What couples should know about how genetics can affect their baby

If you have any Jewish ancestry or are a couple of mixed heritage, you may be a carrier of a very serious genetic disease.

Jewish Genetic Disease Consortium

JGDC
It is not just Tay-Sachs. There are now carrier tests for many more diseases.

Couples with any Jewish ancestry, even those with mixed heritage, should have carrier screening.

Prior to testing consult with a genetic counselor to review the best options for screening.

The best time to be screened is before pregnancy – carrier couples have multiple reproductive options to build a healthy family.

Explore your options for up-to-date testing before any pregnancy. Screening panels improve over time and diseases are often added.

If you have had any genetic testing, it is important to review your results with a genetic counselor.
**Jewish genetic heritage**

There are different genetic concerns for people of Ashkenazi Jewish background (Eastern /Central European), and people of Sephardic (Mediterranean) or Mizrahi (Middle Eastern/ North African) backgrounds. Some testing panels are targeted for individuals of Jewish ancestry and some include a broader range of diseases that are common among all ethnic backgrounds (pan-ethnic).

While many diseases are more commonly found in certain ethnic groups, there is still a chance that individuals from other backgrounds might be carriers, as well.

**What it means to be a carrier**

A carrier is an individual who does not develop the disease, but can pass the gene with a mutation to his or her children.

In the case of most Jewish genetic diseases, which are termed “autosomal recessive,” if both parents are carriers of the same disease they can have an affected child.

Some individuals may discover during the course of carrier screening that they are at-risk for having symptoms of one of the less severe or late-onset diseases.

**How most Jewish genetic diseases are inherited**

If two carriers of an autosomal recessive disease have children, with each pregnancy there is a 1 in 4 chance that their child will be affected with that disease, a 2 in 4 chance that the child will be a carrier, and a 1 in 4 chance that the child will be neither affected nor a carrier.

**How is testing done?**

Carrier screening for all of the Jewish genetic diseases by DNA analysis can be done using blood or saliva. For Tay-Sachs disease screening, enzyme analysis using blood is optional, as new research has established that comprehensive DNA sequencing of the Tay-Sachs gene is just as accurate as enzyme analysis.

All genetic tests should be ordered and the results read by a knowledgeable physician or genetic counselor trained to interpret such results. Pre-test education and post-test counseling are recommended with any form of genetic testing. Please be aware that not all genetic testing companies provide these services. Individuals should request a copy of their lab report and keep it in a safe location. Diseases are often added to testing panels, so be sure testing is up-to-date before any pregnancy.

There are two approaches to carrier screening: ethnicity based and expanded carrier screening.

In ethnicity based screening, you are tested for disorders based on your ethnicity or family history. Over time, we have learned
that ethnicity based screening has many limitations. Today, with expanded carrier screening, many disorders can be screened for without focusing on either race or ethnicity. Most national genetics and reproductive medicine societies support the use of these expanded carrier panels for all individuals. As a result, highly accurate carrier screening is now available for Jews of all backgrounds – Ashkenazi, Sephardic and Mizrahi – as well as couples of mixed heritage.

Options for carrier couples

There are multiple reproductive options that enable carrier couples to build a healthy family. Each couple needs to make their own informed decision, based on the disease or diseases for which they are carriers, and other personal considerations. Genetic counselors can explain options in depth.

During pregnancy

A carrier couple may choose to become pregnant and test the fetus early in the pregnancy, either by chorionic villus sampling (CVS) or amniocentesis. In the case of an affected fetus, the couple can decide how to proceed with the pregnancy. In order to test the fetus, the parents’ carrier statuses must be known.

Before pregnancy

A carrier couple may choose…

- To undergo In-vitro fertilization (IVF) with pre-implantation genetic diagnosis (PGT). In this procedure, the egg is fertilized outside the body and embryos are tested at a very early stage for the disease(s) carried by the parents. Only unaffected embryos are implanted in the womb.
- To use egg or sperm from a screened non-carrier donor
- To adopt
- Not to have children
- Not to continue the relationship

Where can carrier screening be done?

Currently there are several ways to obtain screening: a hospital or clinic-based medical genetics program, online genetics programs that offer education, counseling and screening, or your doctor’s office. For assistance in selecting a genetic counselor, visit nsgc.org. For screening resources and a description of Jewish genetic diseases, visit JewishGeneticDiseases.org/resources.

Who should be screened?

Everyone considering having children should be screened. While many diseases are more commonly found in certain ethnic groups, there is still a chance that an individual from a different background might be a carrier, as well.

At least one member of every couple should be screened. If his or her results are positive, the other member of the couple should then be screened. Some couples decide to be screened simultaneously, as to avoid the waiting between results, especially if they are already pregnant.

There may be some diseases on the testing panel for which only females are screened. Therefore, if one partner is tested first, it is recommended that the female be first.

When is the best time to be screened?

Screening before pregnancy is optimal. This allows a carrier couple the greatest number of reproductive options. It is important to note that if the couple discovers their carrier status once they are already pregnant their only options, if the fetus is affected, are to continue or to terminate the pregnancy. Diseases are often added to testing panels, so be sure testing is up-to-date before any pregnancy.

Is testing covered by insurance?

Although every insurance plan is different, many cover testing for Jewish genetic diseases. Contact your insurance company for coverage information. A medical genetics program can be helpful in determining coverage. If screening is not covered, the JGDC may be able to provide referrals to low cost screening options or provide a letter of necessity for your insurance company.
The **Jewish Genetic Disease Consortium** (JGDC) increases awareness about Jewish genetic diseases and encourages timely and appropriate carrier screening for all persons who have any Jewish ancestry, as well as couples of mixed heritage. The JGDC is an alliance of non-profit organizations sharing the common goal of combating Jewish genetic diseases. While each JGDC member organization has its own individual mission, the JGDC unites these organizations so that we may jointly strengthen public education and awareness and appropriate carrier screening.

Through its Medical Grand Rounds Program, Rabbi Education Program and Jewish Community Program, the JGDC educates physicians, rabbis, Jews of all backgrounds and couples of mixed heritage about Jewish genetic diseases. The goal is to decrease the incidence of Jewish genetic diseases and assure healthy Jewish families by increasing preconception carrier screening rates and promoting the understanding of reproductive options available to carrier couples.

**Member Organizations**
- Bloom's Syndrome Foundation
- Canavan Foundation
- FD Now
- Genetic Disease Foundation
- Mathew Forbes Romer Foundation
- National Tay-Sachs & Allied Diseases Association
- Neuromuscular Disease Foundation
- Usher IF Collaborative

**Supporting Partners**

For more information visit [JewishGeneticDiseases.org](http://JewishGeneticDiseases.org)

For additional copies of this brochure or other JGDC materials, e-mail: info@JewishGeneticDiseases.org or call 855.642.6900.

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