

Medicine: A Clinical and Applied Orientation. 1999. Vikas Publishing House Pvt. Ltd., New Delhi.

2. National Family Health Survey 1998-1999, India 2001; International Institute for population Sciences, Mumbai.

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## Carrier Parents of Tay Sachs Disease and $\beta$ -Thalassemia

Tay-Sachs disease and  $\beta$ -thalassaemia are transmitted in autosomal recessive manner. These diseases differ remarkably, in their etiology and clinical presentation. We report two siblings affected by these two different disorders. Both the parents were diagnosed to be carriers for Tay-Sach's disease as well as  $\beta$ -thalassemia.

The couple in the discussion brought their eighteen months old male child with complaints of regression of milestones for six months, attack of generalized tonic clonic seizure 2 months back, and respiratory distress of five days duration. He was born of non-consanguineous marriage and an uncomplicated full term vaginal delivery. No major antenatal or postnatal problems were noted.

This was their 2nd child, the 1st being a female child who died at the age of three and a half years. That child's illness had also started with similar complaints of regression of milestones and convulsions. That child had died undiagnosed.

On examination, growth was within normal limits but there was global mental retardation, developmental age corresponding to 6 months. General examination was normal. There were no dysmorphic features, neurocutaneous syndromes and also no apparent gross congenital malformation. Respiratory system examination revealed

bilateral wheezing. Abdominal examination showed no organomegaly. In central nervous system examination, the child was conscious, with no cranial nerve palsy. The only positive findings were hypertonia and hyperreflexia of all four limbs with intermittent scissoring of both the lower limbs with exaggerated startle response. The fundus examination showed bilateral cherry red spots in the macula.

Hematological and radiological studies were normal. EEG suggestive of primary generalized epilepsy was reported. Tay-Sachs disease was suspected and blood lymphocyte enzymatic studies for hexoseaminidase levels of both parents were carried out. This showed B-hexoseaminidase (Total) 300 nmoL/hour/mL of protein in father, and 264 nmol/hour/mL of protein in mother, against the normal levels of  $801 \pm 90$  nmol/hour/mL of protein. The B-hexoseaminidase-A levels were 44% in father, 40% in mother, against the normal values of 55 to 72%.

Thus, both the parents were diagnosed to be carrier of Tay-Sachs disease. Also, with the combination of the clinical and the investigative data, diagnosis of Tay-Sachs disease was confirmed in the child. The child was treated symptomatically and anti-epileptics were given for the control of convulsions. However, the child died at the age of 30 months.

The parents were explained about the nature of the disease and genetic counseling was also done, also prenatal diagnosis during further pregnancies was also advised emphatically. So, when the mother was

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  $1908 \pm 900$  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

$\beta$ -thalassemia major and parents were investigated for carrier. The parents were stamped as  $\beta$ -thalassemia trait and the diagnosis of  $\beta$ -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.