UW Medicine
UNIVERSITY OF WASHINGTON MEDICAL CENTER

產前檢驗指南

瞭解你的胎兒的健康

本章描述產前檢驗，這些檢驗可以提供有關你的胎兒的健康資訊。是否進行這些檢驗由你自己決定。請和你的醫生談談，以瞭解更多情況，幫助你決定這些檢驗中是否有一些是適合於你。

如果你已經做過任何這些檢驗，我們將要求你閱讀每項檢驗的詳細內容。我們還會要求你閱讀並簽署每項檢驗的同意書。

你在懷孕期間可以做很多事情來使你和胎兒保持健康。服用產前維生素、吃健康食品、鍛煉、保證充分的睡眠都是很重要的。

人體是複雜的。即便你在孕期所做的一切都是"正確"的，胎兒也並不總是能夠正常發育。約 3% 至 5% 左右（100 個人中有 3 至 5 個人左右）的胎兒在出生時會有某種健康問題。

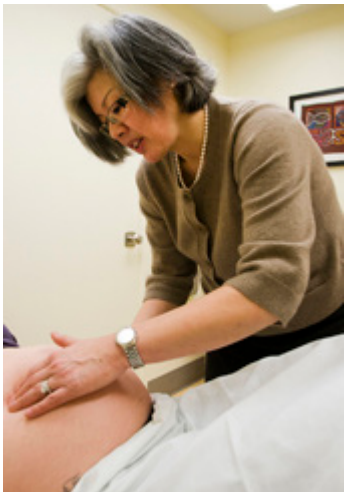
本材料提供有關這些檢驗的一些基本資訊，幫助你作出最適合你的決定。

有些什麼檢驗?

有兩種基本類型的檢驗：

- **篩檢** - 預測你的胎兒是否有某種出生缺陷的*機會*。
- **診斷檢驗** - 告訴你你的胎兒是否有某種出生缺陷。

下一頁中的表格列出了這些檢驗以及做這些檢驗的時間。表格還對每項檢驗給出簡短的說明以及這些檢驗會給你什麼資訊。本章的其餘部分對這些檢驗作了更詳細的說明，你應該先閱讀它們，然後再和你的醫生談。



產前檢驗可以提供有關你的胎兒的健康資訊。

篩檢

檢驗名稱	何時檢驗	檢驗說明	檢驗目的
後頸皮下透明層 (NT) 超聲波	11～14 周	腹部超聲波，測量胎兒後頸透明層的厚度。	你的胎兒患有染色體問題的*機會*。
綜合性篩檢	11～14 周 *和* 15～22 周	NT 超聲波，加上 2 次單獨的血液採樣。	你的兒患有唐氏綜合症、18 號染色體三體綜合症，或脊柱裂症狀的*機會*。
四聯法篩檢 (Quad screen)	15 ～ 22 周	1 次血液採樣。	你的胎兒患有唐氏綜合症、18 號染色體三體綜合症，或脊柱裂症狀的*機會*。

診斷檢驗

檢驗名稱	何時檢驗	檢驗說明	檢驗目的
絨毛膜採樣 (CVS)	11～14 周	透過陰道或腹部對胎盤進行採樣。	檢查胎兒*是否有*染色體問題和其他基因疾病。
羊水診斷 (用超聲波)	16～22 周	透過腹部抽取胎兒周圍的液體樣本。	檢查胎兒*是否有*染色體問題、脊柱裂症狀和其他基因疾病。

其他檢驗

檢驗名稱	何時檢驗	檢驗說明	檢驗目的
解剖超聲波	18～22 周	腹部超聲波，檢查胎兒生長和發育情況。	檢查*是否有*任何不正常現象，以及是否需要進一步檢驗。

篩檢

後頸皮下透明層 (NT) 超聲波

此項篩檢在孕期的 11～14 周進行，利用超聲波來測量胎兒的長度，以確定預產期。此外，超聲波還被用來測量胎兒頸後皮下的小空間。這個空間叫作*後頸皮下透明層* (NT)。這個液體空間越大，胎兒有染色體問題的*機會*就越大。NT 超聲波檢驗必須由經過專業訓練的人執行。

