A CASE OF A RECIPROCAL TRANSLOCATION BETWEEN THE Y AND NO. 1 CHROMOSOMES

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Summary A reciprocal translocation between the Y and No. 1 chromosomes was found in a male infant with psychomotor retardation and infantile spasms. The karyotype was designated as 46,X,t(1; Y) (q21; q11) on the basis of G- and Q-banding analyses. Previously reported cases with Y/autosomal translocations are reviewed.

INTRODUCTION

Since banding techniques were introduced, many cases of structural abnormalities of the Y chromosome have been described; isochromosomes of the long arm (Book *et al.*, 1973; Magnelli *et al.*, 1974) and short arm (Siebers *et al.*, 1973), pericentric inversions (Grace *et al.*, 1972; Zeuthen and Nielsen, 1973), deletion of the long arm (Meisner and Inborn, 1972) and dicentric Y chromosomes (Cohen *et al.*, 1973). However, relatively few cases of translocation between autosomes and Y chromosomes have been reported. This report describes a case of a *de novo* Y/1 translocation.

CASE REPORT

The proband was a 4-month-old male infant, born at term to a 21-year-old primigravid mother, whose pregnancy, labor and delivery were uneventful. The father was 26 years old. The birth weight was 3,050 g. Family history was not contributory. The infant exhibited neonatal hyperbilirubinemia which was treated by phototherapy. During the newborn period he had feeding difficulties and vomited frequently after sucking. At the age of 4 months, he was referred to us because of poor weight gain, psychomotor retardation and massive myoclonic spasms. Physical examination showed a small malnourished infant but no obvious malformations. His height was 60.0 cm (10th percentile), weight 4,490 g (less than third percentile) and head circumference 39.5 cm (10th percentile). The external genitals were those of a normal male. His psychomotor development was grossly retarded with no head control or visual following. Neurologic examination indicated muscular hypotonia, hyperreflexia and dysphagia. Dermatoglyphic study showed eight

loops, one whorl and one arch on the fingertips. Simian creases were present on the both hands.

The laboratory data (complete blood count, urinalysis, serum electrolytes, serum cholesterol, blood urea nitrogen, serum transaminase and alkaline phosphatase, serum thyroid and growth hormone values) were all within normal ranges. Cerebrospinal fluid protein and cell count were normal. Urinary screening was negative for metabolic disorders. X-ray examinations of the chest, skull and hip joints and an intravenous pyelogram were normal. Electroencephalography showed a modified pattern of hypsarrhythmia and computed tomography of the head indicated diffuse cortical atrophy of the brain with mild dilatation of both lateral ventricles. Since the age of 3 months, seizures of infantile spasms had occurred several times a day. These seizures had been refractory to usual medications and ACTH therapy of 4 weeks duration was initiated at the age of 6 months. He was discharged at the age of 8 months, when seizures had been controlled and electroencephalographic improvements had been obtained. However, there had been little somatic and psychomotor development during the admission.

CYTOGENETIC FINDINGS

Peripheral blood was cultured in a usual manner. Chromosome preparations

Fig. 1. Q-banding karyotype of the proband. Note a reciprocal translocation between the No. 1 and the Y chromosomes.

Fig. 2. G-banding karyotype of the proband.

were submitted to conventional Giemsa staining, G-banding (Seabright, 1971, with minor modifications) and Q-banding (Caspersson *et al.*, 1970). Conventional staining showed that in all 30 metaphases examined a No. 1 chromosome and a Y chromosome had been replaced by two unusual chromosomes; one was an acrocentric chromosome somewhat larger than those of D group, the other was a submetacentric one approximating to No. 6 chromosome in form and size. QM-fluorescence study showed a reciprocal translocation between the distal four-fifths of the long arm of the No. 1 chromosome and the distal half of the long arm of the Y chromosome (Fig. 1). All the other chromosomes showed normal G-banding patterns (Fig. 2). G-banding analysis made more precise identification of the break-points possible. The breaks appeared to occur at the q21 band of the No. 1 chromosome and at the q11 band of the Y chromosome. The latter break-point was considered to be just above the junction of the fluorescent and nonfluorescent part of long arm of the Y chromosome. According to the Paris Conference (1971) nomenclature, the karyotype was designated as 46,X,t(1;Y) (q21;q11). Karyotypes of both parents were normal.

Table 1. Cases of Y/autosomal translocations.

