



Screening For Genetic Problems

OB-GYN, P.C. offers the following screening options to all pregnant patients.

- These tests are OPTIONAL
- Please read this document and the additional resources given BEFORE your first visit with your doctor or midwife.
- Please call your insurance company and/or Natera to discuss cost and coverage. Information to help with this process is on the reverse side and in your packet.

Screening Your Baby

The following tests are optional during each pregnancy:

- **Panorama – available to all women, most helpful for women older than 35**
 - One blood draw anytime after 10 weeks.
 - Looks at the baby's DNA (genetic material) in your blood sample.
 - Screens for Down syndrome (Trisomy 21), Trisomy 18, Trisomy 13, sex chromosome abnormalities, and triploidy.
 - >99% detection rate* for Down syndrome with a 0.03% false positive rate**
 - We recommend another blood test in the 2nd trimester to screen for possible neural tube defects in the baby.
 - More information and FREE counseling are available at natera.com. Please visit this website if you are considering Panorama.
- **Sequential Screen – available to all women, recommended for low-risk women**
 - 2 Steps: 1) 1st trimester ultrasound and blood draw; 2) 2nd trimester blood draw
 - Part 1 must be done at the Bronson Maternal Fetal Medicine office between 10 weeks and 13 weeks pregnant.
 - Risk assessment for Down syndrome (Trisomy 21), Trisomy 18, and open neural tube defects.
 - 95% detection rate* for Down syndrome with a 0.1% false positive rate**.
 - Results may also suggest other genetic issues.

*Detection rate – How often does the test find babies who really do have the problem?

**False positive rate – How often does the test come back abnormal when everything is really fine?

Note: These tests screen for increased or decreased risk of these conditions. They cannot tell you for sure if your baby is affected. More testing would be needed to be sure. Normal Sequential or Panorama results do not guarantee a healthy pregnancy or baby. Sometimes, results are uncertain. If this happens you might need more testing.



Screening For Genetic Problems

Screening You

Genetic carrier screening is OPTIONAL blood testing that looks at **your** genes to see if you are a carrier of certain genetic conditions. If you are a carrier, you are at increased risk to have a baby with the genetic condition. At OB-GYN, P.C. we offer a test called **Horizon** for carrier screening. No test can detect 100% of genetic carriers, but Horizon will detect more than 95% of carriers in most cases.

Horizon Carrier Screen

- Horizon 4 includes testing for the following disorders:
 - Cystic Fibrosis (CF) – leads to lung problems, digestive problems, poor growth, infertility, and high risk of infection.
 - About 1 in every 45 adults is a CF carrier
 - Spinal Muscular Atrophy (SMA) – causes damage to nerve cells in the spinal cord, leading to very weak muscles; the most common form causes death by age 2.
 - About 1 in every 35-65 adults is a SMA carrier
 - Fragile X Syndrome – can cause intellectual disability, autism, and/or behavior problems.
 - About 1 in every 250 adults is a Fragile X carrier
 - Duchenne Muscular Dystrophy (DMD) – damages muscle cells leading to severe weakness; over time it damages the heart and the muscles used to breathe.
 - About 1 in every 500 adults is a DMD carrier
- Horizon offers larger screening panels that may be better for some patients based on family history and ethnicity.

****We STRONGLY recommend genetic counseling if you are considering Horizon. Natera offers this to our patients by phone for FREE.**

- This is especially important if you have family history of a genetic disease (including your children) OR if you are of Ashkenazi Jewish descent.
- Schedule at 844-706-9530 or my.natera.com/services/genetic_information.

Codes and Coverage

- Sequential – Call your insurance company to discuss coverage
 - Procedure: 76813-Ultrasound; 84163-first lab draw; 84702-second lab draw
 - Diagnosis: Z36.0
- Panorama and Horizon – Contact Natera to discuss cost, coverage and options
 - 877-869-3052 or my.natera.com/billing.