family communication, and emotional processing to occur over time and result in an understanding of healthy genetic behaviors prior to testing age (18+).

2.6 LIMITATIONS

Like many cohorts in research on targeted populations, our sample was not geographically, socioeconomically, or ethnically diverse. The majority of respondents were from the southern region of the United States and identified as a Reform Jew, despite the intentional effort to contact various organizations from all regions of the country and reach people from all denominations of Judaism. An additional limitation includes the unreliability of self-reported AJ ancestry. Additionally, the study’s sample size was small in comparison to the estimated population of Ashkenazi Jewish individuals, many of which are likely to be young adults in our targeted age group.

2.7 FUTURE DIRECTIONS

We recommend an introductory module to AJ genetic health concepts be incorporated into the bar/bat mitzvah curriculum by Hebrew schools and Jewish Day schools. Follow-up studies may include the identification and validation of assessment items, preference for resource material, community uptake, measurement of progress, and many more.

Another follow-up study could consider implementing the participant’s suggestions for various types of survey advertisements and measuring the most useful strategies for education outreach. A study of this type would be informative to assess the utility of their suggestions. Similarly, identifying the most successful places for
education and types of educational materials could further direct future educational efforts. A study with this design would benefit from utilizing a pre- and post-test knowledge assessment. In general, studies assessing AJ young adult’s familiarity, risk perception, and knowledge are necessary to conduct on a continuum in order to measure the success of educational efforts and identify improved practices.
CHAPTER 3: CONCLUSION

The impacts of Jewish genetic health education reach beyond a single individual to an entire community. People with AJ ancestry often feel a sense of pride regarding their heritage and traditions and emphasize the importance of passing on tradition to younger generations in order to keep the community thriving. Partnering with Hebrew schools and Jewish Day schools to begin familiarizing AJ adolescents about Jewish genetic health concepts as they prepare for bar/bat mitzvah around age 11 will better equip their future selves to make autonomous, informed medical decisions and healthy genetic behaviors.

The collaboration of healthcare providers, religious leaders, schools, organizations, and family members are a vital component towards the pursuit of “Ashkenazi Jewish Genetic Health Literacy.” Educational efforts should not cease once genetic health literacy has been achieved, as genetic information and technology are ever-evolving. Updated, long-term strategies utilizing the suggestions of the young adult AJ population are the key to future educational efforts.
REFERENCES


Daly, M. B., Pal, T., Berry, M. P., Buys, S. S., Dickson, P., Domchek, S. M., Elkhanany, A., Friedman, S., Goggins, M., Hutton, M. L., CGC, Karlan, B. Y., Khan, S., Klein, C., Kohlmann, W., CGC, Kurian, A. W., Laronga, C., Litton, J. K., Mak, J. S., ... Dwyer,


Department of Health and Human Services.


https://doi.org/10.1080/07347330802359776


https://doi.org/10.1177/1363459312442420

Ashkenazi Jewish population. *Journal of the American College of Medical Genetics, 19*(5), 529-536. https://doi.org/10.1038/gim.2016.154
APPENDIX A: RECRUITMENT INVITATION

A.1 Email to Leadership at Organizations (i.e. Chabad on Campus, Hillel, JCC, Rabbis, etc.)

A.1.1 Email Content
Hi ____,

I am a graduate student at the University of South Carolina in the genetic counseling program. I have been contacting Jewish organizations across the country looking for the perspective of young adults (ages 18-27) with Ashkenazi Jewish background to participate in a 10-15 minute survey to support my thesis research regarding Jewish genetic health. Those who complete the survey are eligible to enter to win one of five $100 gift cards, thanks to the generous donation from the Columbia Jewish Federation.

Your organization’s participation in this project would greatly help contribute to its success! Do you have a staff member at your organization who oversees your organization’s newsletter, social media accounts, or other forms of communication? I would like to connect with them in hopes of getting the word out about the survey.

I have attached a Word document with pre-written descriptions and information regarding various ways to distribute the questionnaire, including a flyer to print or post on social media, copying the survey link in your next newsletter, or any other way that works best for your organization. Please see the ‘Survey- Distribution Information’ attachment.

I am happy to provide any additional information about the project. I look forward to hearing from you!

Kind regards,
Hayley Granger

Thesis Committee:
Janice Edwards, MS, CGC, Program Director- UofSC Genetic Counseling Program
Karen Grinzaid, MS, CGC, CCRC, Executive Director- JScreen at Emory University
Adele Schneider, MD, FACMG, Professor of Pediatrics- Thomas Jefferson University; Clinical Geneticist- Wills Eye Hospital
Caroline Say, MS, CGC, Clinical Cancer Genetic Counselor- UofSC & Prisma Health
A.1.2 Attachment: ‘Survey Distribution Information’ Document

Jewish Genetic Health Survey: Distribution Information

Newsletter/ Email List:
Do you have Ashkenazi Jewish background? Are you between ages 18-27? Take the 10-minute survey regarding your knowledge about Jewish genetic health and enter to win one of five $100 gift cards! Click here to begin the survey: http://bit.ly/jewishgh

If you know an organization that would be interested in sharing the survey or have further questions, please contact Hayley Granger (Hayley.Granger@uscmed.sc.edu).

Thank you!

