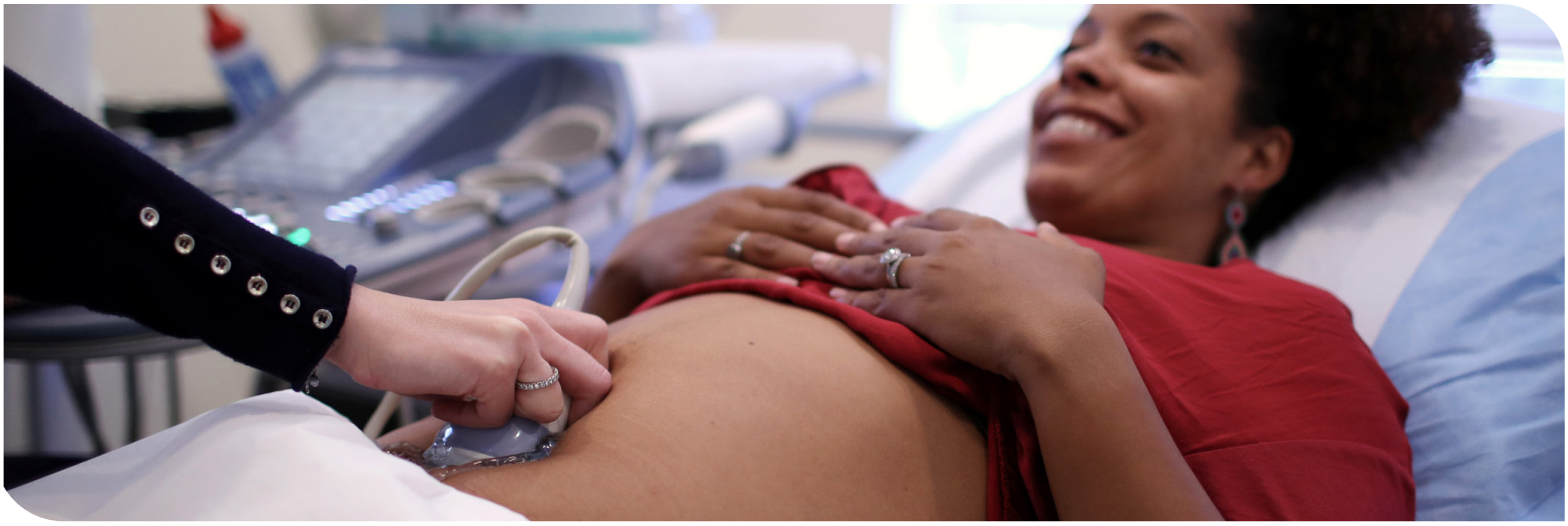
Prenatal Visits & Screenings



Prenatal Testing Timeline

In an effort to keep you informed and involved in your care, we have provided a timeline of prenatal testing that will be offered to you throughout your pregnancy. While this is a standard list of testing, please remember each pregnancy is unique and may require additional testing.

GESTATIONAL AGE | 6 – 12 WEEKS

- Prenatal profile – including blood type, initial blood count, Hepatitis B, Rubella, RPR and HIV
- Gonorrhea and Chlamydia testing
- Urine culture
- Pap smear – if you are due
- Early ultrasound – will confirm viability and take measurements for a final due date

These are nationally recommended tests; they will help your providers offer appropriate surveillance to you and your newborn.

GESTATIONAL AGE | 20 – 22 Weeks

- Anatomy ultrasound – structural survey of the fetus

If your pregnancy has been deemed high risk, you will be referred to our Maternal Fetal Medicine department for a Level II ultrasound.

GESTATIONAL AGE | 24 – 28 Weeks

- Repeat blood count
- Repeat RPR
- Gestational diabetes screen

GESTATIONAL AGE | 36 – 40+ Weeks

- Group Beta Strep (GBS) culture – performed once around 36 weeks
- Cervical exams

Genetic Screening is an option for all of our obstetric patients. Please see our Genetic Testing FAQ flyer for testing options, time lines and CPT codes for insurance purposes.

