



Preconceptional Genetic Counseling^{©2004}

What is Genetic Counseling?

Genetic Counseling is a professional assessment of a patient's risk factors regarding their family history and/or a current or future pregnancy. Many couples want to know what their chances are of having a healthy baby, once they become pregnant. Preconceptional genetic counseling is a way to identify any risk factors that may increase the risk for birth defects once a pregnancy is achieved. This information is important when considering options such as using one's own gametes (eggs or sperm), versus choosing a donor, or whether to undergo preimplantation genetic diagnosis. Genetic counseling is provided for egg donors for these same reasons (i.e. to determine genetically whether the donor is a good candidate from a genetic standpoint).

The goal of genetic counseling is not only risk assessment, but also to explain cause/inheritance of a disorder, availability of testing, prognosis, medical management, treatment and options. Genetic counseling sessions typically last one hour, or longer, depending on the complexity of the case.

What is a Genetic Counselor?

Genetic Counselors have a minimum of a master's degree in genetic counseling or a related field. The former includes extensive training in both human genetics as well as psychology. Board Certification is offered through the American Board of Genetic Counseling. All Genetic Counselors at Emory Genetics are Board-Certified and work under the supervision of a Board-Certified M.D. Clinical Geneticist.

Reasons to Refer Patients for Preconceptional Genetic Counseling:

PREGNANCY FACTORS

- Infertility cases where either parent is suspected of having a chromosomal abnormality
- Couples who have had two or more spontaneous abortions (miscarriages), stillbirths, or other pregnancy losses on a repetitive basis.
- Couples requiring assisted reproduction techniques to achieve pregnancy, or individuals donating eggs or sperm for those purposes.
- Couples interested in undergoing preimplantation genetic diagnosis.
- Couples with difficulty achieving a pregnancy due to male factor infertility.
- Maternal Age 35 years or greater at delivery.
- Maternal serum screening (AFP with or without HCG, uE3) indicating an increased risk for neural tube defects (spina bifida, anencephaly), Down syndrome or trisomy 18.
- Abnormal prenatal diagnostic results or abnormal prenatal ultrasound examination.
- Maternal factors such as schizophrenia, depression, seizures, alcoholism, diabetes, thyroid disorder and others in which fetal abnormalities may be associated either with the disease process or with common medications prescribed for the disease.
- Fetal or parental exposure to potentially teratogenic, mutagenic or carcinogenic agents (i.e. drugs, chemicals, radiation, infections).
- Paternal age over 55 years at time of conception.

FAMILY HISTORY FACTORS

- Previous child or family history of: mental retardation, neural tube defects, chromosome abnormality (i.e. Down syndrome, trisomy 18, Klinefelter syndrome, etc.), cleft lip/palate, congenital heart defects, short stature, single gene defects (i.e. cystic fibrosis, muscular dystrophy, hemophilia, PKU, etc.), hearing or visual impairments, learning disabilities,

psychiatric disorders, cancers, multiple pregnancy losses (miscarriages, stillbirths, early infant deaths) or other disorders which could be genetic.

- Either parent with an autosomal dominant disorder, or any disorder seen in several generations.
- Both parents carriers for an autosomal recessive disorder, diagnosed either by the birth of an affected child (i.e. cystic fibrosis) or by carrier screening (i.e. sickle cell anemia, Tay Sachs, etc.).
- Mother, known, or presumed carrier of an X-linked recessive disorder (i.e. hemophilia, Duchenne muscular dystrophy).
- Either parent a known carrier of a balanced chromosome abnormality.

OTHER FACTORS

- Persons in specific ethnic groups or geographic areas with a higher incidence of certain disorders, such as Tay Sachs disease, sickle cell disease or thalassemias.
- Extreme parental concern or fear of having a child with a birth defect.
- Cases of consanguinity or incest where a pregnancy is involved.
- Premarital or preconception counseling in couples at high risk for genetic disorders based on family or personal medical history.