

The Perinatal Centers

SIDE 1: Summary of Genetic Carrier Testing Options

This page (front and back) lists the most common carrier tests and chromosome testing options offered in our center. Please review the lists and complete the information at the bottom of each side of this sheet. Keep in mind that **these tests are optional**. This sheet is for information purposes only.

All individuals have a chance of being a carrier for a genetic condition. Carriers usually do not have symptoms of the condition and both parents often need to be carriers in order to have a child with the condition. Couples typically have no family history of the condition.

1. **Alpha- and Beta-Thalassemia** - People with thalassemia may have weakness, a large spleen, bone and heart problems, and slower growth. These conditions are more common in individuals of African American, Mediterranean, Hispanic or Asian descent.
2. **Cystic Fibrosis (CF)** - People with CF have problems with breathing, digestion, and fertility. It is a life-limiting condition. There is no known cure for CF, but there is lifelong treatment. This condition is more common in individuals of Caucasian and Jewish ancestry.
3. **Fragile X syndrome (FXS)** - People with FXS may have severe learning problems, autism, fertility problems, and tremors. This condition can affect all ethnicities.
4. **Sickle Cell Anemia (SCA)** - People with SCA have problems with joint and bone pain, anemia, and organ damage. SCA is more common in individuals of African American, Mediterranean and Hispanic descent.
5. **Spinal Muscular Atrophy (SMA)** - People with SMA have problems with their muscles and development. Babies with SMA typically die between 2-4 years of age. This condition can affect all ethnicities.
6. **Jewish Carrier Screening** – Some conditions, such as Tay Sachs disease, are more common in individuals of Jewish descent. Children with Tay Sachs have problems with development and pass away by 2-3 years of age.
7. **Expanded Carrier Screening** – This test looks for 85 genetic conditions, including most of those listed above.

Please check one of the following:

- I would like more information about the following tests (list numbers): _____
- I would like genetic counseling to discuss appropriate testing options in greater detail.
- I am not interested in further information about the above tests at this time.

➔➔ See Other Side ➔➔

HCP Reviewer Signature _____ Date _____ Time _____



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SIDE 2: Summary of Chromosome Testing Options

All pregnant couples have a chance of having a baby with a birth defect or chromosome condition, but the chance increases as a woman gets older. Examples of chromosome conditions include Down syndrome, trisomy 18 and trisomy 13. People with Down syndrome can have problems with their heart, developmental delays, and learning problems. Trisomy 18 and Trisomy 13 are conditions that cause severe complications and most babies do not survive past one year of life. Couples typically have no family history of these conditions.

- A. **Screening for chromosome conditions** - These tests give a probability, or chance, that your baby could have a chromosome condition. They involve a blood sample and possibly an ultrasound. There are many options that differ by detection rate, number of conditions tested, gestational age, and other factors.
- B. **Chorionic Villus Sampling (CVS)** - This test is done between 11 – 14 weeks of pregnancy. This test will determine whether your pregnancy has a chromosome condition or other genetic conditions. It involves placing a needle through the abdomen, or a tube through the cervix, to take a sample of the placenta. This test is diagnostic, but carries a small risk for complications that may include miscarriage.
- C. **Amniocentesis** - This test is done between 15 – 23 weeks of pregnancy. This test will determine whether the pregnancy has a chromosome condition or other genetic conditions. It involves inserting a needle through the abdomen to get a sample of the fluid that surrounds the baby. This test is diagnostic, but carries a small risk for complications that may include miscarriage.

Please check one of the following:

- I would like more information about the following tests (list letters): _____
- I would like genetic counseling to discuss appropriate testing options in greater detail.
- I am not interested in further information about the above tests at this time.

These tests are optional. Coverage of these tests varies based on your insurance policy. We can provide you with information you may need to check with your insurance carrier for coverage.

The Perinatal Center physician may refer you for genetic counseling if ultrasound findings, family history, or test results warrant the service.

Patient's Signature _____ Date _____

HCP Reviewer Signature _____ Date _____ Time _____



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