Hayley Granger
Genetic Counseling Student

Paper Flyer- Print ‘Survey Image’ (see attachments) and display in common areas within your meeting place

Social Media- Instagram (Post & Story) / Facebook/ Twitter, etc.

INSTAGRAM STORY
Re-share from USC-Genetic Counseling Program account

1. Search “uscgcp” → Account name: USC Genetic Counseling Program
2. Look for light and dark blue image posted on 10/30/22 (see “Survey Image” attachment in email for reference)
   - Link: https://www.instagram.com/p/CkWQP7UXy/?igshid=MDJmNzVkJmY=
3. Share to ‘Story’...
   - Select ‘paper airplane’ symbol near the ‘heart’ symbol
4. Select ‘Add post to your story’
5. Add ‘Link’ sticker...
   - Click ‘post-it-note with smiley face’ symbol in top right corner
   - Select ‘link’ symbol (2nd column, 4th row)
   - Copy and paste shortened survey link into URL space provided- http://bit.ly/jewishgh
6. Add caption
   - Suggested caption: “It only takes 10-15 minutes (: Click the link to begin the survey! → [insert link sticker]”
7. Post to Story
   - **Please consider posting to your Instagram Story more than once!
INSTAGRAM POST

Make an Original Post

1. Include “Survey Image” provided (see attachments)
2. Use caption provided below:

   “Do you have Ashkenazi Jewish background? Are you between ages 18-27? A genetic counseling student at the University of South Carolina welcomes you to participate in a 10-15 minute survey regarding Jewish genetic health. Those who complete the survey are eligible to enter a raffle to win one of five $100 gift cards! Winners will be selected in February 2023.

   Type “bit.ly/jewishgh” into your web browser to begin the survey!”

   - Add the following to the caption provided above: “Link can also be found in our bio.”
   - **Why is this important?? - Captions on Instagram posts do not allow clickable links or copy and paste. This option makes it easy for people to directly access the survey.**
A.2 Social Media Post

A.2.1 Facebook Groups

Hi, all!

I am a graduate student at the University of South Carolina in the genetic counseling program. I am looking for young adults (ages 18-27yo) with Ashkenazi Jewish background to participate in a 10-15 minute survey to support my thesis research regarding Jewish genetic health.

Those who complete the survey are eligible to enter to win one of five $100 gift cards! Winners will be selected in February 2023.

Click here to begin the survey: http://bit.ly/jewishgh

A.3. Phone Call Script

Hi! My name is Hayley Granger. I am a second year genetic counseling student at the University of South Carolina. I am looking for young adults with Jewish background to participate in a survey for my thesis project.

[If speaking to human]

Is there someone I can talk to about posting a flyer in your community building or sharing about the survey in your newsletter or email list?

[If sent to voicemail]

I would like to connect with the person at your organization who handles community involvement, social media, or a newsletter, if you have one. Please call me back to talk further about the project or answer any questions you may have. My number is ***.***.****. Thank you
APPENDIX B: PARTICIPANT QUESTIONNAIRE

Start of Block: Introduction

University of South Carolina School of Medicine
Genetic Counseling Program

Greetings, participants!

I am a master’s degree student in genetic counseling at the University of South Carolina. I would like to invite you to complete the brief survey attached, the goal of which is to identify how much Ashkenazi Jewish (AJ) individuals know about Jewish Genetic Diseases and BRCA1 and BRCA2 cancer predisposition genes. Your responses will help healthcare providers, educators, religious leaders, and community leaders better understand the information that is widely known by AJ individuals and identify factors that contribute to this knowledge.

The survey will take approximately 15-20 minutes to complete. As a thank you, you will have the chance to enter your email address to win one of five $100 Visa gift cards at the completion of the survey. We do not ask for additional contact information.

Please know that your responses throughout the survey will be used for this research project only and kept anonymous and confidential. At any time, you may withdraw from the study by closing the browser.

Thank you for your time in participating in this survey. If you have questions regarding the research, you may contact either myself or my faculty advisory, Janice Edwards, MS, CGC, using the contact information provided below. If you have any questions about your rights as a research participant, you may contact the Office of Research Compliance at the University of South Carolina at (803) 777-7095.

Hayley Granger, B.S.
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Hayley.Granger@uscmed.sc.edu

Janice Edwards, M.S., CGC
Program Director
University of South Carolina, School of Medicine
Two Medical Park Rd, Suite 208
Columbia, SC 29203
Janice.Edwards@uscmed.sc.edu
Q53 Please verify you are not a robot.

End of Block: Introduction

Start of Block: Eligibility/ Demographic Questions

We would like to ask you a few questions about your background. Some of these questions will be used to make sure you are eligible for participation in the survey.

