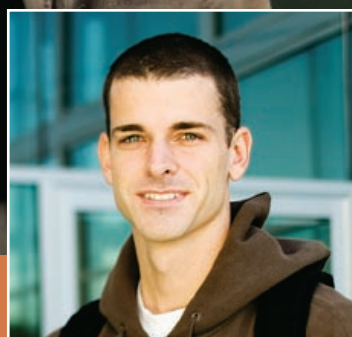


# Getting Tested: Why College Jews Need To Know Now



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Freshman year, **Shoshana Rosen (Nu '04)** took a test that determined her future.

While working as a camp counselor in Pennsylvania, Rosen participated in a genetic screening program run by the Victor Center for Jewish Genetic Diseases in Philadelphia. The test revealed that Rosen carries the gene for Cystic Fibrosis (CF), an inherited chronic disease that affects the lungs and digestive system of

about 30,000 people in the United States. CF is one of many fatal genetic diseases prevalent among Ashkenazi Jews (Jews of Eastern European descent). As a carrier, Rosen has a 25 percent chance of producing an affected baby if her future husband is also a carrier for CF. Rosen's plans to marry Jewish put her at an increased risk for passing on the genetic disorder.

That's because 1 in 4 Ashkenazi Jews is a carrier for a Jewish genetic disease which could be as treatable as Gaucher's Disease Type 1 or as devastating as Tay-Sachs Disease for which there is no cure. Babies born with Tay-Sachs usually die before the age of 5. Without prior screening, young couples run the risk of

being genetically incompatible and facing heartbreaking consequences.

When Rosen learned all this, she took the cause of genetic testing awareness to heart and in 2006, her senior year at the University of Pittsburgh, she brought the Victor Center to campus to educate students about Jewish genetic diseases (see sidebar for more information). Nearly 200 young people, including Rosen's boyfriend, received genetic counseling and were screened for nine different diseases free of charge. Rosen's boyfriend tested negative for the CF gene.

Rosen is now 24 years old and working as the youth director at a Reform temple in Delaware. She and her college boyfriend,

Jonathan Finger, are still dating and can consider marriage free of fear from transmitting CF to their children.

“The good thing about getting testing young is that it’s of such low consequence,” says Rosen. “What about couples who get tested when they are already engaged? What do they do then?”

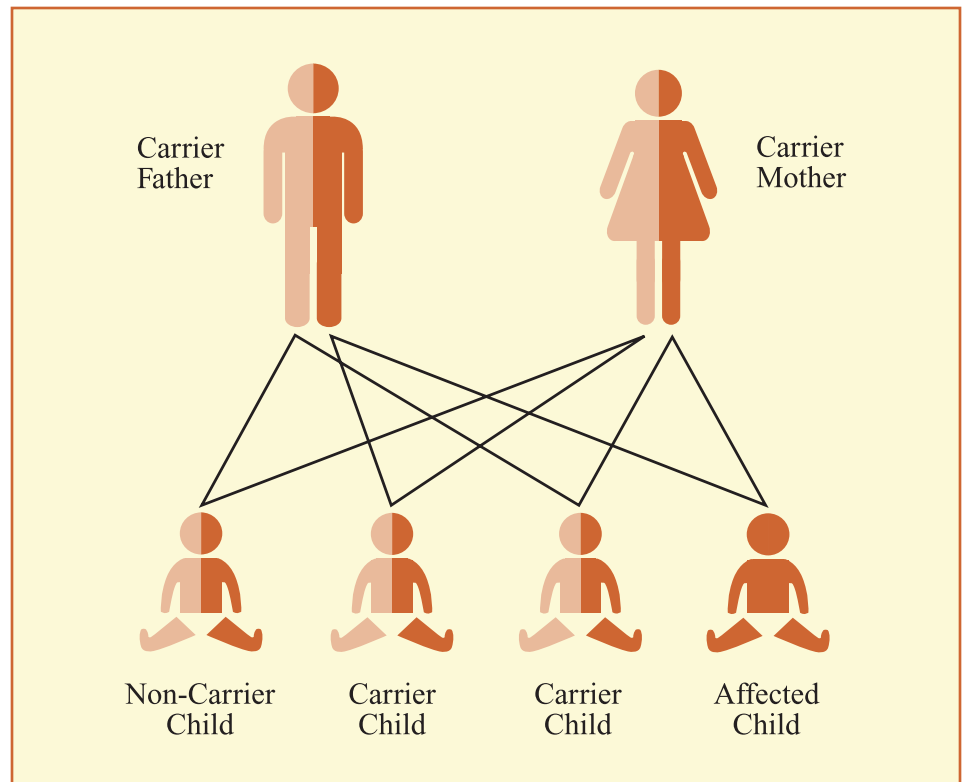
Dr. Adele Schneider, clinical director of genetics at the Victor Center says that promoting awareness early about Jewish genetic diseases is the first step toward eradicating them. She’s worked with many Hillels to sponsor genetic counseling sessions for Jewish students on campus. Penn Hillel hosted the Victor Center for a cross-campus genetic screening for Jewish students from University of Pennsylvania as well as nearby Temple University and Drexel University. Awareness campaigns by several Jewish organizations have also been launched at schools in New York, Arizona, California and Massachusetts. And Yeshiva University offers screening for its students through Dor Yeshorim, an anonymous testing program developed by an Orthodox rabbi in the 1980s.

