Balanced Translocation (10;13) in a Father, Ascertained Through the Study of Meiosis in Semen, and Partial Trisomy 10q in his Son

Characterization of the Region Responsible for the Partial Trisomy 10q Syndrome

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Summary. We describe a reciprocal translocation (10;13) in a man, ascertained through the study of meiosis in semen, and a partial trisomy 10q in his abnormal son. The phenotypic anomalies of the partial 10q trisomy syndrome are probably due to the presence in triplicate of the region q25→qter of chromosome 10.

Introduction

Trisomy for the distal region of the long arm of chromosome 10 is unusual. However, the phenotypic characteristics of the syndrome have been delineated by Yunis and Sanchez (1974).

In this paper, we describe a balanced reciprocal translocation (10;13) in a man, ascertained through the study of meiosis in semen (Sperling and Kaden, 1971), and a partial trisomy 10q in his son, and characterize the region responsible for the syndrome as 10 (q25→qter).

Case History

A 34-year-old Caucasian male who was the father of a severely retarded child was seen in an infertility clinic, and the andrologist requested a study of meiosis in his semen. The meiotic data (Fig. 1a,b) indicated the possible existence of a quadrivalent. As a result, the andrologist karyotyped the patient and his abnormal son. The karyotype of the father showed a balanced reciprocal translocation (10;13)(q25;p12) (Fig. 2). Q-banding studies indicated that the extra material in the short arm of the abnormal chromosome 13 did not correspond to the satellites (Fig. 2). The karyotype of the child was 46,XY,13p+ with a partial trisomy for the region (q25→qter) of chromosome 10 (Fig. 2).

Clinical Findings

The child with a partial trisomy for the long arm of chromosome 10 was a 7-year-old white boy (Fig. 3) with severe growth and mental retardation, hypotonicity, and nervous anomalies. He had a flat and round face, a spacious forehead, arched and wide-set eyebrows, an anti-mongoloid slant of the eyelids, a small nose with a depressed nasal bridge, a bow-shaped mouth with a prominent upper lip, an arched palate, low-set ears, a short neck, cryptorchidism, abnormal feet with hammer toes, and a bilateral simian crease in the palms. The clinical data are summarized in Table 1.

Discussion

With the exception of a single case in which the partial trisomy for the long arm of chromosome 10 was due to recombination aneusomy in a pericentric inversion present in the mother (Dutrillaux et al., 1973), all other cases have resulted from reciprocal translocations involving chromosomes 1, 12, 13, 14, 15, 16, 17, 21, or 22 (Borgaonkar, 1977). To our knowledge, only one case of translocation (10;13) has ever been published (Mul-
Fig. 1a and b. Meiosis in semen from the father of the patient: (a) Pachytene; arrow points to possible quadrivalent; (b) Diakinesis with 21 bivalents and one quadrivalent; small arrow points to sex bivalent

Fig. 2a. G-banded chromosomes from the father with balanced translocation (10;13)(q25;p12); (b) Q-banded chromosomes from the father showing light fluorescence of the translocated region that does not correspond to satellites; the bright region 13p11 indicates that the break has taken place at 13p12; (c) G-banded chromosomes from the child with partial trisomy 10q (q25–qter)

cathy et al., 1974), but the break point in chromosome 10 was at q24 and the translocation affected the long arm of chromosome 13 t(10;13)(q24;q34). On the other hand, only one case of partial trisomy for the region q25–qter of chromosome 10 has been described (Kroyer and Niebuhr, 1975), but it was due to a (10;18) translocation. Thus, ours is probably the first case of partial trisomy (q25–qter) of chromosome 10 due to a reciprocal translocation to the short arm of chromosome 13, detected through a study of meiosis in semen.

To determine the effect of the presence in triplicate of this region (Yunis and Sanchez, 1974; Yunis and Lewandowski, 1977; de Grouchy and Turleau, 1977) we have compared the phenotypic characteristics of our patient with those of other cases of partial trisomy 10q of different size, including the distal half of the long arm

Fig. 3. Phenotype of the trisomic child at 7 years of age