OB Genetic Testing

The genetic code: Most cells in the human body contain 46 chromosomes arranged in 2 pairs of 23 chromosomes. Each chromosome contains multiple genes. Genes are responsible for the traits of an individual which are derived from the mother and father. A sperm carries 23 chromosomes and an egg carries 23 chromosomes, resulting in a normal 46 chromosome fetus. The sex chromosomes are the X and Y chromosomes, with the X chromosome coming from the egg and the X or Y chromosome coming from the sperm, resulting in an XX fetus (female) or XY fetus (male). Trisomies are by definition an extra chromosome ie. Trisomy 21 or Down's Syndrome.

Testing: All obstetrical patients will be offered one form of testing, description to follow. Special consideration is given to any pregnant patient that will be 35 years of age or older at the time of delivery. She is considered AMA (advance maternal age - terminology provided by the American College of Obstetrics and Gynecology) and her risk of trisomies is increased so that those individuals can proceed directly to invasive testing if desired. Non-invasive testing can still be performed with AMA patients with the understanding that although detection rates increase to ~95%, the false positive rates increase from 5-10% to ~25%.

Non-Invasive genetic testing:

1) Ultrascreen: (first trimester screening - usually performed between 11-13 weeks) is testing done out of a facility designed to perform specific measurements of the fetus and blood calculations, usually a Maternal Fetal Medicine office (MFM). Calculations are then performed to determine risks of trisomies (extra set of chromosomes) such as trisomy 21 (Down's Syndrome), trisomy 18 (Edward's Syndrome) and trisomy 13 (Pateau's Syndrome) - with a false positive rate of ~5% and a detection rate of ~85%. If a positive result occurs, a CVS would then be offered and done by a specialist.

2) Quad Screen: Is blood testing done between 14 6/7 - 20 6/7 weeks. There are 4 chemicals that are measured and a computer calculation is performed to determine the risk of the above mentioned trisomies with a false positive rate ~10% and a detection rate of ~80%. If a positive result occurs, an amniocentesis would then be offered and is usually performed by your OB/GYN.

Invasive genetic testing:

1) CVS: chorionic villus sampling is usually done in the first trimester (10-12 weeks). Cells are taken from the placenta during a speculum exam with a concomitant ultrasound being done. A pipelle is placed through the cervix and then behind the placenta. The rate of miscarriage is quoted at ~1/200 to 1/300 - slightly higher than amniocentesis.

2) Amniocentesis: is a sampling of the amniotic fluid which is done around 16 weeks. Cells are taken from the amniotic cavity (where the fetus is at). A very fine needle is passed through the maternal abdomen, below the umbilicus and into the uterus and amniotic cavity, under direct ultrasound guidance. A small sample of fluid is withdrawn and sent to the lab for the cells to grow to determine the chromosomes of the fetus. The rate of miscarriage is quoted at 1/300 or slightly less.

Genetic Testing for Specific Diseases

Cystic Fibrosis (CF) - CF is a genetic disease related to specific populations, predominantly European Caucasian and Ashkenazi Jewish, as well as others. Individuals found to be carriers of the gene can be as high as 1 in 29. The disease is manifested when both parents pass the gene to the fetus. The disease is characterized as a transport mechanism problem where heavy mucous secretions become a problem in the functioning of major organs, especially the lungs and gastrointestine. A number of infections can occur where the individuals tend to fight these off their entire lives. Blood testing is offered during pregnancy or even before conception. If the mother is found to be a carrier, her partner will be offered the testing as

well. A geneticist may be consulted and amniocentesis may also be offered. The cost of testing may range from $100 - $300, depending upon the company and insurance conditions.

Sickle Cell Disease - Is a genetic disease found primarily in African Americans. It is manifested when an abnormal hemoglobin causes the red blood cells to take on the appearance of a sickle. As a result, these cells do not function as well leading to oxygenation problems throughout the body. A blood test - hemoglobin electrophoresis - is recommended to determine carrier status. If the mother is found to be a carrier, her partner will be offered testing. If both are found to be carriers, a geneticist may be consulted and amniocentesis may also be offered.

Thalassemia - Is a genetic disease found in certain populations of Mediterranean descent, usually Italians and Greeks and some individuals of Asian descent. Beta thalassemia is associated with those of Mediterranean descent and alpha thalassemia is associated with those of Asian descent. The disease is related to anemia, causing a production problem with normal hemoglobin. Suspicion of the disease results when certain abnormal values are found in a CBC (complete blood count) which is included in routine blood work on all new OB patients. When thalassemia is suspected, additional blood work - hemoglogin electrophoresis - is ordered. A specialty consult may be requested with a geneticist or maternal fetal medicine.