Prenatal Genetic Testing

What options are available?

The prenatal tests for certain birth defects which are currently available are Screening tests and Cell-Free DNA tests. There are no tests that can predict all types of birth defects, including ultrasound.

Side-by-Side Comparison of Tests	
Screening (AFP, Quad screen or Integrated screen):	**Cell-free DNA (Harmony, MaterniT21, others):**
Maternal blood draw, or 2 blood draws and 1 ultrasound	Maternal blood draw (one)
Integrated screen requires exposing baby to ultrasound	Baby does not have to be exposed to ultrasound
Must be done early in pregnancy	May be done any time after 10 weeks
Accuracy is poor-moderate, only estimates level of risk	Accuracy is very high
Screens for Down Syndrome (Trisomy 21), Trisomy 18, and open neural tube defects.	Tests for Down Syndrome (Trisomy 21), Trisomy 18, Trisomy 13, not for open neural tube defects.
Does not detect fetal gender	Does detect fetal gender
Costs between $100-500, insurance may cover most	Costs between $25-125 for Harmony after insurance

What are the 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. The more complex screening tests, such as the Integrated Screen (which requires an early ultrasound which only measures the thickness of the nuchal translucency at the back of baby's neck) are more accurate, but also more expensive/invasive.

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.
- Most screening tests do not detect Trisomy 13, a serious random defect which affects about 1 in 10,000 babies.

What are the Cell-free DNA tests?

These tests are done with a single blood draw from the mother, any time from 10 weeks to birth. Circulating placental DNA is isolated from the blood sample and tested for aneuploidy, an indication of certain genetic anomalies in which there is an abnormal number of chromosomes; Trisomy 21 (Down Syndrome), Trisomy 18 (Edwards Syndrome), Trisomy 13 (Patau Syndrome), or a sex-chromosome abnormality. Accuracy is extremely high, without the false positives of the older screening tests which cause so much anxiety for parents. These tests do not detect open NTDs, but do detect the gender of baby.

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. Trisomies 18 and 13 result in multiple severe defects, which complicate life or cause death in early infancy.

How accurate are these tests?

Screening tests 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 based upon levels of chemicals in the mother's blood, and are moderately accurate for NTD. Your pregnancy dates need to be very accurate or the test results will be meaningless. Reasons for an abnormal screen can include; twins, an incorrect estimation of your due date, 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.

Cell-free DNA tests are highly accurate, with extremely low false-positive rates, and they can identify more specific chromosomal defects than do the screening tests. Again, they cannot detect all birth defects.

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 genetic testing?

- Abnormal results tend to be followed with more invasive and expensive tests
- 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 (some insurances resist paying for Cell-free DNA tests)
- The screening tests do not identify all cases and can incorrectly identify some (less true of the Cell-free DNA)

What are some benefits of screening?

- Testing is optional. The decision to test is entirely yours.
- Normal test results provide reasonable assurance that your baby is free of the specific defects listed, 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).

By initialing the Informed Choice Checklist, you agree to the following and select your option:

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. I understand that my insurance may not pay for Cell-free DNA testing.

(Please also circle your choice here, for your own recollection/records.)

I choose to HAVE / NOT HAVE a prenatal genetic Screening test.

I choose to HAVE / NOT HAVE a prenatal genetic Cell-free DNA test.