While instances of Jewish genetic diseases have declined significantly since the Jewish community first embraced screening in the 1970s, there still remains a great lack of awareness within the Jewish community according to Michelle Gilats, genetic counselor for Children’s Memorial and the Chicago Center for Jewish Genetic Disorders.

“We want young people to know that this opportunity [for early screening] exists,” explains Gilats. “Judaism is not just a religion, but an ethnic background,” she says. “All ethnic groups have higher rates of different illnesses.”

Because Ashkenazi Jews were an isolated population for nearly 1,000 years, tending to marry among each other with little migration in or out, genes mutated at a higher rate. The result is higher concentrations of certain diseases and disorders within Ashkenazi ancestry.

Robin Fiddle Posnack and her family have been struggling with one Jewish



genetic disease for more than two years. Posnack’s son, Jack, was the 601st child to be diagnosed with the Jewish genetic disease Familial Dysautonomia (FD), also known as Riley-Day disease, in February 2006. Posnack, already a mother to a healthy son from a previous marriage, told a Westchester County, New York, newspaper that her doctor didn’t screen for FD when she became pregnant again by her second husband in 2005. A blood test for FD had been developed four years earlier. As it turns out, both Posnack and her identical twin sister Mandy are carriers. Neither one had any idea until Jack started showing symptoms at birth. Posnack noticed that her son did not shed tears, not even during his bris (ritual circumcision). A barrage of tests over the next eight weeks revealed the devastating news.

Children with FD are lacking a vital protein that affects the sensory and nervous systems. Without that protein, basic functions such as swallowing and breathing are a struggle. FD patients, like Jack, cannot regulate their own body temperature and blood pressure, something a healthy person does automatically. Jack also feels no pain, which makes him vulnerable to a host of

other health issues. The good news is that new research has found babies diagnosed in 2006 (as Jack was) have a 50% chance of living to age 40 (the average lifespan for an FD patient is 15 years).

Leah Metz, 23, and Avi Buchbinder, 24, want to avoid the pain Jack’s family has endured. The couple will be married this summer and underwent genetic counseling in December at the urging of a friend through the Chicago Center.

Because Metz, a graduate of UC Irvine and Buchbinder, a Northwestern University graduate student, are of Ashkenazi descent (though Metz’s mother converted to Judaism) there was some concern about the health of their future children. To their relief, bloodwork on Buchbinder determined he is not a carrier for any genetic diseases and so even if Metz is, there is no risk of producing an affected child together.

“We wanted to be prepared so that we don’t have to worry when we’re ready to have children,” says Metz.

*For more information about setting up a screening on your campus please contact the Victor Center at (215) 456-8722.*

# Jewish Genetic Diseases

## Bloom Syndrome

Individuals have short stature, sun-sensitive facial lesions, an increased susceptibility to infections and respiratory illness, and an increased predisposition to certain cancers and leukemia. Men with Bloom syndrome are usually infertile, and fertility appears to be reduced in women.

## Canavan Disease

Canavan disease is a severe degenerative disease of the central nervous system. Most children with it appear normal at birth. It is not until three to five months of age that parents might notice subtle difference such as a lack of ability to performing motor tasks, such as rolling over. Grasping and visual inattentiveness or tremors are frequently noted.

## Cystic Fibrosis

CF is a genetic disease that is not limited to the Ashkenazi Jewish population. It affects about 30,000 children and adults in the United States. CF makes your body produce an abnormally thick, sticky mucus. This is what happens when cells in the lungs and pancreas fail to transport



salt in the body where it needs to go. The thick CF mucus also blocks digestive enzymes from reaching the intestines where they're supposed to break down and digest food.

## Familial Dysautonomia

Also known as Riley-Day Syndrome, FD is a disease that causes the autonomic and sensory nervous systems to malfunction. The hallmark of FD is the lack of overflow tears with emotional crying. Children with FD may have difficulty feeding. They may also be unable to feel pain.

## Fanconi Anemia

An inherited disorder which is characterized by a reduced number of all types of blood cells in the body, Fanconi anemia leads to bone marrow failure. Symptoms associated with the disease may include missing bones in the thumbs and arms, increased risk for cancer and leukemia, brown coloring to the skin, kidney problems and smaller than usual stature.

