How Is Genetic Testing Prior To Pregnancy Helpful?

If someone knows they have an increased chance to have a child with a genetic condition, then they may have the option to consider additional options not otherwise available or known to them. For example, they may consider preimplantation genetic testing (PGT) with in-vitro fertilization (IVF) which is genetic testing of embryos. This allows them to only implant embryos that do not have the genetic condition. Alternatively, the person may want to test their pregnancy in order to determine if the baby has the condition. If so, then they know to provide treatment as soon as the baby is born or to consider options such as adoption or termination.

Some people do not wish to know this kind of information. It is a personal decision, and a healthcare professional such as a genetic counselor, can help someone figure out which testing, if any, is best.

WHO WE ARE

Modern Reproduction's mission is to facilitate the discussion on the current technologies and genetic testing options available prior and during pregnancy by educating, empowering, and embracing modern reproduction. This handout does not serve as medical advice. Please refer to Modern Reproduction's Disclaimer and Terms and Service.

FOLLOW US

Instagram
@modernreproduction

FIND US

www.modernreproduction.org

Genetic Testing Prior to Pregnancy

Modern Reproduction

CARRIER SCREENING

Carrier screening can let a couple or individual know if they are at an increased chance to have a child with an inherited genetic condition. By assessing specific genes through a blood or saliva sample, carrier screening can identify if certain genes are not functioning as expected, potentially causing a genetic condition if passed down. Carriers, typically, do not have any symptoms of the condition.

If someone or a couple are identified as carriers, then they may have a 25% - 50% chance to have a child with that condition. Testing embryos, testing during pregnancy, or testing after a baby is born can be considered.

Carrier screening covers a range of genetic conditions, including common ones like Cystic Fibrosis, Sickle Cell Disease, and Spinal Muscular Atrophy. The number of conditions to be screened is a personal choice, and healthcare providers can assist in selecting the appropriate panel for you.

PREIMPLANTATION GENETIC TESTING (PGT)

Preimplantation genetic testing is performed on embryos created through in-vitro fertilization (IVF). An embryo is created after an egg is fertilized by a sperm. After fertilization, embryos develop from a single cell into a cluster of around 200 cells. PGT involves extracting a few cells from the embryo for genetic analysis. The goal of PGT is to implant an embryo that is predicted to not have a genetic condition. There are three main types of PGT.

PGT-A: assesses the number of chromosomes in the embryo to ensure the presence of the typical 46 chromosomes.

PGT-SR: assesses the number of chromosomes as well, but PGT-SR is specifically for someone with a chromosomal rearrangement. A chromosomal rearrangement increases the person's chance to have embryos or pregnancies with chromosomal abnormalities.

PGT-M: assesses for a specific, inherited genetic condition such as Sickle Cell Disease or Fragile X. It is suitable for individuals or couples who have an increased risk of passing on a particular genetic condition.

PERSONAL GENETIC TESTING

CHROMOSOMAL TESTING

Assesses a person's chromosomes to ensure they have the typical number of chromosomes and that the chromosomes are not rearranged. Commonly requested for couples experiencing multiple unexplained miscarriages.

CANCER SCREENING

Cancer screening tests genes associated with increased cancer risk due to gene malfunctions. This can be considered for someone who has had a personal cancer diagnosis and/or multiple family members diagnosed with similar cancers.

PERSONAL DIAGNOSIS

If someone has been diagnosed with a genetic condition by a physical exam, then there may be the option to have genetic testing to confirm the diagnosis by identifying a causative gene. If the gene is identified, then preimplantation genetic testing or prenatal diagnosis can be considered to test future embryos or pregnancies.