綜合性篩檢

這項檢驗是利用 NT 超聲波掃描和 2 次驗血的結果進行。第一次血液樣本是在 11 至 14 周之間抽取，通常是在進行 NT 超聲波掃描的同一天。第二次血液樣本是在 15 至 22 周之間抽取。驗血是為了檢查是否存在與某些出生缺陷相關的蛋白質及荷爾蒙。

綜合篩檢告訴你胎兒患有唐氏綜合症、18 號染色體三體綜合症或脊柱裂的*機會*有多大。（參看第 34、35 和 36 頁上側邊欄中更詳細的說明。）該項檢驗並不對這些病症作出診斷。大多數綜合篩檢結果異常的婦女依然能夠生下健康的嬰兒。

綜合篩檢可以檢測出：

- 100 例中 90 例 (90%) 的唐氏綜合症
- 100 例中 90 例 (90%) 的 18 號染色體三體綜合症
- 100 例中有 80 例 (80%) 的脊柱裂

但是，它**不能**檢測出所有病例的這些出生缺陷。而且，它不是用來檢驗任何其他健康問題。

四聯法篩檢

這種篩檢至檢測在 15 至 22 周之間抽取的血液樣本。如同綜合篩檢一樣，它也檢查是否存在與某些出生缺陷相關的蛋白質與荷爾蒙。

四聯法篩檢告訴你胎兒患有唐氏綜合症、18 號染色體三體綜合症或脊柱裂的*機會*有多大。該項檢驗並不對這些病症作出診斷。大多數綜合篩檢結果異常的婦女依然能夠生下健康的嬰兒。

什麼是染色體問題？

染色體是基因指令包，是從父母那裡繼承過來的，負責控制身體的生長和發育。染色體遍佈身體的各個部位，只能透過顯微鏡才能看到。

大多數人擁有 46 個染色體，但是有些人多一個染色體（即 47 個），或少一個（即 45個），或者某個染色體脫落了一塊，或者增加了一塊。

染色體的變化往往會造成出生缺陷和發育遲緩，但並非總是如此。

四聯法篩檢可以檢測出：

- 100 例中 85 例 (85%) 的唐氏綜合症
- 100 例中 75 例 (75%) 的 18 號染色體三體綜合症
- 100 例中 80 例 (80%) 的脊柱裂

但是，它**不能**檢測出所有病例這些出生缺陷。而且，它不是用來檢驗任何其他健康問題。

如果你要到第 4 個月才開始產前護理，或者如果沒有 NT 超聲波掃描檢查，四聯法篩檢便是一種很好的檢驗。

使用遊離 DNA 測量法的高級非整倍數篩檢

你也許在新聞中聽過或在互聯網上看過有關一種可以篩檢唐氏綜合症的血液檢驗。這種檢驗叫做"*使用遊離 DNA 測量法的高級非整倍數篩檢*"。它使用母體的血液樣本，這些血液樣本是在懷孕 10 周開始抽取。它可以篩檢出胎兒的染色體異常。

每個人的血液中都有一些遊離（不是包含在細胞內）的 DNA。當你懷孕時，那些遊離 DNA 大部分來自於你，但有一些則來自於你的妊娠。此項檢驗是要測量你血液中第 21 號、18 號和 13 號染色體的遊離 DNA 總數。

像其他篩檢一樣，此項檢驗不是要告訴你胎兒是否有染色體問題。但是，如果這些染色體中的其中一種 DNA 數量增加了，那麼胎兒患有該種染色體的染色體三體綜合症的可能性便會很高。

目前，只有那些所懷胎兒有較高風險患上唐氏綜合症、第 18 號染色體三體綜合症或第 13 號染色體三體綜合症的婦女能夠做這項檢驗。如果你已經有一個孩子患有其中一種三體綜合症，或者如果你做過另外一種篩檢並且結果不正常，可能會讓你做使用遊離 DNA 測量法的高級非整倍數篩檢。

