

Awareness and Acceptability in Indian Women of Triple Test Screening for Down's Syndrome

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Citation

A Baxi, M Kaushal. *Awareness and Acceptability in Indian Women of Triple Test Screening for Down's Syndrome*. The Internet Journal of Gynecology and Obstetrics. 2007 Volume 9 Number 2.

Abstract

Aims: This prospective study was to summarize the experience of women regarding the decision making to undergo triple test for Down's syndrome screening and how many of them make use of the triple test and to evaluate its outcome.

Methods and Material: This study was conducted among pregnant women booked in a private antenatal clinic of Indore (M P). All antenatal women underwent prenatal detailed counselling regarding screening for Down's syndrome and were allowed to make voluntary choice.

Results: Of the 745 women included in the study, 106(14.2%) were aware about triple test. Acceptability rate was 16.6% and most of them were those who were either aware of triple test or who had past or family history of congenital or chromosomal disorder. Those who accepted had an attitude of undergoing triple test in future pregnancy.

Conclusions: There is lack of awareness regarding Down's syndrome screening among general population especially regarding various methods and their availability. There is need for a systematic approach aimed at better informing and counselling pregnant women about the implications and limitations of the triple test.

INTRODUCTION

Down syndrome (trisomy 21) is the most commonly recognized genetic cause of mental retardation¹. Many children born with trisomy 21 has a wide range of mental retardation. There are several other effects associated with Down syndrome that include mild to severe developmental delay, heart defects, epilepsy, respiratory problems, susceptibility to infection, celiac disease, Alzheimer's disease etc. At birth the incidence of the syndrome is 1.3 per 1000, with the risk rising with increasing maternal age². Because of the morbidity associated with Down syndrome, screening and diagnostic testing for this condition should be offered as optional components of prenatal care.

Until recently screening for Down's syndrome in India was confined to women over an arbitrary age limit of 35 to 37 and entailed amniocentesis or chorionic villus sampling. In 1988 triple test was introduced as a screening test for Down's syndrome. Three biochemical markers were used together with maternal age as a method of screening, and this has been widely adopted. The triple test is usually

performed at 15 to 18 weeks of gestation. The maternal serum levels of each of these proteins and of steroid hormones vary with the gestational age of the pregnancy. With trisomy 21, second-trimester maternal serum levels of AFP and unconjugated estriol are about 25 percent lower than normal levels and maternal serum hCG is approximately two times higher than the normal hCG level.

The triple test can detect 60 percent of trisomy 21 pregnancies; it has a false-positive rate of 5 percent^{3,4,5}. The likelihood of a foetus having trisomy 21 in a patient with a positive test is about 2 percent. A normal result reduces the likelihood of trisomy 21 but does not exclude it. Detailed counselling is required before offering triple test and it should expose the couple to all information regarding the false positive results of the test and a need to undergo amniocentesis if test is positive.

Prenatal diagnosis is available in first and second trimesters, chorionic villus sampling (CVS) and amniocentesis, respectively, to women categorized at increased risk as a result of prenatal screening, advanced maternal age (35 years

or older) and/or personal/family history of a genetic condition₆. Prenatal diagnosis of trisomy 21 allows parents the choice of continuing or terminating an affected pregnancy.

SUBJECTS AND METHODS

This prospective cohort study was done in a private obstetric clinic from October 2004 to September 2006. In this 2-year duration 745 women of age group 22 to 43 years were registered for Antenatal Care. As study place is an infertility clinic 525(70.5%) women were those who conceived after treatment of infertility and for them pregnancy was extremely precious. A questionnaire was developed and pretested. Language and sequence of the questions were changed to improve the feasibility of the study. Counselling sessions were planned.

All pregnant women along with their spouse were called at around 12 weeks of pregnancy for prenatal counselling of Down's syndrome. As per set protocol Performa was filled that include maternal age, socioeconomic status, education, gravidae, parity, past and family history of congenital & chromosomal disorder. Each couple was asked about there knowledge of Down's Syndrome & Triple Test. Detailed questionnaire was done whether they heard about this test for the first time or if they knew, what was the source of information.

Counselling was done regarding Down's syndrome including social aspects, various tests available for prenatal diagnosis, gestational age at which they can be done. All were made aware of triple test and the correct time. Various medical and ethical issues involved with this were also discussed. A nondirective approach was used when presenting women with various options.

All were allowed to make a voluntary choice and were called after 4 weeks. Those who were interested in triple test, at 15th to 18th week of pregnancy, after taking consent, a sample of blood was drawn from the mother and send to laboratory to measure three basic things: Alfa-fetoprotein (AFP), unconjugated estriol and human chorionic gonadotropin(hcg).

Exact gestational age was calculated by ultrasonography. The level of each serum marker is measured and the three results were adjusted by a computer program to give an estimate of the risk of having a foetus with Down syndrome. It is reported as a multiple of the median (MoM) for women with pregnancies of the same gestational age as that of the

patient's.

The likelihood of trisomy 21 is calculated on the basis of each of the serum marker results and adjusted for mother's age, weight, and ethnicity; if she is diabetic; if she is having twins or other multiples; and the gestational age of the foetus. A composite estimate of the risk of trisomy 21 is reported to the clinician. A standard risk cutoff is used to determine when the test is considered "positive."

RESULTS

Out of 745 women enrolled in the study 472(63.3%) were primigravidae & 273(36.7%) were multigravidae. Mean maternal age was 27.2 years. A total of 648(86.9%) women were less than 35 years whereas 97(13.1%) women were more than 35 years Almost all of them were educated enough to understand the significance & importance of triple test. Almost all belong to middle class who can easily afford this test. Three (0.4%) women had previous history of Down's syndrome and 24(3.2%) mothers had family history of congenital & chromosomal disorder. Of all, 436(58.5%) couple had knowledge regarding Down's syndrome and out of them only 106(14.2%) knew about triple test while rest heard about it for the first time when it was counselled to them.

