What are Jewish Genetic Diseases?
Jewish genetic diseases are a group of disorders that occur with higher frequency in the Jewish population. The Ashkenazi Jews, those whose ancestors were from Central and Eastern Europe (i.e. Poland, Russia, Germany, Lithuania, etc.) are at higher risk than the general population for several genetic diseases. This is primarily due to the fact that the Jewish communities in Europe were small and isolated, and members tended to marry within those communities. Since it is estimated that all individuals carry a small number of gene changes (called mutations), the ones present in those small groups of European Jews became more prevalent in future generations.

What are the diseases that are more prevalent in the Ashkenazi Jewish population?
Face the Facts, it’s not just Tay-Sachs! Currently, there is carrier testing for 18 preventable genetic diseases affecting Ashkenazi Jews, most of which are life threatening. Below are brief descriptions of each disorder with the approximate carrier rate (the proportion of Ashkenazi Jews who have a single copy of the specific recessive gene mutation) in parenthesis. Below are the Ashkenazi Jewish genetic diseases most commonly screened for:

1. **Bloom Syndrome** - Characterized by short stature, a sun-sensitive skin rash, an increased susceptibility to infections and higher incidence of leukemia and other cancers. (1 in 100)

2. **Canavan Disease** - A neurodegenerative disorder that presents with normal development until 2-4 months and then there is a progressive loss of skills. Those affected typically die in childhood but may live into adolescence. (1 in 40)

3. **Cystic Fibrosis** - Causes the body to produce thick mucus that accumulates in the lungs and digestive tract, resulting in lung infections and poor growth. (1 in 25)

4. **Familial Dysautonomia** - Causes the autonomic and sensory nervous system to malfunction, affecting the regulation of body temperature, blood pressure and stress response, and causes decreased sensitivity to pain. Frequent pneumonia and poor growth may occur. (1 in 30)

5. **Fanconi Anemia Type C** - Associated with short stature, bone marrow failure, and a predisposition to leukemia and other cancers. Some children have limb, heart or kidney abnormalities and learning difficulties. (1 in 89)

6. **Gaucher Disease Type 1** - A variable condition both in age of onset and symptoms. It may present with a painful, enlarged spleen, anemia, and low white blood cell count. Bone deterioration is a major cause of pain and disability. Treatment is available. (1 in 14)
7. **Glycogen Storage Disease, Type 1a** - A metabolic disorder that causes poor blood sugar maintenance with sudden drops in blood sugar, growth failure, enlarged liver and anemia. Disease management involves lifelong diet modification. (1 in 71)

8. **Maple Syrup Urine Disease** - A variable disorder of amino acid metabolism. Named for the characteristic maple syrup smell of urine in those with the disorder. With careful dietary control, normal growth and development is possible. If untreated, it can lead to poor feeding, lethargy, seizures and coma. (1 in 81)

9. **Mucolipidosis IV (ML4)** - A progressive neurological disorder with variable symptoms beginning in infancy. Characteristics include muscle weakness, severe intellectual disabilities and eye problems. (1 in 125)

10. **Niemann-Pick Disease Type A** - A progressive neurodegenerative disease in which a harmful amount of fatty substance accumulates in different parts of the body leading to death by age two to four years old. (1 in 90)

11. **Tay-Sachs Disease** - An apparently healthy child begins to lose skills around 4-6 months of age and there is a progressive neurological decline leading to blindness, seizures and unresponsiveness. Death usually occurs by the age of 4-6. (1 in 25)

12. **Dihydrolipoamide Dehydrogenase Deficiency (DLD Deficiency)** - Presents in early infancy with poor feeding, frequent episodes of vomiting, lethargy and developmental delay. Affected individuals develop seizures, enlarged liver, blindness and ultimately suffer an early death. (1 in 96)

13. **Familial Hyperinsulinism** - Characterized by hypoglycemia that can vary from mild to severe. It can be present in the immediate newborn period through the first year of life with symptoms such as seizures, poor muscle tone, poor feeding and sleep disorders. Medical or surgical management can control glucose levels. (1 in 66)

14. **Joubert Syndrome** - Characterized by structural malformations of the cerebellar vermis. The most common features of Joubert syndrome in infants include abnormally rapid breathing, hypotonia, jerky eye movements (oculomotor apraxia), developmental delay, and ataxia. Kidney and liver abnormalities can develop, and seizures may also occur. (1 in 92)

