

## Translocation (9;10) Pat, in a Male Child Leading to Monosomy 9p13→9pter and Trisomy 10q24→10qter

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**KEY WORDS** Familial t(9;10); multiple congenital anomalies; monosomy 9p13→9pter, trisomy 10q24→10qter.

**ABSTRACT** A rare case of paternal/familial translocation (9;10) leading to monosomy 9p13→9pter and trisomy 10q24→10qter in a 3 month old male child born with multiple congenital anomalies is reported. Family studies revealed the presence of balanced translocation (9;10) in father, paternal grandmother, three paternal aunts and a paternal cousin.

### INTRODUCTION

Translocation (9;10) as well as 9p monosomy and 10q trisomy syndromes have been reported. (Fraise et al. 1977; Hoo 1986; Huret et al. 1988; Jenkins et al. 1976; Junien et al. 1982; Kroisel and Rosenkranz 1990; Rutten et al. 1978; Young et al. 1982). This report appears to be unique case of rcp t (9; 10) leading to partial monosomy of 9p and trisomy of 10q in a male child.

### CASE REPORT

Proband, male, 3 months old, 4<sup>th</sup> birth order was born to non consanguineous parents. At the time of conception father was 35 years and mother 29 years. Mother had 2 first trimester abortions and a normal female child. Because of threatened abortion during the pregnancy of proband mother was on hormonal treatment from 2<sup>nd</sup> to 9<sup>th</sup> month. Proband was born full term and proband's mother had normal delivery at a hospital.

Proband was asphyxiated at birth and had to be resuscitated. He was 56 cms in length and

weighed 2.5 kgs. He had hypotonia, small flat dysmorphic face, microcephaly, thin sparse scalp hair, large forehead, arched eye brows, small nose, flat nasal bridge, antverted nostrils, long philtrum, small bow shaped mouth, thin lips, high arched palate, low set malformed ears, widely spaced nipples, long fingers and toes, increased distance between 1<sup>st</sup> and 2<sup>nd</sup> toes, flat feet and deep plantar creas ( Fig. 1 )

Proband died at 8 months following a respiratory infection.

Family history revealed that paternal grandmother had 12 pregnancies of which two were abortions. Paternal aunt, 3<sup>rd</sup> birth order, had 3 children the first two had Down Syndrome features ( not confirmed by chromosomal analysis ) and the third resembled the proband. Another paternal aunt, 6<sup>th</sup> birth order had one first trimester abortion and a male child. Two paternal uncles of 4<sup>th</sup> and 5<sup>th</sup> birth order were considered not normal? mental retardation. Rest of the family history was normal.

### Cytogenetic Analysis

Chromosomal analysis of the lymphocytes of the proband revealed ( Fig. 1) the karyotype to be 46,XY,-9,+der(9),t(9;10)(p13;q24); leading to monosomy and trisomy of certain portions of the short and long arms of the 9 and 10 i.e. monosomy for 9p13--9pter and trisomy 10q24--10qter.

Mother and the elder sib' Karyotypes were normal (46,XX). Father had balanced rcp t(9;10) (9qter→9p 13:: 10q24→10qter;10pter→10q24:: 9p13→9pter).

Paternal grandmother and 9 of the 10 living sibilings of the father were Karyotyped. Balanced t(9;10) was observed in the grand mother and three paternal aunts. One of the proband's paternal cousin, male child, is also a

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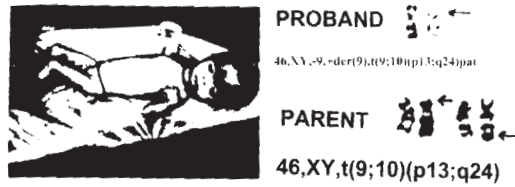


Fig. 1

balanced (9;10) translocation heterozygote.

### DISCUSSION

Knowledge of the critical regions in the chromosomes is always useful in correlating the genotype and the phenotype. Karyotyping determines the critical regions. Moreover, deletion or monosomy of a chromosome is considered to be deleterious than the duplication.

In this report a rare case of paternal/familial rcp t(9;10) leading to a combination of monosomy 9p13→9pter and trisomy 10q24→10qter, in the proband, because of which, the abnormal phenotype and the mortality could have occurred. Mental retardation, ambiguous genitalia along with deletion of part of 9q has been reported (Jenkins et al 1976) in a male (46,XY, t(9;10) (9pter→9q12.5::10qter→10pter→10q25::9q21--9qter). Present case had normal genitalia.

Mental retardation with features of 10q trisomy syndrome in two sibs have been observed (Fraise et al. 1977). It was a case of paternal balanced t(9;10) (q34;q24) leading to trisomy 10q24→10qter. There was no evidence of 9q monosomy.

Patient with multiple congenital anomalies with monosomy 9p24 and trisomy 10q26, resulting from a mat t(9;10) (p24;q26) has been mentioned (Junien et al. 1982).

In all the above cases and in the cases reported in literature, the breakpoints as well as the chromosomal segments involved in the t(9;10) were different from the present case. On

comparison, it was seen that the proband's features were a combination of 9p monosomy and 10q trisomy syndrome (Rutten et al. 1978; Hoo 1986; Huret et al. 1988).

Paternal grandmother, inspite of the carrier status has had 12 pregnancies. In segregation (Kroisel and Rosenkranz 1980), proband's father may have had adjacent II type of meiotic segregation, leading to the deficiency in 9p and duplication in 10q.

To conclude, during genetic counselling, risk of recurrence especially pertaining to the meiotic segregation products of the chromosomes involved in rcp t(9;10) has been emphasized. Also a regular follow up of the family members has been advised.

### REFERENCES

- Fraise J, Lauras B, La Selve A, Freycon F 1977. Deux nouveaux cas de trisomie 10q24--10qter chez deux soeurs par translocation paternelle t(9;10) (q34;q24). *Ann Genet*, **20**,2: 128-131.
- Hoo JJ 1986. Karyotype phenotype analysis: 9p deletion versus 10q duplication. *Ann Genet*, **29**,4: 266-268.
- Huret JL, Leonard C, Forestier B, Rethore MO, Lejeune J 1988. Eleven new cases of del (9p) and features from 80 cases. *JMG*, **25**: 741-749.
- Jenkins EC, Chaganthi RSK, Wilbur L, German J 1976. Ambiguous genitalia and mental retardation associated with a translocation 46, XY, t(9;10) and deletion in 9q. *Birth Defects Original Articles Series*, **XII** (5): 169-173.
- Junein C, Despoisses S, Turleau C, Grouchy J de, Bucher T, Fundele R 1982. Assignment of phosphoglycerate mutase (PGAMA) to human chromosome 10. Regional mapping of GOT1 and PGAMA to subbands 10q26.1 (or q25.3). *Ann Genet*, **25**,1: 25-27.
- Kroisel PM, Rosenkranz W 1990. High resolution banding of an unusual reciprocal translocation in recurrent abortions. *Clin Genet*, **37**: 230-234.
- Rutten FJ, Hustinx TWZ, Dunk - Tillemans AAW, Scheres JMJC, Tjon YST 1978. A case of partial 9p monosomy with some unusual clinical features. *Ann Genet*, **21**,1: 51-55.
- Young RS, Reed T, Hodes ME, Palmer CG 1982. The dermatoglyphic and clinical features of the 9p trisomy and partial 9p monosomy syndromes. *Hum Genet*, **62**: 31-39.