



Reproductive Options for Carriers of Genetic Diseases & Chromosome Rearrangements ©2004

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.

1. 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 and the couple can elect to test a baby at birth (if desired) to determine if he/she has a genetic disease or chromosome abnormality.
2. **Prenatal diagnosis** is available to diagnose a chromosome abnormality or certain genetic diseases prior to birth. There are two routinely used options for prenatal diagnosis, and these include chorionic villus sampling (CVS) and amniocentesis.

A CVS procedure is routinely performed between 10-12 weeks of the pregnancy. The complication rate including miscarriage, associated with CVS is 1%. Amniocentesis is routinely performed after 15 weeks gestation. The complication rate including miscarriage, associated with amniocentesis is 0.5%. Contact your doctor or genetic counselor for more specific information regarding prenatal diagnostic options.

If the pregnancy is affected, the parents have an opportunity to prepare to have a child with special needs and/or have the option of terminating the pregnancy (until 26 weeks gestation in the state of Georgia).

3. **Preimplantation genetic diagnosis (PGD)** is available to test an embryo prior to implantation. PGD is accomplished by combining genetic and in vitro fertilization technology. The eggs are fertilized in the laboratory and the embryos are allowed to grow until 4 to 8 cells are present. Then, one cell is removed from each embryo and genetic testing is performed. Only the unaffected embryos are transferred back into the uterus for implantation. Laboratory charges for testing the embryos vary and may require a blood sample from the parents prior to embryo testing. PGD is available for many chromosome rearrangements but only a few genetic diseases. You can discuss the logistics and costs 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.
4. **Gamete (egg or sperm) donation** is also available. A couple may choose to use an anonymous or known egg or sperm donor. We recommend appropriate genetic testing and screening for any gamete donor.
5. **Adoption** is another option to consider. There are many private and government adoption agencies available.