CASE REPORT

A Case of Trisomy 13, Patau Syndrome

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Abstract

Trisomy 13 or Patau syndrome is a chromosomal abberation due to non-disjunction of chromosome 13.The majority of affected children have multiple malformation and succumb during the first few months. An extra chromosomal 13 causes central nervous system defect and mental retardation together with cleft palate and lip, polydactyly and abnormalities of heart and genitalia. Our case was a male pre term baby of 34 weeks gestation with weighing 2.9 kg with multiple congenital anomalies. The baby succumbed to death after 6 hours after delivery.

Introduction

This syndrome has an incidence of 1 in 12000 and 60% are female. The affected children show intrauterine growth retardation. There may be marked microcephaly. Structural malformations of the brain are common and this may alter facial development. Instead of two cerebral hemispheres with lateral ventricle, a single forebrain with single ventricles may form. This malformation sequence is known as Holoprosencephaly. More usually there is marked hypotelorism with small nose and cleft lip and palate. Ocular abnormalities such as micro opthalmos are common, often reflecting the underlying cerebral abnormality.

In the hands postaxial polydactyly is frequent. Flexion contractures and "Rocker Bottom" feet may also be present. Scalp defects are characteristic that may be helpful diagnostically as rarely appear in other abnormalities. Internally in addition to cerebral malformation, renal and cardiac abnormalities are also present.

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Case history

Baby of Mrs.Mahmuda 25 years old lady, from pallabi, Mirpur, Dhaka delivered on 16-06-2004 at 8:55 p.m. by Caesarean Section. The baby was pre term (34 score) male with birth weight 2.9 kg. The baby had severe birth asphyxia with APGAR of 1 at 3 minutes and 5 at 6 minutes. The baby had multiple congenital anomalies as well. The mother was Rh (-ve) with no history of Anti Rh-D prophylaxis. She was third gravida with 1 living child and history of 1 MR. There was no history of consanguinity. Caesarean Section was done for Pre-Eclampsic Toxaemia with ventriculomegaly which was diagnosed prenataly at 34 weeks by ultrasonography.

Congenital anomalies were cleft lips and palate, polydactyly, Rocker bottom feet, micropenis with hypospadias with undescended testes mid line scalp defect was also seen and confirmed by X-ray.

The baby was flabby with severe respiratory distress. He was managed with suction, saline, oxygen inhalation and antibiotics.

But the baby succumbed to death at 5:45 am on 17-06-2004 i.e. 6 hours after birth.

Discussion

Patau's syndrome (Trisomy 13) is a rare congenital abnormalility and it is incompatible with life. The few cases reported with longer survival have all shown severe retardation. There may be some maternal age factors, but probably not as other trisomeies. It is advisable to counsel mothers regarding the chance of recurrence and offer for amniocentesis.

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