OFFICE ROUTINE FOR OBSTETRICAL CARE

One of your early visits with the doctor will include a complete physical examination and laboratory work including blood work, urinalysis, a Pap smear, and cultures if needed. The results of this lab work will be reviewed with you at your next visit. The Provider and Medical Assistant will each spend time with you discussing your obstetrical care and any particular questions you may have. This patient resource section of our website contains an abundance of information about the office and the Family Maternity Center.

Your baby's "due date" is a day 40 weeks past the first day of your last menstrual period. In some cases we will utilize the ultrasound equipment in our office to pinpoint your due date. Please keep in mind that your baby may come a few days before or after this due date, as babies are not very proficient at keeping calendars! First pregnancies, in particular, tend to go beyond their due dates.

After your initial OB workup, monthly visits will include a "tummy check" to measure your baby's growth. The Medical Assistant will check your weight and blood pressure. The provider will have time to answer any questions you might have.

You will not be asked to provide a urine specimen at each prenatal appointment. If indicated, due to symptoms you report, we may request a urine specimen to check for protein (the spilling of protein into the urine is one of the symptoms of toxemia), sugar (some women will exhibit signs of diabetes only when they are pregnant), and nitrates (which can indicate a possible non-symptomatic urinary tract infection).

You may take one prenatal vitamin daily. These can be purchased over-the-counter at any pharmacy. If you have special needs, other vitamin supplements may be recommended; otherwise, we suggest the most economical brand of prenatal vitamins your pharmacy carries. If you take "Gummi" vitamins, most of them do not include iron, so take an over-the-counter iron supplement along with them, as iron supplementation is important during pregnancy.

Throughout your third trimester, your birth plan will be discussed. You will have some decisions to make at this time. You will need to tell us who your baby's pediatrician will be, if you would want a son circumcised, and whether or not you plan to breast feed.

Anesthesia will be discussed, and we will double-check that you've signed the Consent for Care forms with the hospital. You can do this by stopping by the desk at the Family Maternity Center after 28 weeks of pregnancy.

GENERAL INFORMATION

The providers at EvergreenHealth Obstetrics & Gynecology Care, Coral perform their patients' deliveries when they are on call and/or if you have a scheduled c-section. Should you go into labor and your doctor is not available, your baby will be delivered by the provider who is on call for our clinic. The providers at EvergreenHealth Obstetrics & Gynecology Care, Coral share many of the same beliefs when it comes to pregnancy and delivery. You may also see one of our Nurse Practitioners or Physicians Assistant for your initial pregnancy visit, infection checks, special education, some routine prenatal visits, or if your provider is not available.

Prenatal Education

There are many different prenatal classes available on the Eastside, they are especially valuable if this is your first pregnancy. If you have taken a class during a previous pregnancy, refresher courses are available and highly recommended.

EvergreenHealth Family Maternity Center (FMC) offers a variety of prenatal classes covering breastfeeding, nutrition, prenatal fitness and exercise, childbirth classes and other related topics. We suggest that you tour the Family Maternity Center (FMC) at EvergreenHealth so you are familiar with the facility. For more information on classes and tours at EvergreenHealth, please call 425-899-3000 or visit the website. You will need to register for classes early as they fill quickly, and those who wait may not get their first choice.

Shared Decision Making Re: Trial of Labor After Cesarean Section

We invite you to learn about the options for delivery after having a previous Cesarean Section. There is a class held monthly at EvergreenHealth where you will learn about the delivery options available to you and how to make the best decision to support your care and the care of your baby during delivery. Ask your provider if you are eligible for a Trial of Labor After Cesarean Section and if you should attend this class.

OB SCHEDULING

Name: _____

Estimated Due Date: _____

*Virtual Option May be available

Weeks	Visit	Lab/Ultrasound	Appointment Date/Time
8-10	First OB visit with Provider	Prenatal Lab work, bedside ultrasound	_____
12-13	Optional Nuchal Translucency Ultrasound & Provider visit	Genetic testing (optional)	_____
16*	Provider Visit	Optional AFP	_____
20-22	Ultrasound & Provider visit	Comprehensive anatomy	_____
24*	Provider Visit		_____
28	Provider Visit	Diabetes, Anemia/Antibody Screen (if RH negative)	_____
32*	Provider Visit		_____
34*	Provider Visit		_____
36	Provider Visit	Group B Strep culture	_____
37	Provider Visit		_____
38	Provider Visit		_____
39	Provider Visit		_____
40	Provider Visit		_____
41	Provider Visit	Possible Induction	_____

Updated 6/20/23

PRENATAL APPOINTMENT OVERVIEW

Your baby's "due date" is a day 40 weeks past the first day of your last menstrual period. An ultrasound will help to confirm your due date. Please keep in mind that your baby may come a few days before or after this "due date", as babies are not very proficient at keeping calendars!

Routine Appointments

First appointment: 8-11 weeks
Following appointments:

Once a month from 12 to 32 weeks
Every two weeks from 32 to 36 weeks
Once a week from 36 weeks till delivery

Visit Routine

At each appointment we will check your weight and blood pressure. If your pregnancy is far enough along to hear your baby's heartbeat, usually by 12 weeks, we will check fetal heart tones at each visit, as well. As your pregnancy progresses, your provider will measure uterine growth with a tape measure on your abdomen.

Ultrasounds

Confirmation Ultrasound: 8-11 weeks, routine
You will receive an ultrasound at one of your first prenatal appointments to verify your gestation (if this has not already been done), that the pregnancy is in the uterus, and that there are no complications at this time. This ultrasound is performed using a vaginal probe. No preparation is necessary.

Nuchal Translucency (NT) Ultrasound: 11-13.6 weeks genetic testing
This ultrasound helps to identify a potential for chromosomal abnormalities and possible cardiac anomalies. This is an abdominal ultrasound and having a full bladder is requested.

