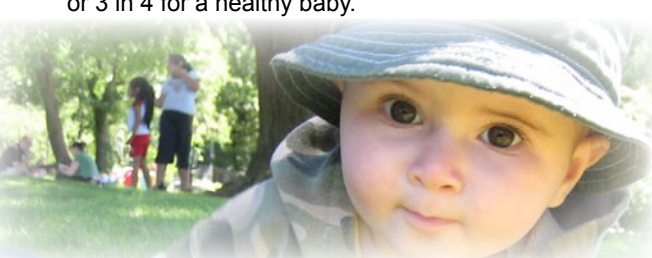


# CARRIER Testing

## Commonly Asked Questions

### Carrier Testing For Genetic Disease

All around the world certain ethnic groups have been found to have an increased risk for particular genetic diseases. In Jewish people whose ancestors are of Eastern European (Ashkenazi) background, several such inherited diseases are known. These are recessive diseases which means couples are at risk for children with the disorder only when each parent is a carrier of the mutant (disease) gene. The diagram below shows the different genetic combinations that can be passed to children when both parents are carriers, each having one normal gene (N) and one disease-causing (C) gene. If both parents are carriers, there is 1 chance in 4 for the child to have the disease, or 3 in 4 for a healthy baby.



Genetic testing is very efficient for identifying carriers. The compelling reason for carrier testing is that prenatal diagnosis is available for carrier couples to determine whether or not their fetus has the disease.

	MOM (N)		MOM (C)	
DAD (N)	Non Carrier N   N	Carrier N   C	Carrier N   C	Affected C   C
DAD (C)	Carrier C   N	Affected C   N	Affected C   N	Affected C   C



### Genetic Diseases For Which Carrier Testing Is Recommended?

**Bloom Syndrome**, is a rare disorder, is characterized by growth retardation and a poor immune system. Affected individuals usually die of cancer before age 30. Approximately 1 in 100 Ashkenazi Jews is found to be a carrier and between 95-97% of carriers are detected with testing.

*Number of Mutations 1*

**Canavan Disease**, like Tay-Sachs, is a degenerative neurological disorder fatal in childhood. Jews of Ashkenazi descent have a 1 in 40 chance of carrying a Canavan gene mutation. Genetic testing can identify approximately 98% of Canavan carriers.

*Number of Mutations 4*

**Cystic Fibrosis**, the most common lethal genetic disease in the Caucasian population, causes progressive lung disease and digestive complications. The symptoms and course of the disease are variable. One in 26 - 29 Ashkenazi Jewish individuals carries a cystic fibrosis mutation and 97% of these carriers can be identified by DNA testing.

*Number of Mutations 31*

**Familial Dysautonomia**, (FD) is a neurological disease characterized by general weakness, feeding difficulties and impairment of those autonomic body functions that control body temperature and blood pressure. Children with FD are frequently hospitalized and have a shortened life span. The carrier rate is 1 in 30 for individuals of Ashkenazi Jewish background. Genetic testing can identify 99% of carriers of FD.

*Number of Mutations 2*

**Fanconi Anemia**, with an Ashkenazi Jewish carrier rate of approximately 1 in 90, results in severe anemia as well as learning disabilities and/or mental retardation, and an increased risk of cancer. DNA analysis can confirm a diagnosis of Fanconi Anemia or identify carriers in the Ashkenazi Jewish population at a detection rate greater than 99%.

*Number of Mutations 1*

**Gaucher Disease**, is a common disorder among Ashkenazi Jews with 1 in 15 found to be carriers. Symptoms which include anemia, bleeding tendency, and bone pain may be severe in some patients, but mild in others. Gaucher disease can be successfully treated with enzyme replacement therapy.

*Number of Mutations 5*

**Glycogen Storage** (Type 1A), or glucose-6-phosphatase deficiency, renders individuals unable to release glucose from the glycogen that is stored mainly in the liver. This causes abnormal amounts of glycogen to build up in various tissues, leading to low blood sugar. Treatment consists of a strict diet and continuous tube feedings of glucose. The carrier rate in the Ashkenazi Jewish population is 1 in 71. Genetic testing can detect 99 percent of the carriers.

*Number of Mutations 1*

**Maple Syrup Urine Disease (MSUD)**, with an Ashkenazi Jewish carrier rate of approximately 1 in 81, presents early in infancy. Mental retardation, physical disabilities, seizures and death can occur if left untreated. Treatment is a lifelong adherence to a special diet to control the buildup of amino acids in the blood.

