

## The health of her unborn child is often a pregnant woman's greatest concern.

Prenatal (before birth) screening can help deal with this concern. Prenatal screening helps find out if the fetus is at greater risk of certain birth defects. This brochure should help you understand the Integrated Screen test and what the next steps might be if your results show the fetus is at increased risk.

### Who should have prenatal screening?

The American College of Obstetricians and Gynecologists recommends that prenatal screening for Down syndrome, trisomy 18, and neural tube defects be offered to all pregnant women, regardless of their age.

### Do I have to have prenatal screening?

You should discuss your options with your health care provider. But, the decision about whether or not to have prenatal screening is up to you. The following information can help you decide.

### What is Down Syndrome?

Down syndrome (trisomy 21) is a condition caused by an extra copy of chromosome number 21. Babies with Down syndrome have a distinct physical appearance and some degree of mental retardation. They will often have other birth defects. About 1 in every 600 babies is born with Down syndrome. The chance of having a child with Down syndrome increases as a woman gets older. But a woman of any age can have a baby with Down syndrome.

### What is trisomy 18?

Trisomy 18 is a condition caused by an extra copy of chromosome number 18. Babies with trisomy 18 have severe mental retardation and multiple birth

defects. Ninety percent of babies born with trisomy 18 die within their first year of life. Trisomy 18 occurs in only 1 out of 7,000 births.

### What is a neural tube defect?

The neural tube develops into the spine and brain of the baby very early in pregnancy. If the neural tube doesn't develop properly, the result can be a defect in the baby's spine and/or brain. Neural tube defects occur in 1 to 2 out of 1,000 births and are not associated with the mother's age.

### What is the Sequential Integrated Screen?

It is a test that provides information about the fetus's chance of having Down syndrome, trisomy 18, or a neural tube defect. It can detect:

- About 92% of fetuses with Down syndrome
- About 90% of those with trisomy 18
- About 80% of fetuses with a neural tube defect

### How is the Sequential Integrated Screen done?

It is performed in two steps. The first step involves taking a small sample of blood from your arm in the first trimester of pregnancy. This sample is used to measure the level of two substances in your blood. Additionally, an ultrasound is done to take a measurement at the back of the fetus's neck. This measurement is called nuchal translucency.

The results from step 1 are used to identify pregnancies with a high risk of Down syndrome.

If the results from step 1 indicate the fetus is at high risk, your doctor may suggest diagnostic testing. If the results from step 1 do not indicate a high risk, no result is reported until another blood sample is taken from your arm in the second trimester. The level of four substances are measured. This is step 2.

The measurements from step 1 are combined with the measurements from step 2 to determine the fetus's risk.

### What if my result is "screen negative"?

After step 1, "in process" means that so far the fetus is not known to be a high risk of Down syndrome. You will proceed to step 2.

After step 2, "screen negative" means that the fetus probably doesn't have Down syndrome, trisomy 18, or a neural tube defect. But, a negative screen does not guarantee the birth of a healthy baby.

### What if my result is "screen positive"?

After step 1, it means that the fetus has a high risk of having Down syndrome. It does not mean that the fetus is definitely affected. You will not proceed to step 2. Instead, you need to consider additional diagnostic testing. Your health care provider can help you understand the risk and explain the additional tests that may be recommended.

After step 2, it means that the fetus has an increased risk of having Down syndrome, trisomy 18, or a neural tube defect. It does not mean that the fetus definitely has one of these disorders. In fact, the fetus does not have these conditions in the majority of cases. Your health care provider can help you understand the risk and explain the additional tests that may be recommended.

### What are these additional tests?

If your test result is screen positive after step 1, your doctor may recommend chorionic villus sampling (CVS) or amniocentesis.

If your test result is screen positive after step 2, your doctor might order a detailed ultrasound and/or recommend amniocentesis.