Screening Overview for Chromosomal Trisomies & Neural Tube Defects

You may choose to have special tests done to screen for a chromosomal problem or neural tube defect during pregnancy. These tests are not required, but they are offered by your Duly physician. Tests screen for neural tube defects which are developmental problems of the brain, spine and spinal cord, (such as spina bifida or anencephaly) as well as for chromosomal disorders caused by an abnormality in the number of chromosomes. For example, a person has 47 chromosomes instead of 46. This is called Trisomy.

Commonly screened trisomies include:

- Trisomy 21 (Down Syndrome)
- Trisomy 13 (Patau Syndrome)
- Trisomy 18 (Edwards Syndrome)

TYPE OF SCREENING	SCREENS FOR	OFFERED TO	TIMING	KIND OF TEST	CPT/ BILLING CODE
First Trimester Screen	Trisomy 21 and Trisomy 18	Women younger than 35, at low risk to have a baby with a chromosomal problem	Between 11 3/7 and 13 6/7 weeks	Ultrasound and Blood test	76813, 36415, 81508, 76801
Cell-Free DNA	Trisomy 21, Trisomy 13, and Trisomy 18. Can also determine fetal gender	Women 35 and older and women with a high-risk first trimester screen	After 10 weeks	Blood test	81420
Alfa Fetoprotein (AFP)	Neural tube defects including spina bifida and anencephaly	All pregnant women	15-18 weeks	Blood test	82105, 36415
Quad Screen (MSAFP)	Neural tube defects including spina bifida and anencephaly, Trisomy 21 and Trisomy 18	Women who did not get a first trimester screen or cell-free DNA	15-18 weeks	Blood test	36415, 81511
Cystic Fibrosis (CF) Carrier	Detirmination of whether parents are carriers of Cystic Fibrosis	All pregnant women and their partners	In the early stages of pregnancy, or prior to pregnancy	Blood test	81220, 36415

Screening Overview for Chromosomal Trisomies & Neural Tube Defects

Types of Screenings

FIRST TRIMESTER SCREEN

Although a routine ultrasound may detect signs of a chromosomal trisomy or neural tube defect, it will not always identify these conditions. This is why some women elect to have a first trimester screen to gain as much information as possible about their pregnancy in order to make the best health decisions (in consultation with their Duly physician) for themselves and their babies.

For example, babies with Trisomy 21 (Down syndrome) are at an increased risk for a variety of health problems including some heart defects. When a diagnosis is made prior to birth, a specialized care plan can be developed for those health needs after delivery.

CELL-FREE DNA TESTING

As women get older, the risk of having a baby with a chromosomal trisomy increases. The risk of having a baby with a chromosome problem at:

- Age 20 years the risk is 1 in 525
- Age 30 years the risk is 1 in 385
- Age 35 years the risk is 1 in 200
- Age 40 years the risk is 1 in 65

Women younger than 35 when they deliver are usually offered a first trimester screen. Women older than 35 when they deliver are usually offered cell-free DNA testing. The first trimester screen is a blood test and also an ultrasound. The cell-free DNA test is only a blood test.

ALFA FETOPROTEIN (AFP)

The risk of neural tube defects does not change significantly with age. Women who want a screening test for neural tube defects are generally offered an Alfa Fetoprotein (AFP) test in the second trimester of pregnancy.

QUAD SCREEN

Women who want to test for a chromosomal problem in the second trimester instead of completing testing in the first trimester can get a Quad Screen blood test. It identifies trisomies as well as neural tube defects and is called Maternal Serum Alfa Fetoprotein (MSAFP).

What is the First Trimester Screening?

The first trimester screen is 85-90% effective in screening for Trisomy 21 and Trisomy 18. These two chromosome abnormalities account for approximately 50% of chromosome abnormalities seen at birth. This test is generally offered to women who are considered low risk for having a baby with a chromosomal problem. The first trimester screen is usually offered to women who will be younger than 35 when they deliver their babies.

