Prenatal Genetic Testing

Each pregnancy has a 3-5% chance of being effected by a physical or mental birth defect. Up to 75% of birth defects have no known cause. About 10% of birth defects are caused by environmental exposures, such as exposure to x-ray, certain illnesses, taking medicines or drugs not safe in pregnancy, or drinking alcohol while pregnant. Genetic problems make up 15-20% of known birth defects. Some genetic defects are inherited and some are random events that happened when the fetus (baby) began to grow. There is no cure for a genetic defect. Testing for birth defects is offered to all Around the Circle Midwifery clients during their pregnancy and you may choose to have testing performed or not.

Genetic counseling is available to help families make decisions regarding genetic testing. Genetic counselors are people who have special training in helping families understand risks of genetic problems, testing options, and treatments for genetic conditions. You may choose to meet with a genetic counselor before deciding on testing options.

There are three different types of tests available: carrier testing, screening tests, and diagnostic tests.

- **Carrier testing** is a blood test that shows if the mother has a gene she could pass to her baby that could cause a genetic disease, such as cystic fibrosis or spinal muscular atrophy. There are tests that are also offered to certain people groups, such as Tay-Sachs testing for people of Jewish decent. Genetic carrier testing may be done before or during a pregnancy. Around the Circle Midwifery offers everyone cystic fibrosis and spinal muscular atrophy carrier screening when they begin care. Other carrier screenings may be chosen after talking with a genetic counselor.

- **Screening tests** predicts who is more likely to have a baby with a birth defects. These tests do not diagnose disease but tell us who should be offered further testing. Testing may be a blood test or an ultrasound. Screening tests look for specific diseases, including Down syndrome (Trisomy 21), Edward syndrome (Trisomy 18), Patau syndrome (Trisomy 13), Neural tube defects, and other rare genetic disorders. There is not one screening test that checks for all the disorders at once, so sometimes families will choose several different screening tests. The following list explains how often some of these diseases happen:

    - Down syndrome: 1 in 700 babies born with increased risk as a mother gets older. This causes mental retardation and physical defects, such as heart disease.
    - Edward syndrome: 1 in 5,000 babies born. This disease is usually fatal with most babies dying before birth. 10% of babies with Edward syndrome will live to 1 year old.
    - Patau syndrome: 1 in 16,000 babies born. This disease is usually fatal with 20% living to 1 year old.
    - Neural tube defects (NTD) 1 in 2,000 babies born. This includes diseases like spina bifida, or open spine, and anencephaly (lack of brain growth). This disease may be mild or severe depending on how the baby is developing.

Optional screening tests include blood samples and ultrasound. Screening tests tell us who should be looked at more closely for diagnostic testing. A screening test may indicate there is a problem when no problem truly exists. This is called a false-positive result. A screening test may also indicate there is no genetic problem when there actually is a genetic problem. This is called a false negative. All tests have a false positive or false negative rate. The following is a list of screening tests offered at Around the Circle Midwifery:

- **Fetal survey ultrasound** uses sound waves to create a picture of the baby and their developing organs. This is done between 20-22 weeks. Neural-tube defects will be detected in 92-95% of babies with these defects. Other genetic diseases listed, like Down syndrome or Edward syndrome will be detected in 80-90% of babies with these diseases. Other diseases and birth defects may also be detected during an ultrasound exam. Ultrasound also gives information about the size of the baby, placenta location, amniotic fluid and sack, as well as the mother's cervical length and ovaries.
- **Maternal serum AFP** detects the presence of neural-tube defects. Approximately 75% of babies with a defect will be detected with this screening, 25% will not be detected. Testing is performed between 15-22 weeks. This test may be combined with an ultrasound exam at 20-22 weeks to increase detection rates. Some families will choose to have this test after an ultrasound exam if timing allows for it.
- **Penta screen** is a blood test that may be performed between 15-22 weeks with the Maternal serum AFP screening. It tests for the NTD as well as Down syndrome, Edward syndrome, and Palau syndrome. The test detects substances in the mother's blood that happen with an increased risk of these diseases. It detects about 60-80% of babies with these diseases and has a 5% false positive rate. A similar test, the **Integrated screen**, is also available slightly earlier but with similar detection rates and false positive rates as the Penta screen. The Integrated screen uses ultrasound at 11-13 weeks and 2 blood tests in first and second trimester. It may be several weeks before test results are available from these screenings.
- **Non-invasive Prenatal Testing (NIPT)** is a blood test that may be performed after 10 weeks of pregnancy. This test looks at fetal cells that are in the mother's blood to check for genetic diseases such as Down syndrome, Edward syndrome, and Patau syndrome. People with a family history of developmental delay may also choose to have an expanded panel that can detect Turner syndrome, Klinefelter syndrome and other sex-linked diseases. This test is able to determine the sex of the baby as well. NIPT detects 99% of babies with these diseases and has a 0.1% false positive rate. It does not test for NTD. It takes 7-10 business days to get the results of this test.

Diagnostic tests tell us if a baby has a genetic disease or not. There is not a diagnostic test that checks for all possible genetic diseases. Diagnostic tests are generally provided to people with a higher risk of having a baby with a disease, like those who had a positive screening test, because there is a 1-2% risk of miscarriage with these tests. Diagnostic tests are available by referral to a specialized physician who offers genetic counseling and testing. Your midwife may provide you with a referral to see one of these doctors one if you desire diagnostic testing.

- **Chorionic villus sampling** collects placenta cells between 10-14 weeks by inserting a tube and needle through the cervix and into the uterus. These cells have the fetus's DNA in them and can accurately show if the fetus has Down syndrome or one of the other genetic diseases listed without the high false positive and false negative rates. It does not test for NTD.
- **Amniocentesis** is performed by inserting a needle through the abdomen and into the uterus to take a sample of amniotic fluid between 15-20 weeks. This fluid has fetal cells in it that may be examined for the genetic diseases listed above. It is also very accurate for genetic disease detection and can detect up to 80% of babies with NTD.

More information may be found by viewing Washington Department of Health information videos on genetic testing found on the Around the Circle Midwifery web page at: http://www.aroundthecirclemidwifery.com/pregnancy.html

Genetic testing may or may not be covered by your insurance. It is your responsibility to determine any costs you may have in obtaining tests by contacting your insurance carrier.

I have read all of the above information and have had my questions answered to my satisfaction. I have had the opportunity to view the informational videos as well. I understand the benefits and risks of testing verses not testing and each of the tests offered.

I elect: (initial one or more):
_____Genetic counseling for more information
_____Genetic carrier screening for (circle choices): cystic fibrosis spinal muscular atrophy
_____Fetal survey ultrasound at 20-22 weeks
_____Maternal serum AFP screen
_____Penta screen
_____Integrated screen
_____Non-invasive Prenatal Testing
_____Referral to a specialist for diagnostic testing

_____I decline speaking to a genetic counselor and all prenatal genetic testing.

Signature:_____

Today's date:_____