On October 25, 1994, Gershon Natan Brickner was born to Debby Duitch and Avi Brickner. Although Gershie was not very active in utero, he was a beautiful little boy born with APGAR scores of 8 and 9. His parents felt doubly blessed—first with Debby’s successful pregnancy at the age of 36, and second with their having a (seemingly) healthy baby. Their lives were soon changed when Gershie was diagnosed with Canavan disease.

Debby had not been screened for Canavan disease prior to her pregnancy. It was not a routine test, and being unaware of any relatives having the disease, she did not ask to be screened for carrier status. Since Gershie’s diagnosis however, Debby’s mother Jean Duitch has worked hard to prevent other families from experiencing the hardship that affected her family. Jean established a successful, free screening program in her home town of Omaha, NE, and has written a manual on how to set up a community program to test for all of the Jewish genetic diseases.

While Gerschie and his family faced many challenges over the years as a result of his disease, they have also experienced a great deal of joy. His parents describe Gershie as “filled with smiles, laughter, and unconditional love.”

On November 1, 2007, in Jerusalem, where the family resides, Gershie became a Bar Mitzvah! Although he broke his leg a month before and could not stand and be called up to the Torah, Gershie proudly sat in his chair and with his head-controlled switch, was able to be an active participant in the ceremony. Gershie’s switch was pre-programmed with his father’s voice. He would turn his head to activate the switch, doing it for the first Bracha, and then for the second Bracha. After listening to the reading of the Torah, Gershie activated the switch one more time to initiate the Bracha for after reading the Torah.

Modern technology (combined with his family’s loving determination) made it possible for Gershie to let the congregation know he was in control and that he was accepting his place among those who had gone before him.
Spotlight On: Mary Gaffney, Administrative Associate

Mary Gaffney was born and raised in the Bronx. After attending Boston University from 1953-55, she returned to New York and worked as a secretary for various companies. In the late 1970s, she obtained her real estate license and sold condos and co-ops in Manhattan. When the real estate market went soft during the mid-1980s, Mary turned her attention to learning computers. She was hired by the College Board as an Office Manager and Marketing Coordinator, and remained there until she retired in 1999.

While at the College Board, Mary worked with Sue Watts, a neighbor of the late Roz Rosen, then President of the Canavan Foundation. Sue knew Mary’s personality and abilities well, and thought that Mary would be a great asset to the Canavan Foundation. Sue recommended Mary and an interview was arranged.

Mary joined the Canavan Foundation in March, 2001 as an Administrative Associate. Working from her home, she manages the database of donors, keeps up the website and creates correspondence. As treasurer Deedy Goldstick confesses, “I don’t think we could function without Mary. She is smart, quick, and just a delight to work with. She certainly keeps the Canavan Foundation machine well-oiled.”

In her free time, Mary volunteers at a food pantry and at the Mercy Center in the Bronx teaching office skills. Her four nieces and eight grandchildren give her a great deal of joy.

The Jewish Genetic Disease Consortium Receives Important Funding

The Jewish Genetic Disease Consortium is made up of 15 different organizations, each dedicated to heightening awareness of one of the 11 genetic disorders affecting the Ashkenazi Jewish population. As one voice, the Consortium has become a powerful force for educating the medical community, the rabbinic community, and the general public about the need for carrier testing for all of these conditions. When couples know their carrier status, they are able to make informed choices when they are planning their families.

The Consortium is extremely fortunate to have Temple Emanu-El’s Rabbi Emeritus, Peter Kasdan, as its Rabbinic Advisor. He has been working at great speed in educating rabbis nationwide and encouraging carrier screening, which can ultimately prevent the diseases.

While Rabbi Kasdan had success in the rabbinic community, the Consortium has been making strides in educating health care providers about carrier testing. The American College of Obstetricians and Gynecologists (ACOG)—the national organization of Ob/Gyns—has policy statements about four of the Jewish genetic diseases, but the most comprehensive testing panels currently contain 11. Since this is obviously insufficient, the Consortium developed a proposal to create an educational initiative—the Grand Rounds Seminar—directed at Ob/Gyn providers.

