Jewish Genetic Screening:

An evaluation of the barriers to utilization and strategies to increase the awareness of young Jewish adults

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Introduction:

Genetic screening in the Ashkenazi Jewish population began in the 1970s with Tay-Sachs disease (TSD) and through its education and counseling efforts has reduced the birth rate of infants with TSD within the Ashkenazi Jewish community by 90% (Schneider). The successful screening programs have incorporated significant education and counseling efforts. Currently, screening is available to test for approximately 16 genetic disorders, four of which (cystic fibrosis, TSD, Canavan disease and dysautonomia) are considered the standard of care (Hegwer). Because TSD screening has been so successful at reducing the numbers of Jewish children born with TSD, many Jews are unaware that they are at increased risk to have an affected child. Ashkenazi Jews have an approximately 14% chance of carrying one of the conditions (Heger). One approach to reducing the risk for having an affected child was established through a program called Dor Yeshorim. Established in 1983, Dor Yeshorim is a program for ultra-orthodox couples who have arranged marriages. It provides pre-marital, anonymous, confidential carrier screening for nine conditions (Loannou). The goal is to avoid an arranged marriage between individuals who are carriers for the same autosomal recessive condition, thereby eliminating the chance for an affected child.

Some controversy has arisen regarding the appropriate timing of screening and the level of participants' knowledge. Though most agree preconception screening is ideal, appropriate timing (i.e. high school, college, early adulthood, pre or post-martial) is still unresolved (Hegner). Screening programs in Jewish high schools in Canada and Australia found that greater levels of knowledge were associated with fewer negative feelings toward the screening process, less anxiety if found to be a carrier, and greater preparation to address future reproductive options. However, others argue that knowledge of one's carrier status may not be relevant or remembered when screening is performed at younger ages , that high school students cannot be adequately informed to give consent, and that the accompanying social stigmas or psychological stress may be too overwhelming (Loannou).

It is apparent that education is a critical factor in any effort aimed at eliminating these preventable disorders, especially as the numbers of Jews with unknown family ancestry continue to increase. These challenges to successful carrier screening in the Jewish community are currently being addressed through the education, screening and prevention programs of the Chicago Center for Jewish Genetic Disorders. The Chicago Center strives to increase awareness of Jewish genetic disorders for the Jewish community, health care providers, religious figures and the general public. In addition to their ongoing work, my research project findings may be used by healthcare providers and other non-profit agencies to more effectively approach and respond to patients.

Background:

The exposure to several Jewish genetic diseases during the Genetics & Neoplasia module, coupled with my personal experience as a member of the Conservative Jewish community, has driven my desire to explore the prevention and management of these disorders in greater detail. After discussing this subject matter with my parents, I learned that they had carrier screening for

TSD prior to conception. Though their tests were negative, I am curious as to how others proceed if one or both members of the couple are found to be carriers, particularly in light of the fact that 14% of participants in screening programs will be carriers of one of the conditions, and 2% screened will be at risk for a child with one of the conditions. In addition to having several friends and family members who are carriers of Jewish genetic diseases, I also have had a long-standing career interest in preventative health and endocrinology, and feel this would be a prime opportunity to explore the reproductive component of the specialty. Though prevalence remains low, no Jewish parent should have to experience the tragedy of having an affected child given the ease with which individuals at risk to have children with these untreatable disorders can be identified.

As an undergrad I completed a medical ethics class, which explored topics such as genetic testing and abortion. Unfortunately, the professor did not address most religious concerns. Outside of class while shadowing an oncologist, I obtained a profound appreciation for the importance of an empathetic, supportive patient-physician interaction that can only be taught through observation and experience. Finally, because this project requires a large Jewish population, I have chosen to do part of the research in Chicago where I have family and friends who will be of assistance with living arrangements. Of additional importance is the fact that Chicago has the closest Dor Yeshorim testing center.

Goals/ objectives:

Screening programs have dramatically reduced the incidence of Jewish genetic disorders. Nevertheless, questions still remain regarding the age at which testing is most appropriate, how much information is required to make informed choices, and the ideal level of education needed to maximize participating while minimizing feelings of fear and hopelessness. It is my goal to determine how the level of awareness and screening rates can be improved among orthodox, conservation and reform Jews. I hope to determine how physicians, counselors and religious figures can maximize their role for the varying Jewish denominations regarding education and prevention, while determining the appropriate time to be screened. Because screening has become so effective, interest in educational programming and screening has declined. I hope to identify those factors which will revive interest in Jewish genetic screening, particularly in the Kansas City area.

