

## Environmental Factors

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Nurses with an Advanced Practice in Genetics, as well as genetic counselors with a Master's Degree, may see families or individuals of any age, referred by a health care provider due to a family history of an inherited diseases or birth defects, a history of infertility or multiple pregnancy losses, an environmental exposure of concern, advancing maternal or paternal age, or any other condition with a genetic predisposition. These genetic professionals take a detailed family history and pedigree, and provide education and information for the patient and family regarding the potential prevention, diagnosis, and/or treatment for the disorder.

Adolescents that are referred for pre-conceptual planning are a relatively new and increasingly important type of referral to genetics centers. According to statistics from the March of Dimes, nearly 13% of all U.S. births were to teens, ages 15-19. Over 1 million teenagers become pregnant in the U.S. each year, and an estimated 485,000 give birth. Approximately 1 in 4 of these teenage mothers have a second baby within the birth of their first baby.

The diet of a typical teenager is inadequate, and may be extremely low in essential vitamins, including folic acid. These younger mothers are more likely than older mothers to have an inadequate weight gain during the pregnancy, which may increase the risk of a low birth weight baby. Pregnant teens have an increased incidence of cigarette smoking during pregnancy. According to Dr. Cathy Melvin, director of Smoke-Free Families, "among white women who did not graduate high school, smoking rates while pregnant averaged 48% in 1998, compared with a smoking rate during pregnancy for other populations of 13%." Exposure to recreational drugs, alcohol, and sexually transmitted diseases during pregnancy are additional factors that dramatically affect the risk of a low birth weight baby, and pregnancy complications, as well as increasing the risk of birth defects in the offspring of teenage mothers. The March of Dimes indicates that in 1987, 7.2% of mothers ages 15-19 received late or no prenatal care, compared to 3.9% for all ages. These teenage mothers are at a greater risk than women over age 20 for pregnancy complications such as premature labor, anemia, and hypertension, especially if they are under 15 years old.

The following is an example (fictional) of a pre-conceptual genetic counseling session, for a teenage couple:

Tina Hunt (age 17) and her boyfriend Johnny Locklear (age 18) have been referred for pre-conceptual genetic counseling because they recently informed Tina's Ob/Gyn of her desire to achieve a pregnancy. The genetic counselor meets with the couple, and obtains a detailed medical, social, and family history. Tina and Johnny are both of Lumbee Indian ethnicity. Tina smokes approximately ½ pack of cigarettes per day, and Johnny smokes approximately 1 pack per day. Tina does not take any multivitamins, and admits that she has been skipping meals to "lose weight before the wedding". The couple indicates that they plan to be married in 6 months, (after graduating from high school) but admit they have "actively been trying to achieve a pregnancy" and therefore, not using any form of birth control for the past 4 months. In the family history, Tina indicates she has one sister that was born with spina bifida. John has one niece with a history of isolated cleft lip and cleft palate.

The counselor first discusses the importance of both members of the couple quitting smoking, prior to conception. She explains that cigarette smoking during a pregnancy, or exposure to second-hand smoke, may increase the risk of Sudden Infant Death Syndrome

(S.I.D.S.) 2 – 3 fold, for the offspring. In addition, cigarette smoking during pregnancy has been associated with an increased risk of a low birth weight infant, an increased risk of miscarriage, decreased fertility, possible increased risk of learning disabilities and/or unexplained mental retardation, and a possible increased risk of certain birth defects, including cleft lip/palate, club foot, and certain congenital heart defects. Mary and Johnny indicate to the counselor that they "had no idea that avoiding cigarettes during pregnancy was so important." The genetic counselor also explains that due to Johnny's family history of a niece with cleft lip and palate, in addition to the couple's American Indian ethnicity, there is an increased risk of this birth defect, which is typically inherited in a multifactorial (many genes plus environmental factors) fashion.

The counselor also explains the importance of good nutrition, both before and during a pregnancy. She recommends Mary having a diet rich in foods that contain folate or folic acid, before she becomes pregnant. Folate is the natural form of folic acid, a B vitamin that is found in foods such as orange juice, leafy green vegetables, beans, peanuts, broccoli, peas, and bread and grain products that have been fortified by the FDA.

Since most women cannot obtain adequate amounts of folic acid through diet alone, the counselor explains that Mary should start taking a daily multivitamin that contains 400 micrograms of folic acid, because research has shown that pre-conceptual use of folic acid, taken at least 3 months prior to conceiving a pregnancy, may reduce the risk of birth defects of the brain and spinal cord, called

neural tube defects (NTD's), by 50-70 %. The neural tube is a structure in the embryo that develops into the brain and spinal cord, and if the neural tube does not close completely, defects in the brain or spinal cord may occur.

The counselor discusses that approximately 1: 1000 babies in North Carolina are affected with this type of birth defect. Since NTD's occur approximately 28 days after conception, before most women realize they are pregnant, it is important to take folic acid before pregnancy. Pre-conceptual folic acid (400 micrograms/day) is currently recommended for all women of childbearing age, since it is estimated that 50% of pregnancies are unplanned.

Since Mary has a family history of one sister affected with Spina Bifida and therefore, Mary has an approximate 2 % risk of a NTD for her offspring, the genetic counselor recommended that Mary speak to her health care provider before she gets pregnant, in order to obtain a prescription for a larger dose of folic acid (4 milligrams), taking it at least one month before pregnancy, and continuing it for the first trimester of pregnancy.

Studies suggest that pre-conceptual use of folic acid may also reduce the risk of other birth defects, including cleft lip/palate as well as congenital heart defects and others. John indicates he will inform his sister that has already had a child affected with cleft lip and palate, of this important information.

The counselor also stresses the importance of early and regular prenatal care with the couple, in order to reduce their risk of pregnancy complications and low birth weight. In addition, the counselor discusses options for future screening and testing during a future pregnancy, which may include blood testing in the second trimester of pregnancy for alpha fetoprotein and free beta hCG, to estimate the risk of NTD's and Down Syndrome, as well as ultrasound and possibly amniocentesis. She explains the benefits, limitations, and risks of each of these prenatal tests.

The couple thanks the counselor for all of the information they learned during the genetic counseling.

While this scenario was a "fictional" one, it is hoped that more health care providers will realize the important impact that pre-conceptual genetic counseling may have for adolescents in preparing them for a healthy pregnancy, and reducing the risk of birth defects and pregnancy complications.

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