

Preconception Genetic Counseling and Screening

By Karen Stebner, CGC, and Philip Buchanan, PhD

Raising a healthy child can begin even before conception of a pregnancy. Preconception genetic counseling and testing are available to provide couples with information regarding health conditions for which their children could be at risk.

Issues that could affect a child's health and can be screened for prior to pregnancy include maternal health conditions, such as diabetes and seizure disorders; a couple's ages; past pregnancy history; and exposures to environmental hazards, such as medications, drugs of abuse, and work-place exposures. A family history of mental retardation, birth defects, and single gene disorders, such as muscular dystrophy, may also increase the risk for an abnormality. These issues can be screened for prior to pregnancy, and a couple can then be offered genetic testing to see if their pregnancies are at risk. Knowledge of a potential risk can provide options for management and treatment of a pregnancy.

Many couples do not have a history of genetic disorders in their families but are still at risk for certain conditions because of their ethnicity. For example, individuals of Jewish descent are more likely to carry the genes for Tay-Sachs (TS) and other disorders. Approximately 1 in 25 Caucasians carries the gene for Cystic Fibrosis (CF). CF and TS are autosomal recessive disorders—therefore the effect of the gene is only seen when a person inherits an abnormal recessive gene from both parents.

Caucasian Couple: *We came for genetic counseling because we know we are more likely to be carriers of CF. We would like to become pregnant but would first like to know our chances for having a child with CF.*

Genetic Counselor: *We can perform a DNA blood test on each of you to see if you carry the gene for CF. If you are both CF carriers, you will have a 25 percent chance of having a baby with CF with each of your pregnancies.*

Couple: *What would be our options for having healthy children?*

Genetic Counselor: *If you decide to become pregnant, prenatal diagnosis is available to test the baby for CF during pregnancy. Chorionic Villus Sampling (CVS) is performed at 10-13 weeks gestation and amniocentesis after 11 weeks. If a diagnosis of CF were made, you would have the option to continue or terminate the pregnancy. Other options for carrier couples are adoption, use of a donor sperm or egg, and pre-implantation diagnosis. Pre-implantation diagnosis involves the technique of in vitro fertilization and testing of an embryo for CF before it is implanted in the uterus.*

Through preconception genetic counseling and testing, couples can gather information so they can make informed decisions about proceeding with pregnancy. To obtain additional information about preconception genetic counseling and testing, talk with your physician and contact a genetic counselor in your area.

Dr. Buchanan is the director of GeneCare Medical Genetics Center. The co-author, Karen Stebner, is a certified genetic counselor. GeneCare provides genetic counseling, screening and laboratory services. For more information, contact their Web site: www.genecare.com