Evaluation of two-year Jewish genetic disease screening program in Atlanta: insight into community genetic screening approaches

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Abstract Improvements in genetic testing technologies have led to the development of expanded carrier screening panels for the Ashkenazi Jewish population; however, there are major inconsistencies in current screening practices. A 2-year pilot program was launched in Atlanta in 2010 to promote and facilitate screening for 19 Jewish genetic diseases. We analyzed data from this program, including participant demographics and outreach efforts. This retrospective analysis is based on a de-identified dataset of 724 screenees. Data were obtained through medical chart review and questionnaires and included demographic information, screening results, response to outreach efforts, and follow-up behavior and preferences. We applied descriptive analysis, chi-square tests, and logistic regression to analyze the data and compare findings with published literature. The majority of participants indicated that they were not pregnant or did not have a partner who was pregnant were affiliated with Jewish organizations and reported 100 % AJ ancestry. Overall, carrier frequency was 1 in 3.9. Friends, rabbis, and family members were the most common influencers of the decision to receive screening. People who were older, had a history of pregnancy, and had been previously screened were more likely to educate others (all $p < 0.05$). Analysis of this 2-year program indicated that people who are ready to have children or expand their families are more likely to get screened and encourage others to be screened. The most effective outreach efforts targeted influencers who then encouraged screening in the target population. Educating influencers and increasing overall awareness were the most effective outreach strategies.

Keywords Jewish genetic diseases · Carrier screening · Genetic education · Community outreach

Introduction

Carrier screening in the Jewish population began in the early 1970s with hexosaminidase A (Hex A) enzyme assay targeting identification of carriers of Tay-Sachs disease (TSD) (Kaplan 1998). Over the last 40 years, genetic screening for TSD has dramatically reduced the incidence of this disease in the worldwide Jewish population, with at least a 90 % decrease in North America (Kronn et al. 1998). The great success of these earlier grass roots community-based genetic screening programs for TSD led to continued acceptance and utilization of carrier screening in Ashkenazi Jewish (AJ) communities worldwide (Rosner et al. 2009).

Technological improvements and an increased understanding of the genes underlying many genetic conditions have revolutionized approaches to carrier screening. The AJ carrier screening panel has evolved from a single enzyme assay test for TSD to DNA-based testing panels that include many more disorders today. In 2004, the American College of Obstetricians and Gynecologists (ACOG) (ACOG 2004) updated its recommended Ashkenazi Jewish carrier screening panel to include three disorders in addition to TSD. Two of these disorders—Canavan disease and familial dysautonomia—also occur at increased frequency in the AJ population. The carrier frequency of cystic fibrosis is comparable in both Ashkenazi Jewish and Caucasian populations and is significantly higher than in other ethnic groups. In 2008, the American College of Medical Genetics (ACMG) (Gross et al. 2008) recommended that an additional five disorders be offered to individuals of AJ descent as a part of preconception/prenatal carrier screening. These include Bloom syndrome, Fanconi anemia type C, Gaucher disease, Mucopolidosis type IV, and Niemann-Pick disease type A.

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Updated ACOG guidelines in 2009 stated that individuals of Ashkenazi Jewish descent may inquire about screening for the five additional diseases (ACOG 2009). The ACMG and ACOG guidelines have been effective in promoting carrier screening to this at risk population.

The majority of Jewish genetic disease programs in the U.S. currently recommend screening for the 9 disorders specified by ACMG and ACOG, as well as 10 other diseases with relatively high carrier frequencies (approximately 1 % or greater) in this population (Klugman et al. 2013). These screening programs and a growing number of medical practices offer carrier screening for these 19 conditions to individuals of AJ descent (Bonham et al. 2010; Strom et al. 2004). All 19 diseases are inherited in an autosomal recessive pattern with a 25 % risk of disease in each child of two parents who carry the same disease gene.

For a variety of reasons, many individuals with AJ ancestry are not effectively reached and offered carrier screening. Of those that are offered screening, many are already pregnant, a time when options for carrier couples are limited (Zlotogora 2009). In some cities, community-based Jewish genetic disease programs are in place. These programs provide educational outreach and serve to facilitate screening for the at risk population prior to conception.

