let's talk about genetic testing

getting to know the basics

Genetic testing can be a helpful tool for many people as they look to build their families. In the context of fertility, genetic testing involves analyzing DNA to identify genetic conditions that may impact fertility or pregnancy, help in making informed decisions about family planning, and reduce the risk of passing on genetic conditions to children.

win.
family-building &
family-wellbeing

carrier screening

When you are preparing to become pregnant, you want everything to go smoothly. While most babies are born healthy, with every pregnancy there is a small chance of having a baby with a genetic disorder. **With genetic carrier screening, you can learn your risk for passing an inherited genetic disorder to your child.**

Genetic carrier screening is a type of genetic test that analyzes your DNA to provide specific information about your child's risk for certain genetic disorders. Most carrier screening is for recessive conditions, and may include X-linked conditions. For a person to be affected with a recessive condition, typically both copies of the gene associated with the condition must have a mutation — one inherited from the egg and one inherited from the sperm.

If a person has only one mutation for a recessive condition, they are known as a carrier. **Carriers of recessive conditions often do not know that they are carriers.** X-linked conditions are associated with mutations in genes on the X chromosome and typically affect individuals assigned male sex at birth.

This test is usually ordered during the beginning of your IVF journey and completed as part of the pre-cycle lab work before creating the embryos.

what it does

Carrier screening determines if an individual carries a gene mutation for a specific recessive genetic disorder, such as Cystic fibrosis, Sickle cell disease, and Tay-Sachs disease.

what you should know

- Carrier screening can be performed when you are planning to get pregnant or during pregnancy.
- Being identified as a carrier is normal, and is typically inherited from a biological parent.
- Involves a blood test or saliva sample from one or both prospective parents.
- Test results can take ~2-3 weeks.
- There are several family-building and reproductive testing options available to patients or couples whose offspring are identified to be at increased risk.

PGT 101

Preimplantation genetic testing, or PGT, is a process used to identify genetic differences in embryos created with in vitro fertilization to significantly reduce the chances of transferring an embryo with a specific genetic condition or certain types of chromosome abnormalities.

There are different types of PGT that can be conducted on embryos, including PGT-A, PGT-SR, and PGT-M. Depending on your situation, your provider will be able to recommend which testing is appropriate to pursue.

type	what it does	what you should know
PGT-A preimplantation genetic testing for aneuploidy	Screens embryos for aneuploidy (extra/missing chromosomes), which typically arises spontaneously.	PGT-A can be performed for any IVF cycle, but may be recommended for many patients who are over 35. There may also be benefits for patients or couples with: • Recurrent implantation failure • Male factor infertility • Recurrent pregnancy loss Test results can take ~7-14 days.
PGT-M preimplantation genetic testing for monogenic disorders	Screens embryos for genetic abnormalities caused by a single gene mutation, with the ability to identify more than 1,000 genetic illnesses.	PGT-M is only performed when the patient, their partner, and/or their donor have abnormal genetic test results that put the embryos at increased risk for a genetic condition. A unique test is used for each case, which requires preparation and family member participation. Test results can take ~14 days.
PGT-SR preimplantation genetic testing for structural rearrangements	Screens embryos for chromosomal imbalances such missing, extra, or rearranged segments of chromosomes.	PGT-SR is only performed when the patient, their partner, and/or their donor have a structural chromosomal rearrangement, such as a translocation or inversion. Test results can take ~7-14 days.

fertility 101

the road ahead

This timeline can give you a sense of what the PGT process during fertility treatment typically entails. Remember, no two fertility journeys are the same!

cycle preparation

- Start off with bloodwork and a vaginal ultrasound
- Order medications and learn how to administer shots

ovarian stimulation

- This part of the process approximately takes 10-14 days
- Daily injections and regular monitoring to stimulate the maturation of a batch of eggs

egg retrieval

- Prompted by trigger shot once follicles are large enough
- About 10-15 minute procedure under light anesthesia

creating embryos

- Fertilized embryos are cultured for 5-7 days before being biopsied and sent for PGT
- The biopsied cells are sent for PGT and the embryos are cryopreserved
- Genetic testing results typically take around 2 weeks.
- If embryos are genetically normal, you can proceed with embryo transfer. If not, your provider will likely talk to you about completing another ovarian stimulation cycle.

embryo transfer

- Typically occurs between days 19 and 21 of the menstrual cycle
- A 5 minute procedure using a catheter to place the embryo in the uterus
- 11-13 days later a blood test will determine if you are pregnant

need on-demand support?

From late night questions and medication assistance to emotional support, our Nurse Care Advocates are available 24/7 through the WINFamily App to help you navigate your fertility journey.

Clinical guidance is a tap away.
Scan to download the WINFamily App and create your account using your company name.