

First Trimester Screening Exams

During your pregnancy, there will be many and varied tests carried out to ensure the safety and wellbeing of you and your baby. The testing required is as follows:

- ABO RH and Antibody screening
- HIV testing
- Rubella screening
- Hepatitis B screening
- RPR testing for Syphilis
- CBC testing for complete blood count – All above carried out at initial Obstetrics visit

Other tests you might want to review and consider, or that may be recommended:

- Cystic Fibrosis
- Sickle Cell
- First Trimester screening

ABO RH and Antibody screening

At approximately 19 weeks gestational age an Anatomy Ultrasound will be performed to check the anatomy of the baby. (There is no guarantee we can identify the sex of the baby at this time.)

The American College of Obstetrics and Gynecology recommend that in order to protect against Rh sensitization, all pregnant women should have a blood test at an early stage of pregnancy.

Therefore, during your pregnancy, you will have a blood test to find out your blood type and whether your blood has the Rh factor. If your blood lacks the Rh factor, it is called Rh negative. If it has the Rh factor, it is called Rh positive. More people are Rh positive. Problems can arise when the fetus's blood has the Rh factor and the mother's blood does not. These problems can be prevented with early treatment. Here we can explain:

- What is the Rh factor
- How problems may occur
- What treatment is needed

What Is the Rh Factor?

Just as there are different major blood groups, such as type A and type B blood, there also is an Rh factor. The Rh factor is the type of protein found on the red blood cells. Most people have the Rh factor—they are Rh positive. Others do not have the Rh factor—they are Rh negative. A simple blood test can tell whether you are Rh positive or Rh negative.

The Rh factor does not affect a person's general health. However, it can cause problems during pregnancy. In most cases, these problems can be prevented by taking a special medication.

When Does the Rh Factor Cause Problems?

The Rh factor causes problems when an Rh–negative person’s blood comes in contact with Rh–positive blood. If this happens, the person with Rh–negative blood may become sensitized. This means he or she produces *antibodies* that fight the Rh factor as if it were a harmful substance.

An Rh–negative woman can become sensitized if she is pregnant with an Rh–positive fetus. If a pregnant woman is Rh negative, her fetus can be Rh positive only if the father is Rh positive. If both the mother and father are Rh negative, there is no chance the fetus will be Rh positive and no risk to the mother that she will be sensitized. If the mother is Rh positive and the father is Rh negative, the sensitization does not occur.

During your pregnancy, you and your baby do not share blood systems. However, a small amount of blood from the fetus can cross the *placenta* into the woman’s system. When this happens, a small number of pregnant women with Rh–negative blood who carry an Rh–positive fetus will react as if they were allergic to the fetus. These Rh–negative women become sensitized and make antibodies that attack the blood of the Rh–positive fetus. When this happens, the antibodies break down the fetal red blood cells. This causes *anemia*, which can lead to a serious illness, brain damage, or even death of the fetus or newborn.

Once formed, these antibodies do not go away. In a first pregnancy with an Rh–positive fetus, the baby often is born before the woman’s body develops many antibodies, so there may be no serious problems.

In a second pregnancy with an Rh–positive fetus, these antibodies are more likely to cause anemia in the fetus. In most cases, the condition becomes worse in later pregnancies.

A woman can be sensitized any time the Rh–positive blood mixes with her blood. This can occur if an Rh–negative woman has once had:

- A miscarriage
- An induced abortion
- An ectopic pregnancy
- A blood transfusion
- Amniocentesis
- Chorionic villus sampling (CVS)
- Bleeding during pregnancy

With any of these instances, small amounts of blood can mix with that of the mother, causing you to become sensitized.

How Can Problems Be Prevented?

A simple blood test can identify a woman’s blood type and Rh factor. Another blood test, called an antibody screen, can show if an Rh–negative woman has developed antibodies to Rh–positive blood. Anemia can be prevented in the fetus if the Rh–negative woman has not yet made

antibodies against the Rh factor. ***Rh immunoglobulin (RhIg)*** is a blood product that can prevent an Rh–negative mother from being sensitized. It prevents her body from responding to Rh–positive blood cells of the fetus.

RhIg can prevent sensitization in almost all cases. However, it is not helpful if the mother is already sensitized.