Over the past 40 years, the Metropolitan Atlanta area has had two community-wide campaigns aimed at promoting carrier screening for the Jewish population. The first was a TSD carrier screening program initiated in 1975 (Marion et al. 1977). This program, coordinated by Emory University School of Medicine, screened >2300 individuals and was highly successful in creating awareness around the importance of screening for TSD. In the years that followed, there was no formal carrier screening program in the Atlanta area. Jews of reproductive age relied on their personal physicians to recommend and facilitate testing. Some were seen by local genetic counselors. Consequently, there were major inconsistencies in panels offered, in cost, and in post-testing follow-up.

In 2010, a two-year pilot program called the Atlanta Jewish Gene Screen (AJGS) was launched with the goal of filling this gap. Its mission was to promote awareness and facilitate preconception carrier screening for the Jewish population in the Atlanta metropolitan area. Based on a population survey of the Atlanta Jewish community in 2006, it was estimated that 3000 babies were born to Jewish families in Atlanta each year (Atlanta 2007). The goal of AJGS was to educate and provide screening options to as many of these individuals as possible, preferably prior to pregnancy, and to update screening for those who had been previously screened with more limited panels.

This study is a retrospective analysis of the AJGS program and was designed to identify key demographic parameters of the at risk population in Metropolitan Atlanta. We also evaluated the effectiveness of marketing and outreach efforts in creating awareness and encouraging screening. Results of this

study will help inform effective outreach strategies for carrier screening programs in the future.

Materials and methods

Program organization

The Atlanta Jewish Gene Screen (AJGS) was a community screening initiative directed by the Victor Centers for the Prevention of Jewish Genetic Diseases. Genetic services for AJGS were coordinated by the Emory University School of Medicine, Division of Medical Genetics. The two-year program ran from July 1, 2010 through July 31, 2012.

The AJGS program included extensive marketing and outreach campaigns and a variety of educational and promotional efforts. Outreach was targeted directly to the at risk population or through groups such as the rabbinic community, Jewish community and young adult organizations, and the medical community (primarily obstetricians and gynecologists). These outreach efforts included participation in local events, formal and informal presentations, and dissemination of program information. Marketing efforts included both traditional and digital approaches.

Individuals who were interested in pursuing screening had three options: community screening events, an office visit to the Emory Clinic, Division of Medical Genetics, or a visit to their personal physician. Education and genetic counseling were routinely provided in the first two scenarios. A multidisciplinary medical team that included physicians, genetic counselors, and outreach specialists worked with the program to ensure the quality of care provided.

The counseling and screening process varied depending on where it occurred. Individuals who participated in a community-based screening event received a complimentary 10-min genetic counseling session and had their blood drawn at the event. The community screenings took place at Jewish organizations, synagogues, and college campuses. At the time of screening, a questionnaire was distributed to participants to collect demographic and other relevant information (669 of the 724 participants were asked to fill out questionnaires). Individuals screened at Emory were seen at the outpatient clinic of the Division of Medical Genetics and received a 30–45 minute long pretest genetic counseling session. In addition to covering the basic information discussed with participants at community screening events, a detailed three generation family history was obtained at these clinic visits, and the counselor interviewed the participant to complete the questionnaire.

Subjects

The program was marketed towards any individual of reproductive age (18–45) with self-reported Ashkenazi Jewish

ancestry (defined as having one or more AJ grandparents), but individuals who were over 45 and wanted to participate in the screening were also accepted. The majority of participants were not pregnant. Women who were pregnant and wanted to participate in the program were accepted but were also required to receive more comprehensive genetic counseling through Emory Clinic Division of Medical Genetics.

