Pregnancy & Childbirth

MAT-SU Women's Health SPECIALISTS

Welcome

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Important Information

Hospital and phone number: Mat-Su Regional Medical Center 907-861-6645

My due date: _____ My blood type: _____

My allergies: _____

_____

_____

_____

My GBS status: _____

Other pertinent info: _____

My Scheduled Appointments

Appointment	Date	Time	Provider	Questions/Notes
8-10 weeks				
11-14 weeks				
15-18 weeks				
19-22 weeks				
23-26 weeks				
27-30 weeks				
30-31 weeks				
32-33 weeks				
34-35 weeks				
36 weeks				
37 weeks				
38 weeks				
39 weeks				
40 weeks				
41 weeks				

Medications in Pregnancy

There are many medications that are safe in pregnancy and many others that are not known to cause harm but the effect is unknown. For this reason, we ask that you refer to medications that are known to be safe and ask your provider about any other medications before you take them.

There are also medications that we wish that you AVOID completely, unless otherwise directed by your provider. These are:

- Asprin or aspirin containing products (BC/Goody's Powder, Alka-Seltzer and Bufferin all contain aspirin).
- NSAIDS (ibuprofen, Advil, Motrin, Naprosyn, Aleve or Ketoprofen).
- Prescription medications that you have not been prescribed (never take medication that was not prescribed for you specifically) or that has not been cleared for use by your provider.

There are many medications that are safe. These are:

- Acetaminophen (Tylenol).
- Sudafed or other antihistamines (Claritin, Alavert, Zyrtec) are safe.
- Prescription medication that has been reviewed by your provider.
- Benadryl
- Milk of Magnesia or MiraLax
- Kaopectate
- Antacids (Gaviscon, Prevacid, Zantac, Pepcid, TUMS, Mylanta).

Please do not hesitate to call us during business hours to ask our staff if medications are appropriate for you.

Treatments to Relieve Discomforts During Pregnancy

Listed on the next page are the over the counter/non-prescription medications and treatments that may be used to relieve discomforts experienced during your pregnancy. Please call your physician before taking any medication other than the ones listed.

Condition/Symptoms	Medications & Treatments
Headache	**Tylenol, Regular or Extra Strength (Do NOT use aspirin or ibuprofen which may be harmful to the fetus.)** | Warm compress to the eyes, temples and nose to relieve a sinus headache. Cold compress to the back of the neck to relieve a tension headache. Reduce stress with relaxation exercises. Rest in a dark, quiet room and get enough sleep.
Allergies	**Benadryl, Tylenol Sinus, Claritin (NOT Claritin-D), Zyrtec (NOT Zyrtec-D)**
Cold symptoms	**Benadryl, Tylenol, Emergen-C, Zyrtec (NOT Zyrtec-D), Sudafed** | Increase fluids. Call physician with fever over 100.4.
Congestion or runny nose	**Benadryl, Robitussin (only regular, NOT DM, CR, etc.), Vicks vapor rub, Ocean or any Saline Nasal Spray/NetiPot, Sudafed** | Use a humidifier and drink plenty of fluids. Elevate your head when you sleep.
Cough	**Cough drops or throat lozenges** | Increase hydration.
Fever	**Tylenol (Regular or Extra Strength)** | Report any fever over 100.4 that does not respond to Tylenol.
Sore throat	**Cepacol Lozenges, Chloraseptic spray or lozenges** | Gargle with salt water.
Constipation	**Chia or flax seeds, glycerine suppositories, Metamucil, Colace, Milk of Magnesia, Miralax** | Drink at least 10 glasses of water each day. Eat foods high in fiber such as bran, raw fruits, raw vegetables
Diarrhea	**Kaopectate** | Try bland or BRAT diet (bananas, rice, applesauce, tea or toast). Call your physician if symptoms persist for more than 24 hours
Gas	Increase walking. Avoid gaseous foods: broccoli, cauliflower, onions, dairy
Heartburn/Indigestion	**Gaviscon, Prevacid, Zantac, Pepcid, TUMS, Mylanta, Maalox** | Do not drink fluids while eating. Avoid spicy, greasy or fatty foods. Avoid chocolate and caffeine. Eat smaller, more frequent meals. Eat your last meal of the day several hours before bedtime. Avoid lying down for 2 hour after eating or use pillows to elevate your upper body.
Hemorrhoids	**Tucks Pads, Witch Hazel, Preparation H, Anusol HC** | Drink at least 10 glasses of water each day. Eat foods high in fiber such as bran, raw fruits, raw vegetables. Sit in a tub of warm water to soothe discomfort.
Nausea/Vomiting	**Ginger, Unisom 12.5mg+Vitamin B6 25mg** | Eat meals high in protein. Eat frequent, small meals. Eat toast or crackers first thing in the morning. Take vitamins with food. Call your physician if vomiting persists for more than 24 hours.
Backache	**Tylenol, Regular or Extra Strength (Do NOT use aspirin or ibuprofen which may be harmful to the fetus.)** | Apply heating pad on medium heat or a warm towel four times a day for 15 minutes or take warm baths. Sleep on your side with a pillow between your legs. Wear shoes with good arch support (not flip flops or sandals). Sleep on a firm bed. Avoid lifting heavy objects or standing for long periods of time. Wear a pregnancy support belt. Have prenatal massage or manipulation (osteopathic/chiropractic).
Leg Cramps/Swelling	**TUMS, magnesium oxide 400mg nightly** | Eat foods rich in calcium and potassium. Wear support hose and elevate your feet when possible. Rest on your left side and stretch your legs before bed. Drink plenty of water and avoid excessively salty foods.
Muscle Aches	**Tylenol (Regular or Extra Strength), Epsom Salt Bath, Tiger Balm (Do NOT use aspirin or ibuprofen which may be harmful to the fetus.)** | Massage, stretching, warm baths. Avoid sitting or standing in one position for long periods of time.
Skin Irritation/Rash	**Benadryl, Claritin (not Claritin D), Caladryl or Calamine Lotion, Neosporin** | Moisturize twice a day. Avoid long, hot baths or showers.
Sleep Problems	**Benadryl, Tylenol PM, Unisom**
Vaginal Yeast Infection	**Monistat 7** | Call the office for signs of infection (itching, irritation, green/yellow discharge with foul odor) or if you may be leaking amniotic fluid. **DO NOT DOUCHE!!!**

Diet and Exercise in Your Pregnancy

What can I eat?

You are what you eat (or at least your baby is what you eat). Try to stick to healthy foods and limit the amount of processed "junk" food you eat. Fresh or frozen fruits and vegetables should be a regular part of your diet. Avoid excessive sweets and empty calories.

Pregnancy is not a time to over eat, but it is also not the time to diet. In general, you should eat about 300-500 more calories per day than the normal recommended amount. (For non-pregnant women, this is usually 1800-2000 Calories/day). You may find that small, more frequent meals are better tolerated during pregnancy. Remember: if you haven't eaten all day, neither has your baby!

Having a balanced diet will help support and grow your baby, your placenta and the extra blood that you need to help you have a healthy pregnancy. We ask that you look at www.choosemyplate.gov to see what a healthy meal should look like. There are also multiple recipes and examples of healthy proteins and complex carbohydrates to help you make appropriate choices. If you are unsure if what you are eating is appropriate, we may have you fill out a diet log/diary and go over this with you at one of your appointments. You can feel free to do a log/diary for your own knowledge of how you are eating.