Anatomy Ultrasound: 20 weeks, routine
This ultrasound is performed to confirm that your baby is growing and developing well. The ultrasound technician will take multiple measurements and check the organs that have developed at this stage. You may also find out the gender of your baby if you choose and if the baby is in a cooperative position. This is an abdominal ultrasound. No preparation is necessary. A vaginal ultrasound to evaluate the cervix may be performed as well depending on the abdominal measurement.

Any other ultrasounds are done only when deemed medically necessary by the Physician.

** We do not allow outside recording of the ultrasounds but we do have the ability to send pictures and small video clips to your phone that you can then share as you wish. We can also print out pictures for you.

CHROMOSOMAL ABNORMALITIES IN LIVEBORNS

Maternal Age
Risk for Down Syndrome
Total Risk for Chromosomal Abnormalities

Testing

The following is very important information to help you make informed decisions about testing in your pregnancy. It is a somewhat unwelcome experience to talk about things that could go wrong the beginning of your pregnancy, when you are excited and just getting ready for parenthood. Please remember, these are rare occurrences; we are fishing for unlikely problems. So, though you need to have a very basic understanding of the risks that can befall pregnancy, you do not need to take them to heart or assume that any of them will affect you. It is far likelier that you will not have a problem in your pregnancy.

If you do choose to have any of this testing, it is important to explore how you would react to, or act upon, the results. Ideally, you should have a general idea about how far you would take the testing or if you would choose to terminate if you found out your pregnancy had a chromosome abnormality. These cannot be treated or prevented.

Some of the most complicated questions we will be asking you will be about genetic testing in your pregnancy. Genetic testing falls under two basic categories: 1) risk of problems for current pregnancy and 2) your general genetic risk. So let's divide and conquer to address these two general topics separately.

Risk of Problems for Current Pregnancy

This generally applies to the risk of your current pregnancy having a chromosome problem. Most people associate this with Down syndrome (which is the most common), but other chromosome abnormalities are also out there. The most common type of testing is risk testing. This means that the tests we offer don't actually test for the problem, but instead test for the *risk* of the problem. The two types of testing offering in this category are the combined screen and the quad screen. The combined screen is a combination of an ultrasound measurement of the nuchal fold (back of the fetus' neck *combined* with a blood test from the mother. This test is done between 11 and 13.6 weeks. You make a separate appointment for this. The quad screen is simply a blood test that is done between 15 and 22 weeks (most accurate between 16 and 18 weeks). We can do this test at the same time as one of your regular appointments. Both of these tests come out with a result that reads as a risk; for example, the result will say that there is a 1/*some# risk* that the pregnancy has a chromosome problem.

These tests give us *risk* results for trisomy 21, or Down syndrome with an occurrence of 4.6/10,000 newborns; trisomy 13, or Patau syndrome, with an occurrence of 1/10,000 newborns; and trisomy 18, or Edwards syndrome, with an occurrence of 1/6,000 newborns. The *risk* for having these syndromes increases with increasing maternal age. Tests have to have a cutoff to determine when we say the *risk* is high or low. Truly, this risk will be different for each individual, but the experts have determined that if your risk as shown by one of these tests is greater than about 1/270, it is an abnormal test result.

This means only that we will offer you further testing and possibly diagnostic testing. Diagnostic testing means that we do an invasive test to obtain actual cells from either the amniotic fluid or placenta. If we have cells from these sources, we can analyze them in order to determine what the chromosome number is rather than just the risk. The invasive tests are amniocentesis and chorionic villi sample (CVS). We do not perform these tests in our office. We refer you to EvergreenHealth Maternal-Fetal Medicine (MFM) services. MFM is our group of specialists who deal with the more specialized diagnoses and procedures related to pregnancy. They are specially trained to do these procedures, so we leave them in their capable hands.

Why is 35 years old elderly?
When all of these tests were originally being developed, it became apparent that the medical field did not have the resources to offer them to everybody. Specifically, there were not enough trained individuals available to offer amniocentesis to every pregnant woman. As well, it would be a considerable economic burden to do this with every pregnancy. So how to solve this? Experts decided to draw the line for offering these tests at the age at which the risk of Down syndrome (the most common chromosomal abnormality) was the same as the risk of a miscarriage from the amniocentesis. At the age of 35, the risk is about 1/300 for either of these outcomes. Is this appropriate and fair? Are these really equivalent outcomes (Down syndrome vs. miscarriage?) These are difficult questions to answer. But at this time, some kind of decision had to be made with regard to resource use. These days, we basically have the resources to offer all tests to anybody who desires them. And in fact our field's professional organization, (the American College of Obstetrics and Gynecology, or ACOG) has decreed that we must at least offer some form of genetic risk assessment to all patients. The challenge is that all of these tests cost money. So which should be paid for at what age?

Most insurance companies continue to use the age of 35 to determine that extra or more advanced testing will be paid for in a pregnancy. So it breaks down as follows. To the best of our knowledge, all or most insurance companies will pay for the quad screen for all pregnancies. Some will pay for the combined screen for all pregnancies, but most will pay only after age 35. Amniocentesis and CVS are mostly paid for if you are over 35 years of age or if one of the other tests show "high" risk (1/250). Despite all of that, you do have the right to have any of the testing; this includes going to a genetic counselor. You should just be aware that your insurance may or may not cover your request, depending on your individual risk. It is up to you to call your insurance company to determine coverage if you want one of the more advanced tests.

