Genetic Carrier Screening

Planning a family? Screening can help.

Carrier screening is recommended for all couples who are planning a family, as anyone can be a carrier for a genetic condition. Some conditions, like Tay-Sachs disease, are more common in Jewish families. Even without a family history, there is still a risk. Screening helps you understand your risks, explore family-building options, and plan ahead with confidence.

What is carrier screening?

Carrier screening is a simple genetic test that analyzes your DNA to determine whether you are a carrier for certain genetic conditions.

- A carrier is typically a healthy individual who shows no signs or symptoms of the condition.
- Even with no personal or family history of the condition, you can still be a carrier.
- Depending on your and your spouse's results, there may be an increased chance your child will inherit a genetic condition.

When and how should we do it?

- Carrier screening is ideally performed before pregnancy to provide the most options.
- Both partners can be tested simultaneously to make the process faster.
- Testing is simple and convenient – it is performed with a blood sample or cheek swab.

How do we get screened? What kind of test should we ask for?

- Ask your OB/GYN doctor for carrier screening. Insurance often covers the cost, particularly for those planning a pregnancy.
- Historically, screening for Jewish couples was limited to a few specific conditions, like Tay-Sachs and Gaucher disease.
- Experts now recommend asking for an **expanded carrier screening panel** for a more comprehensive analysis.
- Note: Direct-to-consumer genetic tests like 23andMe and AncestryDNA are not substitutes for this medical-grade screening.

What do our results mean?

- A **positive** carrier result is common, as most individuals carry at least one genetic variant.
- This result alone does not mean there is a high risk for your children.
- The risk is typically elevated only if **both** partners are carriers for the same condition, though there are exceptions.
- Your health-care provider will use the results to determine whether you are a low-risk (little chance of having a child with the condition) or high-risk (an increased-chance) couple.

What happens if we are a 'high-risk' couple?

A high-risk result indicates an increased chance (typically 25% or 50%) that your child could inherit a genetic condition. A genetic counselor can help you understand these results and explore your options:

Conceive Naturally – With the option to test the pregnancy early

IVF with PGT-M – Eggs are retrieved and fertilized to create embryos and test them for the genetic condition

Conceive with Donor Eggs or Sperm

Adoption

Carrier screening reduces the risks but does not test for everything

If you are concerned about a specific condition, ask your doctor for a referral to a genetic counselor, who can determine whether additional testing is needed. Screening tests do not cover BRCA1/2 and Lynch, which are more common in Jewish populations.

Additional Resources

May you be blessed in building a healthy and joyful family; l'dor vador

JewishGenetics - Watch a video about the basics of screening

JScreen - Order screening (includes common Jewish conditions; doctors may offer larger panels)

NSGC - Find a certified genetic counselor to discuss your results