Prenatal Genetic Screening and Testing Options and Consent

Many families have questions about prenatal genetic tests. This handout will help you understand available screening and diagnostic tests. For more information, please speak with your midwife, a maternal fetal medicine specialist, or a genetic counselor.

Pregnant people have options:

1. No genetic testing.

2. Non-invasive screening tests.

- ✓ **There are several non-invasive screening tests available.** These tests involve a blood draw, or a blood draw plus an ultrasound.
- ✓ **Screening tests can estimate the likelihood that your fetus has:**
  - A **neural tube defect**, or an opening in the spinal cord or brain. **Spina bifida** is an example of a neural tube defect.
  - An **aneuploidy,** or a variation in the number of chromosomes in each cell. Examples include **Down syndrome, Edwards syndrome,** and **Patau syndrome**.
- ✓ **Your results will either be "low-risk" or "high-risk."**
  - Positive results DO NOT mean your fetus has a genetic anomaly— false positives are common. If your result is "high risk," you may choose to pursue **invasive diagnostic tests.**
  - "Low risk" results do not guarantee that your fetus is unaffected– false negatives are possible.
  - The older you are, the more accurate these tests will be.
- ✓ **The fetal sex can be detected by many non-invasive screening tests.**

3. Invasive diagnostic tests.

- ✓ These tests extract fetal cells from the placenta or **amniotic fluid** inside your uterus.
- ✓ These tests almost always accurately detect **aneuploidies**. One of the tests described below can also detect **neural tube defects**.
- ✓ Unlike non-invasive screening tests, these tests can detect some **genetic disorders**, including **Tay-Sachs disease, cystic fibrosis,** and **sickle cell anemia.**
- ✓ **The fetal sex can be detected by diagnostic tests.**

Families choose or decline prenatal testing for many different reasons.

- Some families undergo testing because they would choose to terminate a pregnancy affected by certain anomalies.
- Others want to know if their child will be born with a health problem so they can plan the most appropriate place of birth, and prepare emotionally and financially.
- Others decline testing for equally valid reasons: because testing causes worry and stress, or because a positive test result would not change any of their decisions regarding the pregnancy.

There is no right or wrong choice. The midwives at BBC will support you no matter what you choose.

Some points to keep in mind as you consider your options:

- For all screening tests, there is at least 95% chance of having negative / normal results.
- All BBC clients will have an **anatomy sonogram** in their 2nd trimester. **Neural tube defects** and certain physical anomalies can usually be identified by an anatomy sonogram. This sonogram may also identify **soft markers** (possible indicators) for genetic anomalies.
- No test can guarantee the birth of a healthy baby. A negative result can be reassuring, but no test is 100% accurate. 2 – 3% of all babies are born with a physical or developmental issue. Many of these issues cannot be detected by prenatal tests.
- Prenatal genetic tests check for **chromosomal anomalies, neural tube defects,** and **certain hereditary disorders.** Most of these conditions **cannot be cured—** though some neural tube defects can be minimized with surgeries performed during pregnancy or infancy.

Non-Invasive Screening Tests

Non-Invasive Blood Tests

Brand Names:	Harmony, Panorama, Verifi, MaterniT21, Counsyl, ClariTest
Screens For:	Trisomy 13 (Patau syndrome) Trisomy 18 (Edwards syndrome) Trisomy 21 (Down syndrome) Fetal sex, the number of X and Y chromosomes **Some tests screen for other rare chromosomal anomalies.**
When It's Done:	Most non-invasive blood tests can be performed when you are 9 – 10 weeks pregnant and beyond.
How It's Done:	The pregnant person undergoes a blood draw. Pieces of the fetal DNA are found and analyzed.
Accuracy:	99% sensitive for Trisomy 21 96% sensitive for Trisomy 18 90% sensitive for Trisomy 13 and Monosomy X Less than 1% "false positive" rate for all conditions
Advantages:	Non-invasive Accurate In rare cases, these tests detect a chromosomal anomaly or cancer in the pregnant person.
Disadvantages:	Does not test for neural tube defects and other disorders May not be covered by insurance unless a risk factor is present Less accurate if the pregnant person is of high weight Not recommended for twin pregnancies 4 – 8% of tests do not provide a result

