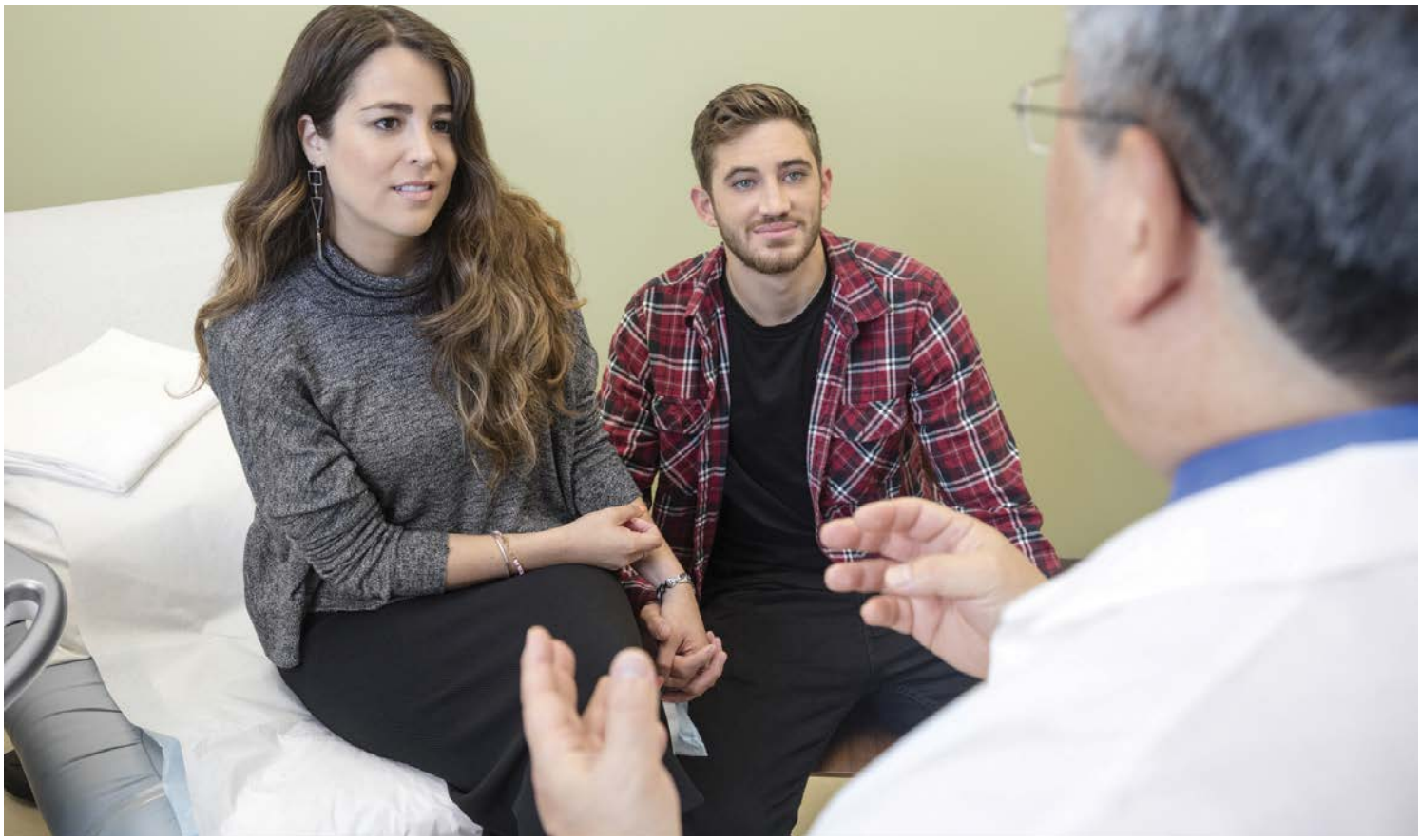
Stanford MEDICINE

Fertility and Reproductive Health

Preconception Genetic Testing Options



Carrier Screening

Carrier screening is genetic testing, ideally performed before a pregnancy occurs. The purpose of testing is to identify if there is an increased risk of having a child with specific genetic diseases.

It is very common to be identified to be a carrier through these expanded carrier screening panels (more than 100 conditions are tested for). Approximately 50 to 60 percent of people are identified as a carrier after testing. Carrier screening panels typically include both recessive and X-linked conditions.

Recessive conditions: Being a carrier (having one genetic change or "mutation") of a recessive condition rarely means anything for that particular person's health. It is only if both the egg and sperm provider are carriers for the same condition that this would cause an increased risk (25 percent) of having a child with the condition.