1. Through what organization did you find this survey?
   - Chabad On Campus ...................................... 1
   - Facebook Group: ______ (included in 8)
   - Hillel ............................................................ 2
   - Jewish Community Centers (JCC) ...................... 4
   - Sorority/ Fraternity ........................................ 5
   - Synagogue or Rabbi ....................................... 6
   - Other (please specify) __________________________
     Campus Org (outside Chabad/ Hillel) ............... 3
     Personal Referral .......................................... 7
     Social Media (including FB group) .................... 8

2. Do you have Ashkenazi (Central/ Eastern European) Jewish background? (i.e. at least one of your grandparents are Ashkenazi Jewish)
   - Yes ............................................................... 1
   - Yes, I think so ................................................. 2
   - No ................................................................. 3
   - I don’t know .................................................... 4

   *Skip to: Early Close If Q2= ‘No’ or ‘I don’t know’.*

3. To which denomination of Judaism do you identify? Check all that apply.
   - Reform .......................................................... 1
   - Conservative .................................................. 2
   - Reconstructionist ........................................... 3
   - Orthodox ....................................................... 4
   - Other ______ ................................................... 5
4. In which state do you currently reside?

□ Alabama (1) ... I do not reside in the United States (53)

Northeast ...................................................... 1
    CT, MA, ME, NH, NJ, NY, PA, RI, VT
Midwest ............................................................ 2
    IL, IN, IA, KS, MI, MN, MO, NE, ND, OH, SD, WI
South ................................................................. 3
    AL, AR, DE, DC, FL, GA, KY, LA, MD, MS, NC, OK, SC, TN, TX, VA, WV
West ................................................................. 4
    AK, AZ, CA, CO, HI, ID, MT, NV, NM, OR, UT, WA, WY
International .................................................... 5
    I do not reside in the United States

5. What is your age? ______

**Skip to: Early Close If Q5 is <18 or >27.**

6. What is your gender?

○ Female .............................................................. 1
○ Male ................................................................. 2
○ Transgender Female ............................... 3
○ Transgender Male ...................................... 4
○ Other ______ ..................................................... 5

**IF Gender= ‘3’, ‘4’, or ‘5’= Gender Minority .... 3

7. What is the highest level of education you have completed?

○ High school .................................................. 1
○ Associate’s degree ......................................... 2
○ Bachelor’s degree ........................................... 3
○ Graduate/ professional .................................... 4
○ Medical school .............................................. 5
[Early Close]: Thank you for your interest in the study. Based on your initial responses, you are not eligible to participate. If you have any further questions, please email us at usc.gcsurvey@gmail.com.

End of Block: Eligibility/ Demographic Questions

Start of Block: JGD Awareness

JGD Awareness

Here are a few terms that may be helpful when answering the upcoming questions:

**Jewish Genetic Diseases (JGDs):** A group of inherited medical conditions that affect Jewish individuals at a higher frequency compared to the general population (i.e. Tay-Sachs Disease, Gaucher Disease, etc.).

**Carrier Screening:** A type of genetic test performed on an individual to determine if they are a carrier for specific genetic diseases. This test is most often used by couples who are considering becoming pregnant to determine the risk for their child to inherit a genetic disease. If a couple is found to be carriers of the same genetic disease, they may choose to reduce the risk to have an affected child by utilizing in vitro fertilization (IVF) with preimplantation genetic diagnosis (PGD), prenatal diagnosis, non-carrier sperm or egg donation, or adoption.

8. How much have you heard about Jewish Genetic Diseases (JGDs)?

   ○ I have never heard of JGDs. ................................................................. 1
   ○ I have heard of JGDs, but I do not know much about them .............. 2
   ○ I know a fair amount about these diseases ......................................... 3
   ○ I know a great deal about these diseases ............................................ 4

9. How much have you heard about the association between Ashkenazi Jewish (AJ) ancestry and specific genetic diseases?

   ○ I have never heard of this association ................................................ 1
   ○ I have heard of the association, but I do not know much about it ...... 2
10. Where did you learn about this association?

Please check all that apply and fill in the blank with additional information.

- At home/ parents ................................................................. 1
- Friends/ family outside of the home ...................................... 2
- Social media: _____ .................................................................. 3
- Jewish organization: ______ ..................................................... 4
- Non-profit community-based education organization: _____ ...... 5
- Physician (please identify specialty) _____ ............................... 6
- Other Healthcare Provider (please identify specialty) ______ ...... 7
- Other: ______
  (School) .................................................................................. 8
  (Entertainment i.e. TV)................................................................. 9
  (Advertisement i.e. PSA on train) .............................................. 10

11. At what age did you learn about this association?

- Before age 10 ................................................................. 1
- 11-14 ...................................................................................... 2
- 15-17 ...................................................................................... 3
- 18-22 ...................................................................................... 4
- After age 22 ................................................................. 5

12. Have you and/or your family members undergone carrier screening for JGDs (i.e. Tay-Sachs Disease)?

- No ................................................................. 0
- Yes ................................................................. 1
12a. Please identify who in your family has had carrier screening (i.e. 1 parent, 2 siblings, 3 cousins, etc.): _____________

- Self........................................ 1
- 1 Family Member ............ 2
- 2+ Family Members........ 3

- I don’t know ........................................ 2

Skip to: Q14 if Q12 = No
Skip to: Q14 if Q12 = I don’t know

13. Have you and/or your family members received any of the following results?

Check all that apply.