First Trimester Combined Screening

Screens For:	Trisomy 18 (Edwards syndrome) Trisomy 21 (Down syndrome)
When It's Done:	10 weeks, 5 days – 13 weeks gestation
How It's Done:	The pregnant person undergoes a blood draw. Blood is analyzed for levels of 2 hormones: PAPP-A (pregnancy associated plasma protein A) and beta hCG (human chorionic gonadotropin). An ultrasound is also performed. The sonographer and provider analyze the size of the nuchal fold on the back of the fetal neck.
Accuracy:	85% sensitive for Trisomy 21 and Trisomy 18 5% "false positive" rate
Advantages:	Non-invasive Commonly available and covered by most insurance plans Can be done in combination with an early ultrasound for dating
Disadvantages:	Requires both blood draw and ultrasound Only detects Trisomy 18 and Trisomy 21 Less sensitive than other available tests

Sequential Screening

Screens For:	Trisomy 18 (Edwards syndrome) Trisomy 21 (Down syndrome) Neural tube defects Abdominal wall defects
When It's Done:	11 – 21 weeks gestation
How It's Done:	The pregnant person undergoes two blood draws— one at 11 – 13 weeks and one at 15 – 21 weeks. Blood is analyzed for PAPP-A (pregnancy associated plasma protein A), beta hCG (human chorionic gonadotropin), and Inhibin. An ultrasound is also performed on the day of the first blood draw. The sonographer and provider analyze the size of the nuchal fold.
Accuracy:	94% sensitive for Trisomy 21 93% sensitive for Trisomy 18 5% "false positive" rate
Advantages:	May detect rare anomalies not detected by blood testing alone. Hormones analyzed are associated with certain pregnancy complications, and may signal a need for closer monitoring.
Disadvantages:	Must have two blood draws at specific times in the pregnancy, and ensure they are processed by the same laboratory

Alpha-fetoprotein (AFP)

Screens For:	Neural tube defects Abdominal wall defects
When It's Done:	15 – 21 weeks, 6 days gestation Often offered in conjunction with first trimester screen / NIPT
How It's Done:	The pregnant person undergoes a blood draw. Blood is analyzed for alpha-fetoprotein.
Accuracy:	75% sensitive for spina bifida 90% sensitive for anencephaly
Advantages:	Commonly available and covered by most insurance plans Useful option if screening was not done earlier in pregnancy
Disadvantages:	Does not test for any chromosomal anomalies Less sensitive than other available tests

Quad Screening

Screens For:	Trisomy 18 (Edwards syndrome) Trisomy 21 (Down syndrome) Neural tube defects
When It's Done:	15 – 22 weeks gestation
How It's Done:	The pregnant person undergoes a blood draw. Blood is analyzed for alpha-fetoprotein, hCG (human chorionic gonadotropin), Estriol, and Inhibin-A.
Accuracy:	85% sensitive for Trisomy 21 with a 5% false positive rate 60% sensitive for Trisomy 18 with a 0.2% false positive rate 79% sensitive for spina bifida with a 3% false positive rate 88% sensitive for anencephaly
Advantages:	Useful option if screening was not done earlier in pregnancy
Disadvantages:	Less sensitive than other available tests

Invasive Diagnostic Tests

Chorionic Villus Sampling

Tests For:	Trisomy 13 (Patau syndrome) Trisomy 18 (Edwards syndrome) Trisomy 21 (Down syndrome) Many **hereditary disorders**, including **Tay-Sachs disease, cystic fibrosis,** and **sickle cell anemia—** specific disorders are tested upon request based on your family history or carrier status.
When It's Done:	10 – 13 weeks gestation
How It's Done:	A needle is inserted through the pregnant person's cervix or abdomen. The needle is used to extract cells from the **chorionic villi**, or tiny fingerlike projections on your placenta.
Accuracy:	Nearly always diagnoses specified anomalies.
Advantages:	Diagnoses anomalies, rather than just indicating risk status Can be used to diagnose certain hereditary disorders
Disadvantages:	Does not test for neural tube defects Small risk of miscarriage (0.5% to 0.002%), infection, fetal injury Takes several weeks to obtain results

Amniocentesis

Tests For:	Trisomy 13 (Patau syndrome) Trisomy 18 (Edwards syndrome) Trisomy 21 (Down syndrome) Neural tube defects Many **hereditary disorders**, including **Tay-Sachs disease, cystic fibrosis,** and **sickle cell anemia—** specific disorders are tested upon request based on your family history or carrier status.
When It's Done:	16 – 20 weeks gestation
How It's Done:	Under ultrasound guidance, a needle is inserted through the pregnant person's abdomen. The needle extracts **amniotic fluid**, which contains fetal skin cells. These cells are analyzed for DNA.
Accuracy:	Nearly always diagnoses specified anomalies
Advantages:	Diagnoses anomalies, rather than just indicating risk status Can be used to diagnose certain hereditary disorders
Disadvantages:	Small risk of miscarriage (0.5% to 0.002%), preterm labor, premature rupture of membranes, infections, fetal injury Takes several weeks to obtain results

Review of Genetic Screening from Client Record:

Clients who transfer their care to the Brooklyn Birthing Center during or after their second trimester often have already completed some genetic testing with their previous provider. Below we have listed the tests (if any) that you have already performed.

