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MEDICAL UPDATE

Genetic Screening Options for Couples Trying to Conceive

Updated 2017 Recommendations

There are many indications that make genetic testing prior to conception and prior to an embryo transfer necessary. Depending on each couple's unique situation the recommendations vary, giving patients a range of options when trying to achieve a healthy pregnancy. Understanding the options available is important when determining the best recommendation for each patient.

Expanded Carrier Screening of Both Partners

Generally completed via blood draw or saliva testing prior to conception, expanded carrier screening offers women trying to conceive and their partners greater insight into their own genetics. The insights gained by testing both partners offer the ability to identify possible genetic diseases that may be passed on to offspring prior to conception.

In February 2017, the American College of Obstetricians and Gynecologists (ACOG) formally made the recommendation to expand carrier screening to women beyond the traditional ethnic-specific screening historically offered prior to conception. The recommendations now also include regular screening for cystic fibrosis, spinal muscular atrophy (SMA), and a complete blood count to assess risk of hemoglobinopathy.

In the past, genetic screening was very expensive and as a result out-of-reach for most patients. Over the last couple of years, advances in screening technology have allowed patients the ability to screen for over 100 genetic diseases at a low out-of-pocket cost to patients. As a result, more are taking advantage of this testing.

WHEN TO INITIATE EXPANDED CARRIER SCREENING:
Prior to conception

WHAT CARRIER SCREENING TELLS PATIENTS:
Which, if any, genetic diseases they are carriers for and their probability of having a child affected by that specific genetic disease.

TREATMENT OPTIONS IF A GENETIC DISEASE IS FOUND:
IVF with preimplantation genetic diagnosis (PGD)

Genetic Screening of Embryos

When embryos are cultured outside of the body, as in the case with in vitro fertilization, the ability to genetically test each embryo prior to transfer becomes available.

APPROXIMATELY
70%
OF MISCARRIAGES ARE CAUSED BY GENETIC ABNORMALITIES IN THE FETUS. GENETIC TESTING DECREASES RISK OF MISCARRIAGE BY IDENTIFYING THE NORMAL EMBRYOS.

When proceeding with genetic testing, a few cells from each embryo are removed after several days of development, ideally when the embryo has reached the blastocyst stage. Each embryo is then frozen and the cells are sent to a genetic testing laboratory for evaluation, which takes between 10-14 days. Currently there are two different types of testing available for embryos, preimplantation genetic diagnosis (PGD) and preimplantation genetic screening (PGS).

PREIMPLANTATION GENETIC DIAGNOSIS (PGD) looks at the specific genes related to a known genetic disorder. This technology is generally used in situations when a couple has been confirmed to carry a gene related to a specific disease, generally determined through carrier screening. Identifying the embryos that are not affected for transfer is the couple's best chance of not passing on the disease.

PREIMPLANTATION GENETIC SCREENING (PGS) evaluates each embryo for abnormalities in the number of chromosomes. By looking at the overall make up of each embryo according to its number of chromosomes, either a missing or extra chromosome can be identified.

Once the results are available, genetically normal embryos are identified and the process begins to prepare her body for a frozen embryo transfer (FET) cycle where one (or more) of the genetically normal embryos is transferred into her uterus.

Additional Options for Patients

Should no normal embryos result, the couple can elect to initiate another fresh IVF cycle to create additional embryos that are genetically normal or, in some cases, consider the use of donor egg or sperm treatment. The donor chosen would not be a carrier for the genetic disease, thereby decreasing the chance of a chromosomal abnormality.

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Indications for Genetic Screening

Genetic screening is helping men and women who are having trouble conceiving to maintain a healthy pregnancy. Whether the couple is a carrier for a genetic disease, has experienced recurrent pregnancy loss, or is at a higher risk of chromosomal abnormalities associated with advanced maternal age, today's genetic screening options can be the answer to a healthy pregnancy and child.

The American College of Obstetricians and Gynecologists (ACOG) recently updated their recommendations for expanded carrier screening. These recommendations, paired with advances in IVF technology, are giving couples more options. Understanding the screening options available prior to conception and during IVF treatment is an important step when guiding patients.

LEARN MORE ABOUT GENETIC TESTING ▶

Expanded Carrier Screening

- Family history of genetic disease
- Ethnic-specific screening
- Proactive screening

Preimplantation Genetic Diagnosis (PGD)

- Known carrier of a specific genetic disease
- Recurrent pregnancy loss (RPL)
- Chromosomal translocation

Preimplantation Genetic Screening (PGS)

- Recurrent pregnancy loss (RPL)
- Advanced maternal age

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