- Diagnosis of a JGD........................................... 1

13a. Please identify who in your family received this result (i.e. 1 parent, 2 siblings, 3 cousins, etc.): _____________

- Self........................................ 1
- 1 Family Member ............ 2
- 2+ Family Members........ 3

- Carrier for a JGD ........................................... 2

13b. Please identify who in your family received this result (i.e. 1 parent, 2 siblings, 3 cousins, etc.): _____________

- Self........................................ 1
- 1 Family Member ............ 2
- 2+ Family Members........ 3

- None of the above ........................................... 3
- I don’t know.................................................. 4
14. How do you perceive your chance of being a carrier of a genetic disease compared to that of a non-Jewish individual?

- Very low .................................................. 1
- Low ............................................................ 2
- Neutral ....................................................... 3
- High .......................................................... 4
- Very High .................................................... 5

15. There are reproductive options for couples that are found to be carriers of JGD.

- Disagree ..................................................... 1
- Somewhat Disagree ..................................... 2
- Neutral ....................................................... 3
- Somewhat Agree ......................................... 4
- Agree .......................................................... 5
- I don’t know ................................................ 6

End of Block: JGD Awareness

Start of Block: JGD Knowledge Questions

The next few questions are about your opinions and beliefs regarding JGDs. *This is not a test.*

Please indicate whether you agree or disagree with each statement.

16. JGDs are only found in the Ashkenazi (Central/ Eastern European lineage) Jewish population.

- Agree .......................................................... 0
- Disagree ....................................................... 1

17. If both parents are carriers of a JGD, all of their children will be affected.

- Agree .......................................................... 0
- Disagree ....................................................... 1
18. All JGDs are fatal with no known treatment or cure.
   - Agree ................................................................. 0
   - Disagree ........................................................... 1

19. Children of interfaith couples are not at risk of JGDs.
   - Agree ................................................................. 0
   - Disagree ........................................................... 1

Page Break

20. If a family has no known history of Jewish genetic disorders, future generations will not be at risk.
   - Agree ................................................................. 0
   - Disagree ........................................................... 1

21. Carriers of an autosomal recessive disorder typically have no symptoms.
   - Agree ................................................................. 1
   - Disagree ........................................................... 0

22. If the carrier test is negative, you have no risk of having a child with any of these conditions.
   - Agree ................................................................. 0
   - Disagree ........................................................... 1

23. Which of the following puts a child at risk to inherit a recessive genetic disease?
   - Only one partner is a carrier. ............................................. 0
   - Both partners are a carrier, but for different genetic diseases. ....... 0
   - Both partners are carriers for the same genetic disease. ............... 1
   - I am not sure. .................................................................. 0

24. Approximately how many American Jews of Ashkenazi descent are a carrier for at least one JGD?
Here are a few terms that may be helpful when answering the upcoming questions:

**BRCA1 and BRCA2 (BRCA1/2):** These are two of many tumor suppressor genes that work to protect our body from developing cancer. Most individuals have two working copies of both BRCA1 and BRCA2 genes. When a change, or mutation, is found in one of the two copies of either BRCA1 or BRCA2, the person has an increased chance to develop certain cancers often at a younger age. Therefore, BRCA1/2 are often referred to as ‘cancer susceptibility genes.’

**Pathogenic Variant (mutation, change):** A genetic alteration identified through genetic testing that is known to increase an individual’s likelihood to have or develop a certain disease.

**Variant of Uncertain Significance (VUS):** A type of result that can be identified from genetic testing. It indicates the finding of a genetic change but the impact on the individual’s cancer risk is not well understood.

25. Do you have a personal or family history of breast, ovarian, prostate, or pancreatic cancer?

   ○ No ................................................................. 0
   ○ Yes ................................................................. 1
   ○ I don’t know..................................................... 2

26. Prior to this survey, how much had you heard about BRCA1 and BRCA2 cancer susceptibility genes?

   ○ I have never heard of these genes. ........................................ 1
   ○ I have heard of these genes, but I do not know much about them. .... 2
   ○ I know a fair amount about these genes. ................................ 3
27. Where did you learn about BRCA1 and BRCA2 cancer susceptibility genes?

Check all that apply and fill in the blank with additional information.

- At home/ parents .............................................. 1
- Friends/ family outside of the home ................................ 2
- Social media: ______ ........................................... 3
- Jewish organization: _____ ........................................ 4
- Non-profit community-based education organization: _____ .......................... 5
- Physician (please identify specialty) _____ ........................................ 6
- Other Healthcare Provider (please identify specialty) _____ ............ 7
- Other: ______
  - (School) ................................................................. 8
  - (Entertainment i.e. TV shows) ........................................ 9
  - (Genetic Testing Services) ............................................. 10

28. At what age did you learned about BRCA1 and BRCA2 cancer susceptibility genes?

- Before age 10 ................................................. 1
- 11-14 ................................................................. 2
- 15-17 ................................................................. 3
- 18-22 ................................................................. 4
- After age 22 ..................................................... 5
29. Have you and/or your family members undergone testing for changes in the *BRCA1* or *BRCA2* genes?