Specific Foods to Avoid

- **Unpasteurized dairy products** These may contain Listeria (a potentially harmful bacterium). Raw milk and some soft cheeses like feta, goat, and brie may not be pasteurized. Raw eggs should be avoided as well.
- **Raw meats** Be sure your beef, chicken and pork or other meats are cooked thoroughly. Red meat should be medium well to well done. This includes raw seafood.
- **Tilefish, Shark, Mackerel and Swordfish** The FDA has recommended you limit your fish and shellfish consumption to 12 ounces weekly. There are some concerns about the amount of mercury found in commercial fish. Canned light tuna contains less mercury than white albacore tuna. Wild caught Salmon and Halibut are fine, just keep your consumption to 12 ounces per week.

What vitamins do I need to take?

We recommend that you take a prenatal vitamin during your pregnancy. Make sure that the vitamin you choose has at least 400mcg of folic acid and includes calcium. You should take 1200mg of calcium daily. This can be combined from your diet and prenatal vitamin. If your diet is low in calcium, you may take a supplement as well.

We may prescribe extra iron to be taken during your pregnancy. When taking iron, take it with a Vitamin C or a glass of orange juice to help your body absorb it. Do not take it with a cup of coffee because caffeine decreases the absorption of iron.

Remember to take your prenatal vitamin with food. Nausea is a common side effect of taking vitamins, especially on an empty stomach. You can also take them in the evening before bed to help prevent nausea.

Please do not take any additional supplements INCLUDING HERBS without first talking to your provider to see if it is appropriate in pregnancy. There are many over-the-counter herbal supplements that are not okay in pregnancy.

24 Hour Diet Recall

Time	Meal (check one)	Food	Amount	How was it prepared?	How did you feel? (i.e. nausea, vomiting, heartburn, diarrhea)
	☐Breakfast ☐Snack ☐Lunch ☐Snack ☐Dinner ☐Snack				
	☐Breakfast ☐Snack ☐Lunch ☐Snack ☐Dinner ☐Snack				
	☐Breakfast ☐Snack ☐Lunch ☐Snack ☐Dinner ☐Snack				
	☐Breakfast ☐Snack ☐Lunch ☐Snack ☐Dinner ☐Snack				
	☐Breakfast ☐Snack ☐Lunch ☐Snack ☐Dinner ☐Snack				
	☐Breakfast ☐Snack ☐Lunch ☐Snack ☐Dinner ☐Snack				
	☐Breakfast ☐Snack ☐Lunch ☐Snack ☐Dinner ☐Snack				

How much weight should I expect to gain in pregnancy?

This is a question that we often get from our expecting mothers. The table below provides the most accurate information available. Depending on your Body Mass Index (BMI), you may need to gain more or less weight during pregnancy. We will calculate your BMI at your initial visit.

Institute of Medicine Weight Gain Recommendations for Pregnancy

Prepregnancy Weight Category	Body Mass Index*	Recommended Range of Total Weight (lb)	Recommended Rates of Weight Gain† in the Second and Third Trimesters (lb) (Mean Range [lb/wk])
Underweight	Less than 18.5	28–40	1 (1–1.3)
Normal Weight	18.5–24.9	25–35	1 (0.8–1)
Overweight	25–29.9	15–25	0.6 (0.5–0.7)
Obese (includes all classes)	30 and greater	11–20	0.5 (0.4–0.6)

*Body mass index is calculated as weight in kilograms divided by height in meters squared or as weight in pounds multiplied by 703 divided by height in inches.

†Calculations assume a 1.1–4.4 lb weight gain in the first trimester.

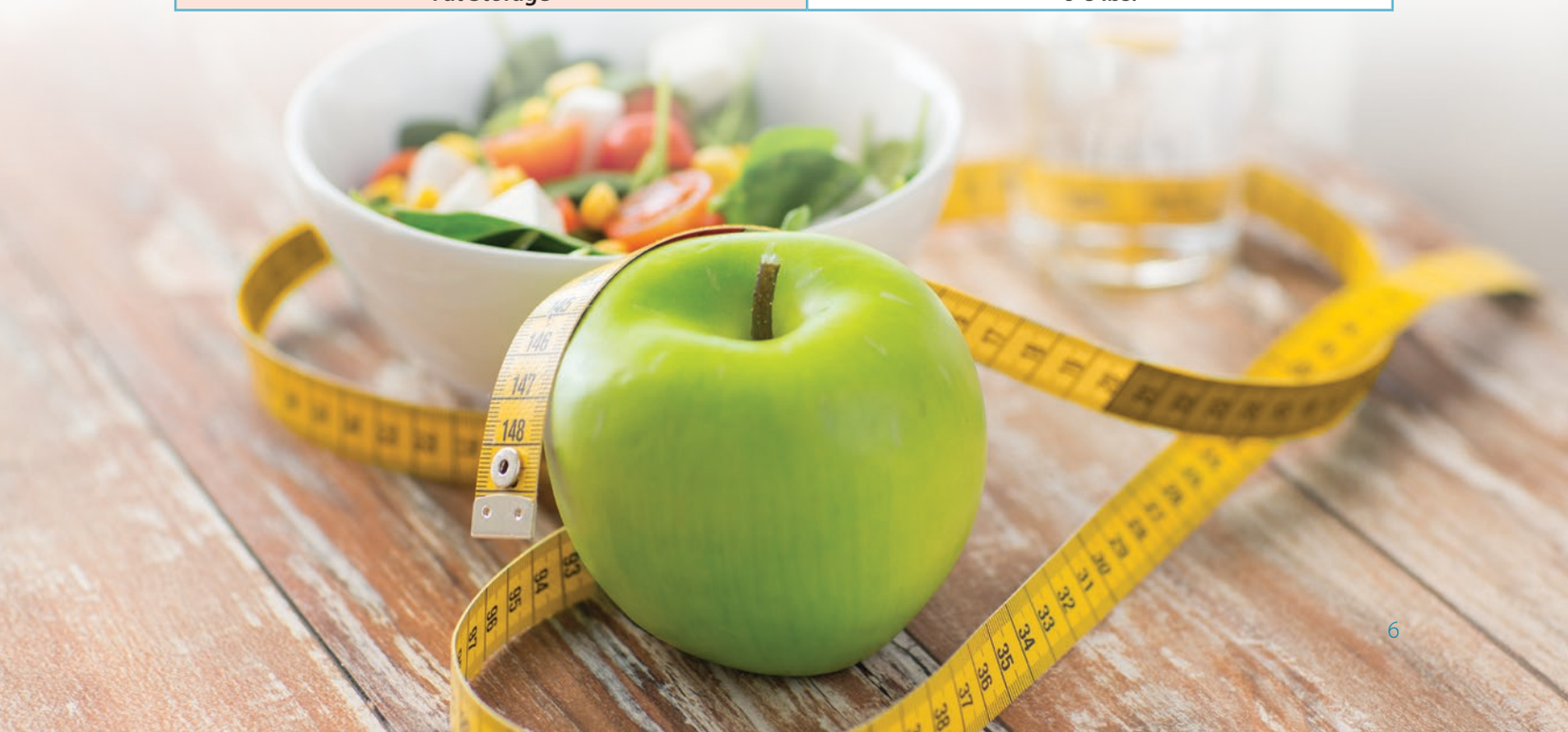
Modified from Institute of Medicine (US). Weight gain during pregnancy: reexamining the guidelines. Washington, DC. National Academies Press; 2009. ©2009 National Academy of Sciences.

