

## Understanding Maternal Serum Screen 4 (MSS4)

### ◆What is the Maternal Serum Screen 4 (MSS4)?

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The MSS4 is a blood test performed during pregnancy to help you and your physician learn more about your developing baby. Its purpose is to SCREEN for possible neural tube defects, Down syndrome or trisomy 18 in the developing baby. The laboratory will measure four substances in your blood: Alpha-fetoprotein (AFP), human chorionic gonadotropin (hCG), estriol, and inhibin A.

AFP is a substance made by the baby that enters the amniotic fluid (the bag of water surrounding the baby) and the mother's bloodstream. A small amount of AFP is normally found in the amniotic fluid and the mother's blood. When the amount is high, it is a signal to your physician to look further for the possibility of a neural tube defect.

Estriol, hCG, and inhibin A come from the developing baby and placenta and can be measured in the mother's blood. A woman who is carrying a baby with Down syndrome may have lower blood levels of AFP and estriol and higher blood levels of hCG and inhibin A than women with unaffected baby. A woman who is carrying a baby with trisomy 18 may have lower blood levels of AFP, estriol, hCG, and inhibin A than women with unaffected babies. The MSS4 detects the same number of neural tube defects and trisomy 18 cases as other currently available maternal serum prenatal screens.

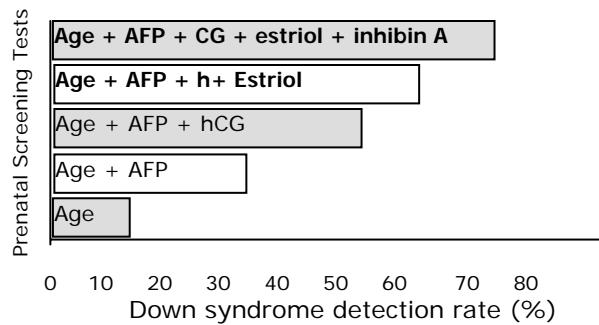
When inhibin A is used with AFP, hCG, estriol, and the mother's age, approximately 10 - 15% more babies with Down syndrome can be detected before birth.

Remember that not even the MSS4 can detect all babies with Down syndrome before they are born.

The chart below shows that the MSS4 can detect about 75 % of unborn babies with Down syndrome. The chart below shows greater than that of other maternal serum screening tests.

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### Maternal Serum Screen 4



### **◆Why should someone consider having a MSS4?**

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MSAFP (Maternal serum AFP) may lead to the detection of up to 85% of open neural tube defects, when used in conjunction with diagnostic procedures such as ultrasound and amniocentesis. Abnormal MSS4 results followed by ultrasound and amniocentesis may lead to the increased detection of Down syndrome pregnancies and many trisomy 18 pregnancies. In addition to providing information about potential neural tube defects, Down syndrome and trisomy 18, the MSS4 may provide information that could help to identify twins, find certain other abnormalities that may be present, and alert your physician to increased risks for other pregnancy complications.

A normal MSS4 is a SCREENING test and does not guarantee that you will have a healthy baby. The test is simply not able to detect every pregnancy with a neural tube defect, Down syndrome, or trisomy 18. Thus, some women with a normal MSS4 result may still have a baby with a neural tube defect, Down syndrome, or trisomy 18.

### **◆Who should have the MSS4?**

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MSS4 screening may be offered to all pregnant women, regardless of their maternal age or family history. In most cases, the MSS4 provides reassurance that the baby is developing normally. It is important for you to understand the benefits and the limitations of the MSS4. Discuss any questions or concerns with your physician.

### **◆When should I have the MSS4 test?**

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The MSS4 can be performed any time between 14 and 21 weeks after the first day of your last menstrual period. The highest detection rate for open neural tube defect is 16 -18 weeks.

The results of the test, with a full explanation, are generally available to your physician within 48 - 96 hours.

### **◆What is a Neural Tube Defect?**

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The neural tube is part of the unborn baby that develops into the spine and brain. In approximately one in every 500 developing babies, there is a defect in the development of the neural tube, resulting in either spina bifida or anencephaly. Spina bifida means, "open spine". Children born with open spine require surgery to close the opening. They also may have various medical problems, such as trouble with bowel and bladder control, walking, and learning. The degree of disability varies from one child to the next depending on the size and location of the opening. Anencephaly, a more severe abnormality involving incomplete development of the brain and skull, usually results in death before or shortly after birth.

### **◆Am I at risk for having a baby with a Neural Tube Defect?**

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Anyone can have a baby with neural tube defect. If someone in your family was born with a neural tube defect, you need to discuss this with your physician, since your baby has a greater risk. If you have no family history, then your risk is no greater than the general population risk. However, you should be aware that most babies with neural tube defects are born to parents with no family history to such problems.

### **◆What is Down Syndrome?**

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Down syndrome is a disorder caused by an extra chromosome, which is a structure that contains genetic material that determines physical and mental characteristics. Children with Down syndrome have abnormalities that may include mental retardation, heart defects, and other health problems.

### **◆What is Trisomy 18?**

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Trisomy 18 is another disorder caused by an extra chromosome. Children with trisomy 18 also have mental retardation, heart defects, and other health problems but are more severely affected and generally die in early childhood. Trisomy 18 is much less common than Down syndrome.

### **◆Am I at risk for having a baby with Down Syndrome or Trisomy 18?**

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As with neural tube defects, anyone can have a child with Down syndrome or trisomy 18. The risk of having a baby with Down syndrome or trisomy 18 depends on your age. As a woman gets older, her risk increases. In general, a woman 35 years old or older is offered prenatal testing (amniocentesis or chorionic villus sampling) based on her age alone.

### **◆What if my result is positive for Down syndrome or Trisomy 18?**

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Low MSAFP and estriol combine with high hCG, and inhibin A concentrations may be found in pregnancies with Down syndrome. Low levels of MSAFP, estriol, hCG, inhibin A are often found in pregnancies with trisomy 18. These abnormal MSS4 results also may be due to a pregnancy that is less far along than what was thought previously. Following an abnormal result, your physicians may recommend an ultrasound to verify the baby's age or amniocentesis to study the baby's chromosomes.

### **◆What if my MSAFP result is elevated?**

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An elevated MSAFP result does not necessarily indicate that the baby has a neural tube defect. Since the levels of MSAFP depend, among other factors, on the age of the developing fetus, a test result may appear to be high, but may not be, if the baby's age has been miscalculated. There are also other possible causes, including twin pregnancy, vaginal bleeding, and the presence of less common birth defects. Occasionally, MSAFP results are elevated for no apparent reason.

### **◆What does a negative screen mean?**

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A negative screen means that your baby probably does not have a neural tube defect, Down syndrome or trisomy 18. Further testing is not required. A negative screen, however, does not guarantee that your baby will not have some form of birth defect.

### **◆Summary**

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A negative prenatal screen means your baby probably does not have a neural tube defect, Down syndrome, or trisomy 18; however, it does not guarantee the health of your baby.

A positive prenatal screen means additional tests are needed to determine whether or not your baby has a birth defect. If a birth defect is found, your doctor or a genetic counselor will help you make decisions in the best interest of you, your family, and your baby.