Carrier screening assay

The AJGS used several Clinical Laboratory Improvement Amendments (CLIA) certified clinical genetic testing laboratories over the course of the program. All of the laboratories offered DNA-based allele specific carrier screening with assay for common Jewish mutations in the disease genes. In the first 10 months of the program, the disease panel included 18 of the most common Ashkenazi Jewish genetic diseases for which testing was available at the time. In 2011, screening for a Jewish founder mutation in the *POMT1* gene for Walker-Warburg syndrome (WWS) gene became available, and WWS was added to the panel. Starting in May 2011, the full 19 disease panel was offered to new participants. Previous participants were contacted and offered complimentary WWS testing through the AJGS program. The 19 diseases include Tay-Sachs disease, Canavan disease, familial dysautonomia (FD), Bloom syndrome, Fanconi anemia type C, Gaucher disease type I, mucopolysaccharidosis type IV (ML4), Niemann-Pick disease type A, glycogen storage disease type 1a (GSD1a), maple syrup urine disease (MSUD), dihydrolipoamide dehydrogenase deficiency (DLD), familial hyperinsulinism (FHI), nemaline myopathy (NM), Usher syndrome type III, Usher syndrome type IF, Joubert syndrome (*TMEM216* gene), and Walker-Warburg syndrome (WWS), all of which are more frequent in Ashkenazi Jews. Cystic fibrosis (CF) (Kornreich et al. 2004) was included based on ACOG and ACMG guidelines, and spinal muscular atrophy (SMA) was included based on ACMG guidelines (Table 2).

Both molecular and enzyme testing (Hex A assay on leukocytes) were performed for TSD screening (Grody et al. 2001). All AJGS testing was billed through participants' third party insurance for those who were covered, with maximum out-of-pocket costs delineated for all participants. The cost of the lab testing to the patient decreased over time. The AJGS program offered financial assistance for those who were unable to pay.

Study design

This study is a retrospective analysis based on a de-identified dataset. This dataset was created from a clinical database that included information from medical chart reviews and questionnaires. The key to match these datasets is held by an IT specialist, and those who looked at the data directly did not

have access to this key. Emory's Institutional Review Board (IRB) reviewed the retrospective analysis protocol and determined that it was exempt from human subjects review.

Measures

The de-identified database included patient ages, gender, testing laboratory, screening venue, screening results including mutations identified, partner results (if available), and answers from the questionnaire. The questionnaire collected information that included basic demographics, marital status, childbearing history, carrier screening history, number of Ashkenazi Jewish grandparents, number of Sephardic and non-Jewish grandparents, the way in which the participant heard about the AJGS program, and the participants' answers about whether they were willing to educate others about the importance of screening.

Data analysis

The statistical analysis was performed in SAS version 9.3 (SAS Institute, Cary, NC, USA). Standard descriptive statistics were used to determine the carrier frequency for each of the 19 diseases included in the AJGS program. The Fisher exact test was used to compare these carrier frequencies to those reported in the literature.

Demographic information, including age, marital status, number of AJ grandparents, and childbearing history, as well as documentation of screening venue and how participants heard about the program were summarized using descriptive statistics. For continuous variables, the number of observations, mean, and standard deviation were calculated. For categorical variables, frequency and percentages were calculated. Logistic regression models were used to define the relationship between participants' demographic information and screening and follow-up practices. We used a Cochran-Armitage trend test to study the relationship between the percentage of AJ ancestry and the likelihood of being a carrier.

Results

Demographics

A total of 724 individuals were screened through the AJGS program. Of these, 685 individuals underwent carrier screening for at least 17 diseases (some participants opted to forego screening for Joubert syndrome, Walker-Warburg syndrome, or spinal muscular atrophy based on insurance coverage and cost at that time). The rest of the participants (39) were partners of other participants. These partners, many of whom did not have Jewish background, were tested for a limited

number of conditions, most often only the disease(s) for which their partner was found to be a carrier.

Of all the participants, 523 (72.24 %) were screened at community events, whereas 185 (25.55 %) were screened at the Emory Clinic, Division of Medical Genetics. The remaining 16 (2.21 %) individuals were counseled by the AJGS program but chose to be screened at their physicians' offices. The program was not able to collect data on individuals who elected to have counseling and screening through their physicians' offices.

Demographic information was collected during the registration process and questionnaires were administered prior to screening. A total of 669 participants filled out questionnaires. Some participants did not answer all questions; thus, the total number of responses was not equal for every question.

