



What Jewish and Interfaith Couples Need to Know About Jewish Genetic Diseases ►



If you have any
Jewish heritage,
you may be a
carrier of a serious
genetic disease.

Jewish Genetic Disease
Consortium

JGDC

Did You Know?

It is not just Tay-Sachs. There are now carrier tests for many more diseases of concern to people of Jewish heritage.

There are specific genetic concerns based on family origin: Ashkenazi, Sephardic, or Mizrahi.

The best time to be screened is before pregnancy.

There are multiple options for a carrier couple who want to build a healthy family.

Interfaith (Jewish and non-Jewish) couples should be screened prior to pregnancy for the Jewish genetic diseases.

Jewish genetic heritage does not depend upon religious practice or observance.

You may be a carrier for a Jewish genetic disease even though you do not consider yourself Jewish.



Screening Resources and
Descriptions of all Jewish Genetic Diseases

JewishGeneticDiseases.org



Search for a Genetic Counselor
nsgc.org

Talk to Your Doctor About Jewish Genetic Disease Screening

If you or your partner have **any** Jewish heritage and plan to become pregnant, it is important to be screened for the Jewish genetic diseases.

If you are already pregnant, it is necessary for both partners to be screened at the same time, as soon as possible.

Tell your doctor about

Your plans to become pregnant.

All family heritage, whether Ashkenazi (German, French or Eastern European), Sephardic (Mediterranean) or Mizrahi (Persian/Iranian or Middle Eastern).

Any family history of genetic diseases.

If you have been screened before, and your test results are available, bring them to your appointment.

If you do not have a doctor, or if your doctor does not offer genetic screening and counseling, you can contact a hospital based medical genetics program or an online genetics program that offers education, counseling and screening. Screening resources are listed at JewishGeneticDiseases.org. You can also search for a genetic counselor in your area at NSGC.org.

Whether you are currently single, dating, engaged or married, you may be thinking about having children or adding to your family.

If you or your partner have **any** Jewish heritage, you need to consider genetic screening.

If both of you are carriers of the same disease, it is best to have this information before pregnancy. A genetic counselor can discuss your options for building a healthy family.





Jewish Genetic Heritage

There are different genetic concerns for people of Ashkenazi Jewish background (German, French or Eastern European), and people of Sephardic (Mediterranean) or Mizrahi (Persian/Iranian or Middle Eastern) background.

Ashkenazi

It is estimated that nearly 1 in 3 Ashkenazi Jews in the United States is a carrier of at least one of 19 Jewish genetic diseases. Therefore, people of Ashkenazi Jewish Heritage should consider preconception carrier screening.

Sephardic and Mizrahi

There is no single preconception carrier-screening panel for people of Sephardic or Mizrahi background. Carrier screening is dependent upon country of origin. People of Sephardic or Mizrahi background should seek genetic counseling.

What it means to be a carrier

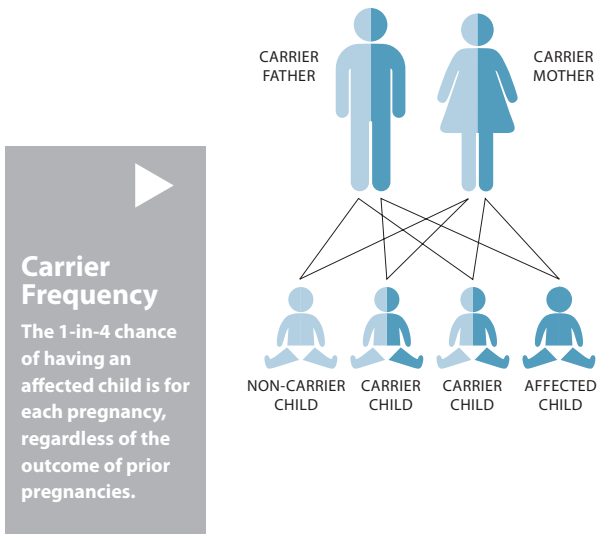
A carrier is an individual who does not develop the disease, but can pass the gene with a mutation to his or her children.

In the case of the Jewish genetic diseases, which are termed “autosomal recessive,” one carrier parent can pass on the carrier mutation, but both parents must be carriers of the same disease to have an affected child.

Some individuals may discover during the course of carrier screening that they are affected by one of the less severe or late-onset diseases.

How Genetic Diseases are Inherited

If two carriers of a gene with a mutation for the same disease have children, there is a 1-in-4 (25%) chance with each pregnancy that their child will be affected with that disease, a 2-in-4 (50%) chance that the child will be a carrier, and a 1-in-4 (25%) chance that the child will be neither affected nor a carrier.



How is the test done?