診斷檢驗

解剖超聲波

此項檢驗是在 18～22 周之間進行。超聲波被用來檢查你的胎兒周圍的液體量以及你的胎盤和子宮，以確定胎兒在生長，全部主要器官均已形成。

什麼是唐氏綜合症？

唐氏綜合症亦稱爲第 21 號染色體三體綜合症。產生這種綜合症的原因是第 21 號染色體多出一個染色體。

唐氏綜合症以不同方式影響人們。患有唐氏綜合症的人看上去總是和其他家人不一樣，總是有某種發育遲緩現象，但遲緩程度因人而異。

患有唐氏綜合症的成年人或許能夠參加體育活動、從事一份基本工作、與人交友等。但是如果沒有幫助，他們通常不能獨立生活。

患有唐氏綜合症的許多嬰兒都有心臟缺陷，有時能夠透過手術來治癒。唐氏綜合症有時也會導致其他健康問題和出生缺陷，但是並不多見。

此階段的胎兒已經足夠發育，使得超聲波能夠發現問題，例如嚴重的心臟缺陷、脊柱裂、腎缺失和嚴重唇裂。儘管這種檢驗不是用來診斷染色體問題，但是它可以顯示出染色體或其他疾病的症狀。

絨毛膜採樣，或叫 CVS

這種診斷性檢驗通常是在 11 至 14 周之間進行。醫生利用一個軟性細針頭或細塑膠管從胎盤上採下一小塊樣本。與此同時進行超聲波檢驗，以便在採樣過程中能夠看到你的胎兒。

胎盤樣本被用來診斷染色體問題。如果你的家族中有基因病症，例如*肌肉萎縮症*或*血友病*，可用這些樣本檢驗胎兒是否患有這些病症。

做完 CVS 檢驗後，流產的機會是 100 個婦女中有 1 至 2 例流產 (1% 至 2%)。

羊水診斷或羊膜穿刺術

這種診斷性檢驗通常是在 16 至 22 周之間進行。醫生利用軟性細針頭從胎兒周圍抽取 2 湯匙液體。與此同時進行超聲波檢驗，以便在採樣過程中能夠看到你的胎兒。

這些液體被用來診斷染色體問題和脊柱裂。如果你家族中有基因病症，例如肌肉萎縮症或血友病，這些液體被用來檢驗胎兒是否患有這些病症。

羊水診斷造成流產的機會是每 400 名婦女中有 1 人流產 (0.25%)。

祖先基因攜帶者篩檢

你的祖先或種族是一種線索，有助於瞭解胎兒是否患有罕見的基因疾病。每個祖先類群都有一些比其他族群更常見的遺傳疾病。第 36 頁的表格上列出了每個祖先類群相關的疾病。

大多數時候，如果父母*兩人*都是*相同*疾病的“攜帶者”，這對配偶只能有一個孩子患有這些疾病中的其中一種。**攜帶者通常不會有任何疾病症狀。**大多數攜帶者也沒有這些疾病的家族史。如果你家中有人患有其中一種疾病，應告訴你的醫生。

什麼是 18 號染色體三體綜合症？

18 號染色體三體綜合症亦稱爲愛德華氏綜合症 (Edwards syndrome)。當一個人的第 18 號染色體多出一個染色體時，便會患上這種綜合症。

患有 18 號染色體三體綜合症的大多數胎兒不能在孕期中幸存下來，而患有 18 號染色體三體綜合症的兒童會有嚴重的腦損傷，通常還會有其他問題，例如心臟缺陷和畸形足。

什麼是脊柱裂?

脊柱裂是一種病症，指胎兒脊柱的一部分未能正常形成，導致脊柱內的神經受損。這種情況會在孕期的頭幾週內發生。

脊柱裂以不同方式對人產生影響。 有些人行走困難，需要使用支架或輪椅。 有些人難以控制自己的大小便。有時，脊柱裂可造成腦損傷和發育遲緩。

如果你和你的伴侶都是某種相同基因疾病的攜帶者，那麼你的胎兒可能會遺傳到該種疾病。如果想在胎兒出生前確定，可做羊水診斷或 CVS。綜合篩檢、四聯法篩檢和超聲波**不是**用來診斷這些失調症。

要查看自己是否是這些遺傳病的攜帶者，你需要提供小量血液樣本。是否做任何或所有這些檢驗是由你自己決定。

此表格改編自“美國國家遺傳顧問學會”於 2005 年發表的“祖先基因疾病攜帶者篩檢” (Ancestry Based Carrier Screening) 一文：

