

pregnant again, chorionic villi sampling (CVS) examination was done to rule out Tay-Sachs disease in the fetus. The report of B-hexoseaminidase (total) 2000 nmol/hour/mL of protein against 1908 ± 900 nmol/hour/mL of protein and B-hexoseaminidase-A 33.33% against 28 to 63%. The results showed that the fetus was not affected with Tay-Sachs disease; as a result pregnancy was allowed to continue.

She delivered a male child normally. This infant was brought for routine visit at 4 months of life in pediatric outdoor department. According to the parents the child did not have any complaints. The child had unremarkable antenatal and postnatal history.

However on examination, the child was found to be underweight but the development of the child was normal. General examination showed that the child was significantly pale, but it was unassociated with icterus, edema, skin hemorrhages, and rashes. Abdominal examination revealed a palpable spleen 7 centimeter with liver rounded margin with span of 8.5 centimeters. Child's hemogram showed Hb: 6.4 g/dL, Reticulocyte count : 12%, MCV : 66 fl (normal = 82.2 to 97.4 fl), RDW : 35% (normal = 11.6 to 13.7%), HbF : 37%), thus, the reports were suggestive of

β -thalassemia major and parents were investigated for carrier. The parents were stamped as β -thalassemia trait and the diagnosis of β -thalassemia major in the child was confirmed.

For this surviving male child, parents were advised bone marrow transplantation as a curative treatment. The mother is advised to have pregnancy for getting healthy sibling for getting bone marrow.

This report highlights importance of screening of all prospective parents for beta thalassemia trait. Also, it should be noted that normal report of prenatal diagnostic test for one disorder does not rule out the possibility of other genetic disorders.

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Chorionic Villous Sampling for Prenatal Diagnosis in Beta Thalassemia

Prenatal diagnosis of thalassemia by chorionic villous sampling (CVS) is now available at few centers in India and we here present our experience in this regard.

Of the 95 thalassemic children registered

for treatment at our center, 26 families opted for prenatal diagnosis in the last 7 years. We found very good acceptability for the prenatal counseling at our center. Of the remaining families, 51 couples didn't want further children and 17 couples have still to plan their next baby. One lady conceived while she was away in the village and could not avail this facility. This child was found to be thalassemic. Among these 26 couples, a total of 37 CVS were done between 10-12 weeks of gestation.

Seventeen mothers went through the procedure only once. Of these, only 3 fetuses were found affected and these were terminated. Of the remaining, 5 mothers underwent a second CVS after the first resulted in an affected fetus. The second baby was found to be unaffected in all 5 cases. Two mothers chose to have a second CVS despite a previous successful attempt (having a normal baby) and the second baby was also normal in all these cases. Two mothers went through this procedure 3 times. One of them had unaffected fetuses in the 1st and 3rd pregnancies. The second pregnancy was terminated as the fetus was affected. The second mother was less fortunate with only one live issue despite 3 CVS samplings. The 1st pregnancy was terminated for an affected fetus, the 2nd ended in miscarriage 5 days post CVS sampling and only the 3rd baby was normal.

Thus a total of 10 pregnancies (out of 37 subjected to CVS), had to be terminated due to affected fetuses. Of the remaining 27 unaffected fetuses, 26 were safely carried to term. One fetus aborted 5 days after the sampling. All the pregnancies carried to term went on to have unaffected babies. On a maximum 7 years follow up, 15 of these are thalassemia carriers and the rest 12 are normal children. Apart from one case of abortion, no other adverse effects of CVS were observed.

Since its inception in the 1980s, CVS has gained popularity as a means of rapid prenatal diagnosis in early pregnancy. Its advantages over a second trimester procedure like amniocentesis include reduced emotional and physical stress in couples at risk, a less obvious pregnancy and therefore more privacy. Termination of pregnancy, if indicated, can be carried out at a safer time in such cases. Old, *et al.*, were the first to report successful diagnosis of beta thalassemia using CVS(1). Since then few studies in the West(2,3) and

also from India(4,5) have demonstrated the high efficiency, safety and acceptability of this procedure prompting experts to suggest that first trimester CVS should be the gold standard for prenatal diagnosis. This message can be propagated through thalassemia clinics and thalassemia welfare societies who have a major role in the creation of awareness regarding the problem of thalassemia.

Acknowledgement

The authors wish to acknowledge the services of Dr. M. Kabra, Associate Professor, Pediatrics, All India Institute of Medical Sciences, New Delhi for carrying out mutation analysis.

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