A new set of tests has been developed. These tests are now commercially available and can be offered to you in your pregnancy if you have an abnormal screening test. The tests take a blood sample from you that we can screen for fetal DNA that is in your bloodstream. Then, since we have that DNA, we can directly test it for chromosome abnormalities. The different tests available to us for your use are listed on a handout enclosed in this notebook. It is important that you call your insurance company regarding coverage and then let your provider know which test you would prefer. The numbers and websites of the various testing companies are also included in the handout. There is a video, as well, done by EvergreenHealth MFM genetic counselors than explains more about these testing options. The link to that YouTube video can be found at **https://www.evergreenhealth.com/prenatal-testing-and-diagnosis** find the link please watch this video first at the bottom of that page.

Your Own Genetic Risks

Everybody brings their own unique set of genetic risks and assets to the table. With increasing technology, we are able to test you for more and more genetic risks. We are going to offer an overview of genetic diseases in the next paragraphs. We are not going to explain them all in this text in order to save space, but note that we've hyperlinked them all if you want more information.

We started out with the general ability to test for cystic fibrosis (www.ncbi.nlm.nih.gov/pubmedhealth/PMH0001167D)
Then we started offering testing for spinal muscular atrophy (www.ncbi.nlm.nih.gov/pubmedhealth/PMH0001991/)
Fragile X (www.ncbi.nlm.nih.gov/pubmedhealth/PMH0002633/)

These are the three basic tests that we offer you in our office. But, you should know that there are now several tests, looking for hundreds of diseases. If you are interested in this type of testing or you know that your family carries a certain disease, please let your provider know. We will refer you to the genetic counselors at EvergreenHealth MFM so they can determine your risk and offer you the appropriate tests.

For those of you reading this and feeling as if you're in a time machine going forward, we can relate. We get new information in our office every day about this test or that test that can now be done! We feel that each patient needs to decide how much of this information they want.

CHROMOSOMAL ABNORMALITIES IN LIVEBORNS

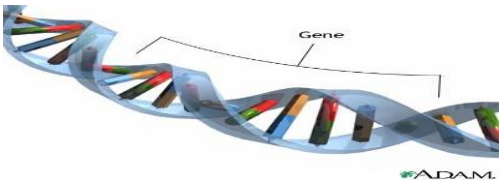
MATERNAL AGE	RISK FOR DOWN SYNDROME	TOTAL RISK FOR CHROMOSOMAL ABNORMALITIES
20	1/1,667	1/526
21	1/1,667	1/526
22	1/1,429	1/500
23	1/1,429	1/500
24	1/1,250	1/476
25	1/1,250	1/476
26	1/1,176	1/476
27	1/1,111	1/455
28	1/1,053	1/435
29	1/1000	1/417
30	1/952	1/385
31	1/909	1/385
32	1/769	1/322
33	1/602	1/286
34	1/485	1/238
35	1/378	1/192
36	1/289	1/156
37	1/224	1/127
38	1/173	1/102
39	1/136	1/83
40	1/106	1/66
41	1/82	1/53
42	1/63	1/42
43	1/49	1/33
44	1/38	1/26
45	1/30	1/21
46	1/23	1/16
47	1/18	1/13
48	1/14	1/10
49	1/11	1/8

Genetic Screening Options

Test	Description	When Test is Done	How Test is Done	Advantages	Disadvantages
Combined	Helps assess risk for Down Syndrome, Trisomy 13 and 18. This is a screening test, not diagnostic.	12.0-13.6 weeks	A blood test and ultrasound which measures the thickness of the fetal neck	Noninvasive to fetus. 85% pick-up of Down Syndrome and 80% pick-up of Trisomy 13 and 18	Not 100% accurate. 5%-10% false-positive rate. Not always covered by insurance plans
Nuchal Translucency Ultrasound (NT)	Helps identify a potential for chromosomal abnormalities and possible cardiac anomalies. This is a screening test, not diagnostic.	12.0-13.6 weeks	Abdominal ultrasound that measures the thickness of the fetal neck	Noninvasive to fetus	Not 100% accurate. Not always covered by insurance.
NIPT Fetal Cell-Free DNA test (i.e. MaterniT21, Panorama)	This test finds fetal cells in maternal blood and can help identify chromosomal abnormalities like Down Syndrome, Trisomy 13, 18, and others.	10-22 weeks	Blood test	Noninvasive to fetus. 99% accurate for picking up DS, 97% accurate for Trisomy 18, and 91% accurate for Trisomy 13	Not 100% accurate. 1-8% false-positive rate. Test is not approved for women under age 35. For women over 35, not always covered by insurance.
Alpha-fetoprotein (AFP)	Signals the potential for open tube defects (spina bifida). This is a screening test, not diagnostic.	15-22 weeks	Blood test	Noninvasive to fetus. Can pick up a suspicion of spina bifida 80-90% of the time. Typically covered by insurance	Not 100% accurate
Chorionic Villus Sampling (CVS)	This is a diagnostic test which assesses the presence of Down Syndrome and other chromosomal abnormalities. Referral to Maternal Fetal Medicine required.	11-13.6 weeks	A perinatologist does a procedure which collects a sample of the placenta through the mother's cervix or abdomen	99% accuracy for picking up chromosomal abnormalities. This test can be done earlier than amniocentesis.	1% chance of miscarriage following the procedure
Amniocentesis	This is a diagnostic test which detects the presence of chromosomal abnormalities like Down Syndrome and others. Referral to Maternal Fetal Medicine required	15-22 weeks	A perinatologist does a procedure with collects a sample of the amniotic fluid through the mother's abdomen.	99% accuracy for picking up chromosomal abnormalities, spina bifida, or other inherited diseases.	Less than 1% chance of miscarriage following the procedure.

Updated 7/1/24

UNDERSTANDING GENETIC TESTING OPTIONS



Interested in learning about genetic testing during pregnancy?
Are you confused about pregnancy genetic testing options?
Are you having a hard time deciding what is right for you and your baby?
Already pregnant? Your Obstetrician recommends that you view this video
Before or during the first several weeks of your pregnancy.