種族	遺傳性疾病	成為攜帶者的機會
非洲裔美國人	B 型地中海貧血	10% （100 人中有 10 人）
	鐮狀細胞病	11% （100 人中有 11 人）
東歐（德系）猶太人	海綿狀腦白質營養不良症	2.5% （100 人中有 2 至 3 人）
	囊腫性纖維化	3% 至 4% （100 人中有 3 至 4 人）
	家族性植物神經障礙	3% （100 人中有 3 人）
	家族性黑蒙性白癡症	3% （100 人中有 3 人）
歐洲白種人	囊腫性纖維化	3% （100 人中有 3 人）
地中海沿岸人	B 型地中海貧血	3% 至 5% （100 人中有 3 至 5 人）
	鐮狀細胞病	2% 至 30% （100 人中有 2 至 30 人）
東亞和東南亞人*	A 型地中海貧血	5% （100 人中有 5 人）
	B 型地中海貧血	2% 至 4% （100 人中有 2 至 4 人）
西班牙人*	B 型地中海貧血	0.25% 至 8% （100 人中有不到 1 人至 8 人）
	鐮狀細胞病	0.6% 至 14% （100 人中有不到 1 人至 14 人）
中東和中南亞人*	B 型地中海貧血	0.5% 至 5.5% （100 人中有不到 1 人至 6 人）
	鐮狀細胞病	5% 至 25% （100 人中有 5 至 25 人）

** 此組資料為估計數字，可能因具體種族而有所不同。*

基因諮詢顧問可以幫助你和你的伴侶作出有關是否做產前檢驗的決定。

決定是否做這些檢驗

選擇是否做任何此類檢驗，或者決定哪些檢驗對你最合適可能會很困難。沒有「絕對正確」的選擇。有些婦女只選擇解剖學超聲波檢驗，而不進行其他檢驗。 其他婦女則可能選擇綜合篩檢和解剖學超聲波檢驗。如果這些檢驗之一的結果異常，她們可進行羊水診斷。有些婦女喜歡 CVS 或羊水診斷，而不作任何篩檢。

作出明智的決定

在華盛頓大學醫學院，我們的目標是與病人和家庭合作，一起爲他們的保健護理作出決定。我們鼓勵你提出問題，以便我們能夠幫助你做出決定。

如果你想做基因檢驗，以下是一些你也許想問自己的問題：

- ❑ 我是否需要任何這類資訊？
- ❑ 在嬰兒出生之前瞭解這些出生缺陷將怎樣幫助我和我的醫生制定計劃？
- ❑ 如果發現有出生缺陷，這類資訊將怎樣幫助我就我的懷孕作出選擇？
- ❑ 做這些檢驗是否會使我感到更放心？

你的醫生可與你進一步討論你的選擇。或者，你可以和“產前診斷診所”預約時間。該診所的基因顧問受過專門訓練，能夠幫助人們考慮分析這些問題，幫助你作出對你最適合的決定。

有任何問題嗎？

你的問題很重要。如果你有任何有關產前檢驗的問題，請在工作時間致電你的醫生。

你也可以致電“產前診斷診所” (Prenatal Diagnosis Clinic)，電話：206-598-4072

Guide to Prenatal Testing – Chinese - Traditional
Published PFES: 07/2003, 12/2007, 12/2012
Clinician Review: 12/2012
Reprints on Health Online: https://healthonline.washington.edu

Guide to Prenatal Testing – Chinese - Traditional
Published PFES: 07/2003, 12/2007, 12/2012
Clinician Review: 12/2012
Reprints on Health Online: https://healthonline.washington.edu

UW Medicine
UNIVERSITY OF WASHINGTON MEDICAL CENTER

Guide to Prenatal Testing

Learning about your baby's health

This chapter describes prenatal tests that give information about your baby's health. It is your choice whether or not to have these tests done. Talk with your health care provider to learn more and to help you decide if any of these tests are right for you.

If you have any of these tests done, you will be asked to read more about each one. You will also be asked to read and sign a consent form for each test.

There is a lot you can do during your pregnancy to keep you and your baby healthy. Taking prenatal vitamins, eating healthy foods, exercising, and getting enough sleep are all important.