The following genetic tests have already been completed and the results are as listed.

	Non-Invasive Prenatal Test	
	First Trimester Screen	
	Sequential Screen	
	Alpha-fetoprotein (AFP)	
	Quad Screening	
	Chorionic Villi Sampling (CVS)	
	Amniocentesis	

Consent / Refusal for (Additional) First and / or Second Trimester Testing:

- I have read the information provided on first and second trimester screening and diagnostic tests. I understand the risks, benefits, and alternatives of testing.
- I have had the opportunity to ask questions regarding testing options. I understand how those options have been explained to me.
- I understand that the tests outlined above do not guarantee that I will have a fetus or baby who is affected or unaffected by a birth defect, and that there is a chance that screening results may not be accurate.
- I do not hold the Brooklyn Birthing Center or its midwives responsible for test accuracy or risks of testing, detection, or the recommendations of the consulting provider(s).
- I understand that if I have an abnormal screening result, I will be referred to a perinatologist for further testing. It is up to me to schedule that appointment during the proper time frame, consent to, or decline further testing.
- In the event of abnormal screening results, Brooklyn Birthing Center Midwifery Group may release my lab results, prenatal records, and imaging reports to their perinatal consultants for the purposes of further assessment.
- I understand that I am responsible for contacting my insurance company to find out if first and / or second trimester screening is a covered benefit. I am also responsible for finding out if perinatal consultation and any / all testing they recommend is covered by my insurance.
- My testing preferences are initialed below:

_____ I consent to the following screening or diagnostic tests:

_____ I decline first and second trimester screening. I understand that if my anatomy scan or another test reveals potential fetal anomalies, non-invasive blood tests and amniocentesis will still be available and potentially recommended.

_____

Signature of client Date

Glossary

Abdominal wall defect (AWD), such as **gastroschisis** and **omphalocele**, are rare, occurring in about 1 out of every 5,000 births. Pregnant teens are about 4 times more likely to have a baby with an AWD compared to pregnant people over 20. Incidence is higher in the Midwest, possibly due to a herbicides used in industrial agriculture.

Neural tube defects (NTDs) are conditions in which an opening in the spinal cord or brain remains after early fetal development. Specific types include: **spina bifida, anencephaly,** and **encephalocele.** NTDs are among the most common birth defects. Spina bifida affects about 0.035% of babies born in the US.

Sex chromosome aneuploidies have a wide range of expression. Examples include **Monosomy X** (**Turner syndrome), Triple-X syndrome, Klinefelter syndrome,** and **XYY syndrome.** However, many sex chromosome aneuplodies are so mild they might not otherwise be diagnosed except via prenatal screening / testing.

Trisomy 13, or Patau syndrome is caused by an extra copy of chromosome 13. Affected babies often have heart problems, brain or spinal cord abnormalities, and other congenital anomalies such as small eyes, cleft palate, and weak muscle tone. Many affected babies die before birth. For babies born alive, treatment is supportive. About 5 – 10% of affected babies survive beyond the first year of life.

Trisomy 18, or Edwards syndrome is caused by an extra copy of chromosome 18. Affected babies are often born small, with heart defects. Other features include a small head, small jaw, clenched fists with overlapping fingers, and severe intellectual disability. Many affected baby die before birth. For babies born alive, treatment is supportive. About 7.5% of affected babies survive beyond the first year of life.

Trisomy 21, or Down syndrome is caused by an extra copy of chromosome 21. Down syndrome is one of the most common chromosomal anomalies. It is associated with multiple health issues, physical growth delays, and mild to moderate intellectual disability. The possibility of having a baby affected by Down syndrome increases from less than 0.1% in 20-year-old pregnant people to 3% in 45-year-olds. 61-97% (depending on population evaluated) terminate their pregnancy once this prenatal diagnosis is made. Non-invasive tests can indicate whether a pregnancy is “high risk,” but amniocentesis is required for definitive diagnosis.

For more information, please explore the following websites:

American College of Obstetricians and Gynecologists: ACOG.org
American Pregnancy Association: americanpregnancy.org
Centers for Disease Control and Prevention: CDC.gov