The AJGS participants were fairly evenly divided with respect to gender, with slightly more females than males (57.3 and 42.7 %, respectively). Participants ranged in age from 19 to 68 with a median age of 31. The age of our screened population was consistent with average reproductive age previously reported for the Jewish population in North America (Laurence Kotler-Berkowitz 2003). Five hundred seventeen (77.28 %) participants reported that they were married or had a stable partner. Forty-four (6.58 %) individuals were pregnant or had a pregnant partner at screening. One hundred twenty-nine (19.28 %) individuals already had children. When asked about Ashkenazi Jewish ancestry, 619 (96.9 %) participants reported that they had at least one AJ grandparent. One hundred sixteen (18.99 %) individuals had received carrier screening for one or more genetic disorders prior to the AJGS program. Four hundred thirty-five (71.19 %) participants denied any screening history and another 60 (9.82 %) reported that they were uncertain. Fewer than 10 individuals reported a family member known to be affected with a Jewish genetic disorder that was included in our panel (Table 1).

Carrier frequencies

The carrier frequencies for the 685 individuals screened for at least 17 diseases are summarized in Table 2 and ranged from 0.0058 (1 in 171; NPD) to 0.0686 (1 in 14; GD). All disease alleles were in Hardy-Weinberg equilibrium. We compared the carrier frequencies calculated from AJGS with those reported by the Victor Center for the Prevention of Jewish Genetic Diseases (Victor Center 2014) by Mount Sinai School of Medicine (Scott et al. 2010) and by a recent study presenting literature combined carrier frequencies (Hoffman et al. 2014). (Table 2) Overall, the carrier frequencies derived from the four studies are consistent. The exceptions are the carrier frequencies for spinal muscular atrophy, which was much less common in the AJGS group than in the Victor Center's sample (1 in 103 vs 1 in 41; $p=0.0179$) and nemaline myopathy,

Table 1 Demographics of the study population

Demographics of participants		
Category	Number	Percent (%)
Gender	<i>N</i> =719	
Female	412	57.30
Male	307	42.70
Age	<i>N</i> =722	
18–21	12	1.66
22–25	83	11.50
26–29	208	28.81
30–33	188	26.04
34–37	144	19.94
38–41	53	7.34
42–45	23	3.19
≥46	11	1.52
Ancestry	<i>N</i> =639	
Jewish ^a	619	96.87
Non-Jewish	20	3.13
Partner/spouse	<i>N</i> =669	
Yes	517	77.28
No	152	22.72
Current Pregnancy	<i>N</i> =669	
Yes	44	6.58
No	625	93.42
Prior pregnancy	<i>N</i> =669	
Yes	129	19.28
No	540	80.72
Prior carrier screening experience	<i>N</i> =611	
Yes	116	18.99
No	435	71.19
Not sure	60	9.82

Note: The total number of answers differs for each question

^a Having one or more Ashkenazi Jewish grandparents

which was much more common in the AJGS group than in other reports (all $p<0.05$).

Through the AJGS program, 188 individuals (25.9 %) were identified as carriers for at least one of the 19 diseases. This carrier frequency of 1 in 3.9 is consistent with the 1 in 4 carrier frequency reported by other screening programs (Victor Center 2014). Of these individuals, 158 were identified as carriers for one disease; 29 were carriers for two diseases, and one was a carrier for three diseases (Table 3).

Three couples were identified in which both members were carriers for the same disease. These couples received comprehensive genetic counseling regarding the disease, risks to their future children, available reproductive options, and the importance of testing on other family members. All three couples elected to pursue prenatal diagnosis in their subsequent pregnancies.

