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使用游離 DNA 測量法的高級非整倍數篩檢

產前檢驗指南

在華盛頓大學醫學院，我們與我們的病人及其家屬合作，幫助他們作出有關其保健護理的決定。

本手冊給出的資訊可以幫助你決定你是否需要做高級非整倍數篩檢。是否做這項檢驗由你自己決定。有些人覺得這類檢驗幫助不大。你可以在任何時候拒絕做這項檢驗。

本手冊對這項篩檢和一些醫學術語作出說明。要瞭解有關這方面的更多資訊，請諮詢你的醫生。

什麼是使用游離 DNA 測量法的高級非整倍數篩檢？

非整倍數篩檢可以在懷孕第 10 周開始做。

*使用游離 DNA 測量法的高級非整倍數篩檢*利用從你手臂上抽取的血液進行測量。它可以在懷孕 10 周時開始進行。這項檢驗用來篩檢胎兒中某個特定*染色體*的變異。染色體是包含在我們細胞中的 DNA“包”。

DNA 的全稱是*脫氧核糖核酸*，這是一種將基因信息從一代人攜帶到下一代人的分子。我們的 DNA 大部分儲存在我們身體的細胞內。*游離 DNA* 卻不是包含在細胞內。每個人 的血液中都有一些游離 DNA。當你懷孕時， 你血液中的大部分游離 DNA 都來自於你，但是有些則來自於你的胎兒。

這項檢驗是在實驗室中測量你血液中第 21 號、18 號和 13 號染色體以及 X 染色體的游離 DNA 總數。如果你血液中這些染色體的其中一種所含 DNA 數量異常，那麼，你胎兒體內該種染色體爲非整倍數的可能性是很高的。

非整倍數是什麼？

非整倍數 是指一個人體內某些染色體的數量多出了或缺失了。

人類有 23 對不同的染色體，對於大多數人，每對染色體都有 2 條染色體，總共 46 條。*三體綜合症*是指在體內的所有細胞中，某對染色體有 3 條染色體。*單體綜合症*是指在體內的所有細胞中，某對染色體只有 1 條染色體。

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- **第 21 號染色體三體綜合症**是指在所有細胞中的第 21 號染色體有 3 條染色體。它是導致稱爲*唐氏綜合症 (Down syndrome)* 這種基因疾病的最常見原因 。
- **第 18 號染色體三體綜合症**是指在所有細胞中的第 18 號染色體有 3 條染色體。這種疾病亦稱爲*愛德華氏綜合症 (Edward syndrome)*。
- **第 13 號染色體三體綜合症**是指在所有細胞中的第 13 號染色體有 3 條染色體。這種疾病亦稱爲*巴陶氏綜合症 (Patau syndrome)*。
- **X 單染色體綜合症**是指 X 染色體只有一條染色體。它是導致稱爲*透納氏綜合症 (Turner syndrome)* 這種基因疾病的最常見原因。

這項檢驗有哪些優點？

- 這是迄今可用來測量染色體非整倍數的最精確的篩檢法。
- 做這項檢驗**不會**對胎兒帶來任何風險。
- 正常的檢驗結果可以使你安心，幫助你減少你可能對你的胎兒健康的憂慮。

這項檢驗有哪些局限性？

- 高級非整倍數篩檢可以檢測出**幾乎**所有唐氏綜合症、18 號染色體三體綜合症、13 號染色體三體綜合症以及 X 單染色體綜合症的病例。**但是，它不能肯定地告訴你你的胎兒是否有任何這些疾病。**
- 有時篩檢結果不正常，即使胎兒的染色體**沒有**非整倍數，但這種情況很罕見。
- 有時篩檢結果正常，即使胎兒的染色體**確實**有非整倍數，但這種情況很罕見。
- 約有 1% 的血液樣本（100 個樣本中有 1 個）無法解釋。如果這種情況發生了，你將得不到結果。
- 高級非整倍數篩檢不是用來檢驗任何其他的染色體異常、其他的出生缺陷或其他的基因疾病。

檢驗結果正常意味著什麼？

正常的檢驗結果表示你的胎兒有非整倍數染色體的可能性非常**低**。

如果檢驗不正常，我如何能夠確認結果呢？

如果你想確認你的胎兒有非整倍數染色體：

- 分娩前，你可以做*絨毛膜取樣* (CVS) 或*羊膜穿刺*測試。請向醫生索取這些測試的說明手冊。
- 分娩後，可以從你的嬰兒身上抽取小量血液樣本，以測試你的嬰兒的染色體。