Eating a balanced, low sugar/low fat diet will help you reach your ideal pregnancy weight. Remember, you will need to consume an average of 300 extra calories/day during your pregnancy. Be mindful that not all calories are created equal. "Empty calories" or those with low nutritional value (Ex: processed foods, white bread, fast food, pop, etc) will cause excess weight gain without providing the building blocks of nutrition your baby needs to grow properly.

Please do not hesitate to ask us any nutrition/dietary questions. We are here to help you have a happy, healthy pregnancy.

Where the Weight Goes

Baby	**6.5-9 lbs.**
Amniotic Fluid	**1.5 lbs.**
Uterus	**2 lbs.**
Heavier Breasts	**2-3 lbs.**
Additional Blood Volume	**3-4 lbs.**
Increased Fluids	**3-4 lbs.**
Fat Storage	**6-8 lbs.**



What can I do?

Exercise is recommended for most pregnant women. Studies have shown that women who exercise regularly during pregnancy, tolerate their pregnancy, delivery and recovery better than women who do not exercise. Even if you do not exercise regularly right now, beginning a light program will help you.

Exercise should not be intense or high impact during your pregnancy (no downhill skiing, snow machining or hockey). Your heart rate should remain in the 140's or lower for the majority of your workout. Walking, swimming, biking, light weights and pregnancy yoga are some excellent choices for pregnancy. Be aware that your exercise tolerance generally drops as your pregnancy progresses. Don't "fight through the wall" if you start to get fatigued. Take the opportunity to rest and re-hydrate. Listen to your body!

We recommend you avoid exercising in extremely hot or humid places (hot yoga) or in low oxygen environments (scuba diving or extreme mountain climbing). Don't exercise on an empty stomach and make sure you hydrate! Avoid abdominal crunches or sit-ups in pregnancy, core work is okay, but avoid over using your abdominal muscles.

If you are exercising make sure you are feeding your body well! You will want to add 100-200 calories of high-quality, nutritious food to your diet for every 30 minutes of exercise you do.

Can I travel?

If you are having an uncomplicated pregnancy, you may safely travel until 36 weeks. This includes air, boat, train or car travel. If you will be traveling for an extended period of time (more than 2 hours), please be sure to take water and stay hydrated.

We also want to prevent you from developing blood clots in your legs. If traveling by car, stop every 2 hours to walk and stretch your legs. If you are flying, get up and walk in the aisle every now and then. Remember, if you are drinking plenty of water you will be going to the restroom very often and having to get up and walk, this is a GOOD thing!

Activity Restriction in Pregnancy

Society for Maternal-Fetal Medicine (SMFM) with the assistance of Christina Davidson, MD, Division of Maternal Fetal Medicine, Baylor College of Medicine, Houston, Texas.

What is restriction of activity in pregnancy?

Restriction of activity, sometimes called "bed rest" or "modified bed rest," is sometimes suggested for a number of potential pregnancy complications. These complications include, but are not limited to, preterm (before 37 weeks' gestation) contractions, a dilated cervix from preterm labor, a short cervix, preterm premature rupture of membranes (when the bag of water ruptures before 37 weeks' gestation and before the onset of labor), elevated blood pressure, preeclampsia (a pregnancy-specific disorder in which women develop elevated blood pressure and protein in the urine), inadequate growth of the baby, placenta previa (a placenta that covers the opening of the cervix), risk of miscarriage, and multiple gestations (for example, twin pregnancies).

The terms "bed rest" and "activity restriction" are often used interchangeably, but in reality, can differ to a great extent. Most women admitted to the hospital for complications of pregnancy are subjected to some degree of "bed rest," with the ability to use the bathroom and bathe. For women at home, "bed rest" or "activity restriction" can have different meanings and different degrees of limitations.

How common is it for obstetric providers to recommend activity restriction or bed rest in pregnancy?

Surveys have shown that both ob/gyns and maternal-fetal medicine specialists prescribe activity restriction and bed rest, even though most of the physicians surveyed do not expect that doing so will actually improve pregnancy outcomes.

Are there risks associated with activity restriction in pregnancy?

Extended periods of activity restriction can result in muscle and bone loss. This is true for pregnant and nonpregnant individuals and is called "deconditioning." These changes can occur after only a few days of immobility. There is not a lot of information about the full impact of these changes in pregnant women.

Pregnancy is associated with an increased risk of developing blood clots in the legs (deep venous thrombosis, or DVT) and movement of clots to the lungs (pulmonary embolism, or PE). Some studies have described an additional increased risk of DVT and PE among pregnant women placed on bed rest compared to pregnant women who were not placed on bed rest. There appears to be an increased risk of blood clots in patients placed on activity restriction.

Some studies suggest that restriction of activity is associated with a higher rate of developing diabetes in pregnancy in women admitted to the hospital for other pregnancy-related complications. More studies are necessary in this area, but elevated levels of blood sugar commonly occur in nonpregnant patients placed on activity restriction.

Along with the potential negative maternal physical effects associated with activity restriction, there is also an increased risk of maternal anxiety and depression, adverse psychological effects on the family, loss of income, and lower birth weights.

What are the benefits associated with activity restriction in pregnancy for prevention of preterm birth?

Based on information from a few well-designed studies, bed rest did not reduce the chance of preterm delivery in women either at risk of or already experiencing preterm labor. One study found that preterm birth was more common in women already at risk of preterm birth when they were placed on any type of work or nonwork-related activity restriction, both at home and in the hospital.

There are no studies to date that have been able to identify any improvements in newborn outcomes in women at risk of preterm birth who have their activity restricted. Studies have shown higher rates of newborn complications including lower birth weight, earlier gestational age at delivery, and a higher risk of developing motion sickness and allergies later in life in women whose activity was restricted during the pregnancy.

Are there other pregnancy conditions that may potentially benefit from activity restriction?

There are no data indicating that activity restriction is of benefit for any obstetric condition. Disorders of pregnancy associated with elevated blood pressure, preterm premature rupture of membranes, multiple gestation, and inadequate growth of the baby are among the most common reasons for hospital admissions during pregnancy and often trigger a recommendation of activity restriction.

With regard to blood pressure disorders, some studies have suggested a benefit from modest restriction of activity (4-6 hours per day of rest), but there is not enough proof of benefit to recommend this practice. With regard to preterm premature rupture of membranes, there are no studies that have examined the impact, if any, that activity restriction or bed rest has on pregnancy outcomes.

Women with multiple gestations are at an increased risk of preterm birth. Hospital bed rest was once offered routinely to such patients in the middle to late pregnancy in an effort to prevent preterm birth but studies have not shown that it improves infant outcomes. In contrast, studies have demonstrated that maternal stress, side effects, and depressive symptoms may increase, and weight gain may be suboptimal in patients placed on hospital bed rest.

Inadequate growth of the baby is often attributed to problems with blood flow to the placenta, and activity restriction and/or bed rest is often prescribed in an effort to improve placental blood flow. Again, studies have failed to show a benefit to this practice.

What are the current recommendations from professional societies regarding activity restriction in pregnancy?

