

## THE TRIPLE/QUAD SCREEN TEST

Your healthcare provider has offered you a blood test called the Triple/Quad Test Screen. This brochure has been designed to answer the most commonly asked questions about the Triple/Quad Test Screen and Prenatal screening.

### WHAT IS THE TRIPLE/QUAD SCREEN?

The Triple/Quad Test Screen is a prenatal screening test that is offered to many pregnant women. The purpose of a screening test is to identify a group of people (in this case pregnant women) at increased risk for certain conditions. A screening test is not capable of diagnosing the conditions. The conditions screened for by the Triple/Quad Test Screen are birth defects called neural tube defects and Down's syndrome. The Triple/Quad Test Screen will detect 80-90% of open neural tube defects, approximately 60-75% of Down's syndrome cases and approximately 60% of cases of Trisomy 18.

### WHAT ARE NEURAL TUBE DEFECTS AND DOWN'S SYNDROME?

Neural tube defects (NTDs) are defects in the normal formation of the spinal cord or brain. Spina bifida is a form of neural tube defect. In this condition, the neural tube does not properly connect with all the nerves in the lower part of the body. Spina bifida can interfere with bowel and bladder control and may also be associated with an accumulation of fluid in the brain (hydrocephalus). In most cases surgery is necessary (often more than one) during infancy and childhood. Neural tube defects occur in about one to two per one thousand pregnancies.

Down's syndrome is caused by a change in the normal number of chromosomes. Chromosomes are small particles inside the cells. Humans normally have 46 Chromosomes, but people with Down's syndrome have 47 Chromosomes. People with Down's syndrome have varying degrees of mental retardation and may also have heart and other defects. Down's syndrome occurs in about one in one thousand births.

### DOES THE TRIPLE/QUAD SCREEN TEST FOR ANY OTHER BIRTH DEFECTS?

Yes. In certain cases, less common chromosome defects are associated with unusual screening results. Other defects called abdominal wall defects (a defect in the normal closure of the abdominal wall) can be screened for as well.

### HOW IS THE TRIPLE/QUAD TEST SCREEN PERFORMED?

The Triple/Quad Screen is a simple blood test that is best performed **between 15 to 20 weeks** in pregnancy. The three/four chemicals, alpha fetoprotein (AFP), human chorionic gonadotropin (HCG), unconjugated estriol, (uE3) and Dimeric Inhibin A (DIA) are naturally produced by the fetus or the placenta. Some levels of these chemicals are always found in a pregnant woman's blood. Once the levels are measured, they are compared with the amounts normally present during pregnancy.

### WHAT DO ABNORMAL LEVELS OF THE PROTEINS IN MY BLOOD INDICATE?

There are several possibilities: your Triple/Quad Test Screen may either indicate an increased risk for neural tube defects (elevated AFP levels) or an increased risk for Down's syndrome (lower levels of AFP and uE3 and a higher level of HCG). Approximately 8% of all pregnancies tested will have abnormal values but very few will actually have a birth defect. The most common reason for the Triple/Quad Test Screen to yield abnormal results are: 1) the pregnancy is either not as far along or is farther along than was thought, 2) there is more than one baby, 3) normal variation in the levels of protein in the blood.

Remember: Abnormal results do not diagnose birth defects; they suggest that follow up tests are indicated for further clarification. Most women with these results have normal, healthy babies.

### WHAT TESTS ARE RECOMMENDED IF THE TRIPLE/QUAD TEST SCREEN SHOWS AN INCREASED RISK FOR BIRTH DEFECT?

You may first be asked to have a sonogram (ultrasound) exam to see if any of the simple causes like twins or different dates can explain the results. Some of the defects (neural tube defects and abdominal wall defects) can be seen on sonogram. Occasionally, a repeat AFP sample will be requested when the first level is elevated.

If the sonogram does not provide an explanation for the test results, you may then be offered genetic counseling and an amniocentesis. Genetic counseling provides you with an opportunity to discuss the screening results and to make a decision whether further diagnostic testing such as amniocentesis would be helpful to you. The decision to do any further testing is yours.

### HOW WILL I LEARN THE RESULTS OF MY TEST?

All results are sent directly to the referring healthcare provider and are generally available in three to seven days. If the test results are abnormal, your health care provider will contact you directly. If it is normal, he or she will give you the results at your next regularly scheduled appointment.

It is important to remember that a normal test result cannot ever guarantee a baby will be born "perfect". Prenatal screening and testing can only assess for certain specific conditions.

The decision to have prenatal screening and the Triple/Quad Test Screen is best made by you. Your healthcare provider and this pamphlet can only offer information and guidelines; if you have any other questions or concerns about the testing, do not hesitate to inquire further so that you have all the information you need to make an informed decision.