有任何問題嗎？

你的問題很重要。如果你有任何問題或疑慮，請致電你的醫生或保健提供者。

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Advanced Aneuploidy Screening with Cell-Free DNA – Chinese - Traditional
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Advanced Aneuploidy Screening with Cell-Free DNA

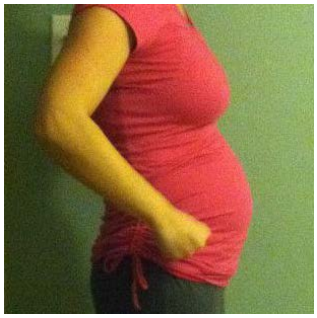
A guide to prenatal testing

At University of Washington Medical Center, we partner with our patients and families in making decisions about their health care.

This handout gives information to help you decide if you want to have an advanced aneuploidy screening test. Having this test is up to you. Some people do not find this type of test to be helpful. You may refuse testing at any time.

This handout explains the screening test and some medical terms. Talk with your health care provider to learn more.

What is advanced aneuploidy screening with cell-free DNA?



Aneuploidy screening can start in the 10th week of pregnancy.

Advanced aneuploidy screening with cell-free DNA is done using blood drawn from your arm. It can be done starting at 10 weeks of pregnancy. The test screens for specific disorders in your baby that are related to *chromosomes*. Chromosomes are the "packages" of DNA contained in our cells.

DNA is *deoxyribonucleic acid*, a molecule that carries genetic information from one generation to the next. Most of our DNA is stored inside the cells of our body. *Cell-free DNA* is not contained within a cell. Everyone has some cell-free DNA in their blood. When you are pregnant, most of the cell-free DNA in your blood is from you, but some is from your fetus.

In this test, the lab measures the total amount of cell-free DNA from chromosomes 21, 18, 13, and X in your blood. If there is an abnormal amount of DNA from one of these chromosomes in your blood, there is a high chance that your baby has aneuploidy for that chromosome.

What is aneuploidy?

Aneuploidy is when a person has extra copies or missing copies of certain chromosomes.

Most people have 2 copies each of 23 different chromosomes, for a total of 46. *Trisomy* is when there are 3 copies of a certain chromosome in all of the cells in the body. *Monosomy* is when there is only 1 copy of a certain chromosome in all of the cells in the body.

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- **Trisomy 21** is when there are 3 copies of the chromosome number 21 in all cells. It is the most common cause of a genetic condition called *Down syndrome*.
- **Trisomy 18** is when there are 3 copies of chromosome 18 in all cells. This condition is also called *Edward syndrome*.
- **Trisomy 13** is when there are 3 copies of chromosome 13 in all cells. This condition is also called *Patau syndrome*.
- **Monosomy X** is when there is only 1 copy of the X chromosome. It is the most common cause of a genetic condition called *Turner syndrome*.

What are the benefits of this test?

- It is the most accurate screening test for aneuploidy available today.
- Taking this test does **not** pose any risk to your fetus.
- Normal results may be reassuring and help lower anxiety you may have about your baby's health.

What are the limitations of this test?

- Advanced aneuploidy screening detects **nearly** all cases of Down syndrome, trisomy 18, trisomy 13, and monosomy X. **But, it will not tell you for sure whether or not your baby has any of these disorders.**
- Rarely, the result will be abnormal, even though the fetus **does not** have aneuploidy.
- Rarely, the result will be normal, even though the fetus **does** have aneuploidy.
- About 1% of blood samples (1 out of 100) cannot be interpreted. If this happens, you will not get a result.
- Advanced aneuploidy screening does not test for any other chromosome disorders, other birth defects, or other genetic conditions.

What does a normal result mean?

A normal result means that the chances of your baby having aneuploidy are very **low**.

If the test is abnormal, how can I confirm the results?

If you want to confirm that your baby has aneuploidy:

- Before you deliver, you can have a *chorionic villus sampling* (CVS) or an *amniocentesis* test. Please ask for handouts that describe these tests.
- After delivery, a small blood sample can be taken from your baby to test your baby's chromosomes.

Questions?

Your questions are important. Call your doctor or health care provider if you have questions or concerns.

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