Beth Israel Lahey Health
Beth Israel Deaconess Medical Center

On behalf of BIDMC Prenatal Genetics
and Maternal-Fetal Medicine

BIDMC Prenatal Genetics Guide to Aneuploidy Screening Options

While most pregnancies are healthy, there is a small chance in any pregnancy for there to be a genetic condition or birth defect. A chromosome abnormality is one type of genetic condition. This type of condition is not usually inherited, but can happen for the first time in any fetus, just by chance. A chromosome is a package of genes or instructions. We have 46 chromosomes in each of our cells, 23 that come from the egg and 23 that come from the sperm. When an egg or sperm has an extra or missing chromosome the resulting fetus is said to have a chromosome abnormality. The most common chromosome abnormality is Down syndrome (extra chromosome #21). Some chromosome abnormalities are more severe and some are less severe than Down syndrome. Chromosome abnormalities happen more often as the pregnant patient gets older. Your provider can tell you the chance for a chromosome abnormality in your pregnancy based on your age, and will also offer you "screening tests." These are non-invasive tests that can better predict the chance for a fetus to have a chromosomal abnormality. These tests are entirely optional. Also available are invasive tests, like amniocentesis, that can tell for sure if a fetus has a chromosome abnormality and can detect a wider range of more rare chromosome abnormalities, but pose a small risk for miscarriage.

The decision about which test to have performed, if any, can be difficult and confusing. On the next page please find a chart comparing the two most common screening tests offered through BIDMC-Lahey Health. An informational booklet can also be found at https://www.bidmc.org/-/media/files/beth-israel-org/centers-and-departments/obstetrics-and-gynecology/pregnancy/screening-chromozomebooklet-horizontal201607.pdf. If you have difficulty making your decision, or you have additional questions about your testing options, a visit with the BIDMC-Lahey prenatal genetic counseling team can be scheduled through your OB/GYN provider.

	1st trimester combined screen	Cell-free DNA screening
What is it?	A combination blood and ultrasound test that estimates the chance for certain extra or missing chromosomes in the fetus by measuring the levels of certain pregnancy hormones and measuring the pocket of fluid behind the neck of the fetus (the nuchal translucency).	A blood test that estimates the chance for certain extra or missing chromosomes in the fetus by reading small pieces of the chromosomes in a person's blood that come from the placenta.
What does it test for?	• Down syndrome • trisomy 18 • trisomy 13 • Turner syndrome • non-specific more rare chromosomal abnormalities • some birth defects	• Down syndrome • trisomy 18 • trisomy 13 • Turner syndrome • extra sex chromosomes • triploidy
When is it done?	Fingerprick at ~9w to 13w6d Ultrasound at ~11w3d to 13w6d	Blood drawn from arm after 10w
Pros	• Results back quickly (2-3 days) • Always covered by insurance • Also detects some birth defects through NT ultrasound	• Very low false positive rate (<1%) • Highest detection rate for Down syndrome (>99%) • Can predict the fetal sex
Cons	• Higher false positive rate (5%) • Somewhat more likely to incorrectly call a pregnancy low risk that is actually affected with Down syndrome, trisomy 13, or trisomy 18.	• Results take longer (7-10 days) • Higher potential cost (depends on insurance) • Sometimes does not work (more often for higher BMI) • Can give an “uncertain” result or find a maternal condition • Does not detect birth defects unless provider also orders NT ultrasound
Consider if…	• You do not have increased risk due to age or history • You have high BMI • Your estimated cost for cell-free DNA analysis is high	• You will be 35 or older at delivery or had a prior pregnancy with a chromosomal abnormality • You are worried about false positive results • Your primary worry is about the chance for Down syndrome and you want the most accurate screen for this condition
Insurance CPT codes	82105, 84704, 84163	81420