- No ................................................................. 0
- Yes ................................................................. 1

29a. Please identify who in your family had testing for *BRCA1/2* (i.e. 1 parent, 2 siblings, 3 cousins, etc.): _____________

- Self............................................................... 1
- 1 Family Member ................................. 2
- 2+ Family Members......................... 3
- I don’t know ................................................. 2

*Skip to: End of Block If Q29 = No*

*Skip to: End of Block If Q29 = I don’t know*

30. If ‘1’ to Q29: What were the results of the *BRCA1/2* testing?

- A pathogenic variant (i.e. mutation) in *BRCA1* or *BRCA2* was identified.......... 1

30a. Please identify who in your family received this result (i.e. 1 parent, 2 siblings, 3 cousins, etc.): _____________

- Self............................................................... 1
- 1 Family Member ................................. 2
- 2+ Family Members......................... 3

- A variant of uncertain significance (VUS) was identified................................. 2

30b. Please identify who in your family received this result (i.e. 1 parent, 2 siblings, 3 cousins, etc.): _____________

- Self............................................................... 1
31. Did the individual(s) meet with a genetic counselor before, during, or after receiving the results?
   - Yes ........................................................................... 1
   - No ........................................................................... 2
   - I don’t know .............................................................. 3

End of Block: BRCA1/2 Awareness

Start of Block: BRCA1/2 Knowledge Questions

The next few questions are about your opinions and beliefs regarding BRCA1/2 cancer susceptibility genes. *This is not a test.*

Please indicate whether you agree or disagree with each statement.

32. Pathogenic variants in BRCA1/2 increase the chance for both men and women to develop cancer.
   - Agree ........................................................................... 1
   - Disagree ........................................................................ 0

33. If a female tests negative for mutations in BRCA1 and BRCA2, then she will not get breast or ovarian cancer.
   - Agree ........................................................................... 0
   - Disagree ........................................................................ 1
34. If a male or female tests positive for mutations in BRCA1 or BRCA2, then they are both at risk to develop breast cancer.

- Agree ................................................................. 1
- Disagree ............................................................. 0

35. A mother can pass down a BRCA1 or BRCA2 mutation to her sons.

- Agree ................................................................. 1
- Disagree ............................................................. 0

36. A father can pass down a BRCA1 or BRCA2 mutation to his daughters.

- Agree ................................................................. 1
- Disagree ............................................................. 0

37. If an individual is a carrier of a BRCA1 or BRCA2 mutation, each of his or her children has a 1 in 4 chance of also having a mutation.

- Agree ................................................................. 0
- Disagree ............................................................. 1

38. If there is no history of breast or ovarian cancer in my family, I am not at risk for carrying a BRCA1 or BRCA2 mutation.

- Agree ................................................................. 0
- Disagree ............................................................. 1

39. Individuals with AJ ancestry may want to consider genetic testing for BRCA1 and BRCA2, even if they have no personal or family history of cancer.

- Agree ................................................................. 1
- Disagree ............................................................. 0

40. Approximately how many American Jews of Ashkenazi descent have a genetic mutation in BRCA1/2 cancer susceptibility genes?
Great! You’re almost finished.

As you may know, individuals with Ashkenazi Jewish (AJ) ancestry have a higher chance to carry genetic changes (i.e. mutations) for certain autosomal recessive conditions and cancer-causing mutations in BRCA1 and BRCA2 genes.

Previous studies show that AJ individuals have limited knowledge regarding their genetic health risk. In an effort to improve educational efforts, we would like to hear your suggestions about how to better educate AJ individuals about their genetic health.

41. What suggestions do you have about where AJ individuals may want to learn about their genetic health risks?

(i.e. a presentation at a synagogue or other religious group, public online modules, etc.) _____________________

Themes (developed post-survey):

At home/ parents ................................................................. 1
Friends/ family outside of the home ..................................... 2
Social Media ........................................................................ 3
Public, Online Modules ..................................................... 4
Jewish organization/ Religious Group ............................... 5
Non-profit, community-based educational Org. & Impacted Families 6
Physician, Researchers, & “Experts” in Genetics................... 7
School ............................................................................. 8
Synagogue ....................................................................... 9
42. What suggestions do you have about how to reach AJ individuals who are unaware of their genetic health risk?

(i.e. social media, flyer in a community building, etc.) __________

Themes (developed post-survey):

Virtual Communication (1,0) vs. Physical Communication (1,0)
Religious Source (1,0) vs. Non-Religious Source (1,0)

43. At what age do you feel it is most appropriate for AJ individuals to learn about their genetic health risk?

- Before age 10 ................................................. 1
- 11-14 ................................................................. 2
- 15-17 ................................................................. 3
- 18-22 ................................................................. 4
- After age 22 ....................................................... 5

End of Block: Wrap-up

Thank you for your participation!

Please fill in your email below if you would like to enter the raffle to win one of five $100 Visa gift cards. Winners will be selected at random in February 2023 and contacted via email.