Table 2 AJGS Carrier frequencies and comparison with published literature

Disease	Carriers	Total	AJGS carrier freq.	Victor Center carrier freq.	Mount Sinai carrier freq.	Hoffman literature combined carrier freq.
Bloom syndrome (BS) ^b	5	685	1 in 136	1 in 100	1 in 133	1 in 130
Canavan disease (CD) ^{a, b}	8	685	1 in 85	1 in 45	1 in 55	1 in 59
Cystic Fibrosis (CF) ^{a, b}	27	685	1 in 25	1 in 26	1 in 22	1 in 25
Dihydrolipoamide dehydrogenase deficiency (DLD)	10	685	1 in 68	1 in 96	1 in 107	1 in 106
Familial dysautonomia (FD) ^{a, b}	16	685	1 in 42	1 in 30	1 in 30	1 in 35
Familial hyperinsulinism (FHI)	7	685	1 in 97	1 in 66	1 in 67	1 in 55
Fanconi anemia group C ^b	8	685	1 in 85	1 in 89	1 in 100	1 in 93
Gaucher disease type 1 (GD) ^b	47	685	1 in 14	1 in 15	1 in 15	1 in 17
Glycogen storage disease type 1A (GSD1A)	8	685	1 in 85	1 in 71	1 in 64	1 in 70
Joubert syndrome (JS)	5	684	1 in 137	1 in 92	N/A ^c	1 in 92
Maple syrup urine disease (MSUD)	5	685	1 in 136	1 in 81	1 in 97	1 in 122
Mucopolidosis IV (MLIV) ^b	11	685	1 in 63	1 in 122	1 in 88	1 in 97
Nemaline myopathy (NM)	11	685	1 in 63	<i>1 in 149 (p=0.0079)</i>	<i>1 in 167 (p=0.0059)</i>	<i>1 in 140 (p=0.0093)</i>
Niemann-Pick disease type A (NPD) ^b	4	685	1 in 171	1 in 90	1 in 115	1 in 112
Spinal muscular atrophy (SMA) ^b	6	629	1 in 103	<i>1 in 41 (p=0.0179)</i>	N/A ^c	1 in 66
Tay-Sachs disease (TSD) ^{a, b}	23	685	1 in 29	1 in 25	1 in 27	1 in 26
Usher syndrome type III (US)	3	685	1 in 228	1 in 107	1 in 147	1 in 120
Usher syndrome type I F	1	685	1 in 685	1 in 141	1 in 119	1 in 130
Walker-Warburg syndrome	8	605	1 in 75	1 in 112	N/A ^c	1 in 150

Those carrier frequencies that are significantly different from published literature are set in italics. *p* value <0.05

^a Diseases included in ACOG guidelines

^b Diseases included in ACMG recommendation

^c Disease not screened in this program

Ashkenazi Jewish ancestry

The AJGS program not only screened those with 100 % AJ ancestry but also those with mixed (AJ as well as Sephardic or non-Jewish) background. The percentage of AJ ancestry was calculated based on the number of AJ grandparents an individual reported on the questionnaire. The relationship between percent AJ ancestry and positive carrier screening results was determined. Out of 639 participants who provided ancestry information, 463 (72.4 %) individuals were 100 % AJ, whereas 20 (3.1 %) reported either no or uncertain AJ ancestry. The remaining 156 participants (24.4 %) reported mixed AJ ancestry (Table 4). The likelihood of being identified as a Jewish

genetic disease carrier significantly increased with the number of AJ grandparents ($p=0.0011$) (Table 4).

Of the 445 individuals who answered that they were married or in a stable relationship, 52 (11.68 %) reported that their partners had no Jewish ancestry. This intermarriage rate is significantly lower than the intermarriage rate previously reported for the Jewish community in Metropolitan Atlanta (50 % in 2006), and for the Jewish population in the U.S. (58 % in 2012) (Atlanta 2007; Tighe et al. 2013).

Screening and follow-up practices

AJGS program participants were asked how they heard about the screening program. Friend ($N=184$), rabbi/synagogue ($N=137$), and family member ($N=116$) were identified as the three most common sources of information. Fifty-five individuals learned about the program from their healthcare providers and 104 heard at community events. The rest of the participants learned about the program through traditional media, social media, or online advertisements (Fig. 1).

Six hundred and eleven individuals were asked about whether they would pledge to educate others about the importance of screening. Participants who were older or had a prior

Table 3 Carriers for one or more disease genes

No. of diseases screened positive	Individuals	Percent (%)
0	536	74.03
1	158	21.82
2	29	4.01
3	1	0.14

Table 4 Carrier frequency based on number of AJ grandparents

No. of AJ grandparents	Carrier ^a		No. of participants	Percent (%)	Likelihood of being a carrier (%)
	Yes	No			
0	3	17	20	3.13	—
1	1	16	17	2.66	5.88
2	19	86	105	16.43	18.1
3	14	20	34	5.32	41.18
4	137	326	463	72.46	29.59
Total	174	465	639	100	27.23

Cochran-Armitage trend test: p value=0.0011^a For one or more of the 19 diseases

pregnancy were significantly more willing to educate others about the availability and importance of carrier screening. Participants who were married or engaged were somewhat more likely to educate others about screening, although this finding was not statistically significant (Table 5).

