GENETIC SCREENING & COUNSELING

BASIC INFORMATION

DESCRIPTION

- Genetics is the study of inheritance and how the characteristics of one generation are passed on to another. Prior to pregnancy, or once pregnancy is diagnosed, a medical and family history of both parents will be taken by the doctor to look for any any conditions that could affect, or be inherited by, the fetus. Most couples are at little risk for transmitting a genetic problem. If there appears to be an increased risk, certain screening tests will be recommended and, possibly, a referral to a doctor who is a genetic specialist for further testing and counseling.
- Genetic counseling is used to obtain and provide expectant parents with as much objective information as possible to help them arrive at a decision that is based on their own desires, values and ethics. Counseling is not used to coerce a couple to take or not take any particular tests or to make any reproductive decisions that are not theirs alone.
- Though some defects are genetically transmitted, other defects are the result of environmental effects as the fetus develops in the womb and many of the defects that occur are spontaneous without traceable cause.

POSSIBLE INDICATIONS FOR GENETIC TESTING

- Advanced maternal age (35 or older).
- Parental chromosome abnormality.
- Couples whose blood tests indicate they are carriers of a disorder.
- Couples who have already had a child with a genetic abnormality.
- Closely related couples such as first cousins.
- A pregnant woman whose routine prenatal tests detected a fetal defect.
- One of the partners has a congenital defect such as congenital heart disease.
- Couples who have a family history of a hereditary disorder or mental retardation of unknown origin.
- A woman with a history of multiple miscarriages (generally 3 or more) or stillbirth.
- Either parent belongs to a race or ethnic group at high risk for a specific genetic disease (e.g., sickle-cell disease in African-Americans, Tay-Sachs disease in Jews of Ashkenazic background, and beta-thalassemia in people of southern European ancestry).
- Either partner has been exposed to high doses of radiation, drugs, occupational exposure to gases or chemicals, or other environmental agents which, could result in congenital abnormalities.

PRELIMINARY SCREENING TESTS FOR GENETIC (CHROMOSOMAL) DEFECT

- Maternal blood test–measures the serum level of alpha-fetoprotein and other hormones which can identify a fetus with high risk for neural tube defect (cranial and spinal abnormalities) or chromosome abnormality such as Down's syndrome. There is a possibility of false-negative or false-positive results.
- Ultrasound–the high-frequency soundwaves produce images of the fetus and placenta. It can detect major defects in the heart, bones, brain, abdomen and spinal cord.

SCREENING TESTS CONCLUSIVE FOR GENETIC (CHROMOSOMAL) DEFECT

- Amniocentesis–a needle is used to take a sample of amniotic fluid (liquid surrounding the baby in the uterus) that is then analyzed for many genetic disorders.
- Chorionic villus sampling (CVS)–a sampling of cells in the chorionic villus (part of the placenta) is analyzed for the presence of various defects or diseases.
- Fetoscopy (rarely used)–an optical instrument with a lighted tip is inserted into the uterus to observe the fetus. A blood sample can be taken at the same time for laboratory analysis.
- Cordocentesis–a sample of fetal blood is obtained from the umbilical cord with a fine needle guided by ultrasound. The blood sample is analyzed for defects or disorders.

OUTCOME OF TESTING

- Tests may indicate no problems.
- Tests may indicate a possible problem that requires further testing.
- Testing may show a definite problem. The parents will need to make a decision about continuing with the pregnancy if the defect is serious. Though the decision is the parents' alone, the medical care team will provide counseling and important information to assist them in a nondirective, but understanding manner.
- The determination of serious genetic defects in a couple's history may present problems about future pregnancies and lead to decisions about permanent sterilization and discussions about other methods of contraception such as donor insemination. Genetic counseling will assist in determining the best alternative options available.

NOTIFY OUR OFFICE IF

You or a family member has questions or concerns about genetic screening or counseling.