

## **SCREENING FOR CHROMOSOME ABNORMALITIES AND OPEN NEURAL TUBE DEFECTS**

**Screening:** A screening test is an evaluation offered to all patients that is designed to identify women who have a higher risk than normal. Most women who “fail” a screening test will be found to have a normal pregnancy. Screening helps identify pregnancies with certain conditions or diseases. The main screening tests for birth defects are:

### **FIRST TRIMESTER SCREENING OR NT TESTING:**

We offer first trimester screening for several chromosome abnormalities (Down syndrome and trisomy 18) and open neural tube defects (Spina bifida). This testing involves a sonogram to measure specific anatomy of the fetus (a “nuchal translucency” or NT, a measure of the nape of the neck). The testing also involves blood samples drawn on the patient (NOT on the baby); typically two blood samples are drawn, one in the first trimester and one in the second trimester. This test is non-invasive and does not impose a risk on the pregnancy.

The SequentialScreen™ is the main type of first trimester screening that we offer. It is a two-part test that gives patients two results- an early, preliminary result and a final, more complete result.

Detection rates:

	Down Syndrome	Trisomy 18	Neural tube defects
Part 1	70%	80%	-----
Part 2	90%	90%	80%

The first part of the SequentialScreen™ includes genetic counseling to evaluate the family history and explain testing options, the NT sonogram, and a blood draw on the patient. This should be completed between 10 weeks 5 days and 13 weeks 5 days after the woman’s first day of her last period (LMP).

The second portion just involves a blood draw; this blood draw must be done at a Saint Luke’s facility so it will route back to the correct laboratory. The second blood draw is done between 15 and 21 weeks post-LMP, with 16 to 18 weeks being preferred.

Patients receive a call from one of our genetic counselors with results from both portions of the test. The referring physician receives a report with each result as well.

### **SECOND TRIMESTER SCREENING:**

This test is also called an AFP test, triple screen or quad screen. It involves a single blood draw at about 15 to 20.9 weeks post-LMP. This test is usually drawn by the patient’s physician. The advantage of this test compared to the first trimester screen is

that it is simpler and can be done in the patient's community. The disadvantage is that no results are available until mid-pregnancy and the detection rates for chromosome abnormalities are a bit lower.

Detection rates:

	Down Syndrome	Trisomy 18	Neural tube defects
Triple Screen	60%	60%	-----
Quad Screen	75%-80%	60%-80%	80%