Dear Friends:

I am pleased to report that with your generous support we've continued to partner with the JGDC this year to give brochures to women planning to start a family – and to educate their doctors. We know it works because a young woman recently told us she took the brochure to her doctor's visit and found it “empowering” to share our testing information.

We would love to find a way to take this provider education program national, and are exploring partnering with other organizations to make this happen. If you are familiar with any local or family foundations in major cities in the U.S. beyond the New York area that might be interested in supporting this effort, please let us know.

This year we also supported the work of Dr. Paola Leone, a leading researcher on Canavan disease, to identify novel mutations for the disease. Dr. Leone hopes to add approximately nine mutations to the public Canavan mutations database, thereby improving prenatal screening.

I do hope you'll be able to join us at “Raisin in the Sun” in April. Look for details in this newsletter and online.

Best wishes for a happy and healthy New Year!

Orren Alperstein

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**Doctors’ offices.** Throughout 2013 the Canavan Foundation, in partnership with the JGDC, continued our program of visiting every OB/GYN office in the New York metropolitan area, offering educational materials to providers and brochures for patients. We are still the only organizations making sure that this vital information gets directly into the hands of Jewish couples at the time when they are making decisions about having children.

An addition to the program in the near future will be a new one-sided checklist of Jewish genetic disease screening recommendations, which will be distributed to all the practices as soon as it’s printed.

When the JGDC brought their educational seminars to Savannah, Detroit and Ann Arbor this year, they took with them a list of OB/GYN offices in those areas; they visited the offices in person, distributing educational materials and consumer brochures.

**Rabbis and Cantors.** We continue to be in touch with the New York area rabbis and cantors we trained several years ago. We send quarterly e-mails offering to replenish the stock of brochures they use during premarital counseling, and we receive dozens of orders for hundreds of brochures every quarter.

We are reaching out to the central organizations of both Reform and Conservative Judaism to offer counseling brochures for use across the country, at the nearly 700 Reform and 400 Conservative congregations outside of the New York region.

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**Jewish Genetic Disease Checklist for OB/GYNs**

- Identify all patients with Jewish heritage. A patient with even one grandparent of Jewish heritage should be screened for Jewish genetic diseases. Patients do not necessarily know to discuss their heritage with you. Ask about Jewish heritage as part of your standard patient intake procedure.
- Discuss screening with couples of mixed heritage. If only one member of the couple is of Jewish heritage, that person should be screened first. If that person is a carrier, the other partner should be screened for the diseases carried by the first partner.
- Stress the need for preconception screening. Genetic carrier screening should ideally be done before conception. Patients who are carriers should have their partner tested and seek advice from a genetic counselor about their family planning options if they are both carriers of the same disease.
- Offer the full panel appropriate for the patient’s heritage.
  
  Ashkenazi (German, Eastern European), there is currently a standard screening panel for 19 diseases that affect persons of Ashkenazi (Eastern European) Jewish heritage. The panel is based in the JGDC-patient-education brochure and on our website.
  
  Sephardic (Mediterranean) and Mizrahi (Persian/ Middle Eastern): There is no standard panel for persons.

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Partial view of checklist given to doctors.

Since launching this program in the summer of 2012 we have distributed nearly 30,000 brochures to more than 900 doctors. Our regular calls and visits have shown that the brochures are used consistently: over 95% of the practices have asked to be replenished on a regular basis.

In order to use our resources most efficiently, we are now alternating in-person visits with phone calls and brochure distribution by mail, but we still make sure to keep close contact with the personnel in each office, giving this program the personal touch.

SAVE THE DATE: 2014 Theater Gala: Raisin in the Sun

The Canavan Foundation's Spring Theater Benefit will be Lorraine Hansberry’s “A Raisin in the Sun,” on Tuesday, April 29, 2014.

The eagerly-awaited production of this beloved play, which takes place in the late ’50s in a South Side Chicago apartment, will feature movie star (and Tony-winning stage actor) Denzel Washington, joined by Diahann Carroll and a star-studded cast, and directed by Tony-nominated Kenny Leon.

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Letter from the president

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Carrier rate study reveals surprising results

An article in *Genetics in Medicine* magazine reports that while genetic disease carrier rates among persons of Ashkenazi Jewish (AJ) heritage were the highest of all population groups screened, a surprisingly high number of carriers of diseases traditionally understood as “Jewish” do not self-report any Jewish heritage. The article, published online last year, reviewed the results of screening more than 23,000 individuals for 108 genetic diseases by means of a universal genetic test.

As expected, persons of AJ heritage were the most likely to be carriers for genetic conditions. However, a significant number of individuals who did not report AJ heritage were found to be carriers of diseases traditionally associated with Ashkenazi genes. For example, of the carriers for Canavan disease, 39.4% reported that they were not AJ.

The carrier rates for these diseases are many times lower among the general population. For Canavan disease the study found only 1 in 683 non-Ashkenazi carriers versus 55 in the AJ population. But the actual number of non-AJ carriers is much closer: for Canavan disease, 28 carriers is much closer: for Canavan disease, 28 carriers are among the non-AJ carriers versus 43 in the AJ population. (The AJ population is 1.5% of the total American population.)

The findings do nothing to diminish the importance of screening for persons of AJ heritage. In fact, people with AJ heritage have even higher carrier rates for some diseases than has been generally reported. But they do point out the utility of universal screening of the general population for a wide range of diseases, which will uncover carriers of serious genetic diseases who otherwise would only learn of their carrier status when they gave birth to an affected child.

Grant helps provider information campaign grow

In early 2013 Counsyl awarded the Canavan Foundation an unrestricted education grant to support our ongoing work educating doctors and patients at OB/GYN offices. Counsyl, a genetic screening company, recently introduced a low cost universal genetic test, which screens for over 100 genetic conditions. To conduct the test, blood is drawn at a doctor’s office and the results are transmitted back to the doctor, who discusses results with the patient.

Canavan Foundation Website Survey

Those of you who have visited the Canavan Foundation website in the past few months may have seen our pop-up survey, which we placed on the site to get an idea of who is visiting, what information they’re seeking, and how well we’re providing it. The survey revealed that our visitors come from a wide range of backgrounds:

- 75% of the visitors are female
- 60% are married, engaged or partnered
- 70% have no known Jewish heritage
- 75% have never discussed genetic carrier screening with a doctor or genetic counselor
- 83% have never been screened

Theater Gala: Matilda the Musical

On April 24th, 2013, 150 supporters and friends of the Canavan Foundation gathered in New York City for our annual Theater Gala. The Gala sold out, which goes a long way toward funding our vital work.

We were treated to a fascinating dialogue between board member Seth Gelblum and producer Michael David, who talked to us about the process of translating the show from London to New York, the challenges of working with a cast of children, and how the theater business has changed during his 40-plus years as a producer.

About the Canavan Foundation

The Canavan Foundation is a non-profit organization dedicated to educating at-risk populations about Canavan disease and other Jewish genetic diseases. We encourage carrier screening and support research that will lead to treatment and a cure for Canavan disease. Your tax-deductible contributions help fund education, outreach, and research programs important to our mission.

How You Can Help

- Write a letter to your temple’s tzedakah fund
- Share Canavan videos with friends and networks
- Ask children and grandchildren about testing plans
- Tell your rabbi about our training program
- Ask your OB/GYN to display our brochures
- Support the Canavan Foundation by donating