



## Optional Testing In Pregnancy

During your pregnancy, we do a number of routine tests. Blood count, blood type and Rh status, gestational diabetes screening are a few of the tests we use to help insure a good outcome for you and your baby.

There are, however, several optional tests we will offer you during your pregnancy. Along with the brochures given in the office and our counseling during your visit, this information is meant to help you make an informed decision about optional testing during your pregnancy.

Here's the bottom line: Do the tests if the results will be helpful for you. If the results of the test won't make any difference for you, don't do the test!

### 1. Triple (or quadruple) Test:

This blood test, done between 15 and 20 weeks gestation, provides screening for two (or three) conditions.

- Spina Bifida (and associated disorders) – spina bifida is a condition that occurs during early development of the baby whereby the spinal bones don't close in a complete circle. This allows the spinal cord to be damaged, to varying degrees. Some children with spina bifida have no obvious problems, others cannot walk or control bowel or bladder function.
- Down's syndrome (also called Trisomy 21) – Rarely is this associated with family history; this is a chromosomal abnormality that occurs very early in the baby's development.
- Cystic fibrosis – In this condition, children accumulate thick secretions in the lungs and other tissues. Breathing problems and infections are the major symptoms.

Since this is a screening test, it is not a perfect predictor of these disorders.

For spina bifida, 5% of the time the test will tell you the baby has the problem when in truth, the baby is just fine.

For Down's Syndrome, the false positive and false negative results depend upon age. The younger you are, the higher your chance of a false result. Combined false positive and false negative rates can be up to 30%.

If you have a positive test (a test that says the infant may be affected), we recommend genetic counseling, a comprehensive ultrasound at a specialist and possible amniocentesis.

### 2. Genetic Testing





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Since the risk of a chromosomal abnormality increases with maternal age, we offer a referral for testing for Down's Syndrome and other chromosomal abnormalities to our patients who will be 35 years or older at the time of delivery.

Why 35? No, it's not that you suddenly reached some scary risk at the age of 35. A decision was made to offer genetic testing to women starting at age 35 because at that age, the risk that you will have an infant with Down's Syndrome equals the risk that an amniocentesis will cause a miscarriage. Are we comparing apples and oranges here? You bet! However, that's the best criteria we've got for offering this testing and more importantly, that's the criteria the insurance companies recognize. (This testing is quite expensive.)

Two options are available to determine if your infant has a chromosomal abnormality:

- **CVS (Chorionic Villus Sampling)** - In this procedure, a tiny sample of tissue from the placenta (placentas are made up of tissue called chorionic villi) is used to provide cells that will grow in the laboratory and be tested for their chromosomal content. For this procedure, we will refer you to an excellent specialist. At his office, you will receive counseling about the risks of the disorder and the risks of the procedure. The tissue is obtained either through the cervix or with a needle through the abdomen. The benefit of CVS is that it can be done in the first trimester. The risk of miscarriage is higher with this procedure.
- **Amniocentesis** – We will refer you to a specialist who performs amniocentesis. A needle is inserted into the abdomen at 12 to 20 weeks gestation, amniotic fluid is withdrawn, and the fetus' skin cells which have sloughed off into the amniotic fluid are cultured (multiplied by growth) in the laboratory to be tested for chromosomal content. If you would plan for termination of a pregnancy if the results were abnormal, a second trimester termination is more difficult. The test is much easier to perform than a CVS and the risks are fewer.

Some patients choose to use two screening tests to help them decide whether to go on for amniocentesis or CVS.

- **Nuchal fold thickness** – There is an area at the back of the neck in first trimester embryos called the nuchal fold that is enlarged in fetuses with Down's Syndrome. This area can be best seen with ultrasound exam at 11 weeks gestation. This screening is still under investigation, and we do not know the absolute predictability of this test, but we are willing to offer this testing if you are interested.
- **Triple Test** – Again, the false positive and false negative results of this test are quite high; yet some of our patients find this a useful aid in deciding whether to undergo amniocentesis.

**As always, we are more than happy to discuss this with you at any time.**

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