15. **Nemaline Myopathy** - This is the most common congenital myopathy. Infants are born with hypotonia and usually have problems with breathing and feeding. Later, some skeletal problems may arise, such as scoliosis (curvature of the spine). In general, the weakness does not worsen during life but development is delayed. (1 in 66)

16. **Spinal Muscular Atrophy (SMA)** - A group of diseases affecting the motor neurons of the spinal cord and brain stem, which are responsible for supplying electrical and chemical signals to muscle cells. Without proper signals, muscle cells do not function properly and become much smaller (atrophy), leading to muscle weakness. Individuals affected with SMA have progressive muscle degeneration and weakness, eventually leading to death. (1 in 41)

17. **Usher Syndrome Type 1F** - Characterized by profound hearing loss which is present at birth, and adolescent-onset retinitis pigmentosa, a disorder that significantly impairs vision. (1 in 141)
18. Usher Syndrome Type III - Causes progressive hearing loss and vision loss. Hearing is often normal at birth with progressive hearing loss typically beginning during childhood or early adolescence. Often leads to blindness by adulthood. (1 in 107)

How are the Ashkenazi Jewish Genetic Diseases inherited?
In each of the cells in our bodies, we have tiny structures called chromosomes. There are thousands of genes on each chromosome. Genes are the basic units of heredity that are passed from parent to child. These genes make up who we are and guide the development of the body. For instance, there are genes for eye and hair color. These genes are in pairs; one is inherited from each parent. Sometimes, a change in a gene, called a mutation, causes the gene to malfunction and can lead to disease.

The diseases mentioned here are inherited in an autosomal recessive manner. This means that males and females are equally likely to be carriers and are equally likely to be affected (have a disease).
Individuals who are affected with one of these diseases have two mutations, one in each copy of the gene, for that disease. Carriers have one copy of the gene mutation and one normal working copy of the gene. Carriers are healthy individuals with no signs of the disease, and they are not at risk to develop the disease. If both partners are carriers of a mutation in the same disease gene, there is a 25% chance of having an affected child, a 50% chance of the child being a carrier like themselves, and a 25% chance of the child being neither affected nor a carrier.

How common is it to carry a mutation for one of the Ashkenazi Jewish genetic diseases?
It is estimated that 1 in 5 Ashkenazi Jews is a carrier of a gene mutation for at least one of the Ashkenazi Jewish genetic diseases.

What is the cost of screening?
The Victor Centers has a special reduced rate as a philanthropic organization so the cost for the 9-disease panel is significantly less than the cost of a similar panel through a commercial laboratory. Students and newlywed couples may be eligible for free or reduced cost screening. Additionally, screening for Glycogen Storage Disease type 1A and Maple Syrup Urine Disease can be obtained at a reduced rate. There is also a small fee for the office visit to cover the cost of consultation, blood draw, specimen handling and follow-up. The office visit fee can be billed to insurance.

Does it matter if I'm Sephardic?
Yes, Sephardic Jews are at increased risk to carry mutations for different diseases than are Ashkenazi Jews. Click here for more information on these diseases.
Does insurance cover the cost of screening?
Each insurance company is different so it is not possible to know which ones cover what testing. Many insurance companies will cover the cost of screening only if a woman is already pregnant. The total cost for screening through a commercial laboratory can be more than $3,000.00. We strongly recommend that you check with your insurance company about coverage before having the screening done through insurance. The reduced cost of screening through the Victor Centers is highly subsidized and cannot be billed to insurance.

When is the best time to be screened?
The best time to be screened is prior to starting a family. Prior to pregnancy, at-risk couples (those in which both are carriers of a mutation in the same disease gene) will have the most reproductive options available to them.

None of these diseases run in my family. Why do I need to get tested?
Carriers of Jewish genetic diseases are healthy individuals, so without genetic testing, it is impossible know if a mutation in any of the genes for these diseases is present in the family. Often, carrier status is only identified after a child is born with the disease. Therefore, most affected babies (those with the disease) are born into families with no prior history of the disease. It is important that Jewish people of Eastern European descent know about their risk for being a carrier of a gene mutation for one of these diseases, so that they can make more informed reproductive decisions.

How is the screening performed?
A simple blood test is all that is necessary for screening, much like any other blood test that your doctor would do. Only a small sample of blood is needed for the entire Jewish genetic disease panel. It is important, however, that all individuals who undergo screening have genetic counseling prior to having their blood drawn, so they understand what they are being screened for and what the implications are of a positive screening result.

If I find out I’m a carrier, what does that mean?
Carriers are healthy individuals so it does not mean anything for your own health. Carriers can pass the gene mutation on to their children, so it is important that your partner be tested as well. It is only a concern if your partner carries a mutation in the same disease-causing gene. Genetic counselors can speak with you about these risks.