Authors	Autosome involved	Phenotypic sex	Karyotype	Main clinical features
van den Berghe <i>et al.</i> (1965, 1973)	No. 2	male	46,X,t(2p-; Yp+)	Hypogonadism
Federman <i>et al.</i> (1967)	D group	female	45,X/45,X,t(Dp+; Yq-)	Mixed gonadal dysgenesis
Nakagome <i>et al.</i> (1968)	(?)	male	47,XY,t(Y; autosome)	Multiple anomalies
Noel <i>et al.</i> (1971)	No. 15	male	46,XY,t(15p+; Yq-)	Normal phenotype
Krmpotic <i>et al.</i> (1972)	No. 14	male	46,X,t(14q-; Yp+)	Multiple anomalies
Frund <i>et al.</i> (1972)	No. 15	male	46,XY,t(15p+; Yq-)	Normal phenotype
Develing <i>et al.</i> (1973)	No. 7	male	46,X,t(7q+; Yq-)	Multiple anomalies
Gilgenkranz <i>et al.</i> (1973)	No. 13	male	46,XY,t(13q+; Yq-)	Multiple anomalies
Hahnemann and Eiberg (1973)	No. 15	male	46,XY,t(15p+; Yq-)	Normal phenotype
Hreidarsson <i>et al.</i> (1973)	No. 15	male	46,XX,t(15p+; Yq-)	Klinefelter's syndrome
Lundsteen and Philip (1973)	No. 22	male	46,XY,-22,+t(Y; 22)(q12; p11 or 12)	not mentioned
Pfeiffer <i>et al.</i> (1973)	No. 15	male	46,X,t(Yq-; 15p+)	Multiple anomalies
Reitahu (1973)	No. 22	male	46,XY,t(22p+; Yq-)	Small genitalia
Hillman <i>et al.</i> (1974)	No. 21	female	45,X,t(21p+; Yq-)	Potter and Turner syndromes
Nielsen <i>et al.</i> (1974)	No. 15	male (5 cases)	46,XY,t(15p+; Yq-)	Normal phenotype
Park <i>et al.</i> (1974)	No. 16	female	45,X,-16,+t(16Yp-)/46,XYq-,-16,+t(16Yp-)	Mixed gonadal dysgenesis
Šubrt and Blehová (1974)	No. 15	male	45,X,t(Y; 15q)	Psychomotor retardation
Dutrillaux and Gueguen (1975)	No. 5	male	46,X,t(5; Y)(p15; q11)	Azoospermia
Nielsen and Rasmussen (1976)	No. 15	male (3 cases)	46,XY,der(15)(15; Y)(p13; q12)	Normal phenotype
		female (2 cases)	46,XX,der(15),t(15; Y)(p13; q12)	Normal phenotype
	No. 22	female	46,XX,der(22),t(22; Y)(p13; q12)	Normal phenotype
Laurent and Dutrillaux (1976)	No. 14	male	46,X,t(Y; 14)(q11; p11)	Azoospermia
Turleau <i>et al.</i> (1976)	No. 7	male	46,X,t(Y; 7)(q122; q111)	Sterility
Warter and Ratel (1976)	No. 16	male	46,XY,ins(16; Y)(q12; q12)	Hypogonadism
Curtis (1977)	No. 13	male	46,XY,der(13),t(Y; 13)(q12; p11)	Oligospermia
Wisniewski and Higgins (1977)	No. 6	male	46,X,t(Y; 6)(q12; q27)/47,X,t(Y; 6)(q12; q27),+derY	Multiple anomalies
Present case	No. 1	male	46,X,t(1; Y)(q21; q11)	Psychomotor retardation

DISCUSSION

Before banding techniques were introduced into chromosome studies, several cases of Y/autosomal translocations (van den Berghe *et al.*, 1965; Federman *et al.*, 1967; Nakagome *et al.*, 1968) had already been reported. Since identification of involved autosomes in Y/autosomal translocations became possible by the use of banding methods, 32 cases of confirmed Y/autosomal translocations including our case have been reported (Table 1); Y/15 (Noel *et al.*, 1971; Frund *et al.*, 1972; Pfeiffer *et al.*, 1973; Hreidarsson *et al.*, 1973; Hahnemann and Eiberg, 1973; Nielsen *et al.*, 1974; Šubrt and Blehová, 1974; Nielsen and Rasmussen, 1976), Y/22 (Reitalu, 1973; Lundsteen and Philip, 1973; Nielsen and Rasmussen, 1976), Y/2 (van den Berghe *et al.*, 1973), Y/5 (Dutrillaux and Gueguen, 1975), Y/6 (Wisniewski and Higgins, 1977), Y/7 (Develing *et al.*, 1973; Turleau *et al.*, 1976), Y/13 (Gilgenkranz *et al.*, 1973; Curtis, 1977), Y/14 (Krmptotic *et al.*, 1972; Laurent and Dutrillaux, 1976), Y/16 (Park *et al.*, 1974; Warter and Ratel, 1976), Y/21 (Hillman *et al.*, 1974). To our knowledge, our case is the first report of a Y/1 translocation. As summarized in Table 1, acrocentric autosomes are frequently involved in Y/autosomal translocations. Y/15 translocation is the most common type and 16 cases have been reported. All but three cases of Y/15 translocations (Hreidarsson *et al.*, 1973; Pfeiffer *et al.*, 1973; Šubrt and Blehová, 1974) showed a normal phenotype. In all of those with a normal phenotype, heterochromatic segment of the long arm of the Y chromosome that is genetically inert was additionally translocated to the short arms of the No. 15 chromosome. This chromosomal duplication-deficiency did not seem to yield any phenotypic effect. In contrast, cases other than those of Y/15 translocations showed some phenotypic abnormalities even where the chromosome changes were apparently balanced. Our case, whose chromosome constitution was an apparently balanced translocation, showed severe psychomotor retardation, infantile spasms and hypotonia. The appearance of these phenotypic abnormalities and the balanced translocation may be coincidental, or the two may be causally related. If causal relationship exists between the two, the abnormal phenotype may be caused by a small microscopically undetectable loss of the involved chromosomes occurring during the translocation process or by a "position effect" on vital genes produced by the translocation.

Finally, chromosome analyses in cases of structural aberrations and translocations of the Y chromosomes have made great contributions to the study of the location of the testis differentiating gene on the Y chromosome. Since individuals with isochromosomes of the long arms of the Y chromosome have a Turner syndrome, the location of the testis differentiating gene has been thought to be on the short arm of the Y chromosome. Krmptotic *et al.* (1972) reported a baby with the karyotype 46,X,t(Yp+; 14q-) who showed a normal male phenotype and suggested that this gene may be located on the centromeric portion of the short arm of the Y chromosome, while Sieber *et al.* (1973) proposed the proximal part of the long arm of the

Y chromosome as the site of the gene from the observation that their proband with 45,X/46,X,i(Yp) karyotype showed a Turner syndrome. The translocated Y chromosome of our case contained both the whole short arm and the proximal half of the long arm of the Y chromosome. Therefore, it is reasonable that our case showed a normal testicular differentiation.

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