The American College of Obstetricians and Gynecologists states that bed rest has not been shown to be effective for the prevention of preterm birth and should not be routinely recommended. The Society of Obstetricians and Gynaecologists of Canada states that increased rest at home in the third trimester or reduction of workload and stress may be useful for women at risk of developing preeclampsia, although strict bed rest in the hospital for women already diagnosed with preeclampsia is not recommended. The National Collaborating Centre for Women's and Children's Health, in collaboration with the Royal College of Obstetricians and Gynaecologists, states that bed rest has not been shown to be of benefit and should not be offered to women with pregnancy-associated high blood pressure or preeclampsia. No other national recommendations exist about use of bed rest or activity restriction in pregnancy.

Conclusion

The practice of activity restriction or bed rest has very little evidence to support a benefit for the mother or infant, but has well-described negative effects on the mother, newborn, and family. In summary, the Society for Maternal-Fetal Medicine recommends against the routine use of activity restriction or bed rest during pregnancy for any indication.

From *Contemporary OB/GYN*

Common Complaints of Pregnancy

Breast tenderness is common and often the first sign of pregnancy. It is usually mild and will resolve by the end of the first trimester. Usually, your breasts will enlarge about one cup size during pregnancy, more once you start breastfeeding. Wear a supportive bra to decrease discomfort.

Fatigue/Tiredness is another common early pregnancy sign. Your body is growing a baby! Get plenty of rest, go to sleep early and nap if needed. Exercise daily to increase your energy level. Eat regular, healthy meals .

Nausea and vomiting of pregnancy (NVP) is quite common. Ninety percent of pregnant women will experience nausea. This is often caused by hormonal changes and increased calorie needs from your changing body. General measures to control NVP are: eating frequent, small meals, avoiding spicy foods, taking vitamins B6 (10-25mg up to three times a day), ginger, drinking clear liquids and getting regular rest. If you have having difficulty keeping any foods down, switch to a "B.R.A.T." diet (bananas, rice, applesauce, toast) and stick to water/sports drinks only. If your nausea is early in the morning, eating something before you get out of bed may help. Also eating every 2-3 hours and eating slowly will keep your blood sugar at a consistent level. If you are still having problems, we will prescribe medications to help you.

Headaches are common, especially from about 10-20 weeks of pregnancy. Usually they will resolve with Tylenol, rest, hydration and time. You can also try ice packs to the back of your neck, or massage. Let us know if they do not resolve.

Spotting or brown discharge is often seen in the first trimester and rarely in the 2nd or 3rd trimester. Usually, the pregnancy is okay, but we request that you notify our office if this is the first time this has happened. Heavy bleeding in the first trimester may indicate a miscarriage is occurring, but that is not always the case. If the bleeding is more than a pad every 1-2 hours, you should call the office or go to the emergency room. If you are having heavy bleeding in the 2nd or 3rd trimester, please notify us by phone and go straight to Labor & Delivery.

Low back pain is another common symptom, especially in the third trimester. Usually rest and Tylenol will help. Topical heat and massage can help as well. Pelvic rock exercises can help stretch the muscles in your back and relieve pain. Avoid lifting heavy objects, when you need to lift, squat down and bend your knees. Use supportive shoes and good support when sitting in a chair. A supportive belly band can also help decrease discomfort by supporting your growing belly.

Round ligament pain usually begins in the 2nd trimester and is characterized by sharp pulling pains on either side of the low abdomen. These symptoms are due to growth of the uterus and the stretching/pulling of the ligaments that hold the uterus in place. Belly bands can be extremely helpful for stabilizing your growing abdomen.

Bleeding gums are more common in pregnancy. It is important to take good care of your teeth by seeing a dentist in pregnancy and brushing and flossing regularly.

Skin changes are also normal in pregnancy. The most common one is a dark line that runs from the pubis to the umbilicus or higher. This is called the linea nigra. You may develop a darkening of the skin over the nose and upper cheeks and this is called melisma or the "mask of pregnancy". Moles may grow or darken. You may also see changes in hair growth. Please notify us of any suspicious moles.

Varicose veins in pregnancy are common due to the increase in blood in your body and the pressure of your growing baby and uterus on your veins, often slowing down the flow in leg veins. Avoid standing or sitting for long periods, move around every 1-2 hours if you have a job that requires you to stand or sit. Avoid crossing your legs for a long period of time. Exercise regularly. Wear supportive hose. We can recommend a consultation with a specialist if your varicose veins cause you too much discomfort.

Leg cramps can be common and bothersome in pregnancy. Be sure you are getting enough calcium in your diet (cheese, milk, broccoli and almonds are rich in calcium). Wear comfortable shoes. Stretch your legs before bed. Talk to your provider about taking a calcium/magnesium supplement if relief measures are not working.

Stretch marks are a concern for many women in pregnancy. This happens because sometimes the skin doesn't have enough elasticity or "stretch" to accommodate the growing belly of pregnancy. You can get them on your abdomen, your thighs and your buttocks. Creams and lotions will not prevent stretch marks, as they are formed under the top layers of the skin, however, moisturizing can minimize itching that may accompany your stretching skin.

Swelling of the feet and legs during pregnancy is also caused by the pressure from your uterus on your blood vessels. This causes fluid retention and swelling particularly in your legs and feet. Try not to stand for long periods of time, put your feet up. Drink plenty of fluid (8-10 glasses/day). Avoid foods high in salt. Wear loose comfortable clothing. Keep your diet rich in protein. Rest on your side during the day to increase the blood flow to your kidneys. You can eat cucumbers, melon and drink iced tea (one cup per day), as these are natural diuretics and can help decrease swelling. If you have swelling in your face or hands, please notify your provider.

Vaginal discharge is a common complaint from women in pregnancy. Increase blood supply to your vagina and increased hormones will cause the vagina to increase the normal amount of secretions. This is normally white or clear, does not smell and does not itch or burn. It can look yellowish when it dries on your underwear or on a panty liner. Wear cotton underwear, avoid tight fitting jeans/leggings. DO NOT DOUCHE!!! Keep yourself clean with water and mild soap if needed. If your discharge becomes foul smelling, green or gray, bloody or causes discomfort, contact your provider so that they can check for an infection.

Braxton-Hicks contractions are mild tightening of the uterus seen in the 2nd or 3rd trimester. Occasionally, they can be painful, but usually they are not. They can occur on one part of the uterus or all over. They are normal, but they alert you to rest and hydrate. Let us know if they do not improve with rest and hydration.

Routine Tests During Pregnancy

Why are tests done during pregnancy?

A number of lab tests are suggested for all women as part of routine prenatal care. These tests can help find conditions that can increase the risk of complications for you and your fetus.

What tests are done early in pregnancy?

The following lab tests are done early in pregnancy:

- Complete blood count (CBC)
- Blood type
- Urinalysis
- Urine culture
- Rubella
- Hepatitis B and hepatitis C
- Sexually transmitted infections (STIs)
- Human immunodeficiency virus (HIV)
- Urine drug screen

What is a CBC and what can the results show?

A CBC counts the numbers of different types of cells that make up your blood. The number of red blood cells can show whether you have a certain type of anemia. The number of white blood cells shows how many disease-fighting cells are in your blood, and the number of platelets can reveal whether you have a problem with blood clotting.

What is blood typing and what can the results show?

