Atrius Health

Prenatal Testing for Chromosomal Abnormalities

There are various approaches to screening for chromosomal abnormalities such as Down syndrome. Some parents want to know all the information they can get about their baby before it is born. Some people do not want this information as it would not change the decisions they make about their pregnancy. There is no right or wrong, each parent needs to decide what is best for them.

If you choose to have screening for the most common chromosome abnormalities, please see our recommendation below – we think this plan provides information on your baby in the best way. Please discuss any questions with your clinician.

Recommended Screening

1. **Ultrasound Screening at 12-14 weeks - Nuchal Translucency**

   This ultrasound checks your due date and looks for findings sometimes associated with chromosomal abnormalities. If there are abnormalities noted at the ultrasound, a clinician will advise you about other options for testing that day. If the screening is normal, you will proceed to the lab for one of the following blood tests.

2. **Screening Blood Tests (performed AFTER the ultrasound)**

   - **Cell free DNA** tests for Trisomy 21 and Trisomy 18 and a few other of the most common chromosomal abnormalities. It will detect 99% of babies with Down Syndrome, but is less accurate for some of other abnormalities. If you request it, this test can also tell you the sex of your baby.

   - **Early Risk Assessment (ERA)** tests for the two most common chromosomal abnormalities Trisomy 21 and Trisomy 18. Five percent of patients will have a positive screen. If the test is positive you and your clinician will decide if you want further testing Often when the test is positive, the baby is still normal.

3. **Ultrasound Screen – 20 weeks – Fetal Survey/Anatomy Scan**

   We also recommend an ultrasound done at around 20 weeks gestation. The baby will be bigger and we will be able to see much more detail. If there are abnormalities noted at that time a clinician will discuss this with you and may recommend further testing including additional ultrasounds.

Diagnostic Testing

Some parents choose to have more invasive diagnostic testing: chorionic villus sampling (CVS) or amniocentesis. This can be because of a family history of abnormalities, an increased risk of a chromosomal abnormality, or because they want the most complete information available. These tests give definitive results but are associated with a small (less than 1%) risk of miscarriage

Insurance Coverage

Your insurance may cover all or some of these tests. You are responsible for finding out which tests your plan will cover and whether there are associated co-payments or deductibles.