For those who undergo screening, I plan to study their motivations for screening at that time, as well as any drawbacks to that timing. This will include any social, psychological and financial concerns. I also hope to assess individuals' perception of their carrier status and how they choose to proceed given their carrier status. I hope to determine the ideal time for screening, acquisition of an appropriate knowledge base, and any other potential improvements which may be made to the screening process. Overall, the project will allow me to gain a better understanding of the issues and current controversies, raise awareness and better prepare me for a future in medicine.

<u>Timeline:</u>

a. Late May to early June (Kansas City): preliminary research, i.e. literature review, conducting interviews with local rabbis, genetic counselors, and patients

b. Early June to early July (Chicago): interview patients during the mass-screening at Children's Memorial Hospital, interview former patients from the February and April mass screenings, interview counselors and rabbis, collect data at the Chicago Center
c. Early July to late July (Kansas City): continue patient and rabbi interviews as needed, analyze data and interview responses for research paper

Methods:

Prior to the summer, I plan to continue reading the history and development of Jewish genetic screening. The Chicago Center for Jewish Genetic Disorders has agreed to allow me to conduct research at their office on their interdisciplinary prevention and educational programming techniques. To achieve these goals, I will be conducting interviews during a mass screening in mid-June with approximately 45 participants of Dr. Joel Charrow and Ms. Michelle Gilats at Children's Memorial Hospital. These interviews will occur between the educational presentations and screening appointments. After participants sign up for the mass screening in March, I will send a letter of introduction, as well as a list of questions. This will allow participants to consider their answers before the face-to-face interview. Should time not allow for all interviews, I will arrange follow up meetings with participants. I will also contact past participants from the January and March mass screenings, consisting of approximately 40 participants each, to obtain a larger population base. Finally, a small advertisement in the Kansas City Jewish Chronicle seeking additional interviewees who have had genetic testing has generated a significant number of responses.

In addition to interviewing patients, I will interview physicians, counselors, religious figures and staff members at the Center both before and after the mass screening to identify the current educational opportunities for families and the barriers to screening. I have also identified over a dozen rabbis from the Orthodox, Conservative and Reform movements, in both the Chicago and Kansas City areas, who have agreed to participate in the project. I also will observe physicians and counselors working with patients in order to gain a better understanding of patients' experiences and to document the strategies used.

When not conducting interviews, I will be assisting the Chicago Center with their prevention strategies. I will also attend educational programming and talk to attendees in order to identify concerns, potential for improvements and their overall response. Michael Begleiter, a genetic counselor at Children's Mercy Hospital in Kansas City, MO, has also agreed to serve as a mentor and help me in whatever capacity is necessary. I will spend the last two weeks analyzing data, compiling the research, creating a presentation and providing feedback to counselors. Analysis will include all aspects of the interview and observation, including demographic information, attitudes and beliefs. Should time allow, I would also like to work on my own educational programming effort most likely targeting college campuses.

Throughout the spring, I will continue to contact counselors, patients and religious figures asking for their cooperation on the project. Due to the importance of education, all clinics, non-profit agencies and religious organizations which I have contacted thus far have expressed excitement with the project, willingness to contribute and interest in the results. The time frame for the project is rather fluid due to my flexible living arrangements in both locations and the possibility that I may lose and add potential

interviewees. Timing will largely depend on when it is most convenient for individuals to meet, scheduling of counseling appointments, and the Center's programming efforts throughout the summer. I am awaiting feedback on my Application for Except Review of Human Subject Research through KUMC, as well as waiting for information from Michelle Gilats regarding obtaining IRB approval through Children's Memorial Hospital. She does not anticipate any problems with approval, as participants in the mass genetic screening are not considered Children's patients, and interviews are typically exempt from the IRB approval process.

