

## **SHOULD I HAVE PRENATAL TESTING DONE?**

Although most babies are born in great condition, worrying that your baby may have a problem is common in pregnancy. A variety of tests are available to help detect some of these problems. This worksheet will help you decide if you want any testing done, and if so, what type. Please review this information and answer the questions at the end **BEFORE** your first visit with the nurse, and bring it with you so we can help you make a plan for your pregnancy.

Prenatal tests are used to screen for problems such as chromosome abnormalities (Downs or Edwards Syndrome for example), or open neural tube defects (spina bifida, anencephaly etc). Test types fall into two categories: screening tests and diagnostic tests. They can be performed in the first trimester at 10 – 13 weeks, the second trimester at 15 – 18 weeks, or in both trimesters.

- Screening tests are used to divide people into lower risk or higher risk groups for certain problems. Screening tests do not tell you if your baby has a problem, they only tell you if you are above or below average risk for them. The advantage of screening tests is that they pose little to no risk for the baby or the mom. The disadvantage is that they don't give you a definite answer, and they have "false positives" (abnormal test, normal baby) and "false negatives" (normal test, abnormal baby). The chance that an affected baby will have an abnormal screening test (abnormal baby, abnormal test) is called the "detection rate".

- Diagnostic tests are very accurate and will almost always give you a definite yes or no answer to your question. Diagnostic tests are nice because you usually know for sure what's happening, but they carry small risks, sometimes causing the miscarriage of a normal baby. So the result of a diagnostic test is more definite than one you get with a screening test, but the risk is higher too.

## **SCREENING TESTS**

### 1) "Combined Test"

This is the combination of a blood test and an ultrasound at 10 – 12 weeks gestation that measures a pregnancy protein in your blood stream (PAPP-A), and the thickness of the tissue on the back of the baby's neck (nuchal lucency). These test results are available early, but the chance of a false positive is higher than with the other screening tests, and therefore more normal babies will be lost due to more diagnostic tests. If you want a screening test and cannot wait until the second trimester for results, the Combined test is your best option.

### 2) "Quad Test"

Four substances in the mother's blood are analyzed at 15 – 20 weeks, and you are classified as "lower than average" or "higher than average" risk for chromosome problems or open neural tube defects. Higher false positive and lower detection rate than Integrated tests. Best option for patients not receiving care in the first trimester

### 3) Integrated Tests ("Serum Integrated" or "Full Integrated")

These tests combine results from two sets of tests, one set done at 10 – 12 weeks, and one set done at 15 – 17 weeks. The results are only available after the second set of tests are drawn. The advantage of these tests is a high detection rate with a low false positive rate. The disadvantage is that results are not available until later than the Combined test.

a) “Serum Integrated”

Blood tests are done on the mother at 10 – 12 weeks (PAPP-A) and again at 15 – 18 weeks (Quad test). The two blood samples are run together after the second sample is obtained. Good choice if you have no insurance coverage for nuchal lucency.

b) “Full Integrated”

Just like the Serum Integrated plus a nuchal lucency ultrasound is done on the baby at 10 – 12 weeks. The results of the ultrasound and both blood tests are analyzed together after the second blood draw. This test has the lowest false positive rate, and therefore will result in the fewest normal babies lost, but the results are not available until 17 – 19 weeks. If you want a screening test done, and can wait until the second trimester for the results, this test offers the highest detection rate and lowest false positive rate of all the screening tests.

Look over this table for more information about the pros and cons of each of the above screening tests.

| TEST             | RESULTS AVAILABLE | DETECTION RATE | FALSE POS RATE (“Screen Pos Rate”) | MISCARRIAGE from diagnostic test (per 100,000 women screened, at 85% detection rate) |
|------------------|-------------------|----------------|------------------------------------|--|
| Combined Test    | 11 – 13 weeks     | 85%            | 5%                                 | 44   |
| Quad Test        | 17 – 20 weeks     | 76%            | 5%                                 | 45   |
| Serum Integrated | 17 – 20 weeks     | 87%            | 5%                                 | 19   |
| Full Integrated  | 17 – 20 weeks     | 94%            | 5%                                 | 9  |

### DIAGNOSTIC TESTS

1) “Chorionic Villus Sampling” (CVS)

A small sample of the placenta is taken by passing a catheter through the mother’s abdomen or cervix between 10 and 13 weeks. The cells are grown to detect chromosome problems, like Down’s syndrome. This test is not available in Lansing, but we work with experienced doctors in Grand Rapids and Detroit and will refer you if you wish.

Miscarriage risk following this procedure is 1 or 2 in 100 procedures done.