Although ACOG was interested in educating their members on the topic, they were unwilling to commit funds to this endeavor in the face of competing priorities. Rabbi Kasdan turned to the generosity of his congregation and approached Marty and Evelyn Bernstein.

The Bernstein Family Foundation made the initial contribution to pilot the program, which has been receiving rave reviews at teaching hospitals and society meetings throughout the New York area. Pleased with its success, the Bernsteins agreed to fund the “rolling out” of the Grand Rounds project nationally.

“If we keep the ball rolling at this stage, the momentum will make it easier for the Consortium to get future funding.”

Mary Gaffney with her great nephews (left to right) Kyle, Thomas, and Christopher Ulmschneider

Foundation machine well-oiled.”
The Canavan Foundation’s 2007 Spring Theater Benefit was held on April 25th. The evening began with a pre-theater buffet dinner at Restaurant Charlotte and was attended by 95 friends of the Canavan Foundation. During dinner there was a Q&A session with Curtains producer Roger Berlind and Seth Gelblum of the Canavan Foundation, which informed and amused the audience with information about what it takes to bring a show to Broadway. Quite a few interesting questions were posed by some of the guests.

Ruth Kreisman and Pat Hirschhorn made a presentation, formally renaming the Foundation’s Clinical Research Training Fellowship the “Rosalind Poss Rosen Clinical Research Training Fellowship.” The new name pays tribute to the inspired work of the late Roz Rosen and her commitment to supporting the research of young scientists in finding treatments and ultimately a cure for Canavan disease.

Lois Neufeld gave a brief overview of the projects the Foundation has been involved with in the past year and noted some of the projects that it will be taking on in 2008.

After dinner, the group was joined by 40 additional supporters to see the Broadway hit Curtains, a musical comedy whodunit starring David Hyde Pierce. Pierce is well-known for his Emmy-winning roll as Dr. Nyles Crane on the TV sitcom Frasier, and won a Tony Award for his performance in Curtains. Pierce’s co-star, Debra Monk is an Emmy Award winner for her role as Katie Sipowicz on TV’s NYPD Blue.

Curtains is set in 1959, backstage of a new musical at Boston’s Colonial Theatre. An untalented leading lady (Monk) dies on opening night during her curtain call, and Lieutenant Frank Cioffi (Pierce) arrives to investigate. While on the case, Cioffi gets “stage-struck” and lured into romance. As a result, he wants the show to become a hit almost as much as he wants the murder solved.

Co-chaired by Deedy Goldstick and Pat Hirschhorn.

Lois Neufeld (far right) and theater benefit attendees applaud Rabbi Peter Kasdan (center, standing with wife Sheila) during a pre-theater buffet dinner at Restaurant Charlotte in New York City. The Kasdans have both played important roles in the Foundation.
Screening Day at the Riverdale Y a Huge Success

On December 2, 2007, the JGDC, in collaboration with UJA-Federation of New York and Montefiore Medical Center, held a free genetic screening program in the Bronx at the Riverdale Y. The entire cost of the program was underwritten by the UJA-Federation of New York.

Despite cold and snowy weather, 101 young people showed up to receive free screening for nine Jewish genetic diseases. Prior to the event, they filled out questionnaires on-line which were reviewed by genetic counselors. The eligible candidates were then assigned a time to arrive. Each participant met individually with a genetic counselor, and had their blood drawn by a phlebotomist.

With the results obtained, the participants will know if they are at risk for passing down a gene mutation for a Jewish genetic disease to an unborn child, and what options they have if they are.

The event would not have been possible without Dr. Adele Schneider of the Victor Center for Jewish Genetic Diseases, who developed the protocol and oversaw the medical aspects of the day. The successful turnout was the direct result of Shari Ungerleider’s efforts in getting the event widely publicized. Shari also pre-registered participants, and made sure that volunteers, phlebotomists, and genetic counselors were well-fed throughout the day. Local newspapers were there to cover the program, which went off without a hitch.