Email: __________
APPENDIX C: ADDITIONAL RESULT TABLES

Table C.1 Demographic Predictors: JGD v. BRCA1/2

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<th>BRCA1/2 Knowledge</th>
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<tr>
<td>Synagogue/ Rabbi</td>
<td>3 (2.5)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Personal Referral</td>
<td>22 (18.5)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Social Media</td>
<td>27 (22.7)</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

* *p value <.001
** **p value <.05
<table>
<thead>
<tr>
<th>Familiarity with JGDs</th>
<th>Frequency (%)</th>
<th>mean (SD)</th>
<th>p value</th>
</tr>
</thead>
<tbody>
<tr>
<td>I have never heard of Jewish Genetic Diseases (JGDs).</td>
<td>24 (20.2)</td>
<td>5.9 (1.7)</td>
<td></td>
</tr>
<tr>
<td>I have heard of JGDs, but I do not know much about them.</td>
<td>66 (55.5)</td>
<td>6.1 (1.8)</td>
<td>.045</td>
</tr>
<tr>
<td>I know a fair amount about these diseases.</td>
<td>24 (20.2)</td>
<td>6.8 (1.4)</td>
<td></td>
</tr>
<tr>
<td>I know a great deal about these diseases.</td>
<td>5 (4.2)</td>
<td>7.2 (0.8)</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Familiarity with association between AJ ancestry and genetic diseases</th>
<th>Frequency (%)</th>
<th>mean (SD)</th>
<th>p value</th>
</tr>
</thead>
<tbody>
<tr>
<td>I have never heard of this association.</td>
<td>24 (20.2)</td>
<td>5.9 (1.8)</td>
<td></td>
</tr>
<tr>
<td>I have heard of this association, but I do not know much about them.</td>
<td>45 (37.8)</td>
<td>5.8 (2.0)</td>
<td>.058</td>
</tr>
<tr>
<td>I know a fair amount about this association.</td>
<td>41 (34.5)</td>
<td>6.8 (1.2)</td>
<td></td>
</tr>
<tr>
<td>I know a great deal about this association.</td>
<td>9 (7.6)</td>
<td>6.8 (1.0)</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Places of Learning</th>
<th>Frequency (%)</th>
<th>mean (SD)</th>
<th>p value</th>
</tr>
</thead>
<tbody>
<tr>
<td>At home/ from parents</td>
<td>59 (49.6)</td>
<td>6.3 (1.5)</td>
<td>.843</td>
</tr>
<tr>
<td>Other/ NA</td>
<td>60 (50.4)</td>
<td>6.2 (1.8)</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Age of Learning</th>
<th>Frequency (%)</th>
<th>mean (SD)</th>
<th>p value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Before age 10</td>
<td>9 (7.6)</td>
<td>7.1 (1.7)</td>
<td></td>
</tr>
<tr>
<td>11-15</td>
<td>31 (26.1)</td>
<td>6.1 (1.7)</td>
<td></td>
</tr>
<tr>
<td>15-17</td>
<td>26 (21.8)</td>
<td>6.3 (1.5)</td>
<td>.518</td>
</tr>
<tr>
<td>18-22</td>
<td>23 (19.3)</td>
<td>6.3 (1.7)</td>
<td></td>
</tr>
<tr>
<td>After age 22</td>
<td>6 (5.0)</td>
<td>5.7 (2.6)</td>
<td></td>
</tr>
<tr>
<td>Never</td>
<td>24 (20.2)</td>
<td>5.9 (1.8)</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Carrier Screening of Relatives and/or Self</th>
<th>Frequency (%)</th>
<th>mean (SD)</th>
<th>p value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Yes</td>
<td>23 (46.2)</td>
<td>6.7 (1.3)</td>
<td>.203</td>
</tr>
<tr>
<td>No</td>
<td>55 (19.3)</td>
<td>6.3 (1.6)</td>
<td></td>
</tr>
<tr>
<td>Unsure</td>
<td>41 (34.5)</td>
<td>5.9 (1.9)</td>
<td></td>
</tr>
</tbody>
</table>

*p value <.001
**p value <.05
**Table C.3** Comparison of JGD Knowledge Assessment outcomes between studies.

<table>
<thead>
<tr>
<th>Questions</th>
<th>Granger et al., 2023 (N=119)</th>
<th>Warsch et al., 2014 (N=412)</th>
<th>( p ) value</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Composite Knowledge Score</strong></td>
<td>5.47 (1.4)</td>
<td>5.69 (-- (^a))</td>
<td>.036**</td>
</tr>
<tr>
<td><strong>Average % Correct</strong></td>
<td>78.2%</td>
<td>81.4%</td>
<td></td>
</tr>
<tr>
<td>1. Jewish genetic disorders are only found in the Ashkenazi (Central/ Eastern European lineage) Jewish population. (Agree/ Disagree)</td>
<td>89 (74.8)</td>
<td>310 (75.2)</td>
<td>.912</td>
</tr>
<tr>
<td>2. If both parents are carriers of a Jewish genetic disorder, all of their children will be affected. (Agree/ Disagree)</td>
<td>91 (76.5)</td>
<td>376 (91.3)</td>
<td>&lt;.001*</td>
</tr>
<tr>
<td>3. All Jewish genetic disorders are fatal with no known treatment or cure. (Agree/ Disagree)</td>
<td>106 (89.1)</td>
<td>391 (94.9)</td>
<td>.022**</td>
</tr>
<tr>
<td>4. Children of interfaith couples are not at risk of Jewish genetic disorders. (Agree/ Disagree)</td>
<td>109 (91.6)</td>
<td>370 (89.9)</td>
<td>.563</td>
</tr>
<tr>
<td>5. If a family has no known history of Jewish genetic disorders, future generations will not be at risk. (Agree/ Disagree)</td>
<td>104 (87.4)</td>
<td>372 (90.3)</td>
<td>.361</td>
</tr>
<tr>
<td>6. Carriers of an autosomal recessive disorder have no symptoms. (Agree/ Disagree)</td>
<td>67 (56.3)</td>
<td>234 (56.8)</td>
<td>.924</td>
</tr>
<tr>
<td>7. If the carrier test is negative, you have no risk of having a child with any of these conditions. (Agree/ Disagree)</td>
<td>85 (71.4)</td>
<td>295 (71.6)</td>
<td>.971</td>
</tr>
</tbody>
</table>

