Patau syndrome - a case report

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Abstract

Patau syndrome is a rare and one of the most severe forms of autosomal trisomies. Having a karyotype of trisomy 13, it is associated with high rate of pregnancy loss and poor infant survival rate. The incidence is 1 in 20,000 live births. The common features associated with Patau syndrome are mental retardation, congenital heart defects, cleft lip and palate, eye defects, polydactyly, scalp defects and holoprosencephaly. We report a newborn with most of the clinical features consistent with Patau syndrome. Fluorescence in situ hybridization (FISH) technique was used to determine the karyotype and confirm the diagnosis.

Key words: Trisomy 13, polydactyly, FISH

Introduction

Trisomy 13 is one of the rarest of all the autosomal trisomies and frequently results in spontaneous abortion and death during the first few days or weeks of life. The condition is less commonly encountered as the anomalies associated are more severe and not compatible with life. Therefore, majority of the cases suspected at birth needs prompt medical attention invariably and by the time cytogenetic analysis is done for chromosomal anomalies, the infants die. The disorder occurs more frequently with advanced maternal age, the additional chromosome being of maternal origin. Majority of the cases occur due to non-disjunction, 5-10% due to Robertsonian / unbalanced translocation and rest due to mosaicism. Although the incidence has been reported as 1 in 20,000 live births, the rate is decreasing as a result of prenatal screening for congenital birth defects. More than 90% of the infants die in the first month after birth and approximately 1% live beyond one year. We are hereby, reporting a case of Patau syndrome the confirmation of which was done by using FISH technique. (FISH - Fluorescence in situ hybridization).

Case Report

A preterm (34 weeks), male with a birth weight of 1.95 Kg, delivered by caesarean section for severe oligohydromnios was admitted in the ICU, Department of Pediatrics, NEIGRIHMS, Shillong. The newborn presented with multiple congenital anomalies and developed respiratory distress since birth. The baby was born of a non-consanguineous marriage, the mother being a 29 year’s old 2nd gravida. The first baby was normal and was delivered five years back by normal vaginal delivery at term. During antenatal period, the pregnancy was monitored regularly and was uneventful with no history of any maternal illness during the antenatal period. There was no history of any drug intake during pregnancy except for iron-folic acid supplementation, two doses of TT injections and one tablet of 500mg paracetamol taken for headache in second trimester with consultation. There was no history of any neonatal death in the family. The newborn was found to have multiple congenital anomalies. Bilateral cleft involving the lip and the jaw (Fig.1D), postaxial bilateral polydactyly in the upper limbs (Fig.1B) and right lower limb (Fig.1A), small head size (<1 Standard Deviation), malformed and low set ears (Fig.1E), upward slanting of eyes and bilateral undescended testes (Fig.1C) were observed. Cardiac anomalies included an atrial septal defect (ASD) of 10mm size and nonrestrictive ventricular septal defect (VSD), detected on echocardiography. Cutis Aplasia (skin defect over back of scalp) was also observed (Fig.1F). The Fluorescence in situ hybridization (FISH) test performed on the nuclei using the Vysis Aneu Vysion TMLEC DNA probe kit, showed trisomy 13 (Patau Syndrome) with normal chromosome 18, 21, X and Y. At the time of admission the baby had mild respiratory distress and was intubated. The baby was evaluated for the cause of respiratory distress keeping the differential diagnoses of mild Hyaline Membrane Disease, congenital heart disease, congenital pneumonia. Infantogram was normal; Ultrasonography - cranium, abdomen and lumbosacral...
region was normal. Ophthalmological examination was normal. MRI- brain was planned but couldn't be done.

On day 6th, the baby developed apnoea and responded to bag and mask ventilation. Again on day 13 of life, the baby had 2 episodes of apnoea and on day 14, the baby was intubated and mechanically ventilated for repeated episodes of apnoea. Repeat sepsis screen and blood culture were negative. Gradually the baby deteriorated on mechanical ventilation and developed feed intolerance, hypoglycaemia, shock and pulmonary haemorrhage and expired on 19th day of life.

Discussion

Trisomy 13 or Patau syndrome occurs most commonly due to disjunctural errors in meiotic or mitotic cell division. The non-disjunction of autosomal chromosomes is one of the major causes of pregnancy loss in humans, being responsible for approximately 50% of spontaneous abortions before 15 weeks of gestation and approximately 50% of these are due to trisomies\(^5\). Only about 2% to 3% of fetuses with trisomy 13 survive up to birth\(^4\). Newborns with trisomy 13 have a median survival of 7 days and only 5% survive beyond 6 months of age\(^4\). The clinical features principally include cleft lip and palate (in 60-80% of cases), cardiac malformations (in 80% of cases), flexed fingers with polydactyly, ocular hypotelorism, bulbous nose, low-set malformed ears, small abnormal skull, cerebral malformation, especially holoprosencephaly, microphthalmia, scalp defects, hypoplastic or absent ribs, visceral and genital anomalies\(^5\). Misanovi et al\(^4\) reported a case of Patau syndrome having multiple congenital anomalies including microcephalia, dolichocephalia, microphthalmia, cheilognatopalatoschisis, polydactyly, and ultrasound changes of the brain, heart and genitourinary
system. The case was confirmed cytogenetically. Different cytogenetic techniques, including FISH can be used to diagnose Patau syndrome. Zhou et al reported a case of Patau syndrome with paternal origin of an extra chromosome 13 due to nondisjunction during the first meiotic division of the father, which was confirmed by FISH. The life expectancy of the infants with trisomy 13 is very low and the evolution is correlated with several factors, including the severity of cardiac and cerebral malformations and of other multiple congenital anomalies. Apnoea, seizures and feeding difficulty arising from the major congenital anomalies are the factors responsible for short life expectancy. Children surviving longer must be suspected for mosaicism. Coco et al reported a case of a two months old child with Patau syndrome with 46, XY, 14-1 (13q14q) + karyotype. Long term survival beyond one year of age is rare, but there are reports of long survival till adulthood. It depends on the cytogenetic finding and severity of the structural anomalies of the patient. The clinical triad of polydactyly, microphthalmia and cleft palate indicative of Patau syndrome as described in some standard texts may not be evident in every case; cytogenetic analysis must be performed in suspected cases to confirm the diagnosis.

Trisomy 13 is a rare genetic disorder and there is no specific treatment or cure for it. Though references citing a better prognosis with use of intensive care including resuscitation and surgical procedures are available, prevention using routine maternal ultrasound and prenatal Chorionic villi sampling (CVS) or amniocentesis in high risk cases can circumvent the existing disease burden in long term. Recurrence risk further should be addressed in confirmed cases by detailed cytogenetic analysis and genetic counseling.

References