Mode of information was newspapers & magazines 33 (31%), Internet 42(39.6%) & medical personal in 31(29.4%). The answers to the questionnaire assessing how they look back on their initial experience regarding the exact meaning of Down's Syndrome and the importance of triple test revealed that less than one fourth were actually aware of there risk of a child with Down syndrome. Reporting early acceptability was significantly associated with a higher education level. The same holds for indecisiveness regarding amniocentesis if triple test reports high risk and pregnancy termination should the amniocentesis have detected a fetus with Down's syndrome.

Total pregnant women who accepted triple test were 124(16.6%) while 621(83.4%) refused. Out of 124(16.6%) women who accepted test 92(12.3%) women were those who were aware of triple test and those who had past or family history of congenital or chromosomal disorder. Motivated percentage of women was only 4.3%. Most common cause for refusal was that the couple does not want to know the fate of the child, as the pregnancy is precious to them. Other reasons include non-acceptance for medical termination of pregnancy and further invasive test if triple test reports high risk.

Acceptance was more among aware population and those with positive family history. Acceptance was low among those women who conceived after treatment of infertility probably because of its preciousness. Overall even after intensive counselling acceptance rate was not high low as most of them had a 'gut feeling' about their pregnancy outcome that everything was fine rather than they expected to learn about whether or not the foetus had Down's syndrome and want to try there luck.

Out of 124 women who accepted triple test, high risk ratio was positive in 12(9.6%) women. Of the 12 women with high risk ratio, 8 pregnant women were > 30 years of age and 4 were of age group 27-30 years and 2 had family history of Down's syndrome. As expected, a large majority of the women reported a high level of distress or worry after the communication of the positive triple test result. All of them were counselled for amniocentesis. Withstanding the high level of distress caused by a positive triple test result all the women underwent amniocentesis for further confirmation. Amniocentesis was done and karyotype was normal in all of them. They were followed till delivery and thereafter and the outcome was normal.

Overall the findings show that retrospectively most women had the feeling that the decision to have triple test was their own decision rather than a professional's counseling. Those who accepted triple test were absolutely convinced for triple test in subsequent pregnancies.

DISCUSSION

In the present study we observed that there is still lack of awareness regarding triple test among the Indian population. Although after counseling the acceptability rate was quiet reassuring. Verloes et al reported in their study that there was major decrease in the incidence of trisomy 21 at birth in south Belgium after acceptability of triple test ⁷. Informed choice for prenatal screening has long been considered an essential aspect of service provision, and has been researched extensively. Jaques et al concluded in their study that health professionals should ensure that all women are provided with suitable information about prenatal screening that is tailored to their level of education and individual needs, and should emphasize that screening is optional ⁸.

A questionnaire based study done in Australia concluded that pregnant women need clearer information about prenatal testing, including the conditions that might be detected ⁹. The majority of women preferred to have both the risk of Down syndrome at the time of screening because they want

to have as much information available as possible so that an informed decision regarding further investigations could be made ¹⁰.

The risk of trisomy 21 is directly related to maternal age ¹¹. The chances of having a baby with Down's syndrome increases with a woman's age although paradoxically more babies with Down's syndrome are born to women under 30 ¹. This is probably because most pregnancies are in this age range and older women are more likely to have diagnostic tests for the condition. However, it is usual for pregnant women, whatever their age, to be offered a test to assess their chance of having a baby with Down's syndrome.

Patients who receive news of high-risk ratio after triple test often experience considerable anxiety. These patients can be reassured by the knowledge that the likelihood of Down syndrome is small, even after a positive triple test ¹². Ultrasound and amniocentesis should be offered ⁵. The risk of fetal loss from amniocentesis should be discussed.

Also a false negative result on prenatal screening seems to have a small adverse effect on parental adjustment evident two to six years after the birth of an affected child ¹³.

Study says that in pregnancy over 35 years a choice should be offered between non-invasive and invasive procedures after being thoroughly informed ¹⁴. There are ethical aspects of prenatal screening for Down's syndrome. Although voluntary participation and nondirective genetic counselling can exclude eugenic purposes. Abortion of affected foetuses isn't among the objectives of prenatal genetic screening but patient's autonomy is supported in decisions concerning the future of the pregnancy. An open debate about the issues of prenatal screening for Down's syndrome could promote the formation of a consensus between professionals and the public ¹⁵.

The findings in the present study emphasis that steps should be taken to make triple test more easily available. Also there is need to increase the awareness regarding Down's syndrome and various screening & diagnostic tests available so that morbidity due to Down's syndrome can be prevented. Further there is need for the development and trailing better methods of giving women and their partner appropriate information in a comprehensible manner. Also cost-effectiveness of the test is to be discussed to make it as an essential component of antenatal testing.

Medical personals should take the responsibility to maintain

equal access to prenatal testing, as to any other health service along with the maintenance of medical standards and evaluation of program performance¹⁶.

The authors feel that medical professionals can play a larger role in informing pregnant women about triple test. Many pregnant women in our study reported that they relied on medical professionals for information regarding antenatal tests. This suggests that strategies on promotion of triple test to screen Down's syndrome should focus on medical professionals too. One of the limitations of our study was that was done upon women attending private antenatal clinic. Hence our study may not be truly representative of pregnant Indian women. In fact this study was planned just to explore educated Indian women who are aware and will accept triple test after counseling. There is need of further studies on these aspects.

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