THE FIRST TRIMESTER SCREEN HAS TWO PARTS:

1. Obtaining blood spots (finger stick or blood draw) on a sample card
2. Ultrasound measurements of:
   - *The fetal length (crown rump length)*
   - *The thickness of an area in the fetal neck*

These measurements and samples must be obtained when a fetus is between 11 weeks, 3 days to 13 weeks, 6 days. The results are then combined with the mother's age in a computer program to estimate the risk for the two chromosome abnormalities (Trisomy 21 and Trisomy 18). These risks are then given to the parent(s) for review (e.g., 1 in 50, 1 in 10,000). A 1 in 300 risk is used as the cutoff for a high-risk result.

WHAT FIRST TRIMESTER SCREENING DOES PROVIDE

An early method of determining risk for the most common chromosome abnormalities:

- Can be obtained at 11 weeks
- Screens for Trisomy 21 and Trisomy 18 which
- make up about 50% of chromosome abnormalities at birth
- An effective method of screening for Trisomy 21 and Trisomy 18 with a detection rate of 85-90%
- A method for effectively screening all pregnancies regardless of maternal age

WHAT FIRST TRIMESTER SCREENING DOES NOT PROVIDE

- It is NOT A DIAGNOSIS
- It does NOT say the fetus has a chromosome abnormality
- It does NOT say the fetus does not have a chromosome abnormality
- It does NOT screen for all chromosome abnormalities
- Only Trisomy 21 and Trisomy 18
- 50% of chromosome abnormalities seen at birth are NOT looked for by this test
- It does NOT screen for Spina Bifida
- It does NOT screen for organ or structural abnormalities (e.g. heart, kidney, gastrointestinal, brain, lung arm, leg etc. abnormalities)

Cell-Free DNA Test & Second Trimester Screening

WHAT IS THE CELL-FREE DNA TEST?

The cell-free DNA test is a blood test that is used to screen for Trisomy 21, Trisomy 13, and Trisomy 18. It can also be used to screen for sex chromosome abnormalities. It is generally recommended as a screening test for women who will be 35 or older when they deliver. It can also be used as a follow-up test for an abnormal first trimester screen.

The cell-free DNA test is a blood test that looks at DNA from the baby and the baby's placenta in the mother's blood. It is drawn just like any other blood test. It can be drawn any time in the pregnancy after 10 weeks.

When a cell-free DNA test is done in women who are at increased risk for a chromosomal problem, for example, women older than 35 or women with a high-risk first trimester screen, it is very likely to detect a chromosomal problem if there is one (90 to 99%). There are some false positives with this test, but the rate is low.

It is not always possible to determine if there is a chromosomal problem when this test is done. Up to 5% of women will have a test failure, usually because there is not enough fetal DNA in the maternal blood sample to make a diagnosis.

The accuracy of this test can be effected by maternal weight. Women who weigh more than 180 pounds have a higher risk of test failure and also a higher risk of false negative results. It is possible to find out the baby's gender with this test by looking for presence or absence of the Y chromosome.

WHAT IS THE SECOND TRIMESTER SCREEN?

In the second trimester, a blood test can be done to screen for neural tube defects like spina bifida and anencephaly. It is also possible to add screening for Trisomy 21 (Down syndrome) and Trisomy 18 (Edwards syndrome) if a first trimester screen has not already been done. This is a blood test that can be done between 15 and 20 weeks gestation.

The Alfa Fetoprotein (AFP) test screens for neural tube defects only. The Quad Screen looks for neural tube defects and also Trisomy 21 and Trisomy 18. The detection rate for this test is 80% for Trisomy 21 (Down syndrome) and for neural tube defects. The detection rate for Trisomy 18 is 60-80%.

The reporting of these tests is similar to the first trimester screen. It does not tell you if the baby does or does not have a chromosomal problem. Rather, it is reported as high-risk or low-risk, with a statistical risk like 1 in 1,000, 1 in 250, 1 in 10, etc.

If the test is done and the result is high-risk, additional testing is recommended. Recommended follow-up tests may include a cell-free DNA test, a detailed ultrasound, or an amniocentesis.

What Types of Birth Defects Can Be Found by the Quad Screen?

