

Prenatal Genetic Screening

What is genetic screening?

Genetic screening is testing that can tell you the chances that your fetus has certain genetic disorders. These include trisomies, or three instead of the expected two chromosomes of each kind.

The three most common trisomies and the ones we can test for are:

TRISOMY 21

Downs syndrome

TRISOMY 18

TRISOMY 13

What is considered a high-risk pregnancy?

- Age at delivery of 35 or older
- Prior baby with abnormal chromosomes
- Presence of fetal abnormalities on ultrasound

Three options for genetic screening:

1. No Screening

2. Sequential Screen | 2 steps

Step 1: Ultrasound and bloodwork, 11-13 weeks 6 days

Step 2: Bloodwork, 15-22 weeks

Tests for trisomy 21 and trisomy 18, abdominal wall defects and neural tube defects (specific defects of the brain and spine)

3. NIPT | Non-invasive prenatal testing

Bloodwork, after 11 weeks

Designed for high risk pregnancies

Tests for trisomy 21, trisomy 18, trisomy 13, +/- gender

If tests need to be repeated additional charges may apply.

Detection rate

Down syndrome or Trisomy 21

20 weeks anatomy ultrasound

50-80%

Sequential screen

95%

NIPT

99%

The bottom line

The sequential screen (option 2) and NIPT (option 3) are screening tests. If you are interested, please check with your insurance. It is important to remember that having a positive result does **NOT** mean your baby has a condition. It means there is an increased risk and further testing can be done to confirm if desired.

Do we want specific contact info on each sheet as a sign off? Consistently displayed? Or is the contact information on the front supposed to “work” for all of these?