PATIENT EDUCATION

Prenatal Screens and Tests

Allina Health

General Information

Most babies are born healthy. However, of all babies born, 3 to 4 percent have some type of birth defect (a physical problem) and 2 to 3 percent are mentally retarded. These risks exist in all pregnancies. Often, the reason for the birth defect is not known.

No test can absolutely guarantee that you will have a healthy baby, but your health care provider will use prenatal screening tests to tell if your baby has a higher than expected risk for some problems. This is called a screen. Remember:

- A positive screen indicates you are in a high-risk category and will be offered more tests to clarify the concern. It does not mean your baby has a problem.
- A normal or negative screen indicates you are in a low-risk category. It does not rule out the problem in question.

If you have a positive screen, there are other tests (diagnostic tests) you will be offered. These will tell whether or not your baby has the problem/disorder about which you are concerned. If you have any questions about your risks or options, ask your health care provider or genetic counselor.

Triple Test (Serum Screening)

The triple test (serum screening) is done between 15 and about 21 weeks in your pregnancy (the time varies with the lab). This test is designed to help better identify pregnancies at high risk for neural tube defects and certain chromosome problems.

Blood is drawn from your arm and measured for three of your baby's proteins in your blood: alpha-fetoprotein (AFP), human chorionic gonadotropin (hCG) and unconjugated estriol (uE3).

High levels of AFP may indicate an increased risk for a neural tube defect such as spina bifida and anencephaly. Other causes of high AFP are inaccurate gestational age, twins or triplets, or placenta problems.

When the levels of AFP, hCG and estriol are considered with your age, the lab can estimate the chance that your baby has Down syndrome and trisomy 18. This is just a more precise estimate on the chance your baby has these, not a firm yes or no answer.

Quadruple Test

The quadruple test (serum screening) is done when you are between 15 and just shy of 20 weeks in your pregnancy. The quad test is similar to the triple test but it measures one more of your baby's proteins: inhibin A.

This test is more likely to alert you to a baby with Down syndrome or trisomy 18 than the triple test. Like the triple test, it provides a new assessment of risk and cannot diagnose or rule out these conditions.

A triple test or a quadruple test is sometimes called a multiple marker test.

First Trimester Screen for Chromosome Abnormalities

The first trimester screen is typically done between 11 weeks and 2 days and 13 weeks and 6 days. This screen is done to help estimate the chance that your baby has Down syndrome or trisomy 18.

This test involves an ultrasound and a blood draw. Two proteins in your blood (PAPP-A and free beta-HCG) will be measured. These proteins come from your baby.

Babies with Down syndrome make lower amounts of PAPP-A but higher amounts of HCG than babies without these conditions. Babies with trisomy 18 make lower amounts of both proteins.

An early ultrasound measurement of your baby's NT (fluid area behind the neck) may help identify a baby who may have certain chromosome problems or heart defects or other physical problems. Babies with certain chromosome problems have larger NTs than usual.

These measurement of the proteins and the ultrasound measurement of the NT will be combined with your age-related risk for a new and more precise assessment of your baby's risk for Down syndrome and trisomy 18. If the test is considered positive you have the option for diagnostic tests such as an amniocentesis or CVS (chorionic villi sample).

Amniocentesis (A Diagnostic Test)

This is a common test for certain types of birth defects, especially chromosome problems and single gene abnormalities but single gene abnormalities are only addressed if there is a unique concern in a family. The test is typically done at 15 to 18 weeks and *may* be an option if you:

- are 35 or older at the time of delivery
- and/or your partner have a family history of an inherited disease or your ancestry puts you at increased risk **and** if carrier tests have shown the pregnancy to be at risk
- or your partner have a family history or an open birth defect such as spina bifida
- or your partner have a child with Down syndrome or other chromosome disorder
- or your partner carries a balanced chromosome rearrangement

- became pregnant by certain assisted reproductive technologies
- have a positive serum screen or an abnormal ultrasound suggesting an increased risk for certain birth defects.

A small amount of amniotic fluid is withdrawn from the sac around your baby under careful ultrasound observation by your doctor. The baby's cells are grown in culture and the chromosomes are studied.

Additional biochemical or genetic tests might be done if your history indicates that it is appropriate to do so. AFP is always measured to screen for spina bifida.

Your doctor or a genetic counselor can talk about the risks with you. The chance of a miscarriage is no more than one-half of 1 percent above the chance that the average woman has in pregnancy.

If the procedure finds a genetic disorder or birth defect, you and your partner are encouraged to talk with your doctor or genetic counselor about your options and to learn more about the baby's problems and the available services to answer your concerns.

CVS (A Diagnostic Test)

CVS is also a common test for certain types of birth defects, especially chromosome problems and single gene abnormalities but single gene abnormalities are only addressed if there is a unique concern in a family.

The procedure is usually done between the 10th and 12th week from the first day of your last menstrual period. It may be an option if you:

- are 35 or older at the time of delivery
- and/or your partner have a family history of an inherited disease or your ancestry puts you at increased risk and

if carrier tests have shown the pregnancy to be at risk

- or your partner have a child with Down syndrome or other chromosome disorder
- or your partner carries a balanced chromosome rearrangement
- became pregnant by certain assisted reproductive technologies
- if you have a positive first trimester screen.

Your doctor will remove a small sample of chorionic villi to test. The chorionic villi are finger-like projections of tissue that transfer oxygen, nutrients and waste between you and your baby (via the placenta). These cells carry the genetic material (chromosomes and genes) of your baby.

This procedure may be better for you than amniocentesis if you have high anxiety or if an early diagnosis is desired for privacy reasons. (CVS is not safer nor more accurate than amniocentesis.)

Your doctor or a genetic counselor can talk about the risks with you. There may be a slightly higher risk of miscarriage with CVS than with amniocentesis.

If the procedure finds a genetic disorder or birth defect, you and your partner are encouraged to talk with your doctor or genetic counselor about your options and to learn more about the baby's problems and the available services to answer your concerns.

Questions

If you have any questions about your pregnancy or about prenatal tests, please talk with your health care provider. He or she may refer you to a genetic counselor.

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