

Read this information with your partner and discuss any questions with your doctor or nurse. In addition you may call 319-356-8892. An AFP nurse will return your call.



Department OB/Gyn
University of Iowa Health Care
200 Hawkins Drive
Iowa City, IA 52242
Telephone: 319/356-8892
Fax: 319/356-7556



Iowa Department of Public Health
Birth Defects Institute
State Coordinator for Genetic Services
Lucas State Office Building
321 East 12th St.
Des Moines, IA 50319
Telephone: 515/281-7584
Fax: 515/242-6384



University Hygienic Laboratory
102 Oakdale Research Campus
#H101 Oakdale Hall
Iowa City, Iowa 52242-5002
Phone: 319/335-4500
Fax: 319/335-4555

Iowa Expanded MSAFP/Quad Screen

Seeing your doctor early to begin prenatal care is an important part of preparing for the birth of your baby. There are tests designed to provide you and your doctor or midwife with information about your pregnancy and your developing fetus. Although most babies are born healthy, about three to four percent are born with a birth defect. Frequently this happens without warning, and families and doctors are not prepared for this difficult experience.

The Iowa Expanded MSAFP/Quad Screen is a test available to all women early in pregnancy. This test can identify a woman with an increased risk to have a fetus with certain kinds of birth defects or to develop a problem later in pregnancy.

What is the Iowa Expanded MSAFP/Quad Screen?

The Iowa Expanded MSAFP/Quad Screen is a single blood test drawn between the 15th and 20th week of pregnancy to measure the amount of AFP and three other markers in the mother's bloodstream. All of the markers are made by the fetus and placenta. The levels of these markers may be altered in a predictable way when a fetus has certain birth defects such as an open neural tube defect, an abdominal wall defect, or a chromosome problem such as Down syndrome or Trisomy 18.

For most women the results of this screen will be negative and no further tests are ordered. This is reassuring, but does not guarantee the fetus is normal. The MSAFP/Quad Screen is the first test in a series of tests that may be done. It cannot directly diagnose birth defects, but can help you and your doctor decide when other tests such as an ultrasound and amniocentesis may be of value.

What is Ultrasound?

Ultrasound (sonogram) uses sound waves to create pictures of the fetus. Careful measurements help to determine the age of the fetus, whether there is more than one fetus, and in some cases may reveal a birth defect.



What is Amniocentesis?

Amniocentesis involves using a thin needle to withdraw fluid from around the fetus for testing. The fetal cells and AFP in this fluid are studied to diagnose birth defects such as neural tube defects and chromosome problems such as Down syndrome or Trisomy 18.

What is AFP?

Alpha-fetoprotein is a substance produced by every fetus as it grows. This substance is passed into the amniotic fluid that surrounds the fetus and from there into the mother's bloodstream. Nothing the mother does alters the level of the protein. It is not related to the protein in the mother's diet, but comes directly from the fetus.

What if I am carrying twins?

Risk for neural tube defects can be evaluated but we do not provide Down syndrome screening for twins. We do not provide screening for pregnancies with more than two fetuses.

What are neural tube defects?

In the first weeks of pregnancy when the fetus is less than one inch long, the brain and spinal cord begin to form a structure along the fetal back known as the neural tube. The top of this tube develops into the head and brain; the rest becomes the spinal cord. Neural tube defects occur when the neural tube does not close properly around the brain or spinal cord.

The two major types of neural tube defects are spina bifida and anencephaly. About one in 1,000 pregnancies in Iowa results in the birth of an infant with a neural tube defect. Half of these have spina bifida and half have anencephaly.

The effects from spina bifida (open spine) may range from mild to severe. The severity of the problem depends upon where the defect in the spinal cord is located. The problems may range from weakness of the leg muscles to actual paralysis. A higher defect on the spine will result in more paralysis in the legs. Because the nerves that direct bowel and bladder functions are at the base of the spinal cord, there may be problems with bowel and bladder control. Over 80 percent of newborns with spina bifida have hydrocephalus (water on the brain). Corrective surgery and physical therapy can help lessen the disability in these children so that some may lead relatively normal lives. In

severe cases the baby is stillborn or dies soon after birth.

Anencephaly (open brain) occurs when the head and brain do not develop normally. Babies born with this condition are almost always stillborn or die soon after birth.

If the defect is in the spinal cord itself, but is covered by skin, the AFP will not leak into the amniotic fluid. These defects will not be detected by the blood test and the effects tend to be less severe.

What is Down syndrome?

Down syndrome is a birth defect caused by an extra number 21 chromosome in every cell of the body. Individuals with Down syndrome are mentally retarded and may have other physical problems such as heart defects. Any woman can have a fetus with Down syndrome, but it is known that Down syndrome and other chromosome problems happen more often as women get older.

What is Trisomy 18?