*Number of Mutations 1*

**Mucopolipidosis Type IV**, is a lysosomal disorder which presents with severe neurological and ophthalmologist abnormalities in the first year of life. The carrier rate in Ashkenazi Jews is 1 in 100, and it is inherited as an autosomal recessive disease. Approximately 96% of Ashkenazi Jewish carriers can be detected by DNA analysis. There is no cure or treatment available for Mucopolipidosis IV at this time.

*Number of Mutations 2*

**Niemann-Pick Disease**, involves progressive mental and physical deterioration with early childhood death. The carrier rate is 1 in 90 in Ashkenazi Jews with a birth incidence in this group of approximately 1 in 32,000. Approximately 95% of carriers can be detected with DNA testing.

*Number of Mutations 4*

**Tay-Sachs Disease**, is characterized by progressive neurological problems leading to death in early childhood. The carrier rate is 1 in 25 for individuals of Ashkenazi Jewish background. Biochemical testing for enzyme activity can detect almost all Tay-Sachs carriers.

*Number of Mutations 5*

EASTERN EUROPEAN JEWISH CARRIER SCREENING				
Disease	Disease Incidence	Jewish Carrier Frequency	Detection Rate	Testing Methods
Bloom Syndrome	1:40,000	1/100	95-97%	DNA
Canavan Disease	1:6,400	1/40	98%	DNA
Cystic Fibrosis	1:2,500-3,000	1/26-29	97%	DNA
Familial Dysautonomia	1:40,000	1/30	99%	DNA
Fanconi Anemia (C)	1:32,000	1/90	99%	DNA
Gaucher Disease	1:900	1/15	95%	DNA
Glycogen Storage Disease	--	1/71	99%	DNA
Maple Syrup Urine Disease	--	1/81	99%	DNA
Mucopolipidosis (IV)	1:40,000	1/100	96%	DNA
Niemann-Pick (A)	1:32,000	1/90	95%	DNA
Tay-Sachs Disease	1:2,500	1/25	94-98%	Enzyme/DNA

- For DNA-based testing, 7 to 10cc should be obtained in lavender top EDTA tubes.
- For enzyme testing, specimen requirements may vary call laboratory for instructions.
- Tay-Sachs carrier testing by enzyme on pregnant women and those on oral contraceptives MUST be done via leukocytes.
- Unless familial mutation is known, DNA testing in the non-Jewish population will have a lower detection rate than in the Jewish population.

For information on any other risk assessment or diagnostic testing contact:



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## Additional Information About Carrier Testing

It is important to understand if any of these hereditary diseases has occurred in your family, your chances to be a carrier are probably higher than the population risk.

*If only one parent in a couple is Jewish, it is usual to test that person first. If he or she is found to be a carrier, the non-Jewish partner could then be tested. If a pregnancy is in progress, it is best to test both partners at the same time.*

If both parents are shown to be carriers of the same genetic disease, then prenatal testing by chorionic villi sampling or amniocentesis can be performed to determine whether or not the fetus is affected. Your health care provider will be able to refer you to the proper center for follow-up.

## For Additional Assistance

This brochure contains general information regarding carrier testing for Ashkenazi Jewish Genetic diseases. However, you may wish to obtain professional genetic counseling prior to having the test.

The individual capabilities and potential of children with Ashkenazi Jewish Genetic diseases are considerations which you may wish to discuss with your counselor or with other healthcare providers. Further information and support are available through groups



### ASHKENAZI JEWISH GENETIC TESTING INFORMED CONSENT

I have read and understand the information in this pamphlet regarding screening for the Ashkenazi Jewish Genetic Testing.

- Yes, I want to have the Ashkenazi Jewish Genetic Testing.
- No, I do not want to have the Ashkenazi Jewish Genetic Testing.

Patient Name: \_\_\_\_\_

Patient Signature: \_\_\_\_\_

Date: \_\_\_\_\_

**IMPORTANT:** Retain Copy in Patient File



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The information included in this pamphlet is not intended as a substitute for personal medical advice. Specific situations always require a personal consultation with your healthcare provider.

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# ASHKENAZI JEWISH PANEL TESTING

## Information for Patients

### For Ashkenazi Jewish Genetic Diseases



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