The health of your 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 your baby is at greatest risk of certain birth defects. This brochure will help you understand the first trimester prenatal screening test and what the next steps might be if your results show your baby is at increased risk.

Who should have prenatal screenings?
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 a prenatal screening?
You should discuss your options with your doctor. But the decision about whether or not to have prenatal screening is up to you. The following information will help you decide.

What is Down syndrome?
Down syndrome (trisomy 21) 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 800 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 6,000 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 second-trimester screening?
The second-trimester screen is a blood test that provides information about your baby’s chance of having Down syndrome, trisomy 18 or a neural tube defect. There are three tests available: the Triple Screen, the Quad Screen and the Penta Screen. These tests measure several substances that may be present in abnormal amounts if your baby is at risk for one of these conditions. The tests are alternatives. Only one of them is needed.

What is the difference between the second-trimester screening tests?
The Triple Screen measures three substances in your blood: AFP, uE3 and hCG. It leads to the detection of:
• 60% to 70% of Down syndrome cases
• 60% of trisomy 18 cases
• 80% of open neural tube defects

The Quad Screen adds a fourth substance called inhibin A. It leads to the detection of:
• 70% to 75% of Down syndrome cases
• 60% of trisomy 18 cases
• 80% of open neural tube defects

The Penta Screen adds a fifth substance called h-hCG. It leads to the detection of:
• 83% of Down syndrome cases
• 60% of trisomy 18 cases
• 80% of open neural tube defects

How is second-trimester screening done?
A small blood sample will be taken from your arm sometime between your 15th and 22nd week of pregnancy. Sometime between 16 and 18 weeks is best. The blood is then tested for the amount of several substances. The results are combined with information about your age, weight, due date, ethnic background, and medical and family history to help your physician interpret the results.

What if my result is “screen negative”? A “screen negative” means that it is unlikely your baby has 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”? If your result is “screen positive,” it means that your baby has an increased risk of having Down syndrome, trisomy 18 or a neural tube defect. It does not mean that your baby definitely has one of
these disorders. In fact, the baby does not have these conditions in the majority of cases. Your doctor 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, your physician may order a detailed ultrasound to get more information about your pregnancy. During amniocentesis, a needle is inserted into the mother’s abdomen and some of the fluid surrounding the baby is taken out. This fluid contains cells and other substances from the baby. The lab can test the fluid and cells to find out if the baby has Down syndrome, trisomy 18 or a neural tube defect.

What if the additional tests are negative?
If the additional tests are negative, your baby is likely to be healthy; however, negative test results do not guarantee that your baby will not have some form of a birth defect.

What if the additional tests are positive?
If the additional test is positive, genetic counseling will help you learn more about your baby’s condition. This will help you make decisions in the best interest of you, your family and your baby.

Summary
Second-trimester screening:
• Helps find out what your chances are of having a baby with Down syndrome, trisomy 18 or neural tube defect.
• Is a blood test that measures three to five different substances.
• Detects up to 83% of babies with Down syndrome, 60% with trisomy 18 and 80% with open neural tube defect.
• Does not guarantee a healthy baby if the results are “screen negative.”
• Is not diagnostic. A “screen positive” result does not necessarily mean your baby has a problem. Additional testing is needed.

For more information, visit www.questdiagnostics.com and talk to your doctor.

Information provided by Quest Diagnostics.