What if two carriers of mutations in the same disease gene want to have children together?
If two partners are carriers of a gene mutation for the same disease, then, with each pregnancy, there is a 1 in 4 chance of having an affected child, there is a 2 in 4 chance of having a child who is an asymptomatic (healthy) carrier, and a 1 in 4 chance of having a child who is neither a carrier nor affected.

If we are both carriers, what are our reproductive options?
There are many reproductive options available to carrier couples, including prenatal diagnosis (chorionic villi sampling and amniocentesis), pre-implantation genetic diagnosis, gamete donation and adoption. Genetic counseling is recommended to learn more about all of your reproductive options. To speak with the genetic counselor at The Victor Center, please call (215)
456-8722. Additionally, your rabbi may be able to provide insight and help in making these decisions.

**What if my partner is not Jewish ... Do I still need to get screened?**
We recommend testing if at least one partner in a couple is Jewish. This is because many of the diseases can occur in the non-Jewish population as well, although at much lower frequencies. In such a situation, it is best to test the person of Jewish background first, and then test the partner only if the first person is found to be a carrier.

**Can I do the screening at home?**
There are several genetics laboratories that can be found on the internet that market Direct-To-Consumer genetic tests for various indications. Due to the complexities of genetic screening and testing, at the present time, it is recommended that any kind of genetic screening or testing be performed only in a medical office with face-to-face genetic counseling. Home test kits are not recommended.

**Do I need a referral from my doctor for screening?**
No. Many individuals who come to The Victor Centers are referred by their rabbis or by their friends who have already been screened. Or they may read about The Victor Centers in a newspaper or Jewish community newsletter.

**How can I organize an on-campus screening at my college/university?**
Contact the Outreach Coordinator for The Victor Centers, for more information. She works closely with college students on all aspects of planning and implementing screening programs on college campuses.

**How can I organize a screening program in my community/synagogue?**
Contact The Victor Centers for more information on how to get started. The staff at The Victor Centers is available to educate your members and to help your group with fund-raising ideas. Additionally, the staff can provide everything needed for the actual screening day, including genetic counselors, supplies, phlebotomists to draw blood, packaging and shipping materials, and follow-up for results.

**Where can I find more information on Jewish Law and screening?**
[www.daneisenberg.com](http://www.daneisenberg.com)

**Are there other diseases that are more prevalent in the Ashkenazi Jewish population?**
Yes, there are other diseases, but because of their complex inheritance patterns, they are not felt to be appropriate for population screening. These include [Torsion Dystonia](#) and Hereditary Breast/Ovarian Cancers. Because these conditions are inherited in a different pattern than those previously mentioned, and because the presence of a mutation in either of these genes has direct health implications on the individual and family, it is strongly recommended that a person undergo more comprehensive genetic counseling before choosing to screen for these two conditions.

There are also some additional recessive conditions found in the Ashkenazi Jewish population,
but several factors play a role into whether or not they are included in population screening, 
including the carrier frequency, severity of the disease, and sensitivity of the testing. The list of 
diseases is constantly changing so it is important to check back often for the most up-to-date 
information.

Other Information Sources

The following web sites are also good sources of information regarding Jewish genetic diseases:

**General information**: American College of Medical Genetics: [www.acmg.net](http://www.acmg.net)
Chicago Center for Jewish Genetic Disorders: [www.jewishgeneticscenter.org](http://www.jewishgeneticscenter.org)
Genetic Alliance: [www.geneticalliance.org](http://www.geneticalliance.org)
Jewish Ethics: [www.jewishmedicaethics.com](http://www.jewishmedicaethics.com)
Jewish Genetic Diseases of Greater Phoenix: [www.jewishgeneticsphx.org](http://www.jewishgeneticsphx.org)
March of Dimes: [www.marchofdimes.com](http://www.marchofdimes.com)
National Organization for Rare Disorders: [www.rarediseases.org](http://www.rarediseases.org)
National Society of Genetic Counselors: [www.nsgc.org](http://www.nsgc.org)
Israeli National Genetic Database: [www.goldenhelix.org/israeli](http://www.goldenhelix.org/israeli)
Genetic Testing: [www.mytestingoptions.com](http://www.mytestingoptions.com)