X-linked conditions: Being a carrier of an X-linked condition means that the egg provider is at increased risk (50 percent) of having a son with that condition. Screening for the sperm provider is not necessary in these cases.

A negative result does not mean there is no risk of having a child with the genetic conditions being tested for; it simply indicates a reduced risk of having a child with the specific conditions covered on the test.

Our clinic will only contact you regarding your carrier screening results in the following cases:

- Both individuals are identified to be carriers for the same recessive condition.
- The egg provider is identified to be a carrier of an X-linked condition.
- The result indicates a risk to your own health management.

We strongly encourage you to sign up for the patient portal through the lab, as you will have access to the full report and free genetic counseling services when results are available.

Preimplantation Genetic Testing

Aneuploidy (PGT-A, previously PGS)

Preimplantation genetic testing is testing for embryos, performed before that embryo is transferred back to a uterus (before pregnancy). Testing for aneuploidy identifies whether embryos are at higher risk of resulting in no pregnancy or miscarriage, by screening for missing or extra chromosomes. Chromosomes are packages of genetic information. Typically, humans have 23 from the egg and 23 from the sperm (total of 46). Any missing or extra chromosomes dramatically increase the risk of a failed transfer or miscarriage.

Age of the egg provider is the greatest factor in the presence of chromosomal abnormalities.

Roughly 25 to 30 percent of eggs retrieved would be expected to develop to blastocyst for biopsy (day5/day6/day7). For each chromosomally normal embryo, there is an approximately 50 percent chance of making it to live birth. Embryos reported to be mosaic (having two or more populations of cells with different genotypes in one individual who has developed from a single fertilized egg) have a 30 to 40 percent chance of resulting in pregnancy. Reportedly abnormal embryos are expected to have a <5 percent chance of resulting in pregnancy. There are currently NO KNOWN HEALTH ASSOCIATIONS between the PGT-A result and outcome of the live birth.

These figures should be used solely for estimation and are in no way an exact prediction of cycle potential.

Age at Time of Egg Retrieval	Percentage of Day5/Day6/Day7 Embryos Expected to Be Normal
Younger than 35	>60%
35 – 37	~50%
38 – 40	~40%
41 – 42	~25%
Older than 42	<20%

Sex information is available through PGT-A. We do not disclose this information unless you ask at the time the PGT-A results are available.

Risks/Limitations

- One percent risk that the embryo may be destroyed in the biopsy process. Biopsy samples are immediately sent to the PGT-A lab and not held at Stanford.
- Cryopreservation (storage at subzero temperatures) of the embryos is required when utilizing PGT. There is a 2 to 5 percent chance that an embryo would not survive the freeze/thaw process and would not be available for transfer.
- Mosaicism: We only sample three to five cells. There is a possibility that abnormalities may not be identified, or that a reportedly abnormal embryo has viability and could have resulted in an ongoing pregnancy.
- It is possible that there are NO normal embryos at the end of an IVF cycle.
- PGT-A does not adequately address the risk of birth defects, developmental anomalies, single gene disorders, adult-onset conditions, autism, or intellectual disability. There is no testing that can identify all of these risks.

- **PGT-A does not guarantee a healthy pregnancy** and should be primarily used as an embryo selection tool to identify a single embryo for transfer. There is a 2 percent chance for misdiagnosis based on next-generation sequencing, either false positive or false negative. Mosaicism is expected to further decrease the overall accuracy, likely closer to 90 percent, although this number is not well validated.
- **Recommended follow-up: Due to the possibility of misdiagnosis, PGT should in no way replace screening in pregnancy.** You should follow your obstetrician's recommendations throughout a pregnancy, including ultrasounds, noninvasive screening for chromosomal abnormalities and birth defects, and invasive procedures if indicated, regardless of whether an embryo underwent PGT prior to transfer. Consultation with a prenatal genetic counselor or obstetrician familiar with prenatal screening is recommended to discuss these options.

Storage
All embryos will be stored, regardless of PGT result, until a signed consent is received that indicates patient approval to remove embryos from storage.

Monogenic Disorder (PGT-M, Previously PGD)

Preimplantation genetic testing is testing for embryos, performed before that embryo is transferred back to a uterus (before pregnancy). Testing for a monogenic disorder identifies whether embryos are at higher risk of inheriting known genetic changes. This means that PGT-M can be performed only in cases where there is a specific genetic change in the family. By screening for that specific change, an embryo not at risk for that specific condition could then be selected for transfer.

Billing:

Cost of testing varies by laboratory and insurance coverage. Please contact our financial coordinators for more information.