Facts about

AFP4 (Quad Marker) Screening

AFP4 is a blood test done during pregnancy. This test checks the developing baby for certain chromosome conditions and birth defects.

When you are pregnant, there are substances in your blood that come from the pregnancy. This test measures four of these substances: alpha-fetoprotein (AFP), human chorionic gonadotropin (hCG), unconjugated estriol (uE3), and inhibin-A (INH). These are made by the placenta and the growing baby. AFP4 uses your test values and your age to estimate the chance for certain conditions in the baby.

How is the test done?

AFP4 is done with a single blood test from your arm. It can be done between 15 and 21 weeks in pregnancy.

What can AFP4 tell me?

AFP4 can tell if your pregnancy has a low risk or high risk for certain conditions, including:

- Down syndrome (trisomy 21): 80% detection rate
- Trisomy 18: 75% detection rate
- Open defects of the spine and belly (spina bifida, abdominal wall defects): 80-85% detection rate

What are the limits of AFP4?

AFP4 only looks for a few conditions. This test does not find all chromosome conditions and does not screen for other birth defects or genetic conditions.

No screening test is 100% accurate. This test can raise false alarms in healthy pregnancies (false positive results). It can also miss a pregnancy that has one of the conditions (false negative results).

DID YOU KNOW?

1 in every 800 babies is born with Down syndrome each year.

1 in every 2,500 babies is born with trisomy 18 each year.

1 in every 1,400 babies is born with spina bifida each year.

1 in every 2,000 babies is born with an abdominal wall defect each year.

These conditions happen by chance and do not usually run in the family. Screening can estimate the chance of these conditions in your pregnancy.

Who can have AFP4?

AFP4 can be done if you are pregnant with one baby or with twins. AFP4 cannot be done with triplets.

How do I get results from AFP4?

Results from AFP4 usually take about 1-2 weeks. Your OB provider will review the result with you at your next visit. You may also get a secure email message or a phone call when the result is ready.

What if my AFP4 result is screen negative (low risk)?

Most people who have AFP4 get a screen negative result. A screen negative result is reassuring. It means there is a low chance for Down syndrome, trisomy 18, or an open birth defect in the baby's spine or belly. This result does not guarantee that the baby has no health problems or birth defects.

What if my AFP4 result is screen positive (high risk)?

A screen positive result means there is a difference in the expected amount of one or more of the substances being tested. This could be due to one of the conditions the test looks for: Down syndrome, trisomy 18, or a birth defect of the spine or belly. The chance is high enough that more testing will be offered. However, this result does not always mean there is a problem with the baby or the pregnancy. Most pregnancies with positive results are normal.

IMPORTANT THINGS TO CONSIDER

AFP4 screening:

...has no risk for miscarriage.

…does not find all pregnancies with Down syndrome, trisomy 18, spina bifida, or abdominal wall defects.

…does not test for all chromosome conditions or birth defects.

…may show a high risk in a healthy pregnancy (false-positive result).

...requires more testing to diagnose a condition in the baby.

What will I be offered if AFP4 shows a screen positive (high risk) result?

If you have a screen positive result you will be offered genetic counseling, ultrasound, and amniocentesis.

- **Ultrasound** uses harmless sound waves to look at the pregnancy. The sound waves are turned into a picture (or "image") on a monitor. This lets the doctor check the size of the baby and look for birth defects. Ultrasound can find more than 90% of open spine and abdominal wall defects during pregnancy.
- **Amniocentesis** can diagnose chromosome conditions by studying fetal cells in the laboratory. The test is done by passing a thin needle through the belly – without touching the baby. A small sample of the fluid around the baby is collected. There is a very small risk of miscarriage (about 1 in 1000).

IT'S YOUR CHOICE.

You can decide if you want to learn about chromosome conditions and birth defects during pregnancy.

Testing may help you prepare for a child with special needs. You might use the information to decide whether or not to continue the pregnancy. But not everyone wants to know about these conditions during pregnancy.

More about…

Down syndrome is a chromosome condition that includes mild to moderate intellectual disability and a characteristic appearance. Babies with Down syndrome also have a higher chance to be born with physical birth defects, such as a heart defect or intestinal problem.

Trisomy 18 is a chromosome condition that include severe intellectual disability and medical problems in many parts of the body. Survival beyond the first year of life is uncommon.

Spina bifida is a difference in the way the fetal spine forms early in pregnancy. Some spine bones (vertebrae) may not close normally. This can lead to nerve damage of the spinal cord. People with spina bifida may have leg weakness or paralysis, and problems with bowel and bladder control.

Abdominal wall defects are a difference in the way the muscles and skin of the belly form. This can leave an opening near the umbilical cord that lets the organs (ex: intestines, liver) move outside the baby's body. The opening is closed after birth by surgery.