ReproMed Fertility Center
Creating Parents and Building Families

**Contact:** (214) 827-8777

# Genetic Screening for Birth Defects

About one in 20 pregnancies result in birth defects. These can be characterized by minor structural abnormalities, or serious genetic diseases and mental retardation. These deformities can be detected early through genetic screening. Genetic screening involves a series of tests that are performed to determine if you are at an increased risk of having a child with a birth defect.

Genetic screening is usually suggested for couples:

- Of certain racial or ethnic backgrounds predisposed to having a high risk for a particular birth defect
- With a family or medical history of a particular abnormality
- Suffering from a condition associated with a birth defect
- With a high risk of being a carrier of a certain abnormality
- In which the woman is of advanced age (over 35 years of age), having a previously affected child or a family history of chromosomal problems and miscarriage.

The birth defects associated with certain racial and ethnic backgrounds include sickle cell disease, cystic fibrosis, thalassemia and Tay Sachs. Those with a familial association include muscular dystrophy, sickle cell, seizures, Huntington's disease, mental retardation, bleeding disorders, unexplained still-births and recurrent miscarriages. Advancement in woman's age during pregnancy is linked to serious chromosomal abnormalities such as spinal bifida or Down syndrome.

Genetic testing can involve the examination of a sample of blood for certain disease markers or chromosomal deformities, using ultrasound image techniques to view any abnormality in the growing fetus, amniocentesis, where amniotic fluid is removed and tested, and pre-implantation genetic diagnosis (PGD), where cells from a particular stage of the developing embryo are removed for analysis of risk factors.