

## 病人教育

产前诊断科



# 综合筛选测试

产前测试简介

华大医疗中心与病人及他们的家属合作、一起为他们的医疗保健一起做决 定。

此小册是为您提供资料以 助您决定是否要做综合筛 选测试。

是否做这测试是由您决定。有些人不认为此类测试有何帮助。您可在任何时间拒绝做测试。

请与您的医护人员讨论以 取得更多的资讯。

### 何谓综合筛选测试?

综合筛选测试是在怀孕期间测试胎儿是否有唐氏综合症、三体性 第 18 对染色体、或脊椎前裂的可能性。综合筛选测试并不能诊断 这些状况。

### 如何做综合筛选测试?

综合筛选测试是包括 3 部分—在怀孕 11 至 14 周之间在不同的时间作一次超音波及 2 次由母亲手臂抽血做血液的测试。

### 超音波

在作超音波时、将测量胎儿的长度以确认预产期。亦使用超音波来测量胎儿颈后皮下的小空间、又称*颈部半透明体* (NT)。一般有唐氏综合症或三体性第 18 对染色体的胎儿此部位较厚。

### 血液测试

第一次血液测试是在怀孕的第一期(头三个月)。通常是在测量颈部半透明体超音波时的同一天。第二次血液测试是在怀孕的第二期即第 15 至 20 周之间。是测试血液内某种由胎儿及胎盘产生的蛋白质及荷尔蒙、每一位孕妇的血液内都会有的。通常有唐氏综合症、三体性第 18 对染色体或脊椎前列胎儿的蛋白质及荷尔蒙的数量是不同的。

### 我能从综合筛选测试得到什么资讯?

综合筛选测试会有 3 个结果、如" 4,000 个例内有 1 例 (0.025%)" 或 "75 例内有一例 (1.3%)"。这些数据代表胎儿有唐氏综合症、三体性第 18 对染色体或脊椎前列的可能性或机遇率。在第二次血液测试后一周内可有结果。



除了综合筛选测试之外还有 其他的选择吗?

如果不能测量到您胎儿颈部 半透明体、您还是可做 2 次 血液的测试而得到结果。它 的精准度会稍微差一些、但 仍然是一个有帮助的测试。

如您怀孕已超过 14 周、请 与您的医护人员征询有关*四 合一*的测试。

### 您有疑问吗?

我们很重视您的提问。如您对综合筛选测试还有其他的疑问时、请在签同意 书前再与您的医护人员讨 论。

华大医疗中心诊所的职员 也可随时给予协助。

产前诊断科: 206-598-8130

妇婴科: 206-598-4070

# University of Washington MEDICAL CENTER UW Medicine

#### **Maternal and Infant Care Clinic**

Box 356159

1959 N.E. Pacific St. Seattle, WA 98195 206-598-4070 综合筛选测试在 10 例中可检测出 9 例 (90%) 的唐氏综合症、三体性第 18 对染色。在 10 例中可检测出 8 例 (80%) 脊椎前列。但它不能检测出所有的先天性残疾、亦无法检测出其他的健康问题。如您怀 多胞胎、综合筛选测试的准确度会降低。但可能仍然有帮助。

### 从综合筛选测试有何益处?

综合筛选测试可为您提供有关您胎儿健康的资讯且不会危及您的妊娠。

### 从综合筛选测试有何风险?

大多数的人在接到综合筛选测试结果不正常时会担心。导致余下的 妊娠期间有压力、尤其是您如果决定不再做下一步的测试时。

### 结果为"阳性"是代表什么?

少数妇女 (约 5%, 或 20 人中有 1 人) 在综合筛选测试结果为"阳性"。此并非意味她们的胎儿有先天性残疾。它仅意味胎儿在任何这 3 种情况之 1 的风险较其他水平的要高。**大多数结果呈阳性的妇女都生了健康的婴儿。** 

如您的医护人员告诉您综合筛选测试结果为阳性但并未告诉您正确的风险度、您可以要求更多的资讯。通常人们知道他们胎儿有先天性残缺的确切风险比例时会较安心。如有唐氏综合症的风险是 10 分之 1(10%), 也可解释为 10 个里有 9 个**没有**唐氏综合症。

如您综合筛选测试结果为阳性、您的医护人员可以请您再回来诊所、如与遗传顾问会谈、再做一次超音波详细的检查胎儿的结构。您也可以选择作*羊膜穿刺*。羊膜穿刺术可以确定的诊断胎儿是否有唐氏综合症、三体性第 18 对染色体或脊椎前列。

### 结果为"阴性"是代表什么?

大多数的妇女(约 95%, 或 20 例内有 of 19 例) 其综合筛选测试结果是"阴性"此并不意味胎儿是完全正常。它仅代表有此 3 类不正常之一可能性较某种指标的可能性为低。少数妇女综合筛选测试结果为阴性、但生的婴儿有**唐氏综合症、三体性第 18 对染色体或脊椎前列**。

## **Patient Education**

Prenatal Diagnosis Clinic



## Amniocentesis

Helping you understand your procedure and the consent form

► Please read this handout before reading and signing the form "Special Consent for Procedural Treatment UH0173."

This handout describes what to expect from an amniocentesis. Included are benefits, risks and complications, and alternatives.

This handout is in addition to the talks you have with your health care providers. It is important that you fully understand this information, so please read these pages carefully.