The human body is complicated. Even if you do everything "right" during your pregnancy, babies do not always develop normally. Between 3% and 5% of babies (between 3 and 5 out of 100) have some kind of health problem when they are born.

This handout gives some basic information about these tests to help you make the best decision for you.

What are the tests?

There are 2 basic kinds of tests:

- **Screening tests** predict the *chance*, or odds, that your baby has a certain birth defect.
- **Diagnostic tests** tell you if your baby does or does not have a certain birth defect.

The tables on the next page list the tests and when they are done. They also give a brief description of each test and what it will tell you. The rest of this chapter gives more details about these tests, if you would like to read about them before you talk with your health care provider.

Prenatal tests can provide information about your baby's health.

Screening Tests

Name of Test	When	Description	What It Tells You
Nuchal translucency (NT) ultrasound	11 to 14 weeks	Abdominal ultrasound to measure small space behind baby's neck	*Chances* your baby has a chromosome problem
Integrated screen	11 to 14 weeks *and* 15 to 22 weeks	NT ultrasound plus 2 separate blood samples	*Chances* your baby has Down syndrome, trisomy 18, or spina bifida
Quad screen	15 to 22 weeks	1 blood sample	*Chances* your baby has Down syndrome, trisomy 18, or spina bifida

Diagnostic Tests

Name of Test	When	Description	What It Tells You
Chorionic villus sampling (CVS)	11 to 14 weeks	Sample of placenta, taken through the vagina or abdomen	*Whether or not* your baby has chromosome problems and sometimes other inherited diseases
Amniocentesis (with ultrasound)	16 to 22 weeks	Sample of fluid from around your baby, taken through your abdomen	*Whether or not* your baby has chromosome problems, spina bifida, and sometimes other inherited diseases

Other Tests

Name of Test	When	Description	What It Tells You
Anatomy ultrasound	18 to 22 weeks	Abdominal ultrasound to check baby's growth and development	*Whether or not* abnormalities are suspected and if further testing is needed

Screening Tests

Nuchal Translucency (NOO-kul trans-LOO-sun-see) or NT Ultrasound

This screening test is done between 11 and 14 weeks of pregnancy. Using ultrasound, your baby's length is measured to confirm your due date. Ultrasound is also used to measure the small space under the skin behind your baby's neck. This space is called the *nuchal translucency* (NT). The larger this space of fluid is, the greater the *chance* your baby has a chromosome problem. An NT ultrasound can be done only by specially trained staff.

Integrated (IN-tuh-grey-tud) screen

This test uses the results of the NT ultrasound and 2 blood tests. The first blood sample is taken between 11 and 14 weeks, usually the same day as the NT ultrasound. The 2nd blood sample is taken between 15 and 22 weeks. The blood tests look for patterns of proteins and hormones that are linked to certain birth defects.

An integrated screen tells you the *chances* that your baby has Down syndrome, trisomy 18, or spina bifida. (See the sidebars on pages 34, 35, and 36 for more details.) It does not diagnose these conditions. Most women who get an abnormal integrated screen result still have a healthy baby.

The integrated screen can detect:

- 90 out of 100 cases (90%) of Down syndrome
- 90 out of 100 cases (90%) of trisomy 18
- 80 out of 100 cases (80%) of spina bifida

But, it will **not** detect all cases of these birth defects. And, it does not test for any other health problems.

Quad Screen

This screening test involves 1 blood sample that is taken between 15 and 22 weeks. It's like the integrated screen, because it also looks for patterns of proteins and hormones that are linked to certain birth defects.

A quad screen tells you the *chances* that your baby has Down syndrome, trisomy 18, or spina bifida. It does not diagnose these conditions. Most women who get an abnormal quad screen result still have a healthy baby.

What is a chromosome problem?

Chromosomes are packages of genetic instructions. We inherit them from our parents. They control how our bodies grow and develop. They are in every part of our body, and they can only be seen with a microscope.

Most people have 46 chromosomes, but some people have an extra chromosome (47), a missing one (45), or a chromosome that has a piece broken off or an extra piece attached.

A change in a chromosome often causes birth defects and developmental delay, but not always.