2) Amniocentesis

A needle is passed through the mother’s abdomen into the uterus under ultrasound guidance, to withdraw amniotic fluid, which is sent for chromosome and biochemical analysis. Miscarriage risk following the procedure is 1 or 2 miscarriage in 200 procedures done.

**FACTS TO CONSIDER**

- Diagnostic tests are less safe but more accurate
- Screening tests are safer but don't give you a "yes or no" answer
- Tests done earlier in pregnancy, such as the Combined test, give you earlier results, but will lead to more amniocenteses or CVS procedures, and therefore more miscarriages of normal babies. Tests that include second trimester blood tests, such as the Serum Integrated and Full Integrated tests, are the most accurate and lead to the fewest miscarriages of normal babies, but the results are not available until later.
- The conditions being tested for cannot be "fixed", even if we know about them in advance, however obstetrical decisions you make might be influenced by the results. For instance, if you knew your baby had a condition that would result in the baby's death within the first few months of life, you might decide against a cesarean delivery for fetal distress.
- Prenatal testing is done to gather information, not to find abnormal babies for pregnancy termination. Many parents who would not terminate an abnormal pregnancy nevertheless have prenatal testing done, in order to be prepared for their newborn's special challenges.
- Many mental and physical handicaps are not due to chromosome abnormalities or anatomic abnormalities, and are therefore not detectable by blood tests, ultrasound, or amnio/ CVS.

**CIRCLE YOUR AGE-BASED RISK**

| AGE AT DELIVERY | DOWN SYNDROME RISK | TOTAL RISK FOR ALL CHROMOSOME PROBLEMS |
|-----------------|--------------------|--|
| 20 – 24         | 1/1490             | 1/500                                  |
| 25 – 29         | 1/1120             | 1/450                                  |
| 30              | 1/952              | 1/417                                  |
| 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                                   |

### QUESTIONS TO ANSWER

• How worried are you about your baby’s health?

Some parents are confident all is well, and need very little reassurance. Others are more worried and would like as much information as we can give them to help. If you are high risk (over age 34 at delivery, family or personal history, exposures to drugs or chemicals, etc) you may be more worried, or perhaps you are low risk, but are just a “worrier”. You have to decide how much reassurance you need and what you are willing to do to get it.

I am very worried about my baby’s health because \_\_\_\_\_

I worry sometimes, but feel things are probably fine

I am confident things are fine and I don’t worry

• How much risk are you willing to take to be reassured about your baby’s health?

To get 100% assurance that your baby’s chromosomes are normal, you must have an invasive test that has a small chance of causing miscarriage. Screening tests are noninvasive, but less accurate. Some parents decide peace of mind is worth the small risk of an invasive test, others are satisfied to live with some uncertainty in return for no risk.

I would accept the small risk of an invasive test (CVS/amnio)

I would accept the risk of an invasive test only if the chance for a problem was high

I would never accept the risk of an invasive test, regardless of how high the chance for problems was

• What level of testing is right for me?

Please select the choice(s) that fit you best. We will discuss this worksheet further at your office visit.

I definitely need to know that my baby’s chromosomes are normal, please schedule me for counseling and invasive testing

I would like a 20 week ultrasound, but am not interested in any other screening or diagnostic tests. I will “take what comes” regardless of how high risk I am

I am interested in a 20-week ultrasound and some screening tests:

I am interested in first trimester screening test (“Combined test”)

I am interested in second trimester screening (“Quad test”)

I am interested in Integrated testing (Serum or Full)

Patient Signature \_\_\_\_\_ Date \_\_\_\_\_

RN Review \_\_\_\_\_ Date \_\_\_\_\_

Doctor Review \_\_\_\_\_ Date \_\_\_\_\_

**OFFICE USE ONLY: CIRCLE APPROPRIATE**

| TEST                    | TARGET GA     | TARGET DATE | DATE DONE | REVIEWED |
|-------------------------|---------------|-------------|-----------|----------|
| Ultrasound only         | 18 – 20 weeks |             |           |          |
| Combined (blood)        | 10 – 12 weeks |             |           |          |
| (NT)                    | 10 – 12 weeks |             |           |          |
| (Optional AFP)          | 15 – 18 weeks |             |           |          |
| Serum Integrated        | 10 – 12 weeks |             |           |          |
|                         | 15 – 18 weeks |             |           |          |
| Full Integrated (blood) | 10 – 12 weeks |             |           |          |
| (NT)                    | 10 – 12 weeks |             |           |          |
| (blood)                 | 15 – 18 weeks |             |           |          |