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# Trisomy 13: Patau's Syndrome (A Case Report from Jammu Region of Jammu & Kashmir)

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**Abstract:**

Trisomy 13 is very rare in live-born children. Only a small number of these children survive the first year and very few cases are reported to live longer. Survival time depends partly on the cytogenetic findings whether full trisomy 13 or trisomy 13 mosaicism and partly on the existence of serious somatic malformations. Patau's syndrome is a very rare congenital anomaly. Here is the report of a male child clinically diagnosed as Patau Syndrome andchromosome study showed free Trisomy 13 to be the cause for this rarecongenital anomaly. The child died within a month after birth.

**Keywords:** Patau syndrome, Trisomy 13 and Congenital anomaly.

## INTRODUCTION

Trisomy 13 is a very rare chromosomal abnormality in the live bornchildren causing Patau syndrome. The extra copy of chromosome 13, eitherfree standing as in the 47, +13 genotype or in a Robertsonian translocationor other rearrangement is responsible for the fatal congenital anomaly(Hook, 1980). Full trisomy 13 is caused by nondisjunction of

chromosomes during meiosis (the mosaic form is caused by non-disjunction during mitosis).The prevalence of Patau syndrome was estimated tobe from 1:12,000 (Hook, 1980; Baty *et.al*., 1994) to 1:29,000(Goldstein and Nielson 1988). The frequency of Trisomy 13 has beenreported to be 100 times higher in spontaneous abortion than in live born(Hook, 1980). Magenis *et.al.*, 1968 showed that 28% of the fewsurviving newborns die in the first week, 44% within first month and 86%within their first year. The median survival of children with Trisomy 13 wasgiven as 89.2 days by Taylor in 1968. Zoll *et.al* (1993)reported further that missingcerebral and cardiovascular malformations probably allowed the longsurvival. At present, Patau syndrome is generally recognized as a specificautosomal trisomy. The condition is less common than Trisomy 18syndrome.

## CASE HISTORY

A 25 days old male infant (Fig.1) was referred for chromosome studybecause of characteristic features suggestive of Patau syndrome. He was 5$^{th}$child of a young, healthy non-consanguineous couple. The mother was 24years of age and father was 27 years old at the time of the child's birth. Brothersand sisters of the Proband were phenotypically normal. The referred child wasborn after 3 years of 4th issue. The Proband was found to have Dysmorphicface, Micropthalmia, Malformed ears, Campactodactyly, Unusual features, overrigid digits, Fingers longer than palm, physically very weak was anemic andSimian lines. However, patient lacked three common features of the Patausyndrome namelymicrocephaly, cleft lip-palate and significant congenital heartdisease. The child was born by a normal vaginal delivery and was referred tous on the 25th day of life for chromosome study.

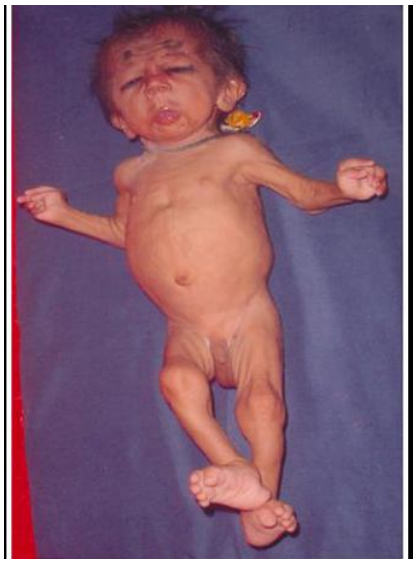

**Fig. 1: Phenotype of Proband**

## CYTOGENETICS

Chromosome study was carried out in the Proband from peripheral bloodlymphocyte cultures. Well spread GTG banded metaphase plates (Fig. 2) were selectedfor their karyo-typing following ISN 1995. Chromosomal analysis of theproband revealed 47, XY$^{+13}$karyotype (Fig. 3) with no evidence of mosaicism.

Thus, the referred child was having free Trisomy 13 and chromosomestudy confirmed the clinical diagnosis.