Down Syndrome

Babies with Down syndrome are born with an extra 21st chromosome. This causes mild to moderate cognitive impairment, specific facial features, and sometimes physical problems, such as heart defects. About half of all babies born with Down syndrome will live to at least age 50.

Trisomy 18

Babies with trisomy 18 have an extra 18th chromosome. This causes multiple physical problems and severe cognitive impairment. Most babies with trisomy 18 do not survive the first year of life.

Open neural tube defects (ONTDs)

Spina bifida and anencephaly are the most common ONTDs. When a baby is born with spina bifida, part of the bone covering the spinal cord does not form correctly and the spinal cord is exposed. Surgery is needed to close the opening.

Spina bifida can cause problems ranging from bowel and bladder control difficulties to paralysis of the legs, hydrocephalus (fluid on the brain) and learning disabilities. Anencephaly occurs when the fetal skull and brain do not develop. Babies with anencephaly cannot survive.

How reliable is the quad screen at finding birth defects?

BIRTH DEFECT	DETECTION RATE
Down syndrome	80%
Trisomy 18	60-80%
Neural tube defects	80%

Deciding to Have a Screening Test for Down Syndrome

duly HEALTH AND CARE

Reasons to consider a screening test

- I want as much information as possible during pregnancy about the health of my developing baby. If my baby has Down syndrome, I want to know while I am pregnant so I can learn as much as possible about the condition before the baby is born.
- I am planning to deliver my baby in a community hospital. If my baby has serious birth defects associated with Down syndrome (e.g., heart or intestinal abnormalities), I would rather deliver at a hospital with a special care nursery.
- I have been anxious since I learned I was pregnant and if I find out that my baby's risk of having Down syndrome is low, I believe it will help ease my anxiety.
- I want to consider all of my options. If my developing baby has Down syndrome, I would want the option to terminate the pregnancy.
- I am not sure what I would do, or how I would feel if my baby has Down syndrome. I am going to take it one step at a time. If my screening test comes back saying I am at increased risk, I will decide at that time if I want to have more testing.

Reasons to choose not to have a screening test

- I have decided that "whatever will be, will be," and I will wait until the baby's birth to find out if the baby is healthy.
- I do not want to be faced with decisions about my unborn baby. Because of religious or personal beliefs, I would never consider terminating an affected pregnancy.
- Because I am certain I would never have a diagnostic test, even with only a small risk of miscarriage, I do not want to have a screening test.
- I want to know for sure if the developing baby has Down syndrome, so I am having a diagnostic test (e.g. chronic villus sampling (CVS) or amniocenteses) rather than a screening test.

Some common myths about screening for Down syndrome

Some of the reasons women decide whether or not to have screening are based on incorrect information, such as:

MYTH My baby won't have Down syndrome because I am young, I exercise, and I am healthy.

FACT A woman of any age can have a baby with Down syndrome, regardless of her health.

MYTH My baby won't have Down syndrome because I do not drink or smoke.

FACT Avoiding alcohol or tobacco during pregnancy is very important for the health of you and your baby; however, it does not affect the chance that your baby will have Down syndrome.

MYTH My baby won't have Down syndrome because no one in my family or the father of the baby's family has Down syndrome.

FACT Down syndrome usually does not run in families. Your baby can be affected even if there is no one else in the family with Down syndrome. If you have a family history of Down syndrome, you should talk to your doctor, nurse, or a genetic counselor to discuss if it will increase your risk of having a baby with Down syndrome.

MYTH I shouldn't have a screening for Down syndrome unless I know that I would terminate the pregnancy if Down syndrome were detected.

FACT Many people who would not terminate their pregnancy choose to have screening. These people want information about their unborn baby's health before birth in order to plan for delivery and newborn care.

MYTH My friend told me that if I have a screening test, it will come back "positive" since most people who have the test end up with a "positive" result.

FACT Most people who have a screening test will have a "negative" result, meaning that the baby has a low risk of having Down syndrome.

Source: Updtodate.com, 2017.

Frequently Asked Questions About Chromosome Screening

Are any of these tests required during my pregnancy?

