

Controversy with Carrier and Prenatal Testing

Prenatal genetic testing has both proponents and opponents. While a personal choice, it carries social, moral and political overtones.

Opposition to termination of a pregnancy is sometimes cited in arguments against genetic testing. *It is important to realize that termination of a pregnancy is never the only option.* There are many benefits to knowing the results of a prenatal genetic test because of all the options that the parents can actually consider. These options are discussed in detail below.

Option #1: Continue with pregnancy and accept this level of risk

Many couples who choose to continue with the pregnancy benefit from carrier testing by being emotionally prepared. Even healthy newborns bring major changes to a family. A newborn with a serious condition can create unique challenges if the family is not emotionally prepared to deal with the stress, sadness, remorse, anger and many other emotions that occur. When parents have time to explore their feelings prior to birth or seek out the counseling that they need, they can begin the process of coming to terms with their situation, and make important decisions that contribute to the quality of life of their child.

With full knowledge of what they can expect, parents may be better prepared for the special needs that their child will have. From a practical standpoint, their new child may need special therapy, medications, diet or assistive/ supportive devices to ensure the best quality of life possible. Knowing the genetic outcome will allow the couple time to get finances in order and prepare their home for their new child. They can also begin building a support network to assist them in the care of their newborn.

Option #2: Adoption

A couple might choose to adopt if preconception carrier testing shows both parents are positive carriers for a fatal genetic disorder such as spinal muscular atrophy. However, as mentioned earlier, most couples do not plan their pregnancy.

Some ordinary families may adopt children with inherited genetic disorders or disabilities. Children with disabilities ranging from childhood to teenagers may be placed for adoption, because the parents are not able to care for them. There are adoption agencies that specialize in placement of children with special needs.

Option #3: Termination of Pregnancy

Termination of the pregnancy is an alternative option. As mentioned before, there are many undercurrents with this option. Certain religions forbid it thereby causing much frustration and guilt for women and men having to face this type of decision. In the U.S. ruminant of pregnancy remains legal but it is not the case in all parts of the world. Even in the U.S., there are communities that will stigmatize a woman who chooses to end a problem pregnancy. There are situations, though, in which the fetus' organs can be harvested for transplant purposes and thus contribute to the life of another baby.

There may be medical reasons to consider termination such as the possibility or likelihood of fetal demise or risk to mother's health.

Option #4: In Vitro Fertilization (IVF) with Pre-implantation Genetic Diagnosis (PGD)

In Vitro Fertilization (IVF) is a method of assisted reproduction in which eggs are removed from a woman's ovary by inserting a needle through the vaginal wall. Fluid is removed and examined by a laboratory to be sure eggs are present. Active sperm are combined with the eggs in a petri dish, and incubated for two to three days or longer.

When the eggs and sperm are combined to form embryos, they start dividing. When the proper stage of division occurs, one cell (called a blastomere) is removed for genetic evaluation. The removal of this cell is technically challenging, so special tools, called micromanipulators, are used so as not to disrupt the remaining embryo. The biopsied blastomere is then sent to the laboratory for testing and the embryo is returned to the incubator.

Some worry that genetic testing will be used to wipe out what some consider weaker genes. This was a basic tenet of the eugenics movement. Others worry that parents will test for certain attributes, such as gender, height, and eye color. Still others base their concern on a belief that genetic testing will increase rates of abortion which is in conflict with their ethics.

Pre-implantation Genetic Diagnosis (PGD) is a method of genetic testing that screens embryos before implantation and pregnancy. Achieving pregnancy through IVF and PGD using healthy embryos can decrease the risk of genetic diseases in offspring.

The genetic laboratory is faced with the difficult task to evaluate a single cell for genetic disorder. For comparison, a laboratory examining various tissues of

an adult person has thousands, if not millions of cells available from patient's blood, biopsied tissue, or amniotic fluid. Thus, with the current available technology, the PGD laboratory cannot truly screen for multiple possible abnormalities. It can only identify the presence or absence of a specific disorder. Only embryos proven to not have a specific disorder are used for implantation. The resulting embryo is then transferred to the woman's uterus to implant and develop naturally. (*Adapted from the Institute for Reproductive Medicine and Genetic Testing website, www.preimplantationgenetictesting.com.)* Disorders that are tested for via PGD from one center include the following:

- Achondroplasia
- Adenosine deaminase deficiency
- Alpha-1-antitrypsin deficiency
- Alzheimer's disease (AAP gene)
- Beta thalassemia
- Cystic fibrosis
- Epidermolysis bullosa
- Fanconi anemia
- Gaucher's disease
- Hemophilia A and B
- Huntington's disease
- Muscular dystrophy (Duchenne and Becker)
- Myotonic dystrophy
- Neurofibromatosis type I
- OTC deficiency
- P53 cancers
- Phenylketonuria
- Retinitis pigmentosa
- Sickle cell disease
- Spinal muscular atrophy
- Tay Sachs disease
- Any genetically abnormal embryos (i.e. Trisomy 21, 13.)

Option #5: Use of Sperm and/or Egg Donor Cells

After determining carrier status, some parents will choose to use sperm/egg donor cells. This is also referred to as “third party reproduction” because a third party has donated sperm or egg cells enabling an individual or couple (usually infertile) to become parents. Sometimes the donor is known and sometimes the donor is anonymous. Egg donation was originally indicated for women with ovarian failure, and the first successful egg donation was in 1984, according to the American Society for Reproductive Medicine. Since then, egg donation has expanded, especially as IVF technology has improved. Now egg donation may be offered to women who are known to be affected by, or a carrier of, a genetic disease, and who would prefer not to pass the genetic disease to the child.

There are screening guidelines for donors recommended by the American Society for Reproductive Medicine that includes an extensive medical questionnaire seeking personal and medical family history, as well as history of communicable diseases and more. Genetic diseases are tested for by ethnicity. Donors of Ashkenazi Jewish descent are tested for Gaucher’s Disease, Tay-Sach’s Disease, cystic fibrosis, and Canavan disease. Caucasians are tested for Cystic Fibrosis, but in most cases they are not tested for SMA.

Option #6: Choice to have no children

Some parents decide to forego testing completely and decide not to have children.

In summary, while there may be other options that are not covered here, it is important to know these options and be prepared to incorporate them into discussions with physicians. Physicians want to provide reproductive options to their patients, and Athena Diagnostics is prepared to help them.

Remember, SMA Carrier testing provides reproductive options to patients