MUSC
MEDICAL UNIVERSITY of SOUTH CAROLINA

Lab Tests and Results

Amniotic fluid methylmalonic acid level

What is this test?

This test measures the amount of methylmalonic acid (MMA) in amniotic fluid. This test is used to diagnose a disease called methylmalonic aciduria in an unborn baby. This is an inherited disorder in which the body is unable to use certain proteins and fats[1][2].

What are other names for this test?

Amniotic fluid methylmalonic acid measurement

Why do I need this test?

Laboratory tests may be done for many reasons. Tests are performed for routine health screenings or if a disease or toxicity is suspected. Lab tests may be used to determine if a medical condition is improving or worsening. Lab tests may also be used to measure the success or failure of a medication or treatment plan. Lab tests may be ordered for professional or legal reasons. The following is a possible reason why this test may be done:

Methylmalonic acidemia

When and how often should I have this test?

When and how often laboratory tests are done may depend on many factors. The timing of laboratory tests may rely on the results or completion of other tests, procedures, or treatments. Lab tests may be performed immediately in an emergency, or tests may be delayed as a condition is treated or monitored. A test may be suggested or become necessary when certain signs or symptoms appear.

Due to changes in the way your body naturally functions through the course of a day, lab tests may need to be performed at a certain time of day. If you have prepared for a test by changing your food or fluid intake, lab tests may be timed in accordance with those changes. Timing of tests may be based on increased and decreased levels of medications, drugs or other substances in the body.

The age or gender of the person being tested may affect when and how often a lab test is required. Chronic or progressive conditions may need ongoing monitoring through the use of lab tests. Conditions that worsen and improve may also need frequent monitoring. Certain tests may be repeated to obtain a series of results, or tests may need to be repeated to confirm or disprove results. Timing and frequency of lab tests may vary if they are performed for professional or legal reasons.

How should I get ready for the test?

An amniocentesis is a procedure that requires your written consent. Review the consent form with the healthcare worker and ask any questions that you have before signing the consent form. Tell the person doing the amniocentesis if you have a history of pregnancy difficulties, such as premature (early) labor, incompetent cervix (a weak or failing cervix), placenta previa (a placenta that is abnormally low, near or over the cervix), abruptio placenta (the placenta is separated from the uterine wall too early), and if you are Rh negative (had a blood type that did not match your child's blood type). Tell the healthcare worker if you have a medical condition or are using a medication or supplement that causes excessive bleeding. You should also report if you have a history of allergic or other reactions to local anesthetics. If ultrasound is used, you will need to drink extra fluids and have a full bladder for the procedure.

How is the test done?

Amniotic fluid is the protective liquid that surrounds the unborn baby while it is in the mother's womb. A sample of this fluid is collected by a procedure called an amniocentesis. For an amniocentesis, you will lie on your back with your legs extended. You may be asked to raise your arms above your head. Usually, an ultrasound will be done at the same time as the amniocentesis. The ultrasound is used to locate your unborn baby, the placenta, and a pocket of amniotic fluid that is suitable for testing. The point selected for needle insertion will be away from your baby and the placenta.

An area of skin on your abdomen will be cleaned with an antiseptic solution, and a sterile area prepared. You will be

given anesthetic to numb your skin. When the area is numb, a needle will be introduced through your skin and into the amniotic sac (the protective sac that surrounds the unborn baby). Using ultrasound as a guide for needle placement, a small amount of amniotic fluid will be withdrawn and thrown away, and then the sample of fluid will be collected.

When enough fluid has been collected, the needle will be withdrawn. More than one needle and syringe may be needed to collect the sample. If your unborn baby moves toward the needle during the procedure, the needle will be withdrawn and the procedure may need to be repeated.

How will the test feel?

The amount of discomfort you feel will depend on many factors, including your sensitivity to pain. Communicate how you are feeling with the person doing the procedure. Inform the person doing the procedure if you feel that you cannot continue with the procedure.

Before an amniocentesis, a local anesthetic is given to the procedure site to numb the area. You may feel mild discomfort or stinging when the numbing medicine is injected. As the procedure needle is inserted through the abdomen, you may feel some discomfort and pressure. You may feel mild cramping in your abdomen and pelvic area during the procedure. The procedure site may be sore for several days.

What should I do after the test?

After an amniocentesis, a bandage will be placed over the site, and pressure applied until the bleeding or drainage has stopped. Rest is necessary. Do not have sexual intercourse, and avoid heavy lifting for at least 24 hours after the procedure.

Contact your healthcare worker if there is redness, swelling, pus, drainage, or pain at the site where the amniotic fluid sample was taken. Alert your healthcare worker immediately if you see bleeding or clear fluid leaking from your vagina, feel severe cramping in your abdominal or pelvic area, or develop a fever. Inform healthcare workers of any change in your baby's movement, such as not moving for a period of time, or suddenly moving more than usual after this procedure.

What are the risks?

Amniotic Fluid: Amniotic fluid is collected by a procedure called an amniocentesis. During an amniocentesis a hematoma (blood-filled bump under the skin) or bleeding at the puncture site may occur. If you have a medical condition, or are using a medication or supplement that causes excessive bleeding, you are at a higher risk of bleeding from the puncture site. It is possible that the needle that is used to collect fluid will injure your baby. After an amniocentesis, a bruise or infection may occur at the puncture site. You may bleed or leak amniotic fluid from the vagina. Rarely, you may develop a fever, have severe abdominal and pelvic cramping, or go into labor. There is a risk that your baby will not survive the procedure. The chances of these risks vary depending on your health status, the reason for having this procedure, and other factors. The person doing this test may need to perform it more than once. Talk with your healthcare worker if you have any concerns about the risks of having an amniocentesis.

What are normal results for this test?

Experts do not agree on what normal results are for this test.

What follow up should I do after this test?

Ask your healthcare worker how you will be informed of the test results. You may be asked to call for results, schedule an appointment to discuss results, or notified of results by mail. Follow up care varies depending on many factors related to your test. Sometimes there is no follow up after you have been notified of test results. At other times follow up may be suggested or necessary. Some examples of follow up care include changes to medication or treatment plans, referral to a specialist, more or less frequent monitoring, and additional tests or procedures. Talk with your healthcare worker about any concerns or questions you have regarding follow up care or instructions.

After an amniocentesis, results are usually available within 7 to 10 days.

Where can I get more information?

Related Companies

- Organic Acidemia Association
- Genetics Home Reference

References

1. Fensom AH, Benson PF, Chalmers RA, et al: Experience with prenatal diagnosis of propionic acidaemia and methylmalonic aciduria. J Inher Metab Dis 1984; 7(2):127-128.
2. Parvy P, Bardet J, Chadefaux-Vekemans B, et al: Free amino acids in amniotic fluid and the prenatal diagnosis of homocystinuria with methylmalonic aciduria. Clin Chem 1995; 41(11):1863-1864.