None of these tests are required to complete during your pregnancy. At your visit, your provider will discuss the different options with you. If you are at an increased risk for any of these disorders, the doctor may recommend that you complete one or multiple tests.

Are these tests covered by my insurance?

Because we care for patients with a variety of insurance providers and plans, our office staff does not know what these tests will cost for each individual patient. It is the patient's responsibility to get in contact with their insurance provider to find out coverage. They will ask for the CPT/ billing codes for the specific test and will then let you know whether or not they are covered and what may be required to pay out of pocket.

What if my insurance company requires a letter of approval from my doctor's office?

If your insurance company requires a letter of approval in order for the genetic screening to be covered, please contact your OB office. Although we cannot write a letter stating that these tests are REQUIRED, we can write a letter stating why the testing was RECOMMENDED for you. This letter is not a guarantee that these tests will be covered for you.

Please keep in mind, that if you are completing the cell-free DNA test with fetal sex, the fetal sex is not required or recommended by our providers, so these letters will not apply to that portion of the test.

Does a normal test result guarantee that my pregnancy does not have a birth defect?

No. These tests do not detect every case of Trisomy 21, Trisomy 18, Trisomy 13, or neural tube defects. Additionally, all pregnancies have a 2 to 3 percent background risk of having a birth defect. These tests screen for the most common chromosomal defects and the alfa fetoprotein test screens for neural tube defects, but these tests do not screen for all birth defects.

My first or second trimester screen came back as "abnormal." What does this mean?

Most pregnancies that have abnormal test results are actually normal pregnancies. False positives occur because screening tests are designed to identify women who are at increased risk to have a baby with certain birth defects. These screening tests are not diagnostic tests. A positive screening test result does NOT mean that your baby has a birth defect; only that he/ she is at increased risk of having one.

What is recommended when a test result is abnormal?

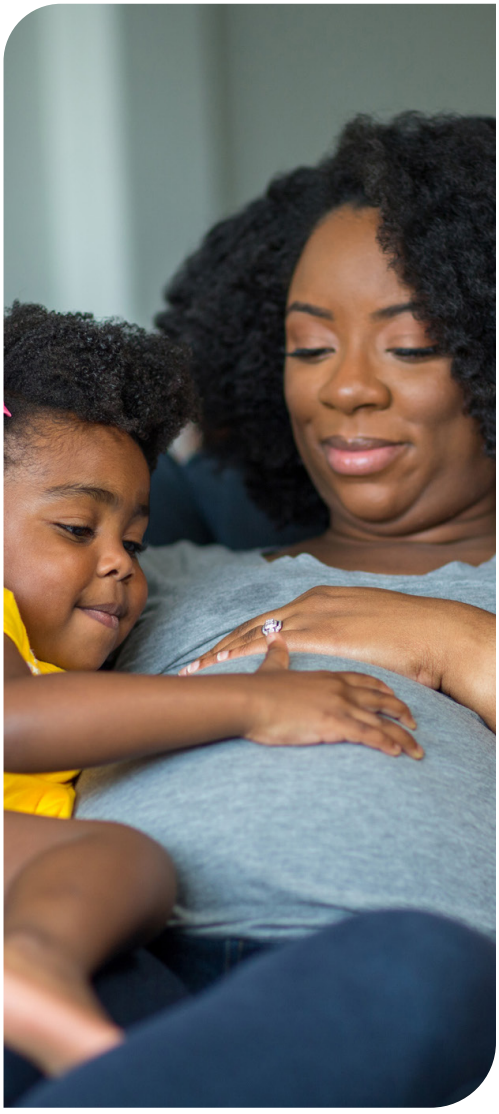
Your doctor or genetic counselor will discuss additional testing that can be done to determine if your baby does or does not have a birth defect. Most often, a detailed ultrasound is recommended. Cell-free DNA or amniocentesis may be offered.

Cell-free DNA is also a screening test, but one that is more sensitive and which has a very low false positive rate. It involves only a blood draw. Amniocentesis involves testing a small amount of the fluid surrounding the baby and allows the laboratory to directly examine the baby's chromosomes to accurately make the diagnosis.