In the video below, EvergreenHealth licensed genetic counselors share their knowledge and expertise on genetic testing and prenatal testing options. They present an overview of the various testing options including first and second trimester screening, genetic carrier screening, CVS, cell free fetal DNA blood testing, amniocentesis and ultrasound.

Please watch this video to prepare for a discussion with your provider on your first OB visit, typically done at 8 to 10 weeks of pregnancy. Testing is done, typically, around your 12th week of pregnancy.

Find the video at the links below.
https://www.evergreenhealth.com/prenatal-testing-and-diagnosis
Find the link please first watch this video at the bottom of that page

Updated 4/22/22

NONINVASIVE PRENATAL SCREENING and GENETIC CARRIER SCREENING

Noninvasive Prenatal Screening

A noninvasive prenatal screen detects whether a pregnancy is at increased risk for certain chromosome conditions including Down syndrome, trisomy 13 and trisomy 18. Your healthcare provider may also choose to screen for conditions involving the sex chromosomes, X and Y, as well as other more rare conditions.

What are the advantages of getting screened?

For patients who screen negative, the noninvasive prenatal screen provides reassurances that their pregnancy has a significantly reduced risk of having one of these conditions. If the results are positive there is a significantly increased chance of having an affected pregnancy. In this case, your healthcare provider will discuss the option of diagnostic testing to determine if the pregnancy is affected and may discuss with you how best to prepare for these conditions.

Genetic Carrier Screening

Carrier screening, as prescribed by your healthcare provider, is a way to identify whether you are a "carrier" of various genetic disorders. Typically carriers are healthy individuals; but when two parents are carriers of the same genetic disorder they can have a child affected with the disorder. Knowing if you and your partner are carriers can help define your risk of having a child with that disorder.

What are the advantages of genetic carrier screening?

- For patients who are not carriers, expanded carrier screening provides reassurance that their child will be at a significantly reduced risk of developing any of the included genetic disorders.
- In most cases, if both parents are found to be carriers for the *same* disorder, there is a significantly increased chance of having an affected child, and this knowledge can help guide future decisions.
- For couples who are found to be at increased for an affected pregnancy:
    - Your healthcare provider can help you understand the medical options available if you are planning on having a family
    - If you are pregnant, you can pursue testing to determine if the pregnancy is affected, as well as work with your physician to learn about how to best care for treatable diseases.

UNDERSTANDING YOUR INSURANCE COVERAGE, BILLING, and YOUR FINANCIAL OBLIGATION

Genetic Testing Options: Noninvasive Prenatal Testing (NIPT)

Cell free fetal DNA testing is not a routine test. Should your physician recommend testing, or should you choose to have testing, you are encouraged to understand what your out of pocket cost may be.

Please do not call your healthcare provider's office, as we cannot assist with questions regarding your insurance coverage for this testing. Each patient's insurance carrier is contracted differently for this type of laboratory testing. Your healthcare provider's office does not bill for these laboratory services. Answers to questions regarding your benefits and coverage are best answered by your insurance carrier's representatives and the laboratory performing the test.

You are encouraged to contact the lab directly to learn about their financial services, quotes of fees and out of pocket responsibilities, discounts, installment plans, and Patient Assistance Programs.

Integrated Genetics	1-844-799-3243	integratedgenetics.com/transparency
Natera	1-844-611-2787	my.natera.com/billing

Once you have made your decision about testing, and have identified the lab you want to use, be sure to schedule an appointment with our office to proceed with the testing. It must be initiated with an order from your provider.

PATIENT EDUCATION

The American College of Obstetricians and Gynecologists
WOMEN'S HEALTH CARE PHYSICIANS

Carrier Screening

*Carrier screening is a type of genetic test that can tell you whether you carry **genes** for certain **genetic disorders**. Some people decide to have carrier screening before having children. Doing so allows you to find out your chances of having a child with a genetic disorder. Although carrier screening can be done during pregnancy, getting tested before pregnancy gives you a greater range of options and more time to make decisions.*

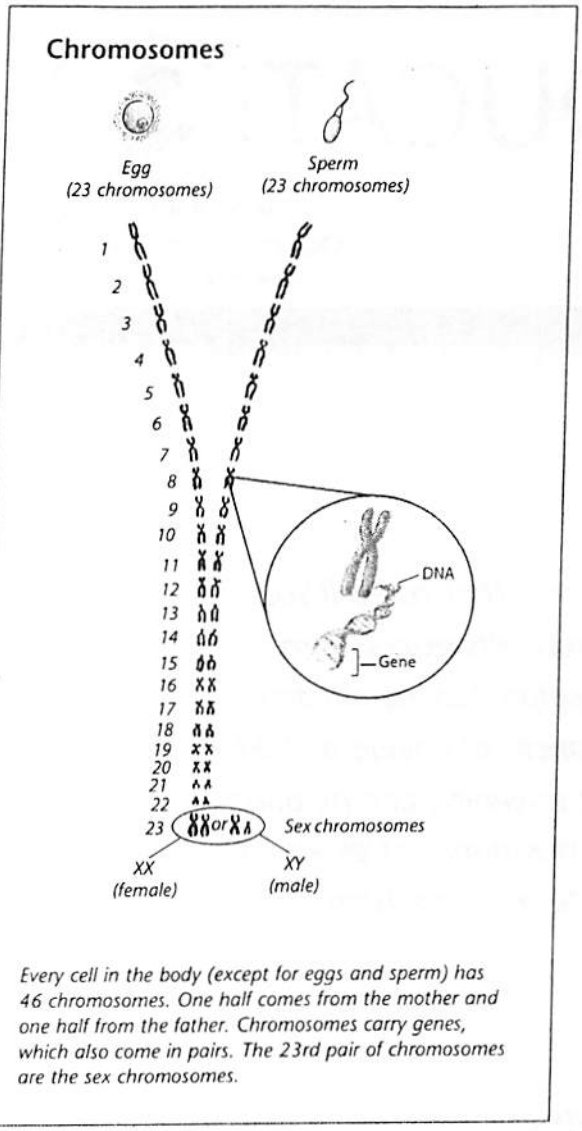
This pamphlet explains

- *how genes work*
- *inherited genetic disorders*
- *how testing is done and what the results mean*
- *timing of carrier screening*
- *approaches to carrier screening*
- *deciding whether to have carrier screening*
- *what to do if you find out that you are a carrier of a genetic disorder*