Of the 188 carriers, 165 reported that they had current partners. Of these, 80 were screened at the same time as their partner and 35 followed up with carrier testing on their partners. The overall rate for partner testing was 69.7 %. For those carriers who did not have a current partner, follow-up was recommended in the future. All carriers were also encouraged to share their carrier status with their at risk family members.

One hundred and sixteen of the participants reported that they had carrier screening for one or more of the 19 diseases prior to the AJGS program and chose to update their screening. Thirty of these individuals were identified as carriers through AJGS. Seven individuals were known carriers. Of the 30, 26 (86.7 %) followed up with carrier testing on their partners. Of the 608 participants who had not been screened before, 158 individuals were identified as carriers. Eighty-nine out of the 158 (56.3 %) carriers chose to follow up with carrier testing on

their partners. Thus, people who had prior carrier screening experiences, whether they tested positive or not, were significantly more likely to follow up with partner testing than those who had no history of carrier screening ($p=0.0018$).

Discussion

Our data indicated a 1 in 3.9 carrier frequency for Jewish genetic diseases, consistent with carrier frequencies reported by other Jewish screening programs in the U.S. (Victor Center and Mount Sinai). The 2014 Hoffman study did not report a cumulative carrier frequency for all 19 diseases. In terms of specific diseases, carrier frequencies were consistent with published literature for 17 of the 19 diseases. The carrier frequencies for SMA and NM were significantly different from published literature, possibly due to small sample size.

It is worth mentioning that two previously undiagnosed patients with Gaucher Disease Type I and one asymptomatic patient with SMA were also detected through this program.

Fig. 1 How participants heard about the AJGS program

Table 5 Factors that affect willingness to educate others about carrier screening

Outcome	Factors	Odds ratio	95 % confidence limits	<i>p</i> value
Pledge to educate	Age	1.046	(1.008–1.086)	<i>0.0167</i>
	Married/engaged	1.441	(0.942–2.203)	0.0927
	Pregnancy history (current and prior pregnancy included)	2.433	(1.285–4.587)	<i>0.0062</i>

Significant results are set in italics

The two patients with GD reported only mild clinical involvement and were referred for evaluation by appropriate specialists. The patient with SMA was counseled about appropriate follow-up as well. All three individuals received genetic counseling regarding implications for themselves and their future children. The detection of three affected individuals highlights the importance of informing participants prior to screening that there is a small chance they will learn that they have a genetic condition.

Our analysis focused on evaluating the effectiveness of the program's outreach and education efforts. Overall, the program achieved its goal of reaching many people in the target population. People who were not pregnant and who were at the highest risk for being carriers (based on ancestry) were captured by the outreach efforts. Only 44 participants (6.58 %) who came through the program were pregnant, which suggests that the program effectively reached the population at the ideal time. The majority of participants had 100 % AJ ancestry, and the intermarriage rate was significantly lower than in the Atlanta Jewish population overall. These characteristics of the screened population suggest that outreach efforts targeting the affiliated Jewish community results in screening those who are more connected to the Jewish community and organizations. Our finding of a positive correlation between number of AJ grandparents and the risk for being a carrier for one of more of the 19 diseases emphasizes the importance of screening this high-risk population. The program was less successful in reaching those who were unaffiliated with Jewish organizations and those who were intermarried.

The most common ways people heard about the screening program were through friends, rabbis/synagogues, and family members. Fewer people were influenced by media, such as newspapers, advertisement, or websites. Our study suggested that participants have more trust in people who are close to them on these sensitive issues such as personal health and genetic testing and that personal contacts are more influential than general advertisements. These results inform future strategies for promoting community carrier screening and suggest that emphasis should be placed on marketing to influencers.