Results from a blood type test can show if you have the Rh factor. The Rh factor is a protein that can be present on the surface of red blood cells. Most people have the Rh factor—they are Rh positive. Others do not have the Rh factor—they are Rh negative. If your fetus is Rh positive and you are Rh negative, your body can make antibodies against the Rh factor. In a future pregnancy, these antibodies can damage the fetus's red blood cells.

What is a urinalysis and what can the results show?

Your urine may be tested for red blood cells (to see if you have urinary tract disease), white blood cells (to see if you have a urinary tract infection), and glucose (high levels may be a sign of diabetes mellitus). The amount of protein also is measured. The protein level early in pregnancy can be compared with levels later in pregnancy. High protein levels in the urine may be a sign of preeclampsia, a serious complication that usually occurs later in pregnancy or after the baby is born.

What is a urine culture test and what can the results show?

A urine culture tests your urine for bacteria, which can be a sign of a urinary tract infection.

What is rubella and what do test results for this disease show?

Rubella (sometimes called German measles) can cause birth defects if a woman is infected during pregnancy. Your blood is tested to check whether you have had a past infection with rubella or if you have been vaccinated against this disease. If you have not had rubella previously or if you have not been vaccinated, you should avoid anyone who has the disease while you are pregnant because it is highly contagious. If you have not had the vaccine, you should get it after the baby is born, even if you are breastfeeding. You should not be vaccinated against rubella during pregnancy.

What are hepatitis B and hepatitis C and what do test results for these infections show?

Hepatitis B and hepatitis C viruses infect the liver. Pregnant women who are infected with hepatitis B or hepatitis C virus can pass the virus to their babies. All pregnant women are tested for hepatitis B virus infection. If you have risk factors, you also may be tested for the hepatitis C virus.

Which STI tests are done in pregnant women?

All pregnant women are tested for syphilis and chlamydia early in pregnancy. Syphilis and chlamydia can cause complications for you and your baby. If you have either of these STIs, you will be treated during pregnancy and tested again to see if the treatment has worked. If you have risk factors for gonorrhea (you are aged 25 years or younger or you live in an area where gonorrhea is common), you also will be tested for this STI.

Why are all pregnant women tested for HIV?

If a pregnant woman is infected with HIV, there is a chance she can pass the virus to her baby. HIV attacks cells of the body's immune system and causes acquired immunodeficiency syndrome (AIDS). If you are pregnant and infected with HIV, you can be given medication and take other steps that can greatly reduce the risk of passing it to your baby.

Which pregnant women should be tested for TB?

Women at high risk of TB (for example, women who are infected with HIV or who live in close contact with someone who has TB) should be tested for this infection.

What tests are done later in pregnancy?

The following tests are done later in pregnancy:

- A repeat CBC
- Rh antibody test
- Glucose screening test
- Group B streptococci (GBS)

When will I be tested for Rh antibodies?

If you are Rh negative, your blood will be tested for Rh antibodies between 28 weeks and 29 weeks of pregnancy. If you do not have Rh antibodies, you will receive Rh immunoglobulin. This shot prevents you from making antibodies during the rest of your pregnancy. If you have Rh antibodies, you may need special care.

What is a glucose screening test and what can the results show?

This screening test measures the level of glucose (sugar) in your blood. A high glucose level may be a sign of gestational diabetes. This test usually is done between 24 weeks and 28 weeks of pregnancy. If you have risk factors for diabetes or had gestational diabetes in a previous pregnancy, screening may be done in the first trimester of pregnancy.

What is GBS and why are pregnant women tested for it?

GBS is a type of bacteria that lives in the vagina and rectum. Many women carry GBS and do not have any symptoms. GBS can be passed to a baby during birth. Most babies who get GBS from their mothers do not have any problems. A few, however, become sick. This illness can cause serious health problems and even death in newborn babies. GBS usually can be detected with a routine screening test that is given between 35 weeks and 37 weeks of pregnancy. For this test, a swab is used to take samples from the vagina and rectum.

What happens if my GBS screening test result is positive?

If your GBS test result is positive, antibiotics can be given during labor to help prevent the baby from becoming infected.

What is the difference between screening tests and diagnostic tests for birth defects?

Screening tests are done during pregnancy to assess

the risk that the fetus has certain common birth defects. A screening test cannot tell whether the baby actually has a birth defect. There is no risk to the fetus with having screening tests.

Diagnostic tests actually can detect many, but not all, birth defects caused by defects in a gene or chromosomes (see FAQ094 "Genetic Disorders"). Diagnostic testing may be done instead of screening if a couple has a family history of a birth defect, belongs to a certain ethnic group, or if the couple already has a child with a birth defect. Diagnostic tests also are available as a first choice for all pregnant women, including those who do not have risk factors. Some diagnostic tests carry risks, including a small risk of pregnancy loss.

What is the first step in screening for birth defects?

Screening for birth defects begins by assessing your risk factors. Early in your pregnancy, your health care professional may give you a list of questions to find out whether you have risk factors, such as a personal or family history of birth defects, belonging to certain ethnic groups, maternal age of 35 years or older, or having preexisting diabetes. In some situations, you may want to visit a genetic counselor for more detailed information about your risks.

What is a carrier test?

A carrier test can show if you or your partner carry a gene for a certain disorder, such as cystic fibrosis. Carrier tests can be done before or during pregnancy. Carrier testing often is recommended if you or your partner have a genetic disorder, have a child with a genetic disorder, have a family history of a genetic disorder, or belong to an ethnic group that has an increased risk of specific disorders. Also, cystic fibrosis carrier screening is offered to all women of reproductive age because it is one of the most common inherited disorders.

What are other types of screening tests for birth defects that can be done during pregnancy?

Screening tests include an ultrasound exam in combination with blood tests that measure the levels of certain substances in the mother's blood.

What are the types of diagnostic tests for birth defects that can be done during pregnancy?

Diagnostic tests for birth defects include amniocentesis, chorionic villus sampling, and a targeted ultrasound exam.

Can I choose whether or not to have testing for birth defects?

Whether you want to be tested is a personal choice. Knowing beforehand allows the option of deciding not to continue the pregnancy. If you choose to continue the pregnancy, it can give you time to prepare for having a child with a particular disorder and to organize the medical care that your child may need. Your health care professional or a genetic counselor can discuss the options with you and help you decide.

Glossary

Acquired Immunodeficiency Syndrome (AIDS): A group of signs and symptoms, usually of severe infections, occurring in a person whose immune system has been damaged by infection with human immunodeficiency virus (HIV).

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.

Antibiotics: Drugs that treat certain types of infections.

Antibodies: Proteins in the blood produced in reaction to foreign substances, such as bacteria and viruses that cause infection.

Bacteria: One-celled organisms that can cause infections in the human body.

Carrier: A person who shows no signs of a particular disorder but could pass the gene on to his or her children.

Cells: The smallest units of a structure in the body; the building blocks for all parts of the body.

Chlamydia: A sexually transmitted infection caused by bacteria that can lead to pelvic inflammatory disease and infertility.

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

Chromosomes: Structures that are located inside each cell in the body and contain the genes that determine a person's physical makeup.

Cystic Fibrosis: An inherited disorder that causes problems in digestion and breathing.

Diabetes Mellitus: A condition in which the levels of sugar in the blood are too high.

Fetus: The developing organism in the uterus from the ninth week of pregnancy until the end of pregnancy.

Gene: A segment of DNA that contains instructions for the development of a person's physical traits and control of the processes in the body. Genes are the basic units of heredity and can be passed down from parent to offspring.