How Genes Work

A gene is a short segment of a chemical called ***DNA***. Genes are the coded instructions for your physical makeup and every process in your body. Genes come in pairs and are located on ***chromosomes***. Chromosomes also come in pairs (one from each parent). Most ***cells*** have 23 pairs of chromosomes, making a total of 46 chromosomes. However, ***sperm*** and ***egg*** cells each have a single set of 23 chromosomes—half the number of other cells. During ***fertilization***, when the egg and sperm join, the two sets of chromosomes come together. In this way, one half of a baby's genes come from the mother and one half come from the father.

A baby's sex depends on the ***sex chromosomes*** it gets. Egg cells only contain an X chromosome. Sperm cells can carry an X or a Y. A combination of XX results in a girl, and XY results in a boy.



Every cell in the body (except for eggs and sperm) has 46 chromosomes. One half comes from the mother and one half from the father. Chromosomes carry genes, which also come in pairs. The 23rd pair of chromosomes are the sex chromosomes.

Inherited Genetic Disorders

Some genetic disorders are caused by a change, or ***mutation***, in a gene. Most mutations are harmless. Other mutations cause diseases or affect a person's appearance or function. If a parent has a mutation, he or she can pass it down to a child. The chance of a child inheriting a mutation depends on whether the gene is dominant or recessive.

Recessive Disorders

Most carrier screening is for ***recessive disorders***. It takes two genes—one inherited from the mother and one inherited from the father—for a person to get a recessive disorder. If a person has only one gene for a disorder, he or she is known as a ***carrier***. Carriers often do not know that they have a gene for a disorder. They usually do not have symptoms or have only mild symptoms.

Some recessive disorders occur more often in certain races or ethnic groups. For example, ***sickle cell disease*** occurs most frequently in African Americans. ***Tay–Sachs disease*** typically occurs in people of Eastern or Central European Jewish, French Canadian, and Cajun descent, but anyone can have one of these disorders. Recessive disorders are not restricted to these groups.

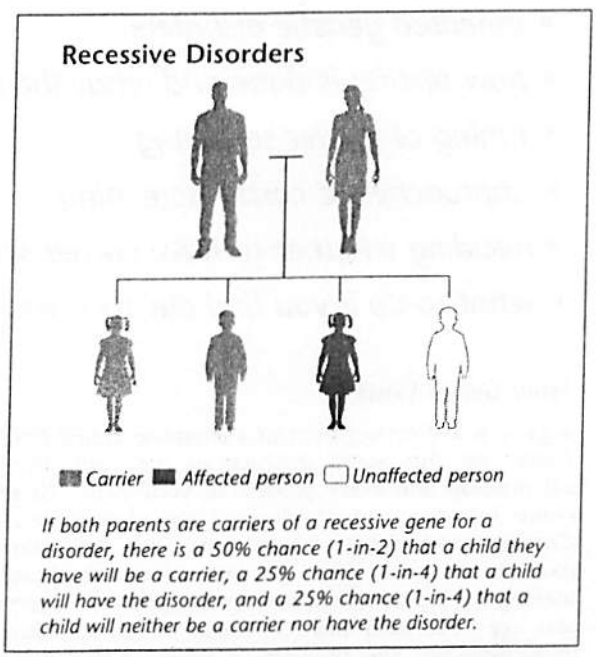
If both parents are carriers of a recessive gene for a disorder, there is a 25% (1-in-4) chance that their child will get the gene from each parent and will have the disorder. There is a 50% (1-in-2) chance that the child will be a carrier of the disorder—just like the carrier parents. If only one parent is a carrier, there is a 50% (1-in-2) chance that the child will be a carrier of the disorder.

Dominant Disorders

In a ***dominant disorder***, only one gene from the mother or father is needed for a person to have the disorder. An example of a dominant disorder is neurofibromatosis, a group of disorders that causes the growth of tumors in the nervous system. Carriers of dominant disorders usually know that they have the disorder because of their symptoms. Carrier tests for dominant disorders usually are not needed.

Sex-Linked Disorders

Disorders in which the gene is carried on the sex chromosomes are called ***sex-linked disorders***. An example of a sex-linked disorder is ***hemophilia***. The gene for the most common type of hemophilia is on the X chromosome. A male child who gets the gene for this disorder will have the disorder. A female child who inherits the gene will be a carrier of the disorder.



If both parents are carriers of a recessive gene for a disorder, there is a 50% chance (1-in-2) that a child they have will be a carrier, a 25% chance (1-in-4) that a child will have the disorder, and a 25% chance (1-in-4) that a child will neither be a carrier nor have the disorder.

How Testing Is Done and What the Results Mean

Carrier screening involves testing a sample of blood, saliva, or tissue from the inside of the cheek. Test results can be negative (you do not have the gene) or positive (you do have the gene).

Negative Test Results

If you are tested for a disorder and your test result is negative, no further testing is needed. But no test is perfect. In a small number of cases, test results can be wrong. A negative test result when you actually have a gene for the disorder tested is called a false-negative result. Because of the possibility of a false-negative result, a negative carrier screening test result does not completely rule out the risk that you are a carrier of a genetic disorder.

