EMORY UNIVERSITY SCHOOL OF MEDICINE

Department of Human Genetics
Division of Medical Genetics
www.genetics.emory.edu

Reproductive Options for Carriers of Genetic Diseases & Chromosome Rearrangements

Individuals who are carriers for genetic diseases, such as sickle cell disease or cystic fibrosis, are at increased risk to have a child with a genetic disease. Likewise, individuals who carry a balanced chromosome rearrangement are at increased risk for miscarriage or having a child with birth defects or mental retardation. There are several reproductive options available to couples who wish to have children but are at increased risk for the above-mentioned concerns.

- **Natural Conception:** Couples in which both are carriers of a genetic disease or one is a balanced translocation carrier may consider a natural conception. With this option, no testing is employed before conception and the couple may either pursue prenatal diagnosis or no testing during the pregnancy. If no testing is performed during the pregnancy, a couple can elect to test a baby at birth (if desired) to determine if he/she has a genetic disease or chromosome abnormality.

- **Prenatal Diagnosis:** Prenatal diagnosis is available during a pregnancy to diagnose a chromosome abnormality or certain genetic diseases prior to birth. There are two routinely used options for prenatal diagnosis:
  1. **Chorionic villus sampling (CVS)** is routinely performed between 10-12 weeks of pregnancy.
  2. **Amniocentesis** is routinely performed after 15 weeks of pregnancy.

  The complication rate, including miscarriage, associated with either procedure is approximately 0.5% (1 in 200). Contact your doctor or genetic counselor for more specific information regarding prenatal diagnostic options.

  If prenatal testing reveals that the pregnancy is affected, the parents have the opportunity to prepare to have a child with special needs, to arrange for adoption of a child with special needs and/or to have the option of terminating the pregnancy. In the state of Georgia, the legal limit for pregnancy termination is 24 weeks gestational age.

- **Preimplantation Genetic Diagnosis (PGD):** PGD is accomplished by combining in vitro fertilization (IVF) and genetic analysis to test an embryo prior to implantation. The eggs are fertilized in the laboratory, one cell is removed from each embryo, and then genetic testing is performed. Only unaffected embryos are transferred into the uterus for implantation. PGD is available for many chromosome rearrangements but only a few genetic diseases. You can discuss the costs and logistics of this option in more detail with a reproductive endocrinologist. Contact your genetic counselor or a center which deals with assisted reproduction to find out more about the availability of PGD for specific conditions.

- **Gamete (egg or sperm) Donation:** A couple may choose to use a known or anonymous egg or sperm donor. We recommend appropriate genetic testing and screening for any gamete donor.

- **Adoption:** There are many private and government adoption agencies available.