



Who Should Be Referred for Genetic Counseling? ©2004

Indications for a genetic counseling referral include:

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 45 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 are 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, is a known, or presumed carrier of an X-linked recessive disorder (i.e. hemophilia, Duchenne muscular dystrophy).
- Either parent is a known carrier of a balanced chromosome abnormality.
- Family history of cancer, particularly at young ages (under 50).

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 thalassemia.
- Extreme parental concern or fear of having a child with a birth defect.
- Cases of consanguinity or incest where a pregnancy is involved.