Positive Test Results

If your test result is positive for a disorder, the next step in carrier screening usually is to test your partner. If the results of both tests are positive, a ***genetic counselor, obstetrician–gynecologist (ob-gyn)***, or other health care professional can explain your risks of having a child with the disorder. Like negative test results, there is a small chance that a positive test result is not accurate. A positive test result when you actually do not have a gene for a disorder is called a false-positive result.

Timing of Carrier Screening

Carrier screening can be done either before pregnancy or during pregnancy. If you have carrier screening before you become pregnant and you and your partner are carriers, you have the following four options:

1. You can become pregnant and then have prenatal ***diagnostic tests*** to see if the fetus has the disorder. Prenatal diagnostic testing involves having ***amniocentesis*** or ***chorionic villus sampling (CVS)***. Diagnostic testing is available for some, but not all, genetic disorders.
2. You can become pregnant using ***in vitro fertilization (IVF)***. You can use your own eggs or sperm or donor eggs or sperm. Tests can be done on the ***embryo*** before it is transferred to the uterus to see if the genetic disorder is present. This is called ***preimplantation genetic diagnosis***.
3. You may choose not to become pregnant.
4. You may choose to adopt a child.

If you have carrier screening after you become pregnant and you and your partner are carriers, your options are more limited. Pregnancy with prenatal diagnostic testing is an option. You also may decide not to have any testing.

Once you have had a carrier screening test for a specific disorder, you do not need to be tested again for the disorder. If new carrier screening tests become available for a disorder that you have not been tested for and for which you may be at risk, you may want to discuss carrier screening for these disorders with your ob-gyn or other health care professional.

Approaches to Carrier Screening

All women who are pregnant or thinking about becoming pregnant are offered carrier screening for certain disorders. These disorders include ***cystic fibrosis***, ***hemoglobinopathies***, and ***spinal muscular atrophy (SMA)***. Additional screening may be recommended based on certain factors.

There are two general approaches to carrier screening: 1) targeted carrier screening and 2) expanded carrier screening. Your ob-gyn, genetic counselor, or other health care professional can help you choose the approach that best addresses your concerns and also meets current recommendations for carrier screening (see Table 1).

Targeted Carrier Screening

Targeted carrier screening involves screening for certain disorders based on your ethnicity or family history. Traditionally, carrier screening has been recommended for people who belong to an ethnic group or race that has a high rate of carriers of a specific genetic disorder. Carrier screening for a specific disorder also may be recommended if you have a family history of that disorder, regardless of your race or ethnicity.

Screening based on your ethnic group or race is called ethnic-based screening. For example, ethnic-based carrier screening for Tay–Sachs disease has been recommended for people of Eastern or Central European Jewish, French Canadian, and Cajun descent.

An advantage of ethnic-based screening is that it is highly accurate. The accuracy of any laboratory test depends on how many people tested actually have the disorder. When you test only people who belong to a group at increased risk, test results are more likely to be accurate.

A main disadvantage of ethnic-based screening is that it has become harder to assign a person to a single race or ethnicity. Many people do not know their ancestry. As the population has become more diverse, carrier screening based on ethnicity or race may not be as accurate.

Expanded Carrier Screening

Advances in technology have resulted in another approach to carrier screening called expanded carrier screening (ECS). In ECS, many disorders are screened using a single sample. ECS is screening done without regard to race or ethnicity.

Companies that offer expanded carrier screening create their own lists of disorders that they test for. This list is called a screening panel. Some panels test for more than 100 different disorders. Screening panels usually focus on severe disorders that affect a person's quality of life from an early age. Many of the disorders cause ***neurological*** problems, physical disability, or early death.

Table 1. Some Genetic Disorders for Which Carrier Screening Tests Are Available

Disorder	*What It Means*	*Who Is at Risk?*
Cystic fibrosis (CF)	CF affects the lungs, digestive system, and pancreas. Symptoms appear in childhood and include coughing, wheezing, loose stools, abdominal pain, and in men, infertility. Some people have milder symptoms than others. Over time the problems tend to become worse and harder to treat. The average life expectancy is 37 years.	Carrier screening should be offered to all women who are considering pregnancy or are currently pregnant.
Spinal muscular atrophy (SMA)	SMA causes muscles to break down (atrophy) and overall weakness. It is caused by a problem with the nerves that control movement. Of the three types, the most severe and most common (Type 1) causes death by age 2 years.	Carrier screening should be offered to all women who are considering pregnancy or are currently pregnant.
Fragile X syndrome	Fragile X syndrome is the most common inherited cause of intellectual disability. Disabilities range from mild (learning disabilities) to severe (autism). It affects both males and females, but some forms affect males more severely.	Carrier screening is recommended for women who have a family history of fragile X-related disorders, unexplained intellectual disability or developmental delay, autism with intellectual disability, or premature ovarian insufficiency.
Sickle cell disease	Sickle cell disease is a blood disorder that causes the red blood cells to have a crescent or "sickle" shape rather than the normal doughnut shape. The sickle cells can get caught in the blood vessels and prevent oxygen from reaching organs and tissues, which causes pain.	Carrier screening should be offered to women of African, Mediterranean, and Southeast Asian descent.
Thalassemias	Thalassemias are several types of blood disorders that cause anemia. Some types are more severe than others and can cause early death if not treated.	Carrier screening for the alpha-thalassemia trait should be offered to women of Southeast Asian, African, Mediterranean and West Indian descent. Carrier screening for beta-thalassemia mutations should be offered to women of Mediterranean, Asian, Middle Eastern, Hispanic, and West Indian descent.
Tay–Sachs disease	Tay–Sachs causes intellectual disability, blindness, and seizures. Symptoms first occur at about 6 months. Death usually occurs by age 5 years.	Carrier screening is recommended for women of Eastern or Central European Jewish, French Canadian, and Cajun descent.
Hemophilia	Hemophilia is a disorder caused by the lack of a substance in the blood that helps it clot. Affected people are treated with factors that help the blood clot and to help prevent excessive bleeding.	Women with a family history of hemophilia may request carrier screening.

