

Anuploidy Screening

DOUBLE MARKER TEST

All pregnancies irrespective of age of the mother are at risk of developing some problems during pregnancy. These problems can affect either health of the mother, like a high blood pressure developing during pregnancy or baby developing some problems like structural defects or genetic problems.

Down's syndrome is one such common condition. This occurs in every 700 births.

Characteristics of Down's Syndrome

People who have Down syndrome have slight to severe abnormalities –

- Eyes can incline upwards and outwards
- A single crease can be seen running across the palm of the person
- Small hands
- Low set ears
- Prone to certain medical conditions like heart problems, defects in the digestive tract and sight and hearing problems
- Cause learning disability
- Life span is less

Why Double Marker Test is Significant?

Today we have various tests available and with double marker test you can detect whether Down's syndrome is present in your unborn or not. The probability of having a baby with chromosomal imbalance or Down's syndrome is likely for elderly pregnant women. It is advisable that all pregnant women should go through this test so that she is sure that the baby is completely normal.

There are different screening tests available in various hospitals.

These tests are considered pretty accurate and are referred by the doctors as well. The accuracy also

What are the risks and side effects to the mother or baby?

Except for the discomfort of drawing blood, there are no known risks or side effects associated with the quad screen test.

When is the quad screen test performed?

The quad screen test is performed between the 16th and 20th week of pregnancy.

All pregnant women should be offered the quad screen, but it is recommended for women who:

- Have a family history of birth defects
- Are 35 years or older
- Used possible harmful medications or drugs during pregnancy
- Have diabetes and use insulin
- Had a viral infection during pregnancy

Have been exposed to high levels of radiation

What does the quad screen test look for?

The quad screen measures high and low levels of AFP, abnormal levels of hCG and estriol, and high levels of Inhibin-A. The results are combined with the mother's age and ethnicity in order to assess probabilities of potential genetic disorders. High levels of AFP may suggest that the developing baby has a neural tube defect such as spina bifida or anencephaly.

However, the most common reason for elevated AFP levels is inaccurate dating of the pregnancy.

Low levels of AFP and abnormal levels of hCG and estriol may indicate that the developing baby has Trisomy 21 (Down syndrome), Trisomy 18

depends on how good the sonography machine is and how talented and trained the test conductor is.

Double marker test looks for levels of two hormones, b-HCG and PAPP-A and depending upon ultrasound findings, maternal age, previous history, all this information is fed in a software which tells what are the chances of specific abnormalities developing.

These tests are either separate or combination of an ultrasound and blood test. However, these are simply screening tests and will tell you how high your risk is and nothing else. You will come to know whether the baby you are carrying in your womb will have Down's syndrome or not. To know more about such tests and the pregnancy weeks during such tests are done, you must consult your obstetrician.

Advantages of Double Marker Test

- It gives you time to think and consult your doctor for another advanced test if your baby is detected with Down's syndrome.
- You can go through the diagnostic test if double marker tests turn positive.
- You have good detection rates.

If your fetus is reported positive after the screening test, the doctor will suggest a diagnostic test (Chorion villous biopsy / amniocentesis) to assess the fetal chromosomal status and depending upon these results, further course of action is decided.

TRIPLE / QUAD TEST

The **quad screen test** is a maternal blood screening test that looks for four specific substances: AFP, hCG, Estriol, and Inhibin-A.

AFP: *alpha-fetoprotein* is a protein that is produced by the fetus

hCG: *human chorionic gonadotropin* is a hormone produced within the placenta

(Edwards Syndrome) or another type of chromosome abnormality.

What do the quad screen results mean?

It is important to remember that the quad screen is a screening test and not a diagnostic test. This test only notes that a mother is at risk of carrying a baby with a genetic disorder. Many women who experience an abnormal test result go on to deliver healthy babies. Abnormal test results warrant additional testing in order to make a diagnosis.

A more conservative approach involves performing a second quad screen followed by a high definition ultrasound. If the testing still maintains abnormal results, a more invasive procedure such as amniocentesis may be performed.

Any invasive procedure should be discussed thoroughly with your healthcare provider and between you and your partner. Additional counselling and discussions with a counsellor, social worker or minister may prove helpful.

What are the reasons for further testing?

The quad screen is a routine screening that poses no known risks to the mother or baby. The quad screen results may warrant additional testing. The reasons to pursue further testing or not vary from person to person and couple to couple.

Performing further testing allows you to confirm a diagnosis and then provides you with certain opportunities:

- Pursue potential interventions that may exist (i.e. fetal surgery for spina bifida)
- Begin planning for a child with special needs
- Start addressing anticipated lifestyle changes
- Identify support groups and resources
- Make a decision about carrying the child to