Organizing a Free Genetic Screening Program in Your Community

Putting together a screening day in any community takes some work but is well worth the effort. The purpose is two-fold: the event itself provides free screening services to those who attend, and the advertising of the event educates people about the importance of genetic counseling and testing, even if they are unable to participate. The message to deliver is clear—since one simple blood test can identify mutations for several Jewish genetic diseases, it is easy for people to learn their carrier status.

The first thing that is needed is an event underwriter—an organization that will cover all costs for testing, counseling, advertising, and any other fees. Family funds, synagogues, and charitable organizations such as the March of Dimes are some examples of funding sources.

The second necessity is a host venue at which to hold the event. Logistically, there should be a place for people to register and fill out paperwork, such as a lobby. Then, two or three rooms close together will enable participants to go to one room to be counseled and then sent on to have their blood drawn in another. It is a good idea to have cookies and juice on hand for people once they are done. Local delis and supermarkets are usually happy to donate these.

The host venue can also be helpful with advertising by including the event in their newsletter, mailings, etc. A flyer is also effective in getting the word out about the program. These can be given to local synagogues, day schools, etc. for them to distribute.

The screening program itself can be done in two ways—a couple screening and an individual screening. The benefit of couple screening is that both people are counseled and tested, so that they are certain of their results and can make decisions based on their results, as a couple. The drawback is that you do not reach as many people, since the testing cost is per individual, not per couple. In counseling and testing individuals, or one spouse of a couple, you are able to screen more people. Then, if one spouse’s results are positive, the genetic counselor can advise them to have the other spouse tested.

More information on instituting a community screening program can be found in Jean Duitch’s manual Testing for Jewish Genetic Diseases. The manual spells out how to prepare a plan and addresses issues such as confidentiality, liability, marketing, fund raising, and how to deal with testing results. To order a copy, contact Jean Duitch at (402) 393-1141 or jduitch@aol.com. The Jewish Genetic Disease Consortium can also provide information on screening programs.
2006 Clinical Research Training Fellowship Update

Co-sponsored by the American Academy of Neurology Foundation and the Canavan Foundation, The Rosalind Poss Rosen Clinical Research Training Fellowship supports research toward the cause, treatment, or cure of Canavan disease. In 2006, Chikkathur N. Madhavarao, Ph.D., was awarded this fellowship, supporting two years of research under the mentorship of Dr. M.A. Namboodiri. The following is an update of Dr. Madhavarao's work from the past year.

My post doctoral research on N-acetylaspartate (NAA) metabolism has led to the important understanding that in Canavan disease, nerve fiber insulation has gone awry. NAA is an important neurochemical produced by neurons. In normal brain development, NAA breaks down into aspartate and acetate, the “building blocks” that work to insulate the nerve fibers. The brains of Canavan patients however, are not able to achieve proper nerve fiber insulation as the NAA-breaking enzyme, does not work.

Dr. Namboodiri and I proposed that if we can supply the brain early on with those building blocks by other means, we may stop the deterioration of the brain. We started testing our treatment strategy, orally supplementing acetate in the animal models of Canavan disease. Since the ASPA gene knockout mouse and the Tremor rat do not have a functioning NAA-breaking enzyme, they both demonstrate the deteriorating conditions of the disease.

The treated mutant animals showed better motor skills in terms of balancing and staying on a speeding rotating rod, and also improvement in some parameters of locomotion. We found that treatment also improves tail health. In the Tremor rat model however, the female mutant rats responded better to treatment than the males, although definitive conclusions would require a larger number of rats in the study. The challenge in breeding a larger number of mutant animals is that the mother rats tend to isolate the mutants from nursing and often kill and eat them.

We continue working to understand what type of lipids/oils are built using the building blocks derived from NAA so that we can improve treatment strategy.

Canavan Foundation: Looking Back

Many years ago, Babs Armour, one of the founders of the Canavan Foundation, had attended the weddings of Charlotte and Hal Chefitz’s two daughters. Both were officiated by Rabbi Peter Kasdan at Temple Emanu-El in Livingston, New Jersey.

