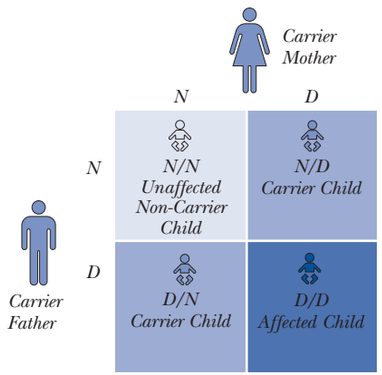
How are genetic diseases passed from one generation to the next?

Genetic disorders are caused by mutations in genes. Every person has two copies of a gene, one inherited from each parent. To have one of the diseases described in this brochure, an individual must have two copies of the abnormal gene. These are called "autosomal recessive" genetic diseases.

What is a carrier?

A carrier is a person who has one normal copy of a gene and one abnormal copy. Having one normal gene is enough to prevent the disease. However, if both parents are carriers of the same abnormal gene, there is a chance that each parent will pass his or her abnormal gene to their baby. If the baby inherits two copies of the abnormal gene, the baby will have the disease. Couples may decide to have carrier testing to find out if they are carriers, and therefore are at risk of having a baby with one of these genetic diseases.



The above diagram shows that if both parents are carriers of the same disease gene (D), each child has a 25% chance of having the disease (D/D) and a 75% chance of not having the disease (N/N, N/D or D/N). The chances are the same for each pregnancy, no matter how many children a couple has, and are also the same for boys and girls.

What if only one partner in a couple is Jewish?

If only one 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 one of these diseases, the non-Jewish partner could then be tested. However, if a pregnancy is already underway, it may be better to test both partners at the same time. Carrier detection rates in the non-Jewish population are lower than those shown in Table 1.

Are there prenatal tests for these diseases?

Yes, if both parents are carriers of the same disease gene, prenatal diagnosis can be performed to determine whether or not the fetus is affected.

Genetic Disease Carrier Screening for Persons of Ashkenazi Jewish Descent

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About Genzyme Genetics

Genzyme Genetics has been a leader in genetic testing and counseling services for more than 20 years.

For more information on our genetic testing and counseling services, please visit our web site at www.genzymegenetics.com.

Additional Resources:

Canavan Foundation
www.canavanfoundation.org

Cystic Fibrosis Foundation
www.cff.org

Dysautonomia Foundation
www.familialdysautonomia.org

Fanconi Anemia Research Fund
www.fanconi.org

FD (Familial Dysautonomia) Hope
www.fdvillage.org

National Gaucher Foundation
www.gaucherdisease.org

National Organization for Rare Disorders
www.rarediseases.org

National Tay-Sachs & Allied Diseases Association
www.ntsad.org

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Learning about your heritage is a beginning.

Around the world distinct ethnic groups are known to have an increased risk for particular genetic diseases. In the Ashkenazi Jewish (Eastern European) population, several inherited diseases are known.

It is considered standard of care for doctors to offer carrier testing for cystic fibrosis, Tay-Sachs disease, Canavan disease and familial dysautonomia to individuals of Ashkenazi Jewish descent.

There are a number of other disorders that occur more often in the Ashkenazi Jewish population for which carrier testing is also available. Interested couples can have a blood test to learn about their chances of having a child affected with one of these conditions.

What are the diseases?

Cystic fibrosis

Cystic fibrosis is a chronic disorder that primarily involves the respiratory, digestive and reproductive systems. Symptoms include pneumonia, diarrhea, poor growth and infertility. Some people are only mildly affected, but individuals with severe disease may die in childhood. The average lifespan today is 32 years, but may improve as scientists search for better treatments. Cystic fibrosis does not affect intelligence.

Tay-Sachs disease

Tay-Sachs disease is caused by an enzyme deficiency that allows a harmful substance to build up in the brain, causing deterioration in both mental and physical abilities. Individuals with Tay-Sachs usually die by age 5. A less common form of Tay-Sachs disease affects adults rather than children. At this time there is no treatment.

Canavan disease

Canavan disease is a disorder which causes brain and nervous system degeneration. Individuals with Canavan disease usually die in early childhood. At this time there is no treatment.

Familial dysautonomia

Familial dysautonomia is a nervous system disorder that causes vomiting, sweating, decreased pain sensitivity, and unstable blood pressure or temperature. Individuals often have normal intelligence, but may have learning disabilities. Symptom management improves quality of life, but only 50% of affected individuals will reach age 30.

Bloom syndrome

Bloom syndrome causes poor growth, poor immune system function and a high rate of cancer. Individuals with Bloom syndrome usually die from cancer before age 30. Bloom syndrome does not affect intelligence.

Fanconi anemia group C

Fanconi anemia group C is a disease that causes anemia, short stature and, oftentimes, abnormalities of the heart, kidneys or limbs. Some individuals have learning disabilities or mental retardation. Patients have a high rate of cancer, especially leukemia.

Gaucher disease

Gaucher disease is caused by an enzyme deficiency. Symptoms are variable and may include fatigue, enlarged liver and spleen, easy bruising and bleeding, bone pain and fractures. The most common form of Gaucher disease is treatable by enzyme replacement therapy. In the most severe form, which occurs much less frequently, the brain and nervous system are also involved.

Glycogen storage disease type 1a

Glycogen storage disease type 1a is a disorder which causes severe low blood sugar, enlarged liver, delayed growth and bleeding. Treatment consists of a strict diet and continuous tube feedings of glucose.

Maple syrup urine disease

Maple syrup urine disease (MSUD) is a disorder which causes certain amino acids to accumulate in the blood. The disease name refers to the characteristic odor of the urine. Without diagnosis and treatment, classic MSUD leads to mental retardation, physical disabilities, seizures and death. Treatment consists of a strict, lifelong special diet to attempt to control the accumulation of amino acids in the blood.

Mucolipidosis type IV

Mucolipidosis type IV affects the brain and nervous system. Symptoms begin in the first year of life, resulting in mental and physical retardation, and impaired vision. At this time there is no treatment.

Niemann-Pick disease type A

Niemann-Pick disease type A causes poor growth, enlarged liver, and mental and physical deterioration. Individuals with Neimann-Pick disease type A usually die by age 4. At this time there is no treatment.

Table 1: Carrier Frequencies and Detection Rates in the Ashkenazi Jewish Population

Disease	Carrier Frequency*	Detection Rate**
Cystic fibrosis	1 in 26	97%
Tay-Sachs disease	1 in 30	98%
Canavan disease	1 in 57	98%
Familial dysautonomia	1 in 30	99.5%
Bloom syndrome	1 in 100	97%
Fanconi anemia group C	1 in 89	99%
Gaucher disease	1 in 15	95%
Glycogen storage disease type 1a	1 in 71	99%
Maple syrup urine disease	1 in 81	99%
Mucolipidosis type IV	1 in 122	96%
Niemann-Pick disease type A	1 in 90	95%

Family History

If someone in your family has one of these genetic diseases or is known to be a carrier, your chances of being a carrier would be higher than the general Ashkenazi Jewish frequencies listed above.

*Carrier frequency is the proportion of individuals in a population who have a single copy of a recessive gene mutation. A carrier frequency of 1 in 26 means that, on average, out of 26 Ashkenazi Jewish individuals, 1 would be a carrier and 25 would not be carriers.

**Detection rate is the percentage of carriers that are identified by the test. A 95% detection rate means that 95% of carriers will have their gene mutation identified by this test and 5% of carriers will have a mutation that cannot be detected by this test.