The quad screen can detect:

- 85 out of 100 cases (85%) of Down syndrome
- 75 out of 100 cases (75%) of trisomy 18
- 80 out of 100 cases (80%) of spina bifida

But, it will **not** detect all cases of these birth defects. And, it does not test for any other health problems.

A quad screen may be a good test to have if you do not start prenatal care until your 4th month or if an NT ultrasound is not available.

Advanced Aneuploidy (ann-you-PLOY-dee) Screening with Cell-free DNA

You may have heard about a new blood test that can screen for Down syndrome. This test is called a*dvanced aneuploidy screening with cell-free DNA*. It uses a blood sample from the mother, and it is done starting at 10 weeks of pregnancy. It screens for specific chromosome disorders in the baby.

Everyone has some free (not contained within a cell) DNA in their blood. When you are pregnant, most of that cell-free DNA is from you, but some is from your pregnancy. In this test, the total amount of cell-free DNA from chromosomes 21, 18, and 13 is measured in your blood.

Like the other screening tests, this test does not tell you if the baby has, or does not have, a chromosome problem. But if there is an increased amount of DNA from one of these chromosomes in your blood, there is a high chance that the baby has trisomy for that chromosome.

Currently, only women who have a high risk of having a baby with Down syndrome, trisomy 18, or trisomy 13 can have this test. If you have already had a child with one of these trisomies, or if you have another type of screen and the results are abnormal, you may be offered advanced aneuploidy screening with cell-free DNA.

Diagnostic Tests

Anatomy (uh-NAT-uh-mee) Ultrasound

This test is done between 18 and 22 weeks. An ultrasound is used to look at your baby, the amount of fluid around him, your placenta, and your uterus. It checks to see that the baby is growing and that all major organs are formed.

What is Down syndrome?

Down syndrome is also known as trisomy 21. It is caused when a person has an extra copy of chromosome number 21.

Down syndrome affects people in different ways. People with Down syndrome always look different than other members of their family. They always have some developmental delay, but the level of delay differs from person to person.

Adults with Down syndrome may be able to play sports, have a basic job, and enjoy friends. But they usually cannot live on their own without help.

Many babies with Down syndrome have a heart defect, which can sometimes be fixed with surgery. Other health problems and birth defects sometimes occur with Down syndrome, but they are rare.

Your baby is developed enough at this age that an ultrasound may find problems such as a severe heart defect, spina bifida, a missing kidney, and severe cleft lip. Although this test will not diagnose chromosome problems, it may show signs of them or other conditions.

Chorionic Villus Sampling (kor-ee-ON-ic VILL-us sam-pling) or CVS

This diagnostic test is usually done between 11 and 14 weeks. The doctor uses either a thin, flexible needle or a thin plastic tube to remove a small sample of the placenta. An ultrasound is done at the same time, so your baby can be seen during the procedure.

The placenta sample is used to diagnose chromosome problems. If an inherited condition such as *muscular dystrophy* or *hemophilia* runs in your family, the sample can be used to test your baby for that condition.

The chance of miscarriage after CVS is 1 to 2 women in 100 (1% to 2%).

Amniocentesis (AM-nee-oh-sen-TEE-sis) or Amnio

This diagnostic test is usually done between 16 and 22 weeks. The doctor uses a thin, flexible needle to take 2 tablespoons of fluid from around your baby. An ultrasound is done at the same time, so your baby can be seen during the procedure.

The fluid is used to diagnose chromosome problems and spina bifida. If an inherited condition like muscular dystrophy or hemophilia runs in your family, the fluid can be used to test your baby for that condition.

The chance that having an amniocentesis will cause a miscarriage is 1 in 400 women (0.25%).

Ancestry-Based Carrier Screening

Your ancestry, or ethnicity, is one clue to help learn if your baby could have a rare genetic disease. Each ancestral group has conditions that can be inherited that are more common in that group compared to other ethnic groups. The conditions that are linked with each ancestral group are listed in the table on page 36.

Most times, a couple can have a child with one of these disorders only when *both* parents are "carriers" for the *same* disorder. **Carriers usually have no symptoms of the disease.** Also, most carriers have no family history of the disease. If someone in your family has one of these conditions, tell your health care provider.