Babs was initially impressed by Rabbi Kasdan, and overjoyed to learn that he required that all of his pre-marital couples were tested for Tay Sachs disease. At the time, Tay Sachs was the only Jewish genetic disease test available, and she herself had facilitated the first community screening for Tay Sachs disease in New Jersey some years before. That this rabbi had decided to insure that all couples he married knew whether or not they carried the gene for Tay Sachs disease was something she would never forget.

Years later, the granddaughter of Babs’ friends Eileen and Arnie Alperstein was diagnosed with Canavan disease. The little girl, Morgan, was blind, confined to a wheelchair, and severely developmentally delayed. She died at seven and a half years old, but was blessed to have devoted parents who were determined to insure that others would not have to face the same tragedy that they did. In the words of her mother, Orren Alperstein Gelblum,

“We loved Morgan fiercely, but wondered whether there was anything we could have done to prevent this condition. At the time there was not, but carrier and prenatal screening became available soon after we adopted our son. Now couples can know whether they have a strong chance of bringing an affected child into the world, and can decide what they want to do in advance. More people should know about this screening.”

Morgan’s parents decided to form the Canavan Foundation. It quickly became apparent that rabbinical education was key in spreading the word about testing for Canavan and other Jewish genetic diseases. Babs Armour remembered the dynamic, red-haired Rabbi Peter Kasdan from Temple Emanu-El and asked him to spearhead the effort. Not only did he agree, but his wife Sheila was so moved by the commitment of the Canavan Foundation that she joined its board. Rabbi Kasdan’s efforts to date have included the formation of an “all-star” rabbinic advisory committee and the initiation of numerous important outreach programs aimed at educating those at risk.
Canavan disease is a fatal, inherited, neurological disorder found most frequently among children of families in which at least one parent is Jewish of Ashkenazi (Eastern European) descent.

Children with Canavan disease are severely disabled, both mentally and physically, as their brains do not produce enough white matter necessary for normal brain function. Generally, they cannot sit, crawl, walk or speak. They have limited ability to move their hands and may gradually lose their ability to see and to swallow food. Children with this degenerative disease have a life expectancy of 3 to 10 years, although some have lived into their early 20s.

There is currently no cure for Canavan disease, but it can be prevented. With one blood sample, parents can be screened for Canavan and other Jewish genetic diseases.

A list of sites that perform carrier screening and prenatal and diagnostic testing can be found on our website www.canavanfoundation.org. The Canavan Foundation does not recommend or endorse the listed labs, is not affiliated with these or any labs, and does not accept responsibility for the administration or interpretation of tests.

About the Canavan Foundation

The Canavan Foundation is a non-profit organization founded in 1992 by families and friends of children with Canavan disease. Our mission is to educate at-risk populations, medical communities and other professional groups, and to facilitate research towards the cure of Canavan disease. We support efforts to ensure that carrier and prenatal testing is available to all who want it. The Canavan Foundation collaborates with other Jewish genetic disease organizations whenever necessary to make greater strides with Congress, doctors, rabbis, and the Jewish community nationwide.

How You Can Help

Increase Public Awareness
Education leads to screening—the first line in prevention. Please help us to spread the word about Canavan disease by contacting your local synagogue, other Jewish organizations, and your friends.

Provide Financial Support
Your tax-deductible contributions help fund the critical research needed to eradicate Canavan disease, and make possible public education materials (such as this newsletter) and our informative website. Donations can be sent directly to the Canavan Foundation with the form below or made on the internet at www.paypal.com.

Contribution

Name (please print)

Address

City, State, Zip

Telephone (day)

Telephone (evening)

Fax

Email

☐ I would like to make a contribution

☐ In memory of  ☐ In honor of

A check is enclosed for $________

Name (please print)

Address

City, State, Zip

Please mail completed form and payment to:

Canavan Foundation, Inc., 450 West End Avenue, #10C, New York, NY 10024