

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 all 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.

	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 C	Affected C C	Affected C C

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.

EASTERN EUROPEAN JEWISH CARRIER SCREENING				
Disease	Disease Incidence	Jewish Carrier Frequency	Detection Rate	Testing Methods
Tay-Sachs Disease	1:2,500	1/25	94-98%	Enzyme/ DNA
Cystic Fibrosis	1:2,500-3,000	1/26-29	97%	DNA
Canavan Disease	1:6,400	1/40	98%	DNA
Niemann-Pick (A)	1:32,000	1/90	95%	DNA
Fanconi Anemia (C)	1:32,000	1/90	99%	DNA
Bloom Syndrome	1:40,000	1/100	95-97%	DNA
Gaucher Disease	1:900	1/15	95%	DNA
Familial Dysautonomia	1:40,000	1/100	99%	DNA
Mucopolipidosis (IV)	1:40,000	1/100	96%	DNA
<ul style="list-style-type: none"> 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. 				
*Numbers may not be exact, use for counseling purposes only.				

Genetic Diseases For Which Carrier Testing Is Recommended?

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](#)

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 29 Ashkenazi Jewish individuals carries a cystic fibrosis mutation and 97% of these carriers can be identified by DNA testing. [Number of Mutations 32](#)

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 27 for individuals of Ashkenazi Jewish background. Genetic testing can identify 99% of carriers of FD. [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](#)

Bloom Syndrome, another 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](#)

Fanconi Anemia, with a Jewish carrier rate of 1 in 100, results in severe anemia as well as learning disabilities or mental retardation, and an increased risk of cancer. [Number of Mutations](#)

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](#)

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](#)

Some 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.

	Mom (N)	Mom (C)
Dad (N)	Non carrier	Carrier
Dad (C)	Carrier	Affected

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.

Can other abnormalities be identified?

Yes. The risk of two other disorders can be estimated. One is Trisomy 18, a rare and usually fatal disorder caused by the presence of an extra number 18 chromosome in the cells of the developing baby. The risk of Trisomy 18 can be estimated using AFP, uE3 and total βhCG, and is reported only when the risk is high. The second is called Smith-Lemli-Opitz syndrome, a genetic disorder caused by an error in the synthesis of cholesterol. Smith-Lemli-Opitz syndrome is associated with many problems in the developing baby, most important are mental retardation and poor growth. The risk of Smith-Lemli-Opitz syndrome can also be estimated using AFP, uE3 and total βhCG, is reported only when risk is high.

Why do you take age into account?

Any woman can have a baby with Down syndrome but the chance of this happening increases as a woman gets older. We use age as one of the factors when working out your risk of pregnancy with Down syndrome. It means that an older woman is more likely to have a result in the higher risk groups (*screen positive*) and be offered a diagnostic test.



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Are there other tests available if I am already pregnant?

Prenatal testing is available for these conditions and sometimes requires testing of the parents as well. Amniocentesis and CVS are the most common types of prenatal diagnostic test. Your healthcare 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, risk assessment for Ashkenazi Jewish Genetic diseases. However, you may wish to obtain professional counseling prior to having the test.

The individual capabilities and potential of children with Ashkenazi Jewish Genetic disease are considered which you may wish to discuss with your counselor or health care providers. For further information or support use our list below or our website www.lenetix.com

Further information and support are available through groups such as your local Down Syndrome Society and Spina Bifida Association as listed below:

March of Dimes www.marchofdimes.com
National Down Syndrome Society www.ndss.org
National Association for Down Syndrome www.nads.org
Smith-Lemli-Opitz Syndrome www.smithlemliopitz.org
Spina Bifida Association www.sbaa.org
Trisomy 18 www.trisomy.org



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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The CARRIER Testing

Information for
Patients

For Ashkenazi Jewish
Genetic Diseases

FIRST STAGE
Final Results Reported

SECOND STAGE
Final Results Reported



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