**Fig. 2: Metaphase Plate (2n=47) of Patau syndrome showing Trisomy 13**

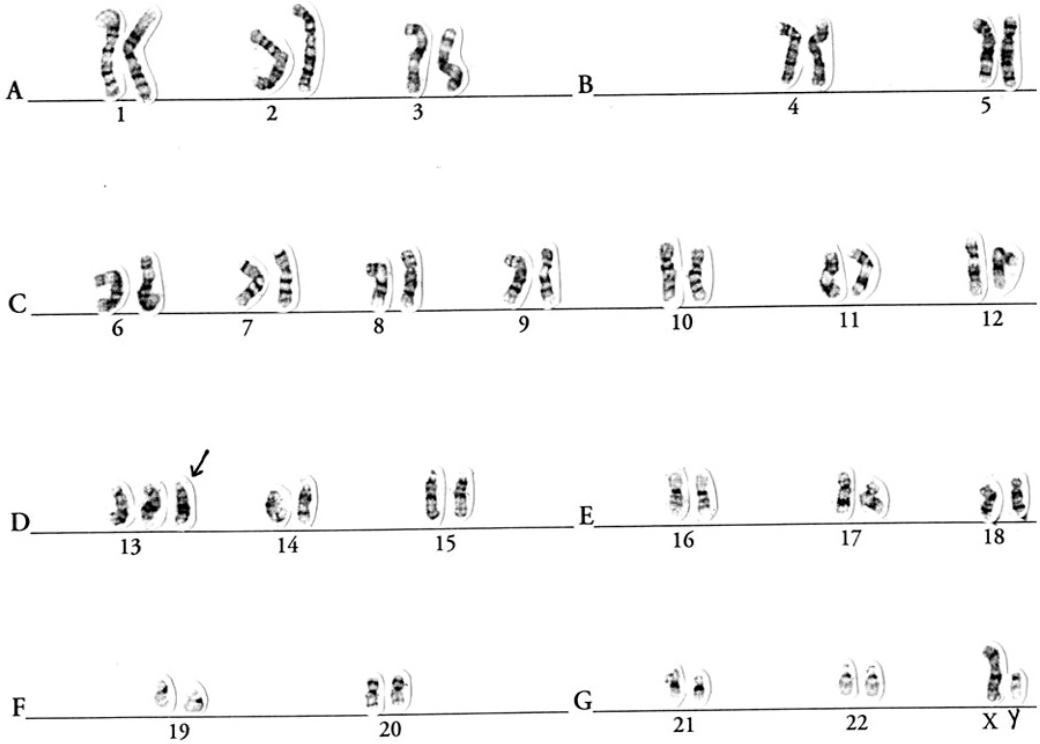


**Fig. 3: Karyotype of Patau syndrome (47, XY$^{+13}$)**

## DISCUSSION

The Proband exhibiting the features of Patau's syndrome had the karyotype47, $XY^{+13}$. Interestingly this case of Trisomy 13 is the only case of its kind inmore than 200 cases of suspected genetic disorders referred to our centre forchromosome study during the past three years thus suggesting the rarity ofPatau syndrome. Our findings on the birth rate of Patau syndrome, thereforesupports the earlier reports (Hook, 1980; Goldstein & Nielson 1988; Baty *et.al.,* 1994). During the present study, it was found again that the incidence rate of Patau syndrome is very low.

Some infants born with Patau syndrome have severe and incurable birthdefects. Best et.al 2004 reported recently that approximately 45% of trisomy 13babies die within their first month of life; up to 70% in the first six months;and over 70% by one year of age. Survival to adulthood is extremely rare. Aijaz*et.al*., (2007) reported a 51 years old Caucasian women with Patausyndrome. Her chromosomal analysis showed partial translocation ofchromosome 13 (unbalanced rearrangement between chromosome 13 andchromosome 14).

Trisomy 13 the main cause of Patau syndrome is due to non-disjunctionof chromosome 13 (Best *et.al.,* 2004). Hara and Sasaki (1975) reportedthat in the live-born 13 trisomic, the non-disjunction at the maternal meiosisand at the paternal meiosis occurred in the ration of 5:1.The clinical features of complete trisomy and partial trisomy for the proximaland distal long arm of chromosome 13 have been defined (Niebuhr, 1977). The patient reported here had no congenital heart lesions commonlyfound in Patau syndrome. He had no evidence of congestive heartfailure.Inspite of the absence of cleft lip and palate, and with no evidence ofconsequent aspiration pneumonia, he died suddenly at the age of 1 month.

## CONCLUSION

In a very few reported cases of Patau syndrome involving Trisomy 13as the main cause, the survival period has been found to be very less.

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