Genetic Counselor: A health care professional with special training in genetics and counseling who can provide expert advice about genetic disorders and prenatal testing.

Gestational Diabetes: Diabetes that arises during pregnancy.

Glucose: A sugar that is present in the blood and is the body's main source of fuel.

Gonorrhea: A sexually transmitted infection that may lead to pelvic inflammatory disease, infertility, and arthritis.

Human Immunodeficiency Virus (HIV): A virus that attacks certain cells of the body's immune system and causes acquired immunodeficiency syndrome (AIDS).

Preeclampsia: A disorder that can occur during pregnancy or after childbirth in which there is high blood pressure and other signs of organ injury, such as an abnormal amount of protein in the urine, a low number of platelets, abnormal kidney or liver function, pain over the upper abdomen, fluid in the lungs, or a severe headache or changes in vision.

Prenatal Care: A program of care for a pregnant woman before the birth of her baby.

Rh Factor: A protein that can be present on the surface of red blood cells.

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

Sexually Transmitted Infections (STIs): Infections that are spread by sexual contact, including chlamydia, gonorrhea, human papillomavirus infection, herpes, syphilis, and infection with human immunodeficiency virus (HIV, the cause of acquired immunodeficiency syndrome [AIDS]).

Syphilis: A sexually transmitted infection that is caused by an organism called Treponema pallidum; it may cause major health problems or death in its later stages.

Trimester: Any of the three 3-month periods into which pregnancy is divided.

Tuberculosis (TB): A disease caused by bacteria that usually affects the lungs but also can affect other organs in the body. If not treated, it can be fatal.

Ultrasound Exam: A test in which sound waves are used to examine internal structures. During pregnancy, it can be used to examine the fetus.

Screening Tests for Birth Defects

What is a birth defect?

A birth defect is a problem that is present at birth, although it may not be noticed until the child is older. Birth defects may affect any part of the body, including major organs such as the heart, lungs, or brain. The defect may affect the baby's appearance, a body function, or both.

What causes birth defects?

Some birth defects are caused by problems with chromosomes. Others are caused by a gene that is passed from parent to child. Some birth defects result from exposure to harmful agents.

What are some examples of chromosome disorders?

Aneuploidy is a condition in which there are missing or extra chromosomes. The most common aneuploidy is called a trisomy, in which there is an extra chromosome. A common trisomy is trisomy 21 (Down syndrome). Other trisomies include trisomy 13 (Patau syndrome) and trisomy 18 (Edwards syndrome).

A monosomy is a condition in which there is a missing chromosome. A common monosomy is Turner syndrome, in which a female has a missing or damaged X chromosome.

What are inherited disorders?

Inherited disorders are caused by defective genes. These disorders are passed down by parents to their children. Some inherited disorders are more common in certain races and ethnic groups, such as sickle cell disease (African American), cystic fibrosis (non-Hispanic white), and Tay–Sachs disease (Ashkenazi Jewish, Cajun, and French Canadian).

What other things can cause birth defects?

Birth defects also may be caused by exposure to harmful agents, such as medications, chemicals, and infections. Some birth defects may be caused by a combination of factors. For most birth defects, the cause is not known.

How can I find out if I am at increased risk of passing on a genetic disorder?

Your health care provider or a genetic counselor can help find out if you are at increased risk of passing on a genetic disorder by asking about your personal and family health history.

What factors may increase my risk of passing on a genetic disorder?

Most babies with birth defects are born to couples without risk factors. However, the risk of birth defects is higher when certain factors are present. You are at increased risk if:

- you have a genetic disorder
- you already have a child who has a genetic disorder
- there is a family history of a genetic disorder
- you belong to an ethnic group that has a high risk of certain genetic disorders

What types of prenatal tests are available to address concerns about birth defects?

The following prenatal tests are available:

- Carrier tests—These screening tests can show if a person carries a gene for an inherited disorder. Carrier tests can be done before or during pregnancy. Cystic fibrosis carrier screening is offered to all women of reproductive age because it is one of the most common genetic disorders.
- Screening tests—These tests assess the risk that a baby will have Down syndrome and other chromosome problems, as well as neural tube defects. These tests do not tell whether the fetus actually has these disorders.
- Diagnostic tests—These tests can provide information about whether the fetus has a genetic condition and are done on cells obtained through amniocentesis, chorionic villus sampling, or, rarely, fetal blood sampling. The cells can be analyzed using different techniques.

What are the different types of screening tests for birth defects that can be performed during pregnancy?

Screening tests are performed during different trimesters of pregnancy. The following table lists the different types of screening tests:

Screening Test	Test Type	What Does It Screen For?	Down Syndrome Detection Rate
Combined first-trimester screening	Blood test for PAPP-A and hCG, plus an ultrasound exam	Down syndrome Trisomy 13 Trisomy 18	82–87%
Second-trimester single screen for neural tube defects	Blood test for AFP	Neural tube defects	85%
Second-trimester triple screen	Blood test for AFP, hCG, and estriol	Down syndrome Trisomy 18 Neural tube defects	69%
Second-trimester quad screen	Blood test for AFP, hCG, estriol, and inhibin-A	Down syndrome Trisomy 18 Neural tube defects	81%
Integrated screening	Blood test for PAPP-A and an ultrasound exam in the first trimester, followed by quad screen in the second trimester	Down syndrome Trisomy 18 Neural tube defects	94-96%
Integrated screening (blood test only)	Same as integrated screening but no ultrasound exam	Down syndrome Trisomy 18 Neural tube defects	85-88%
Contingent sequential	First-trimester combined screening result: • Positive: diagnostic test offered • Negative: no further testing • Intermediate: second-trimester screening test offered	Down syndrome Trisomy 18 Neural tube defects	88-94%
Stepwise sequential	First trimester combined screening result: • Positive: diagnostic test offered • Negative: second-trimester screening test offered	Down syndrome Trisomy 18 Neural tube defects	95%

Abbreviations: AFP, alpha-fetoprotein; hCG, human chorionic gonadotropin; PAPP-A, pregnancy-associated plasma protein A

Do I have a choice between having screening tests or having diagnostic tests?

If a screening test shows an increased risk of a birth defect, diagnostic tests may be done to determine if a specific birth defect is present. Diagnostic testing may be done instead of screening if a couple is at increased risk of certain birth defects. Diagnostic testing also is offered as a first choice to all pregnant women, even those who do not have risk factors. Your health care provider will discuss all of the testing options with you and recommend the tests that best fit your needs.

What are the advantages and disadvantages of diagnostic tests compared with screening tests?

The main benefit of having diagnostic testing instead of screening is that it tells you whether or not the baby will be born with a chromosome disorder or a specific inherited disorder. The main disadvantage is that diagnostic tests can pose some risks to the pregnancy.

Do I have to have these tests?

Although screening tests for birth defects are offered to all pregnant women, it is your choice whether to have them done. Knowing whether your baby is at risk of or has a birth defect beforehand allows you to prepare for having a child with a particular disorder and to organize the medical care that your child may need. You also may have the option of not continuing the pregnancy.

Glossary

Alpha-fetoprotein (AFP): A protein produced by a growing fetus; it is present in amniotic fluid and, in smaller amounts, in the mother's blood.

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.

Aneuploidy: Having an abnormal number of chromosomes.