The main advantage of ECS is that it allows you to be screened for a much greater number of disorders than ethnic-based screening. The cost of expanded carrier screening is about the same as that for single-disorder screening.

ECS also has many limitations. It is important to understand these limitations before choosing ECS:

- The tests on an ECS panel are not individually selected for you based on your personal and family history. For this reason, you may choose to be tested for fewer or additional conditions than those listed on an ECS panel based on your family health history or ethnicity. If you have a family history of a genetic disorder, you may benefit from having targeted genetic testing that looks specifically at your family mutation.
- ECS panels may include genes for disorders that in some people cause a disease but in others do not cause a disease. If a child inherits the gene for one of these disorders from both parents, it is not always certain whether the child will have the disorder.
- Because ECS tests for a large number of disorders, it is common to test positive as a carrier for one or more disorders. Most carriers of genetic disorders do not have any health problems. But in some instances, you may learn through carrier screening that you have the genes for a genetic condition that can affect your future health. If your partner is tested and he screens negative for the disorders you carry, your chance of having a child by the disorders is very low. If your partner screens positive for entirely different disorders than yours, it is unlikely that your child will be affected by any of the disorders.
- Many conditions that are screened are rare. You are probably at low risk of having the gene for many of the disorders that are tested. The likelihood

of having a false-positive result increases when you are at low risk. Also, not a lot of information may be known about rare disorders and how severely a child can be affected.

Deciding Whether to Have Carrier Screening

Carrier screening is a voluntary decision. You can choose to have carrier screening, or you can choose not to. There is no right or wrong choice.

If you do choose carrier screening, you should meet with your ob-gyn or genetic counselor before and after testing. Before testing, you should discuss the benefits and limitations of the screening approaches available. After testing, your health care professional can explain the results to you and help you make decisions if you have a positive result.

If you or your partner have ***consanguinity***, you also may be at an increased risk of certain conditions. In this case, talk with your health care professional about the benefits and limits of carrier screening.

What to Do If You Find Out That You Are a Carrier

If you find out that you are a carrier of a gene for a genetic disorder, you may want to tell other family members. They may be at risk of being carriers themselves. There is no law that states that you have to do so. If you choose to tell family members, your ob-gyn or genetic counselor can advise you about the best way to do this. It cannot be done without your consent.

Many people are concerned about possible employment discrimination or denial of insurance coverage based on genetic testing results. The Genetic Information Nondiscrimination Act of 2008 (GINA) makes it illegal for most health insurers to require genetic testing results or use results to make decisions about coverage, rates, or preexisting conditions. GINA also makes it illegal for employers to discriminate against employees or applicants because of genetic information. GINA does not apply to life insurance, long-term care insurance, or disability insurance.

Finally...

Carrier screening allows you to find out whether you carry genes for certain genetic disorders. There are many approaches to carrier screening. One approach is based on ethnicity or family history. Another is an expanded approach that screens for many disorders without regard to race or ethnicity. In some cases, both approaches can be used to tailor screening to your individual situation.

Glossary

Amniocentesis: A procedure in which amniotic fluid and cells are taken from the uterus for testing. The procedure uses a needle to withdraw fluid and cells from the sac that holds the fetus.

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

Carrier Screening: A test done on a person without signs or symptoms to find out whether he or she carries a gene for a genetic disorder.

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

Chorionic Villus Sampling (CVS): 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. They contain the genes that determine a person's physical makeup.

Consanguinity: A union between two people who are second cousins or closer in family relationship.

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

Diagnostic Tests: Tests that look for a disease or cause of a disease.

DNA: The genetic material that is passed down from parent to child. DNA is packaged in structures called chromosomes.

Dominant Disorder: A genetic disorder caused by one gene.

Egg: A female reproductive cell made in and released from the ovaries. Also called an ovum.

Embryo: The stage of development that starts at fertilization (joining of an egg and sperm) and lasts up to 8 weeks.

Fertilization: A multistep process that joins the egg and the sperm.

Genes: Segments of DNA that contain instructions for the development of a person's physical traits and control of the processes in the body. The gene is the basic unit of heredity and can be passed from parent to child.

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

Genetic Disorders: Disorders caused by a change in genes or chromosomes.

Hemoglobinopathies: Any inherited disorder that affects the number or shape of red blood cells in the body. Examples include sickle cell disease and the different forms of thalassemia.

Hemophilia: A disorder caused by a mutation on the X chromosome. Affected people are usually males who lack a substance in the blood that helps clotting. People with hemophilia are at risk of severe bleeding from even minor injuries.

In Vitro Fertilization (IVF): A procedure in which an egg is removed from a woman's ovary, fertilized in a laboratory with the man's sperm, and then transferred to the woman's uterus to achieve a pregnancy.

Mutation: A change in a gene that can be passed from parent to child.

Neurological: Related to the nervous system.

Obstetrician–Gynecologist (Ob-Gyn): A doctor with special training and education in women's health.

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

Recessive Disorders: Genetic disorders caused by two genes, one inherited from each parent.

Sex Chromosomes: The chromosomes that determine a person's sex. In humans, there are two sex chromosomes, X and Y. Females have two X chromosomes and males have an X and a Y chromosome.

Sex-Linked Disorders: Genetic disorders caused by a change in a gene located on the sex chromosomes.

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

Sperm: A cell made in the male testes that can fertilize a female egg.

Spinal Muscular Atrophy (SMA): An inherited disorder that causes wasting of the muscles and severe weakness. SMA is the leading genetic cause of death in infants.

Tay–Sachs Disease: An inherited disorder that causes mental disability, blindness, seizures, and death, usually by age 5. It most commonly affects people of Eastern or Central European Jewish backgrounds, as well as people of French Canadian and Cajun backgrounds.