\(^a\)Full dataset unavailable; SD cannot be calculated

*Correct answers are in **bold**.
Table C.4 Comparing Descriptive Statistics of JGD Knowledge Studies

<table>
<thead>
<tr>
<th>Variables</th>
<th>Granger et al., 2023 (N=119)</th>
<th>Warsch et al., 2014 (N=412)</th>
<th>p value</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Age (mean, SD)</strong></td>
<td>22.3 (2.7)</td>
<td>24.9 (4.7)</td>
<td>&lt;.001*</td>
</tr>
<tr>
<td><strong>Gender</strong></td>
<td></td>
<td></td>
<td>&lt;.001*</td>
</tr>
<tr>
<td>Female</td>
<td>78 (65.5)</td>
<td>215 (54.7)</td>
<td></td>
</tr>
<tr>
<td>Male</td>
<td>34 (28.6)</td>
<td>178 (45.3)</td>
<td></td>
</tr>
<tr>
<td>Gender Minority</td>
<td>7 (5.9)</td>
<td>--</td>
<td></td>
</tr>
<tr>
<td><strong>Education</strong></td>
<td></td>
<td></td>
<td>&lt;.001*</td>
</tr>
<tr>
<td>High School</td>
<td>57 (47.9)</td>
<td>9 (2.3)</td>
<td></td>
</tr>
<tr>
<td>Associate’s Degree</td>
<td>8 (6.7)</td>
<td>10 (2.6)</td>
<td></td>
</tr>
<tr>
<td>Bachelor’s Degree</td>
<td>45 (37.8)</td>
<td>190 (48.7)*</td>
<td></td>
</tr>
<tr>
<td>Graduate/ Professional Degree</td>
<td>8 (6.7)</td>
<td>127 (32.6)*</td>
<td></td>
</tr>
<tr>
<td>Medical School</td>
<td>1 (0.8)</td>
<td>54 (13.8)</td>
<td></td>
</tr>
<tr>
<td><strong>Religious Denomination</strong></td>
<td></td>
<td></td>
<td>&lt;.001*</td>
</tr>
<tr>
<td>Reform</td>
<td>66 (55.5)</td>
<td>145 (40.6)</td>
<td></td>
</tr>
<tr>
<td>Conservative</td>
<td>30 (25.2)</td>
<td>169 (47.3)</td>
<td></td>
</tr>
<tr>
<td>Orthodox</td>
<td>6 (5.0)</td>
<td>43 (12.0)</td>
<td></td>
</tr>
<tr>
<td>Other/ Unaffiliated</td>
<td>14 (11.8)</td>
<td>55 (13.3)</td>
<td></td>
</tr>
<tr>
<td>Reconstructionist</td>
<td>3 (2.5)</td>
<td>--</td>
<td></td>
</tr>
<tr>
<td><strong>Individuals with a family member known to be:</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Affected by a JGD</td>
<td>1 (0.8)</td>
<td>3 (0.7)</td>
<td></td>
</tr>
<tr>
<td>Carrier(s) of a JGD</td>
<td>5 (4.2)</td>
<td>5 (1.2)</td>
<td></td>
</tr>
</tbody>
</table>

*p value <.001
**p value <.05

*Degree in progress or completed

bp value cannot be calculated due to ‘Affected by a JGD’ N=1 for Granger et al., 2023
### Table C.5  Comparison of BRCA1/2 Knowledge Assessment Outcomes Between Studies

<table>
<thead>
<tr>
<th>Question</th>
<th>Granger et al., 2023 (N=119)</th>
<th>Hardy et al., 2022 (N= 328)</th>
<th>p value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Composite Knowledge Score mean (SD)</td>
<td>3.9 (0.87)</td>
<td>4.0 (0.69)</td>
<td>.074</td>
</tr>
<tr>
<td>Average % Correct</td>
<td>77.1%</td>
<td>80.0%</td>
<td></td>
</tr>
<tr>
<td>1. If a woman tests negative for mutations in BRCA1 and BRCA2, then she definitely will not get breast or ovarian cancer. (Agree/ Disagree)</td>
<td>106 (92.2)</td>
<td>320 (96.7)</td>
<td>&lt;.001*</td>
</tr>
<tr>
<td>2. A mother can pass down a BRCA1 or BRCA2 mutation to her sons. (Agree/ Disagree)</td>
<td>106 (92.2)</td>
<td>308 (93.1)</td>
<td>.085</td>
</tr>
<tr>
<td>3. A father can pass down a BRCA1 or BRCA2 mutation to his daughters. (Agree/ Disagree)</td>
<td>104 (90.4)</td>
<td>301 (91.0)</td>
<td>.161</td>
</tr>
<tr>
<td>4. If an individual is a carrier of a BRCA1 or BRCA2 mutation, each of his or her children has a 1 in 4 chance of also having the mutation. (Agree/ Disagree)</td>
<td>24 (20.9)</td>
<td>73 (22.1)</td>
<td>.636</td>
</tr>
<tr>
<td>5. If there is no history of breast or ovarian cancer in my family, I am definitely not at risk for carrying a BRCA1 or BRCA2 mutation. (Agree/ Disagree)</td>
<td>104 (90.4)</td>
<td>310 (93.7)</td>
<td>.011**</td>
</tr>
<tr>
<td>All Questions Correct</td>
<td>18 (15.1)</td>
<td>62 (18.9)</td>
<td>.357</td>
</tr>
</tbody>
</table>