What happens if the follow-up tests show that the fetus has a birth defect?

If a birth defect is detected, you will be given as much information as possible about the condition.

Several options may be available, including increased surveillance of the pregnancy, arrangements for special care at delivery and/or after the baby is born, or discontinuation of the pregnancy. Your doctor or genetic counselor can discuss your test results and options with you.



Gestational Diabetes Screening

Some women develop diabetes or high blood sugar when they are pregnant. It is important to test for diabetes and initiate treatment, if necessary, to prevent complications to the mother and baby. All women receiving prenatal care are screened for gestational diabetes between 26 and 28 weeks. The test is called the One Hour Glucola Test or Gestational Diabetes Screen.

WHAT TO EXPECT AT YOUR GESTATIONAL DIABETES SCREENING

- This test is not fasting
- You do not need an appointment for this test
- You need at least one hour of time for this test
- You will be asked to drink a sweet liquid
- You must remain in the lab doing quiet activities such as reading until your blood is drawn
- After 1 hour, your blood will be drawn and tested for glucose
- You will be notified if your glucose level is elevated and follow-up instructions will be given
- If ELEVATED you will need to fast for a Three Hour Glucose Tolerance Test to diagnose gestational diabetes. An appointment is required.
- If you are known to be RH Negative we will ask you to schedule an appointment to have a Rhogam injection at this time. This appointment should occur 0-5 days after your blood has been drawn.

What is cystic fibrosis?

Cystic fibrosis (CF) is a genetic condition that affects approximately 30,000 people in the United States.

One in 2,500 Caucasian newborns has CF. Affected individuals have frequent lung infections and difficulty absorbing nutrients from food. Cystic fibrosis does not affect intelligence or appearance, but is usually diagnosed in infancy due to pneumonia and/or poor growth.

Is there a cure for CF?

There is no cure, but treatment improves the length and quality of life of affected individuals by reducing lung damage and optimizing nutrition. CF symptoms and disease severity vary from patient to patient. The average life expectancy is now approximately 38 years.

Digestive problems are treated with daily vitamins and enzymes taken with each meal. Inhaled bronchodialators are utilized to maintain adequate airflow, and chest physical therapy sessions are needed daily to help clear mucous from the lungs and avoid respiratory infections. Even with careful treatment, lung infections can develop, requiring antibiotics, aerosol inhalants, and hospitalization. Lung infections usually worsen over the course of life due to antibiotic resistant bacteria.

Treatments and hospital visits are costly. There is hope that life expectancy will increase through new medications that are targeting the specific defect (mutation) in the CF gene for treatment. The leading cause of death in individuals with CF is respiratory failure from progressive lung damage.

Can my children have CF even if it is not in my family?

Yes. In fact, most couples who have a child with CF have no family history of cystic fibrosis and are surprised to learn that they carry a mutation in the CFTR gene, which causes the condition. Genes are the basic hereditary units determining an individual's traits, such as hair and eye color. CF carriers inherit a single nonfunctional gene from one parent, along with a functional gene from their other parent. Carriers of CF usually have no symptoms, as they have one normal copy of the gene. Both parents must be CF carriers to have an affected child.

Who should consider carrier testing?

An estimated 10 million people in the United States are carriers of cystic fibrosis. Individuals with a relative who is affected with or a carrier of CF should consider testing. The American College of Medical Genetics and the American College of Obstetrics and Gynecology recommend that carrier screening be offered to women who are pregnant or planning a pregnancy and their partners.

Your chance of being a CF carrier depends on your ethnic background, unless someone in your family has CF or is a carrier. Assuming you and your partner are from the same ethnic group and have no family history of CF, the chart below shows the estimated chance of having a child with CF before testing, the test's detection rate, and the chance of having a child with CF if one partner has a normal test result.

Some couples may decide against carrier testing if their ethnic group has a low risk of having a child with CF or because the test's detection rate is low for their ethnicity. Other individuals may forego testing due to lack of insurance coverage or the potential anxiety it may cause.

