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First Trimester Down Syndrome Screening Options

Some facts...

- The vast majority of babies are normal at birth.
- All women, whatever their age, have a small chance (3-5%) of delivering a baby with a physical and/or mental birth defect.
- In some cases, the birth defect is due to a chromosome problem such as trisomy 21, which causes Down syndrome.
- Down syndrome results in mental retardation and occasional other physical abnormalities such as heart defects.

Risks for Trisomy 21 in relation to maternal age

The table below shows how the chance of having a baby with trisomy 21 gets higher as a mother gets older:

Mother's Age (yrs)	Chance of Trisomy 21	
	At birth	At 12 wks of pregnancy
20	1 in 1526	1 in 1018
25	1 in 1351	1 in 901
30	1 in 894	1 in 596
35	1 in 356	1 in 249
40	1 in 96	1 in 64

In the beginning of a pregnancy, the chance that a baby has a chromosome problem is higher than at birth because many babies with chromosome problems die naturally during pregnancy.

The First Trimester Scan

At this point, the baby can usually be seen by abdominal ultrasound. Occasionally, the view is not clear and a vaginal scan is needed.

At the 11-14 week scan we confirm that the baby is alive, and we assess the baby's age by measuring his or her length. This will confirm your delivery date.

We also look for major physical defects and measure **nuchal translucency** (NT) thickness. Measuring the NT allows us to calculate the chance that your baby has certain chromosome problems, including trisomy 21. Generally we cannot tell the baby's sex.

Calculating the chance of Trisomy 21

Nuchal translucency thickness is the black space (fluid) in the neck area of the baby. Findings in over 100,000 pregnancies have shown that most babies have some fluid that can be seen. We use a computer program that combines the NT thickness and your age to calculate an estimate of your risk.

When there is more than the normal amount of fluid, the chance of a chromosome problem, a heart problem, or certain rare genetic syndromes is higher. However, many babies have more than the normal amount of fluid and do **NOT** have a chromosome problem. This is called a **false positive**.

Also, some babies who **DO** have a chromosome problem may have **NORMAL** amounts of fluid in the neck area. This is called a **false negative**.

What next...

If you are comfortable with the results of the NT measurement, you can choose a maternal blood serum screening test. The available tests are called the **combined screen** and the **integrated screen**. If you are worried about the results from the NT measurement, you may feel that you want a more definitive answer. This is available through either

chorionic villous sampling or **amniocentesis** – see over. One advantage of these tests is that you get a definite answer (not another number or probability) about whether or not your baby has certain chromosome problems. One disadvantage is that these tests may cause miscarriage.

The Combined Screen

The **combined screen** is a test that combines the NT and a blood test done at 10-12 weeks. This is another no-risk way to estimate of the chance that your baby has trisomy 21. It is based on the amounts of certain chemicals in your blood. The estimate of the chance your baby has trisomy 21 from this blood test might be very different from that based on the NT measurement alone, but the combination of tests is more accurate and results in less falsely concerning results.

The combined screen is currently only offered by Dynacare laboratories. The first blood test (called the PAPPa and hCG) can be done anytime between 10 weeks 0 days and 12 weeks 6 days. If you have your NT between those times we can send you for the blood test at the same time. Results are generally available about 1 week after the blood test is drawn. At that time you can choose a CVS or amniocentesis if the results are concerning.

The advantage of the combined screen is that the final risk number is much more accurate than either the quad screen or age alone, and results are available early in pregnancy. **The test detects 86-91% of babies with Down syndrome and 5% of women will have a false positive test result.**

The Integrated Screen

The **integrated screen** is similar to the combined screen, but adds another blood test done between 15-21 weeks of your pregnancy. The integrated screen is also currently only offered by Dynacare laboratories.

A final result is not available until about 1 week after the **second** blood test is

completed (about 16-17 weeks of pregnancy). At that time you can choose an amniocentesis if the results are concerning.

The advantage of the **integrated screen** is that the final risk number is slightly more accurate than the combined screen. The disadvantage is that the final result is not ready until 16 weeks at the earliest. **The test detects 85% of babies with Down syndrome. 1% of women will have a false positive test result.**

For women who choose not to have any first trimester screening, the quad screen is available. The quad screen is a blood test done between 15 and 20 weeks that measures the same chemicals as the second part of the integrated screen. The levels of these chemicals are used, in combination with your age, to determine the risk of trisomy 21 in your baby.

Targeted 2nd Trimester Ultrasound

The 11-14 week ultrasound allows us to identify some **but not all** serious physical defects. We therefore recommend that all women have another ultrasound at 18-24 weeks to look more closely at the baby. Even with the best equipment and most experienced operators, however, not all birth defects will be detected by ultrasound.

More information about CVS and amniocentesis

Basic information on CVS and amniocentesis is below. Our genetic counselors are happy to meet with women and families who want to have one of these tests, or who simply want more information.

What is CVS...?

CVS involves taking a small piece of the chorionic villi (placental cells). Both your baby and its placenta (afterbirth) developed from the same cell, and therefore have the same chromosomes. That is why cells of the placenta can be used to check the chromosomes of the baby.

How is CVS done...?

A fine needle is passed into the uterus through the mother's abdomen and a sample of cells taken. The needle is watched carefully by ultrasound so that it does not hurt the baby. CVS takes less than 1 minute. Rarely, the position of the placenta requires us to get a sample of cells via a fine plastic tube passed through the cervix.

When can I expect the results...?

The results are usually available within 2 weeks. A result for Down syndrome may be available in 48 hours if specially requested or indicated because of an especially worrying nuchal scan.

Need to repeat the test...

In about 1% of cases a second CVS or an amniocentesis may need to be done. This is because the cells will not grow in the laboratory or the results are inconclusive.

What are the risks of CVS?

The risk of miscarriage due to CVS is about 1% and has been shown in randomized studies to be as safe as an amniocentesis done at 16 weeks. Some studies have shown that when CVS is done before 10 weeks, there is a small chance of damage to the baby's fingers or toes. Therefore, we do not do CVS before 10 weeks.

Amniocentesis at 15-22 wks

Amniocentesis involves passing a thin needle into the uterus in order to take some fluid from around the baby. The procedure takes less than 1 minute. This test gives the same information as CVS but is done between 15-22 weeks. The results are available within 2 weeks. A result for Down syndrome may be available in 48 hours if specially requested or indicated because of an especially worrying test result. Amniocentesis also allows the lab to test the fluid to detect 98% of spinal defects (spina bifida). Spinal defects are detected with

equal efficiency by an 18-23 week scan. The risk of miscarriage from amniocentesis is usually quoted as 0.8%, but has been shown in randomized studies to be the same as for CVS. It is very rare that an amniocentesis needs to be repeated.

Summary of your options

There are currently **three** different ways to get an estimate of the chance your baby has certain chromosome problems:

- First trimester scan at 11-14 weeks
- Maternal blood test
 - ❖ Combined screen at 10-12 weeks
 - ❖ Quad screen at 15-20 weeks
 - ❖ Integrated screen at 10-12 and 15-20 weeks
- Second trimester scan at 18-24 weeks

These tests estimate the chance— they do not give you a definitive answer

If at any point you feel the estimated chance is high enough that you would like to have a definitive answer, you can choose one of these two tests:

- Chorionic villous sampling at 10-14 weeks
- Amniocentesis at 15-22 weeks

Doing a CVS or an amniocentesis increases your chance of miscarriage by 1.0%.

If you have any questions, suggestions or comments, or if you would like more information, please contact our genetic counselors. They can be reached in either our Kirkland or Bellevue office.



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