



Jewish Genetics & Family Planning

**Getting Screened:
Recessive Genetic Disorders**



Jewish Genetic Disorders & Family Planning: The Basics

- If you OR your partner is of **Jewish descent** (at least one Jewish parent, grandparent, or great-grandparent), you may be at a higher risk for having a child with a genetic disorder.
- At least 100 genetic disorders are more common among **Ashkenazi** (Central or Eastern European) Jews. In fact, 1 in 4 Ashkenazi Jews carries one or more of these conditions.
- While not as common as in the Ashkenazi Jewish population, there are genetic disorders more common in individuals of **Sephardic** (Mediterranean or Middle Eastern) or **Mizrahi** (Middle Eastern) Jewish descent that warrant genetic screening as well.
- Carrier screening plays an important role when you're **starting your family**. The Norton and Elaine Sarnoff Center for Jewish Genetics can help you understand your risk and options.

Screenings: Who, What, When & Where

WHO SHOULD BE SCREENED?

- » All individuals born of Jewish descent regardless of the type of Jew you identify as
- » All individuals who converted to Judaism if their partner is of Jewish descent by birth
- » Non-Jewish partners of Jewish individuals

WHAT DOES SCREENING INVOLVE?

Most carrier screening tests can be accomplished with a saliva sample, but certain scenarios require blood instead of saliva. Laboratories use different methods for screening. The most advanced form includes full gene sequencing as this provides the highest detection rates. Expanded pan-ethnic carrier screening tests include most of the conditions more common in the Jewish population. Your healthcare provider determines which tests to order, so it's important to let your healthcare provider know your heritage.

WHEN SHOULD I GET SCREENED?

Screening is always recommended prior to pregnancy. Knowing your carrier status beforehand gives you the most options when planning your family.

If either you or your partner is already pregnant, you should seek screening right away. If you're both carriers for the same condition, early fetal testing can give you more time for counseling and decision making.





WHERE AND HOW DO I GET SCREENED?

Talk to your primary care provider about genetic screening. Your healthcare provider and/or OB/GYN may order the test for you.

The Norton and Elaine Sarnoff Center for Jewish Genetics offers affordable and accessible screening for all eligible participants. The Sarnoff Center will also help you locate a local resource if you live outside of Illinois.

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Absolutely get tested—so that you can know. Whatever choice you want to make, at least know the cards you’re dealt. I believe that knowledge is power and without knowledge we could not have made the choices that were best for us.”

—A Canavan carrier couple and parents of a healthy girl

Results: What If I’m a Carrier?

CAN THE DISORDER PASS ON TO MY CHILD?

The common Jewish conditions are “autosomal recessive” meaning both you and your partner must carry the same mutated gene to have an affected child.

If you and your partner carry the same disorder, with each pregnancy, your child will have a...



chance of being
a **carrier**



chance of being
affected



chance of being
**neither
affected/carrier**

Other disorders included on the test are X-linked which means the harmful gene is located on the X-chromosome. X-linked disorders typically impact children assigned male at birth and rarely impact assigned females at birth. If the biological mother carries an X-linked disorder, the child will have a...

Male:



chance
of being
affected



chance of
**not being
affected**

Female:



chance of
being a
carrier



chance of
**not being
a carrier**

CAN I STILL HAVE HEALTHY CHILDREN?

Yes! Carrier couples build healthy families every day. Based on your carrier status and when you are tested, you'll have many options. Some options may include: In vitro fertilization (IVF) with preimplantation genetic testing (PGT), prenatal testing, sperm/egg donation, or adoption.

DO I NEED TREATMENT?

In rare instances, carrier screening may reveal a person is at risk of developing a less severe or late-onset form of one of the genetic disorders. In these cases, your healthcare provider can provide the appropriate management options.

Affordable, Accessible Genetic Screening in Illinois

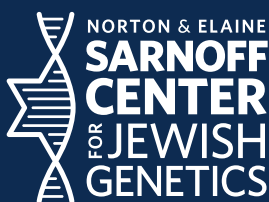
The Norton and Elaine Sarnoff Center for Jewish Genetics provide an accessible screening option for Jewish individuals, couples and interfaith couples. The Sarnoff Center's program allows most participants to get screened right from home... **in 6 simple steps!**

- 01.** Visit www.jewishgenetics.org and register for the program
- 02.** Complete an online education course to prepare for screening
- 03.** Our genetic counseling partner will contact you after completion of the course
- 04.** After receiving the saliva kit in the mail, return a saliva sample using the instructions in the kit
- 05.** Our genetic counseling partner will call you with results 2–3 weeks after the kit is sent in for testing
- 06.** Follow-up appointments will be scheduled as needed with our genetic counseling partner

There is a program fee of \$99, which includes pre-screening education and support through the Sarnoff Center and our healthcare provider. In most cases, the healthcare provider will bill your insurance for the cost of the test. You may request financial assistance from the Sarnoff Center for the program fee and/or genetic test. One of the goals of the Norton & Elaine Sarnoff Center for Jewish Genetics is to ensure that no Jewish individuals, couples, or interfaith couples go without carrier screening due to concerns about cost or ability to pay.

You Could Be at Risk Even If...

- » **Either you or your partner is only “a little bit Jewish”**
Just one Jewish parent, grandparent, or great-grandparent is enough to increase the chance a person is a carrier for a genetic condition common within Jewish community.
- » **Your reproductive partner is not Jewish**
None of the disorders on the test are exclusive to the Jewish population, so individuals who are not Jewish by birth or who have a reproductive partner is not Jewish should still undergo carrier screening; this is the standard of care.
- » **You have no family health history**
Most individuals who are carriers do not have a family history of the disorder. Recessive disorders can only appear when both parents carry the same disease, but the mutation may pass down for generations without producing an affected child. The same can happen with X-linked conditions.
- » **You're starting a family using IVF, donor eggs/sperm, or other methods**
If you discover you're a carrier, you can test donor samples to minimize your chance of having an affected child. In fact, most egg and sperm banks routinely do carrier screening on their donors.



The Norton & Elaine Sarnoff Center for Jewish Genetics provides educational resources related to hereditary cancers and genetic disorders more common among individuals of Jewish descent. We work closely with community members, clergy, healthcare professionals, and partner organizations to raise awareness of these issues and available support options, including our own low-to-no cost genetic counseling and genetic screening programs.

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This information is to be used for educational and informational purposes only. This information does not represent advice regarding medical diagnosis or treatment, referrals to healthcare providers, endorsements of healthcare products or any other recommendations. This information should not be relied upon as a substitute for consultation with your healthcare providers.



Jewish United Fund

TOGETHER for GOOD

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