Whether or not to have CF-carrier testing is a complex personal decision. Some couples may decide to undergo carrier testing if they belong to an ethnic group with a higher chance of having a child with CF. Others may want to learn as much as possible about the health of their future child. The majority of couples undergoing testing will be reassured that their chance of having a child with CF is low. Screening identifies CF-carrier couples who may then make informed decisions about pregnancy options, including prenatal diagnosis.

Ethnicity	Chance of child with CF before test	CF carriers detected	Chance of CF child after normal test result in one parent
Ashkenazi Jewish	1 in 2,300	96%	1 in 53,000
Caucasian	1 in 2,500	92%	1 in 30,000
African-American	1 in 15,100	78%	1 in 67,000
Hispanic-American	1 in 13,500	80%	1 in 66,000
Asian-American	1 in 35,100	55%	1 in 79,000

Spinal Muscular Atrophy (SMA)

What is Spinal Muscular Atrophy (SMA)

Between 10,000 and 25,000 people in the United States live with SMA. This genetic disorder affects the nervous system – impacting muscles used for breathing, swallowing and walking. This is caused by the muscles getting smaller and weaker overtime.

Other complications include:

- **Bone fractures**
  Mobility equipment can support daily activity
- **Malnutrition**
  Some individuals with SMA benefit from a feeding tube
- **Weakened lungs**
  Breathing treatments can help strengthen the lungs
- **Respiratory infections**
  It is extra important for individuals with SMA and their caregivers to get vaccinated against flu and pneumonia.

There are four types of SMA:

TYPE I
Occurs at birth and is the most common type of SMA. Type I is present at birth or becomes apparent within their first six months when they do not meet milestones.

TYPE II
Occurs in children whose nervous system has impacted their daily life. Often Type II children have a shorter lifespan due to their condition.

TYPE III
Occurs in children whose nervous system has not significantly impacted their physical abilities.

TYPE IV
Is a rare adult form that slowly progresses in adulthood.

What causes SMA?

SMA occurs when the SMN1 gene on chromosome 5 is mutated. If both parents carry a mutated gene on their chromosome 5, the risk of their child having SMA is 25%. This risk does not decrease with each pregnancy.

Is there a cure for SMA?

There is no cure, but treatment can help manage symptoms. Research has found that a combination of SMN-based and non-SMN treatments have been the most successful.

Screening for SMA

All pregnant women can receive an SMA carrier screening. A blood test is used to detect whether you are a carrier of SMA. If your results show you are a carrier, it's then recommended your partner is screened as well.

Diagnostic testing for SMA can be performed on the fetus through Chorionic Villus Sampling (CVS) around 10 to 14 weeks or amniocentesis around 16 to 20 weeks of pregnancy.

Who should consider carrier testing?

SMA carrier screenings are offered to all pregnant women. If you are considering having children, carrier screenings can be done before pregnancy.

Can SMA be prevented?

No, but screening for SMA can help you and your family make proactive steps in caring for your child with SMA.

ADDITIONAL INFORMATION

If you would like to learn more about cystic fibrosis carrier or SMA testing, please talk with your healthcare provider or contact a genetic counselor in your area (www. Nsgc.org/resourcelink.cfm).

REFERENCES

1. American College of Obstetricians & Gynecologists www.acog.org (accessed on March 18, 2022)

Maternal Fetal Medicine

Maternal Fetal Medicine (MFM) is a subspecialty of Obstetrics & Gynecology with a focus on both the expectant mother and the growing baby. MFM providers are highly skilled in performing diagnostic exams and treating unforeseen conditions during your pregnancy.

YOU MAY BENEFIT FROM AN MFM REFERRAL IF:

- You are trying to conceive with a preexisting health condition
- You are pregnant with a preexisting health condition
- You had previous complications with a pregnancy
- Your current fetus has developed a health complication
- Your fetus has or potentially has a birth defect
- You are having multiples

HOW MFM COMPARES

MFM providers have two to three years of fellowship training to assess and manage a variety of medical conditions during each stage of pregnancy. Your Obstetrician and MFM providers will work together to co-manage both you and your baby's health throughout your pregnancy.