If an Rh–negative woman is given RhIg, it likely will be injected into a muscle of the arm or buttocks. The most common side effects are soreness where the drug was injected or a slight fever. Both side effects usually go away on their own.

When Is RhIg Used?

During Pregnancy and After Delivery

If a woman with Rh–negative blood has not been sensitized, her doctor may suggest that she receive RhIg around the 28th week of pregnancy to prevent sensitization for the rest of the pregnancy. This prevents problems in the small number of women who can become sensitized during the last 3 months of pregnancy. Sometimes, when a pregnancy has gone past the due date, a doctor may suggest another dose of RhIg.

Shortly after birth, if the child has Rh–positive blood, the mother should be given another dose of RhIg. In almost all cases, this prevents the woman from making antibodies to the Rh–positive blood cells she may have received from her baby before and during delivery. No treatment is needed if the baby is Rh negative.

The treatment is good only for the pregnancy for which it is given. Each pregnancy and delivery of an Rh–positive child requires a repeat dose of RhIg.

An Rh–negative mother may receive RhIg after a birth even if she decides to be sterilized (have her fallopian tubes tied and cut to prevent future pregnancies). In this case, treatment may be given for three reasons:

1. The woman may decide later to have the sterilization reversed.
2. There is a slight chance that the sterilization may fail to prevent another pregnancy.
3. The treatment prevents her from developing antibodies in case she ever needs to be given a blood transfusion in the future. The presence of antibodies makes matching blood types for transfusions harder.

Other Reasons RhIg May Be Given

An Rh–negative woman also should receive treatment after any time the blood of the mother comes in contact with the fetus. This can occur during miscarriage, ectopic pregnancy, or induced abortion. She also should receive it after certain procedures, such as amniocentesis or chorionic villus sampling. This prevents any chance of the woman developing antibodies that would attack a future Rh–positive fetus.

What Happens If Antibodies Develop?

Rh-immunoglobulin treatment does not help a woman who has already developed antibodies. A mother who is Rh-sensitized will be checked during her pregnancy to see if the fetus is at risk.

In some severe cases, a baby with anemia may be delivered early or given blood transfusions while still in the mother's uterus. In less severe cases, the baby may be delivered at the normal time. After delivery, the baby may need a transfusion to replace the blood cells.

Finally...

To protect against Rh sensitization, we recommend all pregnant women have a blood test at an early stage of pregnancy. Rh immunoglobulin should be given to prevent the development of Rh antibodies.

Glossary of terms:

Amniocentesis: A procedure in which a needle is used to withdraw and test a small amount of amniotic fluid and cells from the sac surrounding the fetus.

Anemia: Abnormally low levels of blood or red blood cells in the bloodstream. Most cases are caused by iron deficiency, or lack of iron.

Antibodies: Proteins in the blood produced in reaction to foreign substances.

Chorionic Villus Sampling (CVS): A procedure in which a small sample of cells is taken from the placenta and tested.

Ectopic Pregnancy: A pregnancy in which the fertilized egg begins to grow in a place other than inside the uterus, usually in the fallopian tubes.

Induced Abortion: The planned termination of a pregnancy before the fetus can survive outside the uterus.

Miscarriage: Early pregnancy loss.

Placenta: Tissue that provides nourishment to and takes away waste from the fetus.

Rh Immunoglobulin (RhIg): A substance given to prevent an Rh-negative person's antibody response to Rh-positive blood cells.

Transfusion: Direct injection of blood, plasma, or platelets into the bloodstream.

HIV Testing

All pregnant women should be screened for HIV infection as early as possible during each pregnancy.

HIV screening is recommended for all pregnant patients and they will receive an HIV test as part of the routine panel of prenatal tests unless they decline (opt-out screening).

Rubella Screening

A rubella blood test detects antibodies that are made by the immune system to help kill the rubella virus. These antibodies remain in the bloodstream for years. The presence of certain antibodies means a recent infection, a past infection, or that you have been vaccinated against the disease.

Rubella (also called German measles or 3-day measles) usually does not cause long-term problems. But a woman infected with the rubella virus during pregnancy can transmit the disease to her baby (fetus). And serious birth defects called congenital rubella syndrome (CRS) could develop, especially during the first trimester. Birth defects of CRS include cataracts and other eye problems, hearing impairment, and heart disease. Miscarriage and stillbirth are also possible consequences for pregnant women. The vaccination to prevent rubella protects against these complications.