Contacts:

Chicago Center for Jewish Genetic Disorders Ben Gurion Way 30 S. Wells St. Chicago, IL 60606	Karen Litwack, director
	Michelle Gilats, licensed genetic counselor
	Dr. Joel Charrow, M.D. scientific advisor
Children's Mercy Hospital 2401 Gillham Road Kansas City , MO 64108	Michael Begleiter, M.S. CGC (816) 234-3290 mbegleiter@cmh.edu
The following includes rabbis and other community figures from varying denominations who have agreed to help with the study thus far:	Kansas City: Rabbi Nemitoff, Rabbi Itkin, Chana Itkin, Rabbi Cohen, Rabbi Schuster, Jay Lewis, Rabbi Rockoff
	Chicago: Rabbi Mendel, Rabbi Siegel, Rabbi Conover, Rabbi Sandmel, Rabbi Flinkenstein, Rabbi Moscowitz, Rabbi Zedek, Alison Lewin, Rabbi Lopatin

Conclusion:

Most importantly, this project will benefit the greater Jewish community by assessing how education and prevention efforts can be received with more enthusiasm. In addition to identifying methods through which awareness can be improved, the research will benefit patients participating in screening by assessing optimal timing and level of knowledge. The findings can also be applied to other clinics and non-profit agencies in promoting awareness. The project will supplement my education as a medical student and future clinician by providing exposure to a field of interest, allowing me to acquire a greater sense of patient concerns and cultural differences and experience patients coping with sensitive subject matter. Finally, I would like to thank the Clendening Fellowship Program for allowing me the opportunity to explore an unconventional, yet meaningful humanitarian-based issue within the medical field.

Budget:

a. Round trip transportation to and from Chicago: $300

b. Transportation in Chicago and Kansas City (e.g. gas, parking, public transit): $300 per month
c. 30-day unlimited Chicago Transit Authority (CTA) pass: $86
d. Rent: no charge (provided by friends and family in Chicago)
e. Food: $300 per month
f. Printing and mailing costs: $100
g. Thank you gifts for the Center, counselors and interviewees: $200

Works cited:

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3. Lavin, Norman. "An Introduction to Jewish Genetic Diseases: Part I | Jewish Diseases | Jewish Journal." *Jewish Journal: Jewish News, Events, Los Angeles*. 24 Jan. 2011. 08 Jan. 2012. <http://www.jewishjournal.com/jewish_diseases/item/an_introduction_to_jewish_genetic_diseases_part_i_20110124/>.

4. Remennick, Larissa. "The Quest for the Perfect Baby: Why Do Israeli Women Seek Prenatal Genetic Testing?" *Sociology of Health & Illness* 28.1 (2006): 21-53.

5. Wailoo, Keith, and Stephen Gregory. Pemberton. *The Troubled Dream of Genetic Medicine: Ethnicity and Innovation in Tay-Sachs, Cystic Fibrosis, and Sickle Cell Disease*. Baltimore: Johns Hopkins UP, 2006. Print.

Appendix:

a. Sample interview questions
b. Letter of introduction (Note: the letter will be catered to those participating in the June mass genetic screening and previous mass genetic screenings in Chicago, as well as those who have expressed interest in participating in Kansas City)
c. Email correspondence with the Chicago Center for Jewish Genetic Disorders, several patients in the Kansas City area, and Michael Begleiter

Interview questions:

Orthodox, conservative, reform and mixed-marriage Jewish patients

1. Background demographic information: gender, age, religious affiliation, education level, occupation, brief personal and family history of Ashkenazi Jewish genetic disorders
2. From whom or what resource did you receive information on screening/ testing?
3. What was your motivation to pursue or forego testing at such time? How did you decide on this particular method of testing over other options?
4. Did you have any hesitations and/ or concerns prior to screening?
5. Did you fully understand the screening results and the resources/ options offered? How did you proceed with results (i.e. sharing results, testing other family members, etc.)
6. Did your perception of screening/ counseling change through the process? If so, how has your perspective changed over time?
7. Do you believe awareness is a problem? How, if at all, have you joined the outreach effort to promote awareness? If not, do you propose any ideas to improve prevention?
8. Are you familiar with Dor Yeshorim? If so, what are your feelings regarding Dor Yeshorim?
    a. Should a young person inquire about the carrier status of a potential spouse on a social level before or after meeting?
    b. Should two carriers break up an engagement or refuse to marry?
    c. Must a known carrier divulge their carrier status to their significant other or intended spouse?
    d. Is it acceptable to hope for the best, arrange an abortion, or have an affected child?
9. Describe your overall satisfaction with the process. What additional services, changes or improvements would be beneficial?
10. Is there anything else you'd like to share about the experience?

