shady grove fertility

MEDICAL UPDATE

Counseling Patients about Pre-conception Genetic Testing

Testing Options and Recommendations

Shady Grove Fertility offers two distinct forms of pre-conception genetic testing:

1. **Pre-pregnancy genetic risk assessment** of partners
2. **Genetic testing of embryos** to measure total chromosome content or detect single gene mutations or structural rearrangements

PRE-PREGNANCY GENETIC RISK ASSESSMENT

RECOMMEND TO ALL PATIENTS, REGARDLESS OF ANCESTRY

The American College of Obstetricians and Gynecologists (ACOG) and other professional societies recommend pre-pregnancy genetic risk assessment (also called carrier screening) as part of routine pre-pregnancy testing for ALL patients, regardless of ancestry. This testing, performed using a small amount of saliva or blood, **identifies over 175 disease-causing genetic mutations** (i.e., alleles, mostly recessive) in healthy individuals that, when shared with their partner, can lead to a genetically affected child. Couples who share disease-causing mutations (carriers) for a particular genetic disorder such as cystic fibrosis can pursue measures to minimize the risk of having an affected child (see PGT-M, below).

GENETIC TESTING OF EMBRYOS

RECOMMEND TO PATIENTS EXPERIENCING 2 OR MORE MISCARRIAGES, WITH RISK FACTORS IDENTIFIED DURING GENETIC RISK ASSESSMENT, OR WOMEN OF ADVANCED MATERNAL AGE

In the subset of patients undergoing in vitro fertilization (IVF) in order to conceive, it is possible to test embryos for total chromosome counts. This is called **Pre-implantation Genetic Testing for Aneuploidy (PGT-A)** and can identify embryos with abnormal chromosome counts that will generally not produce a pregnancy or that might lead to a pregnancy loss (or, rarely, a chromosomally affected child such as Down syndrome). Unlike heritable genetic mutations, PGT-A screens embryos for "random" chromosomal errors (extra or missing chromosomes) that all too commonly occur in human embryos and that are more prevalent with increasing maternal age. Embryos found to have normal chromosome counts produce higher live birth rates and lower risk of miscarriage than embryos that remain untested.

For couples with heritable mutations (both partners are carriers), IVF-generated embryos can be screened for the specific mutations in question via **Pre-implantation Genetic Testing for Monogenic/Single Gene Disorders (PGT-M)** to identify embryos that will NOT be affected with that genetic disorder. In this setting, testing includes total chromosome assessment (PGT-A) as well as patient-specific mutation testing (PGT-M) for all of the embryos.

Pre-implantation Genetic Testing for Subchromosomal Structural Rearrangements (PGT-SR) is a variation of PGT-M and is specific to patients with identified chromosomal rearrangements (not entire chromosomal errors) discovered when being treated for problems such as recurrent pregnancy loss.

PATIENT CASE STUDY

PATIENT

34 year old female

Carries BRCA1 gene mutation

DIAGNOSIS

Breast cancer at age 28, underwent double mastectomy

Breast cancer returned at age 29 and began radiation treatment and full hysterectomy

TREATMENT

IVF and froze embryos at age 28

PGT-M to identify embryos without BRCA1

Sister without BRCA1 gene mutation as gestational carrier

SGF PHYSICIAN

Jeanne E. O'Brien, M.D.

OUTCOME

Healthy baby girl without BRCA1 gene mutation

"This was an amazing experience to help a couple facing a devastating diagnosis preserve their ability to not only have children but children that would not face the genetic cancer risk of the BRCA gene."
- Jeanne E. O'Brien, M.D.

Photo courtesy of Erin Silverman

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Who Should Undergo Pre-conception Genetic Testing?

Shady Grove Fertility strongly recommends pre-pregnancy genetic risk assessment because it benefits patients and their future children. If a couple is found to be at risk for passing on a genetic disease, the couple has the option to pursue in vitro fertilization (IVF) with pre-implantation genetic testing to avoid having an affected child.

PRE-PREGNANCY GENETIC RISK ASSESSMENT

The American College of Obstetricians and Gynecologists recommends that all couples planning for pregnancy undergo a genetic risk assessment to identify potential risk factors for heritable disorders.

EMBRYO GENETIC TESTING DURING IVF

Recommended for patients with:

- Risk factors identified by screening or family history
- Recurrent pregnancy loss
- Advanced maternal age
- Significant semen abnormalities
- Chromosomal inversions, translocations, or rearrangements

LEARN MORE ABOUT GENETIC TESTING OPTIONS ▶