What is trisomy 18?

Trisomy 18 is also known as Edwards syndrome. It occurs when a person has an extra copy of chromosome number 18.

Most babies with this condition do not survive the pregnancy. Children with trisomy 18 have severe brain damage and usually other problems, such as heart defects and clubfoot.

What is spina bifida?

Spina bifida is a condition in which part of the baby's spine does not form normally and the nerves in the spine are damaged. This happens within the first few weeks of pregnancy.

Spina bifida affects people in different ways. Some people have trouble walking and may need to use braces or a wheelchair. Some have trouble controlling their bladder or bowel. Sometimes, spina bifida can cause brain damage and developmental delay.

If you and your partner are both carriers for the same genetic condition, then your baby could inherit that condition. If you want to know for sure before birth, an amniocentesis or a CVS can be done. The integrated screen, quad screen, and ultrasound will **not** diagnose these disorders.

To see if you are a carrier for these hereditary conditions, you will need to give a small blood sample. It is your choice whether or not to have any or all of these tests.

This table is adapted from "Ancestry Based Carrier Screening," published by the National Society of Genetic Counselors, Inc., 2005:

Ancestral Group	Hereditary Condition	Chance of Being a Carrier
African-American	Beta Thalassemia	10% (10 out of 100)
	Sickle Cell Disease	11% (11 out of 100)
Eastern European (Ashkenazi) Jewish	Canavan Disease	2.5% (2 to 3 out of 100)
	Cystic Fibrosis	3% to 4% (3 to 4 out of 100)
	Familial Dysautonomia	3% (3 out of 100)
	Tay-Sachs Disease	3% (3 out of 100)
European Caucasian	Cystic Fibrosis	3% (3 out of 100)
Mediterranean	Beta Thalassemia	3% to 5% (3 to 5 out of 100)
	Sickle Cell Disease	2% to 30% (2 to 30 out of 100)
East and Southeast Asian*	Alpha Thalassemia	5% (5 out of 100)
	Beta Thalassemia	2% to 4% (2 to 4 out of 100)
Hispanic*	Beta Thalassemia	0.25% to 8% (fewer than 1 to 8 out of 100)
	Sickle Cell Disease	0.6% to 14% (fewer than 1 to 14 out of 100)
Middle Eastern and South Central Asian*	Beta Thalassemia	0.5% to 5.5% (fewer than 1 to 6 out of 100)
	Sickle Cell Disease	5% to 25% (5 to 25 out of 100)

** Numbers for this group are estimates and may vary depending on exact ethnicity.*

A genetic counselor can help you and your partner make decisions about prenatal tests.

Deciding Whether to Do These Tests

Choosing whether to have any of these tests, or deciding which ones are best for you, can be hard. There is no "right" choice. Some women choose only an anatomy ultrasound and no other tests. Others may choose an integrated screen and anatomy ultrasound. And, if one of these tests is abnormal, they may have amniocentesis. Some women prefer a CVS or amniocentesis without any of the screening tests.

Making an Informed Decision

Our goal at UW Medicine is to partner with patients and families in making decisions about their care. We encourage you to ask questions to help you to make your decisions.

These are some questions you may want to ask yourself as you think about having genetic testing:

- ❑ Do I want to have any of this information?
- ❑ How would learning about these birth defects before my baby is born help me and my health care provider prepare and plan?
- ❑ How would this information help me make choices about my pregnancy if a birth defect is found?
- ❑ Will taking these tests help me feel more reassured?

Your health care provider can talk more with you about your choices. Or, you can schedule an appointment in the Prenatal Diagnosis Clinic. Genetic counselors are specially trained to help people think through these questions. They can help you make the decision that is best for you.

Questions?

Your questions are important. If you have questions about prenatal testing, call your health care provider during office hours.

You may also call the Prenatal Diagnosis Clinic: 206-598-4072

Published PFES: 07/2003, 12/2007, 12/2012
Clinician Review: 12/2012
Reprints on Health Online:
https://healthonline.washington.edu

Published PFES: 07/2003, 12/2007, 12/2012
Clinician Review: 12/2012
Reprints on Health Online:
https://healthonline.washington.edu