Carrier: A person who shows no signs of a particular disorder but could pass the gene on to his or her children.

Cells: The smallest units of a structure in the body; the building blocks for all parts of the body.

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

Chromosomes: Structures that are located inside each cell in the body and contain the genes that determine a person's physical makeup.

Cystic Fibrosis: An inherited disorder that causes problems in digestion and breathing.

Diagnostic Tests: Tests that look for a disease or cause of a disease in people who are believed to have or who have an increased risk of a disease.

Estriol: A substance made by the placenta and the liver of the fetus.

Fetus: The developing organism in the uterus from the ninth week of pregnancy until the end of pregnancy.

Gene: A segment of DNA that contains instructions for the development of a physical trait or control of a process in the body. Genes are the basic units of heredity and can be passed down from parent to offspring.

Genetic Counselor: A health care professional with special training in genetics and counseling who can provide expert advice about genetic disorders and prenatal testing.

Human Chorionic Gonadotropin (hCG): A hormone produced during pregnancy; its detection is the basis for most pregnancy tests.

Inhibin-A: A substance made by the placenta during pregnancy.

Monosomy: A condition in which there is a missing chromosome.

Neural Tube Defects: Birth defects that result from incomplete development of the brain, spinal cord, or their coverings.

Pregnancy-Associated Plasma Protein-A (PAPP-A): A protein made by the fetus and placenta during pregnancy.

Screening Tests: Tests that look for possible signs of disease in people who do not have symptoms.

Sickle Cell Disease: An inherited disorder in which red blood cells have a crescent shape, causing chronic anemia and episodes of pain. It occurs most often in African Americans.

Tay-Sachs Disease: An inherited birth defect that causes mental retardation, blindness, seizures, and death, usually by age 5 years. It occurs mostly in people of Eastern European Jewish (Ashkenazi Jews), Cajun, and French Canadian descent.

Trimesters: The three 3-month periods into which pregnancy is divided.

Trisomy: A condition in which there is an extra chromosome.

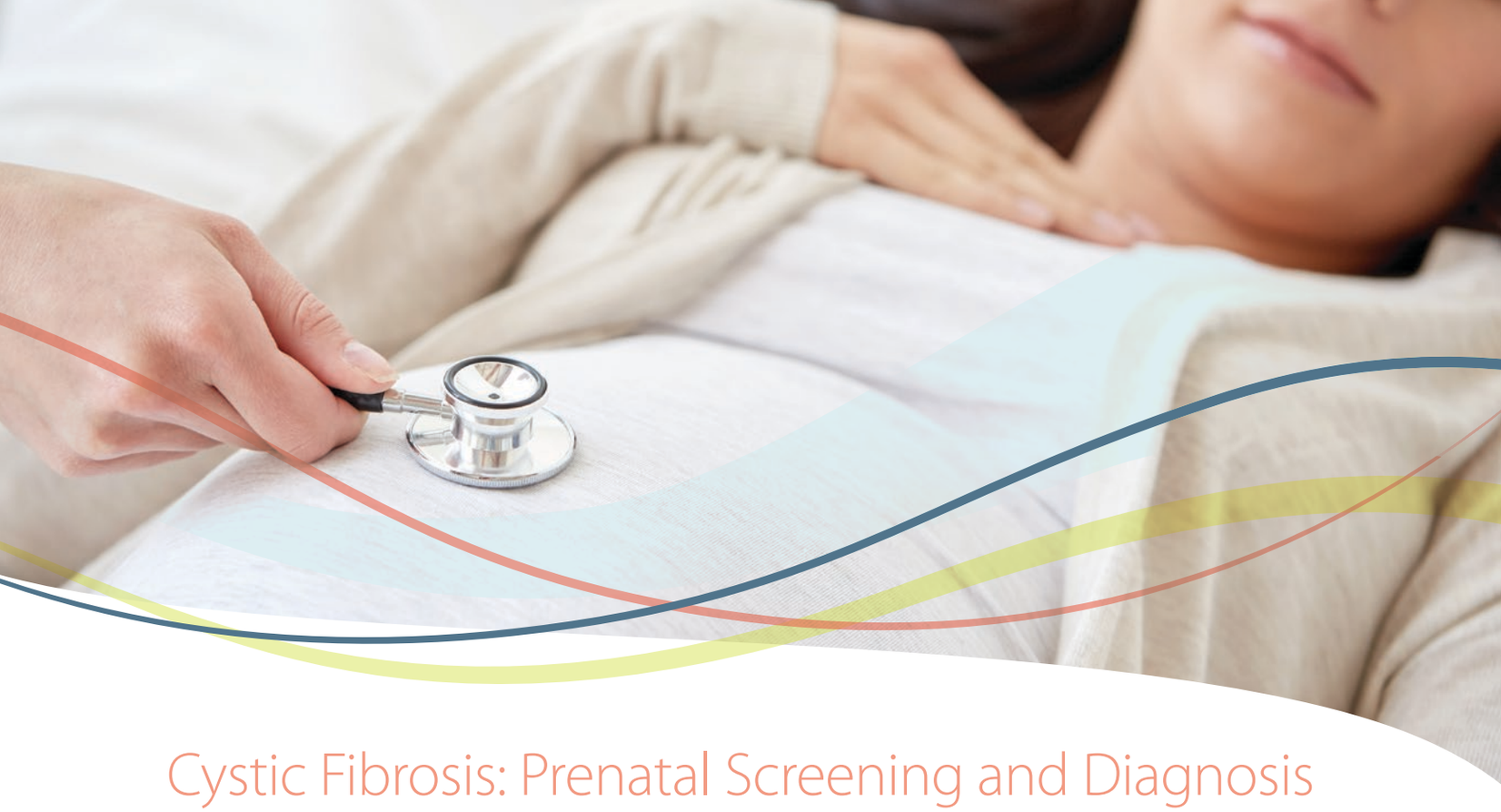
Trisomy 13 (Patau Syndrome): A genetic disorder that causes serious heart defects and other problems with development. Most infants with trisomy 13 die within the first year of life.

Trisomy 18 (Edwards Syndrome): A genetic disorder that causes serious mental and developmental problems. Most infants with trisomy 18 die within the first year of life.

Trisomy 21 (Down Syndrome): A genetic disorder in which abnormal features of the face and body, medical problems such as heart defects, and intellectual disability occur.

Turner Syndrome: A condition affecting females in which there is a missing or damaged X chromosome. It causes a webbed neck, short height, and heart problems.

Ultrasound Exam: A test in which sound waves are used to examine internal structures. During pregnancy, it can be used to examine the fetus.



Cystic Fibrosis: Prenatal Screening and Diagnosis

What is cystic fibrosis (CF)?

Cystic fibrosis (CF) is a genetic disorder. It is caused by an abnormal gene that is passed from parent to child. It is a lifelong illness that affects all of the organs of the body and often causes problems with digestion and breathing. It does not affect a person's looks or mental ability. In some cases, CF poses a serious risk to a person's health and shortens the life span. Despite their physical problems, many people with CF attend school, have careers, and lead full lives.

What causes CF?

Cystic fibrosis is a recessive disorder. In a recessive disorder, both parents must carry a copy of the abnormal gene for the problem to occur in their child. A person who has one copy of an abnormal gene for a recessive disorder is a carrier for that disorder, even though he or she may show no signs of it. If both parents are carriers, each of their children has a 25% chance of having the disorder. Put another way, this couple has a 1-in-4 chance of having a child with CF.