### What is an amniocentesis?

Amniocentesis (am-nee-oh-sen-**tee**-sis) is a procedure that allows your baby to be tested for a variety of health problems. It is also called *amnio*. Amnio is done during the second trimester of pregnancy, usually between 16 and 22 weeks.

### How is an amniocentesis done?

- First, *ultrasound* is used to see your baby's position and to decide the safest way to do the procedure. The ultrasound stays on during the entire procedure, which takes about 2 minutes.
- While watching the baby by ultrasound, the doctor inserts a thin needle into your abdomen and removes about 2 tablespoons of *amniotic fluid*, the liquid that surrounds the developing baby. The needle does not touch the baby.
- You are welcome to have a partner, spouse, friend, or family member with you during your amnio.

Most women are concerned about having an amnio. But, after it is done, nearly all women say that it was much simpler and quicker than they thought it would be.

### What medicines will be used?

The doctor cleans the skin on your abdomen with an *antiseptic soap* before the amnio is done.

If your blood type is Rh negative, you will be given a *RhoGAM injection* after the amnio to prevent problems in future pregnancies.



Prenatal Diagnosis Clinic Amniocentesis

### What can amnio test for?

Amniotic fluid contains cells that have been shed from the baby's skin, which is a normal process. These cells can be used to diagnose some genetic conditions. The baby's chromosomes are examined, which will diagnose conditions such as Down syndrome and trisomy 18. If a hereditary condition such as cystic fibrosis or muscular dystrophy runs in the family, the baby's DNA can be tested to see if the baby has inherited the disorder.

The amniotic fluid also has a protein in it called alpha fetoprotein (AFP). This protein is made by the baby. The amount of AFP in the amniotic fluid is a test for birth defects of the spine and brain, such as spina bifida.

If there is concern that the baby has an infection, the fluid can be tested for some viruses.

It may take up to 3 weeks to complete testing of the samples taken during your amnio.

### What are the benefits of an amniocentesis?

- 1. Amniocentesis allows a clear diagnosis of some health problems. Some people find that knowing a diagnosis is better than being unsure about their baby's health.
- 2. Most of the time, test results are normal. If you have been worried about your baby's health, normal test results may make you feel better about your pregnancy.
- 3. Some women feel that having more information about the baby's health will help them prepare better for the baby's arrival. Others do not want to raise a child with special needs. Having a diagnosis allows parents to make choices, such as choosing an adoption plan or stopping the pregnancy.

## What are the risks and complications of an amniocentesis?

The doctor will take steps to make the risks of the amnio as low as possible, but no procedure is completely risk-free.

The major risk from amniocentesis is miscarriage. A miscarriage may occur if an infection starts in the amniotic fluid, or if the sac surrounding the baby doesn't heal after the procedure, allowing amniotic fluid to leak out.

Without amniocentesis, about 1% to 2% (1 to 2 out of 100) of pregnancies will miscarry between 15 and 28 weeks. Amniocentesis adds an extra 0.25% (1 out of 400) risk of miscarriage. Said another way, out of every 400 amniocentesis procedures done, 399 women (99.75%) do **not** have a miscarriage.

There are some limitations to amniocentesis:

- Many health problems and birth defects are not diagnosed by amniocentesis or any other prenatal tests. Having normal test results from amniocentesis does not guarantee the baby is healthy (see information on the left side of this page).
- It is not always possible to obtain amniotic fluid. Sometimes the doctor can try a second time. If the second try doesn't work, your amnio will be rescheduled for another time.
- Sometimes the lab is not able to examine the baby's chromosomes because cells collected from the amniotic fluid do not grow. This happens less than 0.5% of the time, or less than 1 out of 200 times.

### **Questions?**

Your questions are important. If you have any questions about amniocentesis or the risks, benefits, or alternatives to it, talk with your provider before signing any consent forms.

Prenatal Diagnosis Clinic: 206-598-8130

Maternal and Infant Care Clinic: 206-598-4070

### What are the alternatives to amniocentesis?

You may decide not to have an amnio. Doing an amniocentesis is your choice, and the decision is a personal one. Health problems that can be diagnosed by amniocentesis can also be diagnosed when the baby is born. Some women feel the risk of a health problem is so low that they are not concerned. Others decide they don't want to risk a miscarriage and prefer to learn the diagnosis after the baby is born. Some families feel they can prepare for the birth of a child with special health needs without knowing a diagnosis.

### What follow-up care is needed?

There should be no ill effects from the amniocentesis. You can return to work as long as your job does not require heavy physical activity.

### For 24 to 48 hours after your amnio, we suggest that you:

- Stay off your feet as much as possible.
- Drink extra fluid.

### Also avoid:

- Heavy physical activity, such as running or other exercise.
- Lifting heavy objects (10 pounds or more).
- Sexual intercourse or sexual activity.

After an amniocentesis, it is normal to have:

- Mild, menstrual-like cramping for the first few hours (you can take Tylenol for this).
- Bruising or soreness on your skin where the needle was inserted.

### When to Call Your Doctor

Call your health care provider right away if you have any of these symptoms after your amnio:

- Spotting or bleeding from your vagina.
- Leakage of clear fluid from the vagina.
- Severe or rhythmic cramping or pain in your lower abdomen.
- Fever or chills.
- Diarrhea.
- Flu-like symptoms.



### **Maternal and Infant Care Clinic**