Trisomy 18 is a chromosome problem that is caused by an extra number 18 chromosome in all the cells of the body. Serious problems with growth and development are present before birth and physical problems such as an open spine or heart defect may be present.

What are the risks involved in the screening process?

Anxiety: Waiting for the results of a repeated test is a very anxious time for women who have had the screening test. It is natural to be afraid when considering the possibility of a problem with your baby. The results of all repeated blood tests are telephoned to the doctor's office the day they are complete. Fortunately, most women with a positive MSAFP Quad/Screen will have healthy babies.

Pregnancy Loss: If the screening process leads to amniocentesis there is a small but real risk of pregnancy loss associated with this procedure. Ask the doctor who does your amniocentesis to discuss that possibility with you.

What causes the Iowa Expanded MSAFP/Quad Screen to be positive?

A screen is called positive if the hormone levels are too high or too low compared to the normal levels for that week of pregnancy.

What are the reasons for a positive Iowa Expanded MSAFP/Quad Screen?

1. You may be earlier or later in your pregnancy than you thought. The levels of hormones normally change with each week of pregnancy. After ultrasound, the result of the SCREEN can be refigured if the corrected fetal age was between 15 and 20 weeks at the time the initial sample was drawn.
2. You may be carrying twins.
3. The hormone levels are altered, but the fetus is normal. **Remember this is a screening test** and some women with a normal fetus will have a SCREEN result that is positive.
4. The fetus may be normal, but the placenta may not be functioning properly.
5. The fetus may have a neural tube defect or abdominal wall defect. Any opening in the body of the fetus will allow higher levels of AFP to pass into the amniotic fluid and from there into your blood stream.
6. The fetus may have Down syndrome or Trisomy 18.
7. The fetus may have Smith Lemli-Opitz Syndrome.

What if the Iowa Expanded MSAFP/Quad Screen is positive showing a risk for Down syndrome?

An ultrasound is done.

If the ultrasound changes your due date the result of the SCREEN may now be normal. The blood test is only repeated if the initial sample was drawn before 15 weeks.

After ultrasound, about four percent of women will have a SCREEN that remains positive. They will be offered amniocentesis due to an increased risk for Down syndrome in the fetus. The factors used to figure the risk for chromosome problems include maternal age and weight, fetal age, and the levels of AFP, UE3, hCG and Inhibin A in the maternal blood.

What if the AFP portion of the MSAFP/Quad Screen test is elevated?

An ultrasound is done.

If the AFP result is still elevated after ultrasound confirms or corrects the fetal age, the MSAFP blood screen is repeated. About 40 percent of the repeated MSAFP SCREENS will be normal and no further testing will be needed.

What if the second MSAFP Screen result remains elevated?

Those with an MSAFP SCREEN that remains elevated will go on to have a more detailed ultrasound and possibly an amniocentesis. About one in 30 of these women will have a fetus with a potentially serious birth defect. About half of these will be neural tube defects.

If the amniocentesis results are positive (abnormal) another ultrasound will be done to identify the extent and location of the defect. This information will be needed to provide you with accurate counseling about the problems the fetus may have as a result of the defect.

If the ultrasound and amniocentesis (if done) are normal, but the second MSAFP SCREEN remains elevated, you may have an increased risk for problems later in the pregnancy. These problems are related to the function of the placenta and include early labor, a smaller than average baby and some other pregnancy problems. **Most women with high MSAFP levels will still have healthy babies,** but you should discuss your care with your doctor so you will know what to do if a problem occurs.

Are there other conditions that may be detected because of abnormal levels of these pregnancy markers?

Yes, it has been found that a very low estriol (UE3) may indicate a condition called Smith-Lemli-Opitz Syndrome. Less than one half of one percent of women screened will be positive for this condition.

What is Smith-Lemli-Opitz Syndrome?

Smith-Lemli-Opitz Syndrome is a genetic condition that results in multiple physical and mental problems in the affected individual. There may be a cleft palate, unusual fingers and toes, and cataracts. Malformations of other organ systems including the brain, kidneys, heart and lungs have also been reported. Women with lower levels of estriol will be offered counseling and diagnosis for this condition.

What can be done if a birth defect is detected?

It is a shock to learn that your fetus has a birth defect. You will need information about the specific condition and any treatment that is available. There are two primary options to consider: You may begin to plan for the birth of a baby who may need intensive care at birth or you may elect to end the pregnancy. Any available prenatal treatment options for an affected fetus will be discussed at the time of the evaluation.

SUMMARY

The Iowa Expanded MSAFP/Quad Screen will provide information about your baby before it is born. It is a test available to all women between 15 and 20 weeks of pregnancy. A negative SCREEN provides reassurance that the risk for certain birth defects and pregnancy problems has been reduced. Even though most babies will be healthy, the period of testing is a stressful time. If a problem is detected you will need support and guidance.

Read this information with your partner and discuss any questions with your doctor or nurse. In addition you may call 319-356-8892. An AFP nurse will return your call.