

Sequential Screen for Down Syndrome

Down syndrome, also known as Trisomy 21, is caused by an extra chromosome 21 in all the cells of the body. It is seen in 1 per 800 live births, and usually occurs in women without a family history of genetic abnormalities. It is the most common genetic cause for mental retardation in this country, and can also lead to certain birth defects.

In the past, the only method available for identifying women at higher risk for Down syndrome was “advanced maternal age,” which used to be defined as a mother who was 35 years old or greater. However, by using this cut-off, only 30% of all Down syndrome cases could be detected.

There is now a newer screening option available to you which can increase the detection of Down syndrome to 90%. It is called a sequential screen. This test will not tell you if the baby does or does not have Down syndrome, but it will give you a more accurate estimation of your risk compared to using your age alone.

The sequential screen is a two-part test. Part 1 involves an ultrasound examination between 10.9 and 13.9 weeks to measure the thickness in the back of the baby’s neck (nuchal translucency), along with a blood test from your finger. If the results from Part 1 indicate an increased risk, you will be notified and offered additional testing (see below). If results from Part 1 are reassuring, you will be asked to complete Part 2 of the test, which is ideally performed at 16-18 weeks, and requires a blood sample from your arm. After Part 2 is completed, you will be given a final result, which takes into account your age and the results from Parts 1 and 2. The sequential screen has a very high detection of Down syndrome (90%), but it requires that you return for both parts of the screen. It may also not be available by all laboratories that we are directed to use by your insurance company.

The sequential screen will also detect 90% of fetuses with Trisomy 18, another serious chromosomal abnormality, as well as 80% of neural tube defects such as spina bifida.

In 3.5% of normal pregnancies, the sequential screen will come back positive for Down syndrome. This is known as the false positive rate. In the vast majority of cases (~ 90%), women with a positive screen will not have a baby with Down syndrome. If the sequential screen does come back positive (either in the first or second trimester), you will be offered genetic counseling to discuss your specific risks, and you will be offered a more invasive test, chorionic villus sampling (CVS) or amniocentesis. These tests sample either the placenta (afterbirth) or amniotic fluid, and will tell you for sure if your baby does or does not have Down syndrome.

The sequential screen is available to all pregnant women, regardless of their age. For women over 35, the detection rate for Down syndrome is higher, but more women will have a false positive test result. As the majority will have a negative screen, we encourage women over 35 to consider having the sequential screen.

It is important to remember that the sequential screen is optional. If you would not have a diagnostic test for a positive screen, would not terminate a Down syndrome pregnancy, or simply wish not to be tested, you may decline screening.

As stated by the American College of Obstetricians and Gynecologists (ACOG), all women have the option to have invasive testing by CVS or amniocentesis. Unlike the sequential screen, CVS and amniocentesis are diagnostic tests which will give you a definite answer but are associated with a small risk of miscarriage. For any patient considering diagnostic testing, you would need to speak with a genetic counselor first to discuss the risks and benefits of this testing.

Adapted from Cooper University Hospital, Department of Maternal Fetal Medicine