

## SECOND TRIMESTER MATERNAL SERUM SCREENING

Maternal serum screening (MSS) is an optional screening test to determine if your baby has an increased chance for Down syndrome, Trisomy 18, or open neural tube defects. Approximately 95% of women tested will have a negative (normal) screening result. Of those women with a positive (increased chance) screening result, only a small number of these pregnancies will be found to have a birth defect after further testing. In some cases, a pregnancy with a birth defect will not be identified by MSS.

### HOW DOES IT WORK?

Sometimes referred to as a Quad or Tetra screen, MSS is a blood test drawn between 15 and 20 weeks that measures the amounts of three or four proteins that are normally present in the mother's blood during pregnancy: alpha-fetoprotein (AFP), human chorionic gonadotropin (hCG), unconjugated estriol (uE3), and dimeric inhibin A (DIA). Low and/or high levels of these proteins, along with maternal age, may suggest an increased or decreased chance for the conditions. A result is considered screen positive when the levels indicate a significant chance for Neural Tube Defect, Down syndrome, or Trisomy 18. Since some unaffected pregnancies may be screen positive, further counseling and testing is suggested.

### WHAT DOES IT MEAN TO BE SCREEN POSITIVE FOR NEURAL TUBE DEFECT?

A neural tube defect (NTD) is an opening of the spine (spina bifida) or skull (anencephaly) that may result in serious problems for a baby including paralysis, intellectual disabilities, or even death. Approximately 1 in every 750 babies in South Carolina has a neural tube defect. Approximately 90% of open NTD in a fetus can be identified by the AFP blood screening test (MSAFP).

A result is considered "screen positive for NTD" when the level of AFP in the mother's blood is higher than usual. MSAFP may be elevated for other reasons such as incorrect dating, twin or triplet pregnancy, or placental insufficiency which may result in poor fetal growth or preterm labor. Follow up for an elevated MSAFP may include repeating the test, an ultrasound to check for fetal growth and wellbeing, and/or an amniocentesis to measure the amount of AFP in the amniotic fluid. Measuring AFP in amniotic fluid is a more reliable test for open NTD than the blood screening alone.

### WHAT DOES IT MEAN TO BE SCREEN POSITIVE FOR DOWN SYNDROME?

Down syndrome is one of the most common causes of intellectual disability, affecting one in every 700 babies born. Affected babies have common physical features with mild to moderate intellectual disabilities and may also have other birth defects such as heart problems. Down syndrome does not typically run in families. Approximately 75% of Down syndrome can be identified by MSS.

MSS is considered "screen positive for Down syndrome" when the protein levels indicate a chance greater than 1 in 270 (greater than the risk for the average 35 year old) for the pregnancy to be affected. Ultrasound, amniocentesis and/or noninvasive prenatal screening (NIPS) are suggested follow up tests when a pregnancy is identified as screen positive for Down syndrome.

### WHAT DOES IT MEAN TO BE SCREEN POSITIVE FOR TRISOMY 18?

Trisomy 18 is a chromosome condition that is less common than Down syndrome. It is a very serious condition which includes birth defects, severe intellectual disabilities, and shortened lifespan. Trisomy 18 does not usually run in families. Approximately 75% of Trisomy 18 can be identified by MSS.

MSS is considered "screen positive for Trisomy 18" when the protein levels indicate a chance greater than 1 in 100 for the pregnancy to be affected. Ultrasound, amniocentesis and/or NIPS are suggested follow up tests when a pregnancy is identified as screen positive for Trisomy 18.

### WHO SHOULD HAVE THE TESTING?

Maternal serum screening is offered to all women who do not have a family history of one of these conditions. Women over 35 years of age or with a family history of chromosome conditions are usually offered genetic counseling and additional testing options, such as CVS, amniocentesis, and/or non-invasive prenatal screening (NIPS).

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