

A 25yr old healthy primi was recommended by the obstetrician to take up routine Triple Screen Test at GeneTech. She along with her husband came to GeneTech to give her blood sample and also to understand more about the test.

During the counseling session "What is triple screen test?" asked the patient. "A triple screen test," we answered "is a blood test that is done to look at the risk for a pregnancy having a fetus with common genetic or birth defects. This is *not* a test that tells us the baby does or does not have these defects. This test tells us if the baby is at high risk or low risk for some genetic defects that mostly are random in occurrence. The triple screen measures the levels of three hormones in the pregnant woman's blood stream. These are alpha-fetoprotein, human chorionic gonadotropin, and unconjugated estriol. They reflect the fetal condition to a certain extent. e.g. AFP is low in many cases of Trisomy 21(Down Syndrome)."

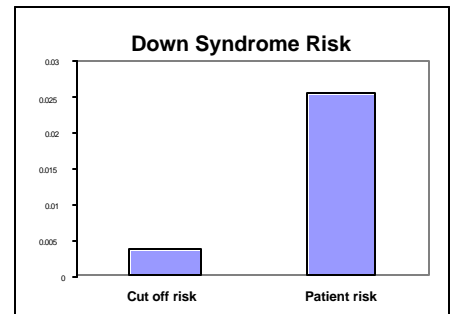
"When we do this test the report will give levels of these three hormones and a calculated risk of you having a baby with trisomy 21, trisomy 18, open neural tube defects. The risks are calculated on the basis of data like maternal age, gestation, weight, family history and diabetic condition. If the test says that the pregnancy is at a high risk, we will offer you the test that does give us a definitive answer; like an amniocentesis with fetal karyotype where fetal chromosomes are studied".

Four days later the triple screen report showed her risk of having a trisomy 21 as 1 in 39. The cut off risk is 1 in 250. She and her husband were counseled about the risk, false positives and false negatives of screening tests, and amniocentesis was recommended. We explained to them that Amniocentesis is the only test that can diagnose the condition accurately.

Age	Weight	Gestation	IDDM	Family history
25 yrs	56 kgs	16 weeks	No	Nil

Marker	Value	units	MoM
AFP	31.8	IU/ml	0.86
uE3	1.34	Ng/ml	0.47
β hcG	45096	mIU/ml	1.9

Condition	Patient Risk	Cut off
Down syndrome	1 in 39	1 in 250
ONTD	1 in 60500	1 in 100
Trisomy 18	1 in 801	1 in 100



Amniocentesis was done immediately after the decision of the family and thirteen days later karyotype of the amniotic fluid revealed trisomy 21 condition in the fetus. In the counseling session following the diagnosis Down Syndrome condition was explained to the patient and her family. As the survival chance and the prognosis of Down syndrome baby would be poor and there is inevitable mental retardation associated the patients opted for termination with their obstetrician.



Two months after termination the patient and her husband came to GeneTech to understand the risk of recurrence. We explained that most of the trisomies are de novo or new events and carry no recurrence risks. For all future pregnancies we strongly recommended them to take up first trimester screening test done at 8-10 weeks of pregnancy and second trimester screening test done at 16th week of pregnancy to screen for common birth defects.

Discussion

This patient is an excellent example of how the triple screen allows us to diagnose Trisomies at a stage early enough to terminate the pregnancy if so desired. While the highest incidence of Trisomy is in women over the age of 34, the largest number of pregnancies with Trisomy occur in women under age 35. This is because the number of women having pregnancies under age 35 is so great. The triple screen offers a non invasive method of determining which of those women under 35 are at increased risk for a Trisomy pregnancy, so that we can offer them amniocentesis. This is valuable information that allows us to start testing these pregnancies and to intervene when indicated. Along with trisomies and open neural tube defects the test also screens for abdominal wall defects, congenital nephrosis, turner syndrome, IUGR and unexplained fetal death. Please call GeneTech's genetic counselor to discuss the case details.