A rubella test is usually done for a woman who is or wants to become pregnant to determine whether she is at risk for rubella.

Hepatitis B Screening

When you come in for your first prenatal visit, you'll be given a series of routine blood tests, including one that checks for the presence of the hepatitis B virus (HBV), which can cause severe illness, liver damage, and even death.

More than a million people in the United States carry the virus, but many have no symptoms and don't know they're carriers. If you're one of them, you could pass the virus to your baby at birth. Determining that you're a carrier would allow your healthcare practitioner to treat your baby immediately after delivery, which would most likely prevent her from becoming infected.

If you test negative for HBV and have not received the hepatitis B vaccine, your practitioner may recommend that you get immunized if you're at high risk for contracting the disease (see below for groups at high risk). The shot is safe for pregnant women and has no effect on your developing baby.

RPR testing for Syphilis

Even though the infection is relatively rare, it's considered vitally important to detect and treat syphilis during pregnancy. The CDC recommends that all pregnant women be screened for the

infection at their first prenatal visit, and some states require that all women be tested again at delivery.

If you live in a community where syphilis is prevalent or you're otherwise at high risk, you should be tested again at 28 weeks and at delivery. You'll also be retested for syphilis if you've contracted another STI during your pregnancy or if you or your partner develops symptoms of syphilis.

Because it takes about four to six weeks after exposure to get a positive result from the blood test, the result may be negative if you're tested too soon. So if you had high-risk sex a few weeks before your test or your partner recently had symptoms, tell your practitioner so you can be tested again in a month. If your screening test is positive, the lab will perform a more specific test on your blood sample to tell for sure whether you have syphilis.

CBC testing for complete blood count

One of the first tests that you'll receive is a CBC (complete blood count). This test checks your hematocrit and hemoglobin, as well as platelet count. Translation for laypeople like us: Hemoglobin and hematocrit measure your potential for anemia.

Why it's important (hematocrit and hemoglobin): Those women who are anemic need to be identified well ahead of delivery so that therapy can improve their low blood count. Physiologic anemia can be problematic to the patient.

Why it's important (platelet count): Because a woman will lose half a liter of blood on average at delivery. The platelet count measures the blood's ability to clot. With a low platelet count, a patient is at risk for bleeding to death.

Cystic Fibrosis Screening

Cystic fibrosis (CF) is a lifelong illness that children can inherit from their parents. It usually becomes noticeable in the first few months of life, and typically affects breathing and digestion. Children that have CF generally need oral medicines to help digest their food and daily respiratory therapy to aid their breathing. Lung infections requiring hospitalizations to receive IV antibiotics are not uncommon and these infections tend to get more severe as the children age. Children with CF have normal intelligence, and both boys and girls are equally likely to inherit the disease. Most people with CF have a shortened life span, with some dying during childhood or adolescence, and others living into their 40's. For reasons not entirely understood, some people with CF have only mild symptoms and minor health problems and would be expected to live rather healthy and complete lives. Unfortunately there is no cure at this time.

The purpose of CF carrier testing is to see if a couple is at increased risk of having a child with CF. If the couple is at increased risk, the developing child can be tested to see if he or she will have CF. Prenatal tests on the developing child, however, cannot always tell how mild or severe

their symptoms will be. CF cannot be treated or cured before birth. The benefit of having this knowledge about the developing baby is so you can prepare yourself for a baby with special needs, or so you can terminate the pregnancy. Carrier testing of the parents requires only a blood draw, and is fully voluntary.

As a background, CF is an autosomal recessive genetic disorder. Each person has two copies of each gene. One gene copy comes from your mother, the other from your father. Genes may not work properly if there is a mutation, or mistake, in them. In some genetic disorders, like CF, both copies of the gene need to have a mistake in them for a person to have the disease. If only one copy is abnormal that person will be healthy, but is called a “carrier” because they carry one mutated gene copy and can transmit the mutated copy to their son or daughter. If two carriers mate, there is a 1 in 4 chance (25%) of both carriers giving their mutated copy to their offspring – so the offspring will have two bad gene copies, and be affected with the disease.