Physicians and genetic counselors:

1. Discuss your goals, as well as the challenges that arise regarding your educational, awareness and prevention efforts.
2. How can educational efforts maximize testing without arousing excessive levels of anxiety?
3. Describe the challenges that arise during the screening/ counseling process.
4. Do you adapt the process to individuals of varying Jewish denominations, and if so, how?
5. What is the optimal time for education and screening?
6. What are the pitfalls of carrier screening (i.e. inconclusive or insensitive results, variable expressivity, misinterpretation of results, genetic discrimination, confidentially)? How do you address and propose to improve these barriers?
7. Who and when should it be decided to pursue testing?
8. Should and what are the limits to pre-implantation, pre-natal and neonatal screening?
9. Who is entitled to screening results and how should genetic information be used?
10. What are your feelings regarding Dor Yeshorim?

a. Should a young person inquire about the carrier status of a potential spouse on a social level before or after meeting?
b. Should two carriers break up an engagement or refuse to marry?
c. Must a known carrier divulge their carrier status to their significant other or intended spouse?
d. Is it acceptable to hope for the best, arrange an abortion, or have an affected child?

<u>Rabbis and religious leaders:</u>

1. If and how was genetic screening/ counseling incorporated into your education, either in rabbinical school or current continued education?
2. How do you personally or do others on staff within your congregation play a role in the education or awareness efforts of Jewish genetic diseases?
3. What would be helpful to you to increase education within your congregation?
4. When is the optimal timing for screening and how could the Jewish community promote this cause (i.e. Hebrew school, Sunday school, young adult programming, etc.)
5. What are your feelings regarding Dor Yeshorim?
    a. Should a young person inquire about the carrier status of a potential spouse on a social level before or after meeting?
    b. Should two carriers break up an engagement or refuse to marry?
    c. Must a known carrier divulge their carrier status to their significant other or intended spouse?
    d. Is it acceptable to hope for the best, arrange an abortion, or have an affected child?

Letter of introduction:

Jewish Genetic Screening:
Evaluation of the barriers and strategies to increase education and awareness for young Jewish adults

I, Rachel Myers, am a first year medical student at the University of Kansas School of Medicine. The Clendening Fellowship Program through the Department of History and Philosophy at the University of Kansas Medical Center offers summer research fellowships for 8-10 first-year medical students to study an area of interest during the summer between the first and second years of medical school under the direction of one of the Department's faculty members.

As a member of the conservative Jewish community with career interests in endocrinology and preventative health, I have chosen to explore Jewish genetic diseases in greater detail, focusing particularly on the means of education, awareness and prevention in the varying Jewish denominations. I plan to conduct research largely by interviewing genetic counselors, physicians, non-profit agencies, religious figures and individuals undergoing carrier screening in both the Kansas City and Chicago area. I also plan to spend time at the Chicago Center of Jewish Genetic Disorders evaluating current means of education and prevention. Finally, I hope to organize a project of my own aimed at increasing awareness and education on college campuses.

Overall, the project will benefit the greater Jewish community by assessing how education and prevention efforts can be received with more enthusiasm. In addition to identifying methods through which awareness can be improved, the research will benefit patients participating in screening by assessing optimal timing and level of knowledge. The findings can also be applied to other clinics and non-profit agencies in promoting awareness.

Participation in the interview is completely voluntary and individual responses will remain confidential. Participation may be withdrawn at any time. After collecting and analyzing responses, results will be documented in a research paper and presented in a Clendening presentation in fall 2012.

Regards,

Rachel Myers

Rachel,
I saw the notice in the Jewish Chronicle. I also spoke with Janet Price (her grandson has Mucolipidosis IV) and Gary Cortes (his wife's sister had Tay-Sachs). Both families have contributed to a fund at Beth Shalom to office free Jewish Genetic Disease Carrier Screening to congregants. Gary told me that he is going to help with your project.
Good luck with the submission. I hope you get funded.
Let me know if there is anything else I can do.
Michael