## Gaucher Disease

There are three types of Gaucher (*pronounced go-shay*) disease (type I, II, and III). Type I is the most common form of the disease. The signs and symptoms of Gaucher disease vary greatly and can appear at any age. The most common symptom of type I is an enlargement of the spleen and/or liver, which is usually painless. Other symptoms may include bruising, bone pain, frequent nosebleeds and a lack of energy. Also, children with type I Gaucher may be shorter than their peers and may have delayed puberty.

## Type 1a Glycogen Storage Disorder

GSD1a is caused when the body is missing an enzyme that is needed by the liver to convert sugar from its storage form (glycogen) to the form that can be used by the body to produce energy (glucose).

## Maple Syrup Urine Disease

MSUD is named for the characteristic maple syrup smell of the urine in those with the disease. In addition, early symptoms include irritability, poor feeding and lethargy. Without treatment, MSUD can lead to neurological damage, seizures, coma and death. With early detection and careful dietary control, normal growth and development are possible. It can be detected by routine newborn screening.

## Mucopolidosis Type IV

Symptoms of ML IV occur when the chemical that typically breaks down certain fatty substances (lipids) in the body is not present. These lipids then accumulate to toxic levels in the body.

## Niemann-Pick Disease

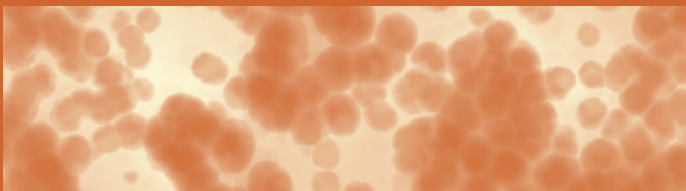
Harmful quantities of a fatty substance accumulate in the spleen, liver, lungs, bone marrow and sometimes in the brain with Niemann-Pick Disease. Children usually have the first signs of the disease at about three to five months of age, which include a progressive loss of early motor skills, feeding difficulties, and a large abdomen.

## Tay-Sachs Disease

Classical Tay-Sachs is an inherited, genetic disorder that causes progressive degeneration and destruction of the central nervous system. Babies born with Tay-Sachs disease appear normal at birth, and symptoms do not appear until about four to six months of age. Children begin to lose previously attained skills, such as sitting up or rolling over. They gradually lose their sight, hearing and swallowing abilities. An estimated one in every 25 Ashkenazi Jews is a carrier for Tay-Sachs Disease.

— from the Victor Center for Jewish Genetic Diseases at the Albert Einstein Healthcare Network

## Jewish Genetic Testing at the University of Pittsburgh



On December 4, 2006, the Victor Center for Jewish Genetic Diseases in Philadelphia, Pennsylvania provided the Jewish students of Pittsburgh with an opportunity to be screened for nine hereditary diseases that are prevalent among Ashkenazic Jews. The effort was spearheaded by **Shoshana Rosen (Nu '04)** and 187 students participated in this first year. That terrific response is due, in large part, to Shoshana's hard work, energy and belief in the importance of this type of screening. "I decided to get involved because I found out that I am a carrier of Cystic Fibrosis, and I wanted to provide my friends with the same opportunity to learn their genetic make-up in order to eradicate this and other terrible and debilitating diseases that are prevalent in the Jewish population," states Rosen.

The screening, which was conducted at the Hillel Jewish University Center, was made possible by a generous grant from Louis Victor, who lost two children to Familial Dysautonomia, and wants to ensure that others do not have to endure the heartache of having loved ones plagued with such diseases. The money has been used to implement screening programs at college campuses nationwide. While the Victor Center in Philadelphia has a partnership with a philanthropic lab in New York, which allows them to screen students at a discounted rate of \$350 per person, the screenings on campus were absolutely free to all students. Without a program such as this, screening costs are a few thousand dollars per person.

The test screens for the following genetic diseases that are either specific to or more prevalent in the Jewish population: Fanconi Anemia- type c,

Cystic Fibrosis, Gaucher Disease – type 1, Glyco, Bloom Syndrome, Caravan disease, Familial Dysautonomia gen Storage Disease- type 1a, Neimann-Pick Disease – type A and Tay-Sachs Disease. Another important aspect of the screening is "by being tested, it increases awareness of these diseases," adds Rosen.

The Pittsburgh Hillel JUC has set an example for future such programs at other college campuses. Many of the staff members and students of the Hillel JUC, as well as several members of **Nu Chapter** at the University of Pittsburgh helped throughout the screening process.

The screening process lasts approximately 20 minutes per person and involves meeting with genetic counselors to ensure informed consent and blood drawing by geneticists. Students were informed of the results one month after the screening. Rosen, who went through this process herself, commented, "People who tested positive for a disease, from what I have seen, are totally ok. They are just grateful that they found out early in their lives, before they are in serious relationships, when getting the information doesn't have a significant impact."