Carrier testing for all the Jewish genetic diseases by DNA analysis can be done using blood or saliva. For Tay-Sachs disease screening, enzyme analysis using blood, in addition to DNA analysis, is required. For Alpha and Beta Thalassemia, a complete blood count (CBC) is done first.

The tests should be ordered, and the results be read by a knowledgeable doctor or genetic counselor trained to interpret genetic tests. Individuals should request a copy of their test results and keep it in a safe location. As new diseases are added to the panel, screening for additional diseases may be necessary before subsequent pregnancies.

Please be aware that there are many laboratories offering Ashkenazi Jewish genetic disease screening with panels varying from 19 to 38 diseases. You should discuss these options with your physician or genetic counselor.

There is no screening panel for people of Sephardic or Mizrahi heritage. Screening is dependent upon country of origin. Those with Sephardic or Mizrahi heritage should seek genetic counseling.

Individuals may also opt to do expanded carrier screening to include disorders not necessarily more common in the Ashkenazi Jewish ancestry (Pan Ethnic Panels). Regardless of the panel chosen, Tay-Sachs enzyme analysis, using blood, must be done, in addition to DNA analysis, for complete screening.

Where can screening be done?

Currently there are several ways to obtain screening – your doctor's office (usually an OB/GYN), Medical Genetics Program, or online genetics program that offers education, counseling and screening. For assistance in selecting a genetic counselor, visit nsgc.org. For screening resources and a description of all Jewish genetic diseases, visit JewishGeneticDiseases.org.

Who should be screened?

If the woman is already pregnant, both partners must be screened simultaneously. If they are both carriers of the same disease, earlier fetal testing can be planned and there is more time for counseling and decision-making.

In a couple where both partners are Jewish, the usual practice is to screen the woman first, if she is not pregnant, generally at her OB/GYN's office. If she is found to be a carrier of a specific disease(s), her partner must be screened for that disease(s) if they want to know if they are at risk for having a child with that disease(s).

If only one partner is of a Jewish background, that partner should be screened first. If he or she is found to be a carrier of a specific disease(s), the other partner must be screened for that disease(s), if they want to know their risk for having a child with that disease(s). Genetic Counseling is recommended.

When is the best time to be screened?

Screening before pregnancy is optimal. This allows a carrier couple the greatest number of 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.



Is the test 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.

Options for Carrier Couples

There are multiple options which 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 counseling can explain options in depth.

A carrier couple may choose to become pregnant and test the fetus early in the pregnancy. In the case of an affected fetus (a 1-in-4 chance for each pregnancy), the couple can decide whether to continue or terminate the pregnancy. In order to test the fetus, the parents' carrier status must be known.

A carrier couple may choose in-vitro fertilization (IVF) with pre-implantation genetic diagnosis (PGD). 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.

A carrier couple may choose to use egg or sperm from a screened non-carrier donor.

A carrier couple may choose to adopt.

A carrier couple may choose not to have children.

A carrier couple may choose not to marry. In this case the couple needs to be screened before engagement or marriage.

IMPORTANT: THIS BROCHURE DOES NOT PROVIDE MEDICAL ADVICE OR GENETIC COUNSELING

The information in this brochure is for informational purposes only. It is intended to give the reader a broad overview of the genetic disease screening process and the Jewish genetic diseases for which screening currently is available.

This brochure is not intended to be a substitute for professional medical advice, diagnosis, treatment, or genetic counseling. Your physician or other qualified health care provider can assist you with any questions you may have regarding your personal situation and the Jewish genetic disease screening process.

The Jewish Genetic Disease Consortium assumes no liability or responsibility for any opinions, advice, procedures, or results provided by any independent individual or entity with respect to Jewish genetic disease screening.

Genetic science is a rapidly developing field. The information in this brochure is subject to update and screening options may change. Please visit JewishGeneticDiseases.org for updates.

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 heritage, as well as couples of interfaith marriage. The JGDC is an alliance of non-for-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 for all persons of Jewish heritage, whether Ashkenazi, Mizrahi, Sephardic, as well as couples of interfaith marriage.

Through its Medical Grand Rounds Program, Clergy Education Program and Jewish Community Program, the JGDC educates physicians, rabbis, Jews of all backgrounds and interfaith couples 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.

Jewish Genetic Disease
Consortium

JGDC

Member Organizations

Bloom's Syndrome Foundation
Canavan Foundation
FD Now
Genetic Disease Foundation
Mathew Forbes Romer Foundation
Mucopolidosis Type 4 Foundation
National Gaucher Foundation
National Tay-Sachs & Allied Diseases Association
Sephardic Health Organization for Referral and Education

Supporters

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For more information visit

JewishGeneticDiseases.org

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