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FIRST TRIMESTER PRENATAL SCREENING & ULTRASOUND EXAM

AKA “Combined Screen” “Sequential Screen” or “Nuchal Translucency”, “NT”

Even though the American College of Obstetrics and Gynecology promotes offering the combined screen to all patients during their first trimester of pregnancy, not all insurance companies cover the screening test.

You should know that the ultrasound portion of the test – which is done in our office – costs approximately $295 (with an additional cost for multiple gestations such as twins). In addition, there will be a charge for the blood test. The blood test is performed by Labcorp. The lab will bill separately.

You can and should call your insurance company to inquire regarding benefits and coverage for the combined screen.

You will need the following information when making this call:

Name of test: First Trimester Screen

ICD-10 Diagnosis Code: Z36.82 – screening for nuchal translucency

CPT (Procedure) Codes: 76813 – Ultrasound done in office
76814 – Ultrasound (if twin pregnancy)
84702 – Blood screening test
86336 – Blood screening test
84163 – Blood screening test

Please confirm your coverage and cost BEFORE you have the test done. Some insurance companies require pre-authorization for this test. Some cover it based on the age of the mother when the baby is born. Some have other restrictions that you should be aware of so that you have no unexpected bills.

LAB WORK

Prenatal Profile and Urine Culture

At your first appointment we will order routine prenatal testing, this verifies your blood type, checks for anemia, checks for immunity to rubella and checks hepatitis, syphilis, and HIV status. We will also send a urine culture to the lab. Optional tests are available should you choose. Included in this notebook you will find more information regarding these tests.

ROUTINE TESTING

Glucose Tolerance Test (GTT): 26-29 weeks gestation

This testing may be done earlier in pregnancy or more than once depending on your history of diabetes and other risk factors. At this stage in pregnancy, your placenta can alter how you metabolize glucose, which can lead to diabetes. Gestational diabetes is a common disease in pregnancy but usually requires treatment and close monitoring to ensure a healthy outcome for both mother and baby. Your provider team will give you complete instructions for your testing at the appointment prior to when your testing is due.

Abnormal GTT

If your initial testing indicates that you have gestational diabetes, you may be referred to EvergreenHealth Nutrition Services for management of your diabetes. A diabetic counselor will educate you on diet restrictions or medication management. Gestational diabetes typically resolves after you deliver. To confirm this, we will order another glucose test at your six-week postpartum checkup. This is an important test to complete to ensure you do not have diabetes that is going unmanaged.

Hematocrit (HCT): 26-29 weeks' gestation

A blood test for anemia is done at this time because this is when the fetus begins to draw iron from your body to store in its liver for after birth.

Normal HCT: 35.0-46.0

Continue taking your prenatal vitamins to maintain your iron level.

Abnormal HCT: <35.0

This is called anemia. You will be instructed to either add iron to your diet or take an additional iron supplement, or both. If you are instructed to take an extra iron supplement, we recommend Slow Fe or any iron supplement with 135 mg of ferrous sulfate (equivalent to 65 mg ferrous iron), which you can find at many pharmacies. We recommend taking this at a separate time from your prenatal vitamin. To maximize iron absorption, avoid dairy products one hour before and after taking your iron pill when possible. Also, taking your iron pill with a 500 mg vitamin C supplement or a glass of juice rich in vitamin C

will help enhance the absorption of iron.

Group Beta Streptococcus (GBS): 35-36 weeks gestation

About a month before your due date, we will test for GBS. This is a bacteria that can be carried in the vagina and rectum and can be passed to the baby during delivery. If the bacteria is passed to the baby, the baby can develop an infection that can potentially cause inflammation of the baby's blood vessels, lungs, brain, or spinal cord. One percent of babies born to mothers who carry GBS become infected with it, and symptoms can present within the first six hours to seven days after birth. We test for this bacteria by collecting a small vaginal and rectal culture with a swab. If you test positive, you will be treated with antibiotics during labor, so your baby is “pretreated” in case they acquire the bacteria during the birthing process. If you test negative, no treatment is necessary. If you are having a cesarean section, we will still test for GBS, but you will receive antibiotics prior to your surgery regardless of results as this is routine with C-sections.

CORD BLOOD BANKING/DONATION OPTIONS

About Cord Blood Banking

Cord blood banking is the once-in-a-lifetime opportunity for patents to save the stem cells found in the blood of their newborn's umbilical cord. The preservation of these stem cells, which are different from embryonic stem cells, allows families the benefit of having them available for current or future medical treatments.

Cord blood banking is completely safe for both the mother and the newborn, since cord blood is collected after the baby is born and after the umbilical cord has been clamped and cut. Currently stem cells are used to treat certain cancers and genetic disorders, but research on other uses is ongoing. The likelihood the stem cells will be used by your baby is very low. You should think of this as you would an insurance policy – nice to have but unlikely to be used.

Cord Blood Banking Options

When deciding what is best for you and your family, it is important to know about all of your cord blood banking options.

- **Family Banking** allows you to store your newborn's cord blood stem cells specifically for your family, making them available immediately should your family ever need them. This service is provided by cord blood banks, which charge a fee for collection, processing, and storage in which you retain ownership of your newborn's stem cells. Research has shown that transplants with related cord blood stem cells have doubled the survival rates of those with unrelated (publically donated) cord blood stem cells.

- **Public Donation** allows your family to offer your baby's cord blood stem cells to the public network at no cost. Your donation may then be made available to any patient requiring a cord blood stem cell transplant. Your family does not retain ownership of the cord blood once it has been donated. As a result, there is no guarantee that it will be available should it be needed by a family member. Testing for transmittable infections will be performed on maternal blood, and completion of a questionnaire will be required. (See Bloodworks pamphlet for more information on donation)

- **Medical Waste** means the cord blood will be thrown out as waste.