Prepare for Your Future—
Learn About Your Heritage

Start Today
Genzyme Corporation
Genzyme Treatment Support
1-800-745-4447
www.gauchercare.com

About Genzyme
Improving the lives of patients is and always will be Genzyme Corporation's top priority. By seeking out unmet medical needs and developing solutions, Genzyme provides hope to patients who otherwise would have no treatment options.

Genzyme’s commitment to patients is about much more than developing and delivering products. It is also about ensuring that patients have access to life-saving therapies regardless of their location or financial circumstances. They work with governments around the world to secure approvals for their products and provide a wide range of support services to patients. For more information about Gaucher disease and Genzyme Treatment Support call 1-800-745-4447, option 3.

References:

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All around the world, distinct ethnic groups have been identified as having increased risks for particular inherited disorders (an inherited disorder or genetic disease is one that is passed down from your parents to you through genes). Since several genetic disorders occur more commonly among individuals of Ashkenazi (Eastern European) Jewish descent, it has become the standard of care for doctors to offer “carrier” testing to this population.

Testing or screening for genetic diseases is a simple way to see if a gene has been passed down to you. Interested couples can have genetic testing to learn about their chances of having a child seriously affected by one of these conditions. Whether or not your parents have been tested, you may want to talk to your doctor or a genetic counselor before planning a family.

The test performed to find out if you are a Gaucher disease carrier is different than the test for determining if you have Gaucher disease. The former tests for DNA mutations, and the latter tests for enzyme deficiency. It is necessary for the carrier gene to be passed from both parents for the individual to be affected with Gaucher disease. You can find a genetic counselor at www.nsgc.org or by consulting your OB/GYN or family doctor. For more information, call the Jewish Genetic Disease Consortium at 1-866-370-GENE (4363).
Will your children be carriers of or be affected by Gaucher disease?

If you and your partner both carry a gene for the same condition, there’s a 25% chance—with each pregnancy—of having an affected child.¹

### Carrier Frequency in the Ashkenazi Jewish Population

<table>
<thead>
<tr>
<th>Condition</th>
<th>Frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gaucher disease type 1²</td>
<td>1 in 15</td>
</tr>
<tr>
<td>Cystic fibrosis³</td>
<td>1 in 26</td>
</tr>
<tr>
<td>Tay-Sachs disease⁴</td>
<td>1 in 30</td>
</tr>
<tr>
<td>Familial dysautonomia⁵</td>
<td>1 in 30</td>
</tr>
<tr>
<td>Canavan disease⁶</td>
<td>1 in 57</td>
</tr>
<tr>
<td>Glycogen storage disorder type 1a⁷</td>
<td>1 in 71</td>
</tr>
<tr>
<td>Maple syrup urine disease⁸</td>
<td>1 in 81</td>
</tr>
<tr>
<td>Fanconi anemia type C⁹</td>
<td>1 in 89</td>
</tr>
<tr>
<td>Niemann-Pick disease type A¹⁰</td>
<td>1 in 90</td>
</tr>
<tr>
<td>Bloom’s syndrome¹¹</td>
<td>1 in 100</td>
</tr>
<tr>
<td>Mucolipidosis type IV³</td>
<td>1 in 122</td>
</tr>
</tbody>
</table>
The following are descriptions of the 11 most common diseases, in order of carrier frequency.

**Type 1 Gaucher disease**
Gaucher disease is an inherited disorder in which a fatty substance builds up in the body, especially in the spleen, liver, and bone marrow, and secondarily in the lungs, kidneys, and intestines. Children or adults may have bruising, fatigue, anemia, nosebleeds, fractures, and enlargement of the liver and spleen. Treatment options are available for type 1 Gaucher disease.

**Cystic fibrosis (CF)**
CF is a disorder of unusually thick, sticky mucus production, primarily affecting the lungs and digestive system. Most individuals with CF require lifelong medical care and experience reduced life expectancy.

**Tay-Sachs disease**
Tay-Sachs disease is the best-known Jewish genetic disorder and is caused by a deficiency of an enzyme called hexosaminidase A (or hex A). Lack of this enzyme affects the brain and the nervous system, causing rapid and progressive deterioration, with death usually occurring by the age of 6. Babies with Tay-Sachs disease begin to lose developmental skills at 3 to 6 months of age.

**Familial dysautonomia (FD)**
FD is a nervous system disorder that commonly includes pain insensitivity, vomiting and sweating episodes, inability to produce overflow of tears, and unstable blood pressure or temperature. Intelligence is often normal, but learning disabilities are common. Symptom management improves quality of life, but only 50% will reach 30 years of age.

**Canavan disease**
Canavan disease is a rare and devastating childhood nervous system disorder. Canavan affects the formation of myelin, or white matter of the brain. Symptoms usually occur within the first few months of life and the disease is fatal in early childhood.
Glycogen storage disorder type 1a (GSD 1a)\(^7\)
GSD 1a is a disorder that, if untreated, results in severely low blood sugar, enlarged liver, growth retardation, and bleeding disorders. Treatment consists of strict diet and continuous tube feeding of glucose (sugar).

*Carrier frequency: 1 in 71*

Maple syrup urine disease (MSUD)\(^8\)
MSUD is a disorder that leads to the buildup of branched-chain amino acids in the blood. Without treatment, classic MSUD results in mental retardation, physical disabilities, coma, and death. Treatment requires dietary restriction of branched-chain amino acids through a special medical formula and intensive monitoring.

*Carrier frequency: 1 in 81*

Fanconi anemia type C\(^9\)
An inherited anemia sometimes accompanied by short stature, skeletal defects, and skin abnormalities. Learning disabilities or mental retardation sometimes occur. The risk of early childhood cancer, especially leukemia, is increased. There is currently no treatment.

*Carrier frequency: 1 in 89*

Niemann-Pick disease type A\(^10\)
Niemann-Pick disease type A is a severe brain and spinal cord disorder in infants. Affected babies experience feeding difficulty, recurrent vomiting, and enlargement of the spleen and liver, which causes the abdomen to appear distended by 6 months of age. There is currently no treatment, and death occurs by 4 years of age.

*Carrier frequency: 1 in 90*

Bloom’s syndrome\(^11\)
Bloom’s syndrome is a disorder characterized by poor growth, sun sensitivity, and high susceptibility to cancer. Death from cancer usually occurs before 30 years of age. Intelligence is normal. There is currently no treatment.

*Carrier frequency: 1 in 100*

Mucolipidosis type IV (ML4)\(^3\)
ML4 is a disorder characterized by severe neurological and eye abnormalities. It usually appears within the first year of life and affected individuals reach the developmental age of 1 to 2 years. There is currently no treatment.

*Carrier frequency: 1 in 122*

*Population of GSD 1a mutation frequency is currently under evaluation. However, published data indicate a frequency of 1 in 71 for individuals of Ashkenazi Jewish descent.*
What if only one partner in a couple is Jewish?
If only 1 partner in a couple is Jewish, it is usual to test that person first. If he or she is found to be a carrier of 1 of these disorders, the non-Jewish partner could then be tested.

Are there prenatal tests for these disorders?
Yes. If both parents are carriers for a gene that causes the same disorder, prenatal testing can be performed to determine whether or not the fetus is affected.

For further questions…
This brochure is intended to provide general information, please speak to your doctor or a genetic counselor for more information.

Resources to learn more
Find a genetic counselor at www.nsgc.org or talk to your doctor about getting tested. For more information about specific conditions find a genetic counselor at www.nsgc.org or talk to your doctor about getting tested. For more information about specific conditions, the following links will connect you to independent Gaucher disease groups and patient support organizations in the United States.*

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