Chance of Being a Cystic Fibrosis Carrier Based on Race and/or Ethnicity

<u>Ethnicity/Race</u>	<u>Chance of Being a CF Carrier</u>	<u>Chance Both Parents are Carriers</u>
European Caucasian/Ashkenazi Jewish	1 in 29 (3.4%)	1 in 841 (0.12%)
Hispanic American	1 in 46 (2.2%)	1 in 2,116 (0.05%)
African American	1 in 65 (1.5%)	1 in 4,225 (0.02%)
Asian American	1 in 90 (1.1%)	1 in 8,100 (0.01%)

If a relative of yours has CF, or is a carrier of CF, your chance of being a CF carrier will be greater than your ethnicity risk shown above.

Because there are over 900 known mutations that can cause CF (and the list is growing) there are some mutations blood testing cannot find. Therefore, if you have a normal test result you could still be a CF carrier for a currently unknown or non-tested mutation, though this risk is very small (less than 1%). Testing can involve the mother first, then the father or both simultaneously. If the mother tests positive, the father should then be tested. If he tests negative, the risk of having an affected baby is very small (1 in 400 or 0.25%), but not zero because the test cannot identify all CF mutations.

To aid you in your decision making on whether to be tested, we offer the following thoughts:

Potential reasons you might want to be tested:

- the chance of being a carrier seems high for you
- you have a family history of cystic fibrosis
- cystic fibrosis appears to be a very serious disease to you

- you and the baby's father would consider testing the developing baby by amniocentesis if you were both found to be carriers (this would allow you to prepare for the birth of a baby with cystic fibrosis, or electively end the pregnancy if you desired)

Potential reasons you might NOT want to be tested:

- the chance of being a carrier seems low to you
- the test is not perfect and cannot identify all mutations, and therefore, all carriers
- some people with cystic fibrosis have only minor health problems and lead normal lives
- cystic fibrosis does not appear to be a very serious disease to you
- you and the baby's father would NOT consider testing the developing baby by amniocentesis if you were both found to be carrier.

www.cff.org/aboutCF/faqs/

Sickle Cell – optional

Sickle cell disease (SCD) is a genetic condition that is present at birth. It is inherited when a child receives two sickle cell genes—one from each parent. A person with SCD can pass the disease on to his or her children.

Sickle cell trait (SCT) is not a disease, but means that a person has inherited the sickle cell gene from one of his or her parents. People with SCT usually do not have any of the symptoms of SCD and live a normal life, but they can pass the sickle cell gene on to their children.

When both parents have SCT, they have a 25% chance of having a child with SCD with every pregnancy.

When both parents have SCT, they have a 50% chance of having a child with SCT with every pregnancy.

First Trimester Screening – optional

First Trimester Screening test for Down syndrome and trisomy 18 and provides patients with a variety of screening options. The information provided by screening tests is beneficial for some families; other families do not feel this information is helpful to have prior to delivery. This information is intended to help you explore these options. Participation in the screening tests is your choice. Your obstetrician can also provide you with more information. This appointment will be scheduled here at our office unless otherwise indicated.

First trimester screening is performed at FABEN at 12 weeks, 4 (\pm 2) days of pregnancy, and will involve both an ultrasound and a blood test.

- **Ultrasound**
Ultrasound measures an area of fluid accumulation at the back of the baby's neck called nuchal translucency (NT). This accumulation of fluid is a normal finding. Increased NT measurements,

however, may indicate increased risk for chromosome abnormalities, congenital heart defects, and other genetic syndromes.

- **Blood test**

The blood test by a finger stick or blood draw from the arm measures the levels of two proteins, freeBeta-hCG and PAPP-A, which are normally found in the blood of pregnant women. The levels of these two chemicals are combined with the NT measurement and the mother's age to provide a risk assessment for Down syndrome and trisomy 18. The combined testing detects approximately 90 percent of Down syndrome and 90 percent of trisomy 18.

Patients determined to be at increased risk for either of these conditions can then be offered diagnostic testing such as chorionic villus sampling (CVS) or amniocentesis. If the screening test does not predict an increased risk, it is recommended that the patient proceed with maternal serum AFP screening in the second trimester to identify increased risks for open neural tube defects and abdominal wall defects.

It is important to remember that a normal screening test does not guarantee a normal baby, nor do abnormal test results definitely mean a baby with a birth defect.

For more information about First Trimester Screening please see the link below:

<https://www.ntqr.org/SM/wfPatientResources.aspx>