

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

I.M. Thomas¹, M. Neelamma, K.S. Reddy², B.G. Gopalkrishna³ and Sayee Rajangam¹

1. Division of Human Genetics, Department of Anatom, St. John's Medical College, Bangalore 560 034, Karnataka, India
2. Quest's Center for Diagnostic Innovation, 33608, Ortega Highway, San Juan, Capistrano, California, 92960, USA
3. Department of Pediatrics St. Martha's Hospital Bangalore 560 009, Karnataka, India

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, 4th 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 2nd to 9th 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 1st and 2nd 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, 3rd 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, 6th birth order had one first trimester abortion and a male child. Two paternal uncles of 4th and 5th 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→9p13::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 grandmother and three paternal aunts. One of the proband's paternal cousin, male child, is also a

Address for Correspondence: Dr. Mrs. Sayee Rajangam, Professor & Head Division of Human Genetics, Department of Anatomy St. John's Medical College, Bangalore 560 034, Karnataka, India, Fax: 91 080 5520777 Phone: 080 2065061 Email: sjmcanat@vsnl.com

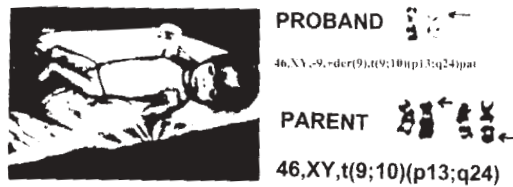


Fig. 1

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.

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