A previous qualitative study suggested that prior experiences and personal situations may influence individuals' perception of the relevance of screening and their interpretation of results (Archibald and McClaren 2012). The AJGS program yielded similar results. Within the screened population,

we found that older age and had a prior pregnancy were all positively associated with people's willingness to educate others about the availability and significance of carrier screening. These findings show that people are more likely to take an interest in testing and share that information with others when they are at a point in their lives when the information is more relevant. Furthermore, we found that people who had been screened previously and knew about the importance of screening were more likely to follow up with partner testing and encourage others to be screened. For carrier screening programs to be effective, appropriate and sufficient genetic education needs to be provided to the target population.

The limitations of this study include a small sample size, a short time period, and a comparatively select population. It is still important to reach out to and screen those who have less Jewish ancestry and those who are distantly connected with Jewish community organizations, as they are still at increased risk for carrying Jewish genetic diseases. In addition, we acknowledge that the participants of the AJGS program are, for the most part, highly educated and well informed compared to other populations. Thus, for the conclusions to be applied to the general populations or to other ethnic groups, further studies are needed.

Future directions

Preconception carrier screening provides people with information that gives them more reproductive autonomy and allows for informed decision-making. The three carrier couples identified from our program chose to pursue prenatal diagnostic procedures in their subsequent pregnancies. Without screening, they would not have known their pregnancies were at increased risk. Both medical professionals and patients perceive the preconception period as the best time for carrier screening, as there are more available reproductive options and less emotional stress at this time (Janssens et al. 2014). The reality is that carrier screening in prenatal settings is much more common. Several studies have attributed the failure of implementing preconception carrier screening in the healthcare system to difficulties in reaching couples before conception, lack of knowledge among healthcare providers, and insufficient resources (Metcalfe 2012).

The lessons learned from the AJGS program provide insight for the development of future carrier screening programs

in the Jewish population. First, outreach campaigns need to target the preconception population in order to promote autonomy and provide the greatest range of reproductive choices. Secondly, many unaffiliated Jews are not reached through marketing to Jewish organizations. In the Greater Atlanta, only 42 % of all Jewish households are communally connected (Atlanta 2007), comparable to national statistics (Tighe et al. 2013). Therefore, future community programs should also take the initiative to reach out to unaffiliated Jews and promote screening for this at risk group. Thirdly, in order to ensure the success of a community-based carrier screening program, educational, and outreach efforts need to concentrate on educating influencers and raising general awareness about the importance of genetic screening.

The success of organized preconception carrier screening programs in Ashkenazi Jewish communities can serve as a model for developing programs targeting other ethnic groups. Education and awareness campaigns should provide information highlighting the importance of carrier screening and should target individuals of reproductive age. Outreach efforts should be customized for each group to best reach those individuals and most effectively capture the target population. Lastly, it is important to provide education and support to influencers in the community, as targeting influencers appears to be more effective than traditional marketing efforts. It is important to acknowledge that leading influencers may vary from group to group.

With recent advances in genetic testing technologies, carrier screening is becoming more comprehensive, less expensive, and more accessible. At the same time, some groups do not have the advantage of coordinated outreach and screening efforts and are calling for equity in access to preconception screening. The changing landscape of carrier screening requires more public health research in how to better reach and effectively capture at risk populations to improve reproductive health on a larger scale.

Conclusions

Our study was designed to evaluate the success of a two-year community-based preconception carrier screening program in the Metropolitan Atlanta area. Our findings suggest that the carrier frequencies in the Atlanta Jewish community are generally consistent with those of other Jewish populations in the U.S. The analysis also profiled the characteristics of screened participants and suggested that outreach efforts are most effective when they target not only potential participants but also influencers whose opinions are highly valued within families or in the broader community. Results of this study will help inform more effective outreach strategies for community screening initiatives in Jewish communities, in other

ethnic groups and in the general population. Additional studies are needed to help design, implement, and evaluate similar preconception carrier screening programs in other communities.

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Compliance and ethics guidelines This retrospective analysis complies with the current laws of the country in which it was performed.

Conflict of interest Yunru Shao, Shuling Liu, and Karen Grinzaid declare that they have no conflict of interest.

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