# Informed Choice for Prenatal Genetic Screening

## What are prenatal genetic screening tests?

The tests are often identified by different labs and providers by a confusing array of names and abbreviations, but are all prenatal genetic screens. All of them involve drawing maternal blood (from mother's arm) to test for the levels of up to four different substances;

1. alpha-fetoprotein (AFP); included in all screens, but not very useful alone
2. human chorionic gonadotropin (hCG)
3. unconjugated estriol (uE3)
4. dimeric inhibin A (DIA); less important, only included in the most complex screens.

The more complex screening tests are more accurate, but also more expensive.

## What birth defects can be detected, and how common are they?

These tests help identify those pregnancies at risk for Neural Tube Defects (NTD) which are open spina bifida and anencephaly, Down Syndrome (Trisomy 21), and Trisomy 18 (Edwards Syndrome). Closed spina bifida will not be detected with screening. These tests do not screen for any other type of birth defect, so parents who have an ethnic or familial risk of other defects should seek professional genetic counseling if they desire additional testing.

- Down Syndrome occurs in about 1 in 600 births. Risks change with maternal age.
- NTD occurs in about 1 in 1000 babies in the Pacific Northwest. Nutrition affects risks.
- Trisomy 18 occurs in about 1 in 3000 live births. This is a random chromosomal defect.

## What kinds of problems do these children have?

The problems a child with spina bifida will have range from mild backaches to severe mental retardation, paralysis, bowel and bladder control problems, and leg deformities. Anencephaly is a failure of the skull and brain to fully develop, and is generally regarded as incompatible with life as there is no treatment or surgery. Down Syndrome results in varying degrees of mental retardation and an increased risk of physical defects, usually of the heart and gastrointestinal tract. Trisomy 18 results in multiple severe defects, which complicate life or cause death in early infancy.

## When are the tests done and how much do they cost?

The optimal time to do the screen is as close as possible to 16 weeks gestation but can be done between 15 and 20 weeks gestation. This time period gives the best measurement of the hormones/proteins, allows adequate time for preparation for caring for this child, and allows for a second trimester termination of the pregnancy. The costs range from about $100 to $500, depending on the lab and the complexity/quality of the test. If you have insurance or DSHS, the costs should be covered. If you have an abnormal result, further testing is usually recommended and adds several hundred more dollars to the costs. Additional testing must be done at the lab, as your midwife does not have all the necessary equipment.

## How accurate are these tests?

These are screening tests only and cannot diagnose the presence (or absence) of an NTD or trisomy or guarantee a “normal” baby. They provide only a risk-estimate for certain types of fetal disorders. If you have an abnormal screen, other more expensive and invasive tests must be done to diagnose the cause. Your pregnancy dates need to be very accurate or the test results will be meaningless. AFP-only tests are moderately accurate for NTD, but need hCG and uE3 added to be moderately accurate for trisomies.

Reasons for an abnormal screen can include; twins, an incorrect estimation of how far along your pregnancy is, a fetal demise, or a molar pregnancy. These tests detect only 75% of cases of NTD, 80% of Down Syndrome, and 60% of Trisomy 18 cases. That means that 20-40% of these defects go undetected by screening (false-negative results). Many cases are also incorrectly identified (false-positive results); in other words, even though the screen is positive, in most cases the fetus does not have these conditions and may be completely normal.

**What happens after the first screening test?**

If the test comes back normal.....No further testing.

If an AFP-only test comes back high.....First; a high-level ultrasound to detect abnormal features.
Second; a more complex test. If still abnormal then,
Third; genetic counseling and possible amniocentesis.

If a complex test comes back positive.....First; an ultrasound to detect abnormal features.
Second; genetic counseling and possible amniocentesis.

**What is an amniocentesis?**

An amniocentesis is done by inserting a large needle through the abdominal and uterine walls of the mother into the amniotic fluid surrounding the baby and withdrawing a small amount of fluid that is sent for analysis. Ultrasound is used to help the provider guide the needle away from the baby, cord and placenta. There is a chance of causing a miscarriage, infection, or other serious complications. Amniocentesis is 99% accurate for chromosomal abnormalities, but does not identify any structural birth defects (which an ultrasound may).

**What if a defect is found?**

The course of action depends greatly on what is found and the confidence we have in the test results. If any test is abnormal, the next step is more testing. Then you must decide if you wish to carry the baby to term, prepare for a birth (potentially by C-section, or at the hospital if the suspected defect precludes home birth), and caring for your baby with this disability. You will have more time to begin your research and contact helpful organizations or providers. You may also decide to terminate the pregnancy. In any of these cases, you will be referred for medical assessment and/or treatment.

**What are some drawbacks of screening?**

- Abnormal results will need to be followed with more invasive and expensive tests for accuracy
- Parents often undergo much worry and stress about the health of the baby while waiting for results
- Some women feel detached from the pregnancy/baby until the results are in
- The financial costs of initial testing and follow-up
- The tests do not identify all cases and can incorrectly identify some
- Tests are not done until 16 week of pregnancy after strong emotional feelings may have developed.

**What are some benefits of screening?**

- Testing is optional. The decision to test is entirely yours, as there is little clinical information that the midwife is likely to receive or be able to use from these tests.
- Normal test results provide reasonable assurance that your baby is free of NTDs, Down Syndrome, and Trisomy 18, especially if the correct gestational age has been used by the lab.
- It may help you decide place of birth and type of delivery, as well as plan for early care of the child
  - It allows you a small window of time to attain a second-trimester abortion of a defective fetus
  - The Inland Northwest Genetics Clinic has counselors to help with your decisions (509-458-7115).

**Informed choice**

*I have read and understand the above information, and had the opportunity to ask questions and do my own research. I understand that the time period for genetic screening is limited, as is the accuracy of the test.*

Please circle your choice: I choose to HAVE / NOT HAVE a prenatal genetic screening test.

Client _____ Date _____

Midwife _____ Date _____