What are the symptoms of CF?

The symptoms of CF can vary in type and severity. Many people with CF produce a thick, sticky mucus in their bodies. This mucus builds up and clogs the lungs, which makes it hard to breathe, and can lead to infection. It also can affect the digestive organs, making it hard for the body to break down and absorb food. Most males with CF are sterile and cannot father children.

Is treatment available for CF?

New drugs and treatments have improved the outlook for people with CF, but it is still a lifelong disease. To treat lung problems, most children with CF need to have physical therapy for about a half hour every day. This therapy helps clear mucus from the lungs. It is easy to do and can be done by parents or other family members.

What are risk factors for CF?

The risk of being a CF carrier is higher in certain races and ethnic groups. It occurs more often in white people than in other racial groups. The risk also is increased in families with a history of CF.

Can I be tested to assess whether I am a CF carrier?

Carrier testing can be done for couples planning a pregnancy or during pregnancy to assess their risk. The test is done on a blood sample. Carrier testing also is available to all pregnant women. If testing shows that a couple is at high risk, more testing can be done during pregnancy to see whether their fetus has CF.

What does it mean if test results for one partner are negative?

If your test results are negative, the chance that you are a CF carrier is small. There are some rare CF gene defects that the test does not detect. For this reason, you could be told your test result is negative, and you could still be a carrier. The likelihood of this is very small.

What does it mean if test results for one partner are positive?

If the test results show that one partner is a carrier, the next step is to test the other partner. Both parents must be CF carriers for the baby to have CF. If one parent has a negative test result, the chance that the baby will have CF is small. Because the risk is small, if one partner is a carrier but the other has a negative result, no further testing is recommended. If the father is not available for a carrier test, a genetic counselor may be able to help you decide whether to have prenatal testing of the fetus to see if it has CF.

What does it mean if test results for both partners are positive?

If two people who are both CF carriers have a baby, there is a 25% chance that the baby will have CF. However, it is more likely that the baby will be a carrier, like the parents, and will have the gene but will not have the disease. It also is possible that although the parents are both carriers, the baby will not be a CF carrier.

If both partners are positive, what follow-up tests are appropriate and what do they assess?

If both partners are CF carriers, further prenatal testing can be done to see if the baby has CF. This testing is not recommended when only one partner is a carrier. Parents may want to know if the baby will have CF so that they can prepare for the care of a child with special health care needs, or they may choose to end the pregnancy.

What prenatal tests can be done to detect CF and other disorders?

Prenatal tests done to detect CF and other disorders are chorionic villus sampling (CVS) and amniocentesis (see FAQ164 "Diagnostic Tests for Birth Defects"). CVS can be performed after 9 completed weeks of pregnancy. Amniocentesis can be performed between 15 weeks and 20 weeks of pregnancy.

What are my options if diagnostic test results show that the fetus has CF?

Two options are available:

1. Continue the pregnancy and prepare for a child with CF. Couples can use this time to learn as much as possible about the disease, current treatment options, and the experiences of other families who have a child with CF.
2. End the pregnancy. Each state has its own laws on pregnancy termination. You should discuss this decision with your health care provider. You also may want to talk with your partner, counselors, and close friends.

What should partners who are CF carriers be aware of when thinking about future pregnancies?

If a test result shows that you are a CF carrier, the result is definite and will not change. If both partners are carriers, it means that in each pregnancy the baby will have a 25% (1-in-4) chance of having CF. If you want to know whether your baby will have CF, you will need to have amniocentesis or CVS in each pregnancy. Other options include the following:

- Adoption
- Using donor sperm or donor eggs (but the donor should be tested for CF carrier status)
- Using in vitro fertilization with your own sperm and eggs, and then using preimplantation genetic diagnosis to see if the fertilized egg has CF or is a CF carrier.

Glossary

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.

Carrier: A person who shows no signs of a particular disorder but could pass the gene on to his or her children. Chorionic Villus Sampling (CVS): A procedure in which a small sample of cells is taken from the placenta and tested. Fetus: The developing offspring in the uterus from the ninth week of pregnancy until the end of pregnancy.

Gene: A DNA "blueprint" that codes for specific traits, such as hair and eye color.

Recessive Disorder: A type of a genetic disorder in which one copy of the defective gene must be passed from each parent for a child to be affected.

Preimplantation Genetic Diagnosis: A type of genetic testing that can be done during in vitro fertilization. Tests are performed on the fertilized egg before it implants in the uterus.

Spinal Muscular Atrophy (SMA) Testing

Spinal muscular atrophy is the most common inherited cause of early childhood death

Normally, healthy nerve cells in the brain called motor neurons send messages to the muscles to tell them when and how to move. But SMA damages these brain cells and prevents those important messages from reaching the muscles. When the muscles don't get direction on how to move, they become inactive, get smaller, and begin to waste away (a condition known as atrophy). SMA does not affect intelligence. There is no cure or treatment.

There are several types of SMA

The most common form, Type 1, which affects about 70 percent of patients with the disease is the most severe. Children with Type 1 SMA usually die from respiratory failure before the age of two. Children with type 2 SMA may be able to sit unaided, but cannot stand or walk unaided. Although they face many challenges, children with type 3 SMA are able to walk unaided and have a normal lifespan. Type 4 is the adult form of the disorder. Most people affected by this type start having symptoms after age 35, and these symptoms slowly get worse over time. Because it develops slowly, many people with type IV SMA don't know that they have it until years after symptoms begin.

About one in every 40 people carries a mutated SMN1 gene and every year, about one in every 6,000-10,000 babies is born with SMA

Please make sure you understand the following points

If you are unclear, ask your healthcare provider before deciding on this test.

- The purpose of this test is to determine whether I am a carrier of one of the common genetic abnormalities that cause SMA
- This test does not detect all carriers of this disease
- The decision to have carrier testing is completely mine
- If I am a carrier, testing my partner will help me learn more about the chance our baby could have SMA
- If one parent is a carrier and the other is not, it is still possible that the baby will have SMA, but the chance is less than 1 percent
- If both parents are carriers, prenatal testing is available to find out whether or not the baby has inherited the abnormal SMA genes

Fragile X Syndrome Testing

Fragile X is caused by a change in the Fragile X Mental Retardation gene

This abnormal gene can be passed from generation to generation. Fragile X syndrome is usually passed on by a gene that is carried by a woman. One in 259 women are carriers. Fragile X is found in all ethnic groups and can occur in families with no history of mental retardation or autism.

Fragile X Syndrome is the most common inherited cause of mental impairment

Fragile X affects approximately 1 in 4,000 males and 1 in 8,000 females. The majority of males with Fragile X have significant intellectual disability. The spectrum ranges from learning disabilities to sever mental retardation and autism. About one third of the females affected with Fragile X have significant intellectual disabilities.

If a mother is a carrier, there is a 50 percent chance to have a child with Fragile X Syndrome.

Please make sure you understand the following points.

If you are unclear, ask your healthcare provider before deciding on this test.

- The purpose of this test is to determine whether I am a carrier of one of the common gene abnormalities that cause Fragile X Syndrome
- This test does not detect all carriers of this disease
- The decision to have carrier testing is completely mine
- If I am a carrier, prenatal testing is available to find out whether or not the baby has inherited the abnormal fragile X gene