**Bloom Syndrome**: Bloom’s Syndrome Foundation: [www.bloomssyndrome.org](http://www.bloomssyndrome.org)

**Canavan Disease**: Canavan Foundation: [www.canavanfoundation.org](http://www.canavanfoundation.org)
Canavan Research Foundation: [www.canavan.org](http://www.canavan.org)
Canavan Research Illinois: [www.canavanresearch.org](http://www.canavanresearch.org)
Jacob's Cure: [www.jacobscure.org](http://www.jacobscure.org)

**Cystic Fibrosis**: Cystic Fibrosis Foundation: [www.cff.org](http://www.cff.org)
Cystic Fibrosis Living: [www.cfliving.com](http://www.cfliving.com)
Live For a Cure: [www.livforacure.org](http://www.livforacure.org)
Cystic Fibrosis.com: [www.cysticfibrosis.com](http://www.cysticfibrosis.com)

**Familial Dysautonomia**: Dysautonomia Foundation: [www.familialdysautonomia.org](http://www.familialdysautonomia.org)
FD Hope Foundation: [www.fdhope.org](http://www.fdhope.org)
Familial Dysautonomia Now Foundation: [www.fdnow.org](http://www.fdnow.org)

**Fanconi Anemia**: Fanconi Anemia Research Fund: [www.fanconi.org](http://www.fanconi.org)

**Gaucher Disease**: National Gaucher Foundation: [www.gaucherdisease.org](http://www.gaucherdisease.org)

**Glycogen Storage Disease Type 1A**: Association for Glycogen Storage Disease:
[www.agsdus.org](http://www.agsdus.org)
The Children's Fund for G.S.D. research: [www.curegsd.org](http://www.curegsd.org)
Maple Syrup Urine Disease: MSUD Family Support Group: [www.msud-support.org](http://www.msud-support.org)

Mucolipidosis Type IV: ML-IV Foundation: [www.ML4.org](http://www.ML4.org)

Niemann-Pick Disease: National Niemann-Pick Foundation: [www.nnpdf.org](http://www.nnpdf.org)

Tay Sachs disease: National Tay-Sachs and Allied Disease Association, Inc.: [www.ntsad.org](http://www.ntsad.org)
National Tay-Sachs and Allied Disease Association of the Delaware Valley: [www.tay-sachs.org](http://www.tay-sachs.org)

---

*Atlanta Jewish Gene Screen*

*Victor Marcus Partnership*

The Atlanta Jewish Gene Screen is a new project for the prevention of Jewish genetic diseases, funded through the generosity of The Marcus Foundation. Led by the Victor Center for the Prevention of Jewish Genetic Diseases, the project will build awareness among doctors, clergy and the community about the current 18 preventable and many life-threatening diseases affecting Ashkenazi Jews and will organize three community screenings in partnership with Emory Genetics Laboratory. Genetic counseling and preconception screening for all 18 diseases will be available to the insured, uninsured and underinsured at the community screenings and throughout the year. Updated screening is recommended prior to every pregnancy.

For more information, please contact Debby Hirshman, project director, at debby@atljewishgenescreen.org or 215-456-5357.

Visit [www.AtlantaJewishGeneScreen.org](http://www.AtlantaJewishGeneScreen.org) to learn about upcoming community screenings in the Atlanta area.

To speak with a genetic counselor or learn more about screenings, contact Karen Grinzaid, genetic counselor, at 404-778-8516 or Karen@atljewishgenescreen.org.

---

**CONTACT INFORMATION – The Victor Centers**

The Victor Centers for Jewish Genetic Diseases
National Coordinating Office
Address: 5501 Old York Road, Levy 2 West
Philadelphia, PA 19141
Phone: 877-401-1093
Website: http://www.victorcenters.org

The Victor Centers for Jewish Genetic Diseases at
Albert Einstein Medical Center

Address: 5501 Old York Road, Levy 2 West
Philadelphia, PA 19141
Phone: 877-401-1093
Email: roensh@einstein.edu
Website: http://www.victorcenters.org

Victor Outreach and Screening Program for Ashkenazi Jewish Genetic Diseases
Floating Hospital for Children at Tufts Medical Center

Address: 800 Washington Street, Box 340
Boston, MA 02111
Phone: (617) 636-7721
Website: The Victor Outreach and Screening Program for Ashkenazi Jewish Genetic Diseases

Victor Center for Jewish Genetic Diseases at
University of Miami Miller School of Medicine

Address: Clinical Research Building
1120 NW 14th Street - 8th Floor
Miami, FL 33136
Phone: 305-243-4524
Email: DWasserman@med.miami.edu
Website: Victor Center for Jewish Genetic Diseases at University of Miami Miller School of Medicine