* p value <.001  
** p value <.05  
*Correct answers are in bold.
**Table C.6** Descriptive Statistics: BRCA1/2 Knowledge Studies with All Participants

<table>
<thead>
<tr>
<th>Variables</th>
<th>Granger et al., 2023 (N=119)</th>
<th>Hardy et al., 2022 (N=331)</th>
<th>p value</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>mean (SD)</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Age</td>
<td>22.3 (2.7)</td>
<td>29.9 (5.8)</td>
<td>.001*</td>
</tr>
<tr>
<td>Gender</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Female</td>
<td>78 (62.1)</td>
<td>199 (60.1)</td>
<td>&lt;.001*</td>
</tr>
<tr>
<td>Male</td>
<td>34 (28.7)</td>
<td>132 (39.9)</td>
<td></td>
</tr>
<tr>
<td>Gender Minority</td>
<td>7 (6.1)</td>
<td>--</td>
<td></td>
</tr>
<tr>
<td>Region</td>
<td></td>
<td></td>
<td>&lt;.001*</td>
</tr>
<tr>
<td>Northeast</td>
<td>21 (17.6)</td>
<td>158 (47.7)</td>
<td></td>
</tr>
<tr>
<td>Midwest</td>
<td>14 (11.8)</td>
<td>44 (13.3)</td>
<td></td>
</tr>
<tr>
<td>South</td>
<td>58 (48.7)</td>
<td>72 (21.8)</td>
<td></td>
</tr>
<tr>
<td>West</td>
<td>25 (21.0)</td>
<td>49 (14.8)</td>
<td></td>
</tr>
<tr>
<td>International</td>
<td>1 (0.8)</td>
<td>2 (0.6)</td>
<td></td>
</tr>
<tr>
<td>Education</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>&lt; College graduate</td>
<td>65 (54.6)</td>
<td>38 (11.5)</td>
<td>&lt;.001*</td>
</tr>
<tr>
<td>College graduate +</td>
<td>54 (45.4)</td>
<td>293 (88.5)</td>
<td></td>
</tr>
<tr>
<td>Relatives with Breast, Ovarian, Prostate, or Pancreatic Cancer</td>
<td></td>
<td></td>
<td>&lt;.001*</td>
</tr>
<tr>
<td>Yes</td>
<td>71 (59.7)</td>
<td>137 (41.4)</td>
<td></td>
</tr>
<tr>
<td>No/ Unsure</td>
<td>48 (40.3)</td>
<td>194 (58.6)</td>
<td></td>
</tr>
<tr>
<td>Relatives tested for BRCA1/BRCA2</td>
<td></td>
<td></td>
<td>&lt;.001*</td>
</tr>
<tr>
<td>Yes</td>
<td>29 (24.4)</td>
<td>72 (21.8)</td>
<td></td>
</tr>
<tr>
<td>No</td>
<td>40 (33.6)</td>
<td>159 (48.2)</td>
<td></td>
</tr>
<tr>
<td>Unsure</td>
<td>50 (42.0)</td>
<td>99 (30.0)</td>
<td></td>
</tr>
<tr>
<td>Results of Relatives’ BRCA1/2 Genetic Testing</td>
<td></td>
<td></td>
<td>.303</td>
</tr>
<tr>
<td>Positive</td>
<td>6 (20.7)</td>
<td>7 (9.7)</td>
<td></td>
</tr>
<tr>
<td>Negative</td>
<td>19 (65.5)</td>
<td>51 (70.8)</td>
<td></td>
</tr>
<tr>
<td>Unsure</td>
<td>4 (13.8)</td>
<td>14 (19.4)</td>
<td></td>
</tr>
<tr>
<td>Familiarity with BRCA1 and BRCA2</td>
<td></td>
<td></td>
<td>.003**</td>
</tr>
<tr>
<td>Never heard of them</td>
<td>34 (28.6)</td>
<td>52 (15.7)</td>
<td></td>
</tr>
<tr>
<td>Did not know much about them</td>
<td>37 (31.1)</td>
<td>128 (38.7)</td>
<td></td>
</tr>
<tr>
<td>--------------------------------</td>
<td>-----------</td>
<td>------------</td>
<td></td>
</tr>
<tr>
<td>New a fair amount about them</td>
<td>37 (31.1)</td>
<td>108 (32.6)</td>
<td></td>
</tr>
<tr>
<td>Knew a great deal about them</td>
<td>11 (9.2)</td>
<td>43 (13.0)</td>
<td></td>
</tr>
</tbody>
</table>

* $p$ value <.001
** $p$ value <.05