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Cytogenetic analysis of 400 sperm from three translocation heterozygotes

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Summary. Sperm chromosome complements were studied in three men who carried reciprocal translocations. A total of 400 sperm were karyotyped after in vitro penetration of hamster eggs: 217 sperm from t(2;9)(q21;p22), 164 from t(4;6)(q28;p23) and 19 from t(7;14)(q21;q13). All possible 2:2 and 3:1 meiotic segregations were observed for t(2;9) and t(4;6); for t(7;14) only 2:2 segregations were observed. For alternate segregations, the number of normal sperm was not significantly different from the number of sperm carrying a balanced form of the translocation in any of the translocations, as theoretically expected. The percentage of sperm with an unbalanced form of the translocation was 57% for t(2;9), 54% for t(4;6) and 47% for t(7;14). There was no evidence for an interchromosomal effect in any of the translocations since the frequencies of numerical abnormalities (unrelated to the translocation) were within the normal range of control donors. The frequencies of X- and Y-bearing sperm did not differ significantly from 50%. Results from a total of 17 reciprocal translocations studied by sperm chromosomal analysis were reviewed.

Introduction

Individuals who are heterozygous for a chromosomal translocation have an increased risk for spontaneous abortions and chromosomally abnormal children. It is impossible to gain information about the meiotic segregation of chromosomes by studying liveborn children because lethal segregations will have already been lost as recognized or unrecognized spontaneous abortions. It is also very difficult to estimate the risk of a chromosomally abnormal liveborn child because of the severe ascertainment bias inherent in families who present at a genetics clinic.

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Analysis of human sperm chromosomes after in vitro penetration of golden hamster eggs has made it possible to examine the products of meiotic segregation in mature sperm directly (Rudak et al. 1978; Martin et al. 1983, 1987). To date, 13 men carrying reciprocal translocations have been studied using the human sperm-hamster egg system (Balkan and Martin 1983; Martin 1984, 1988a; Brandriff et al. 1986; Burns et al. 1986; Templado et al. 1988; Pellestor et al. 1989; Martin et al. 1990). In this report we present our results from the study of three males heterozygous for the following reciprocal translocations: t(2;9)(q21;p22); t(4;6)(q28;p23); t(7;14)(q21;q13).

Materials and methods

Ascertainment of translocation heterozygotes

46,XY,t(2;9)(q21;p22). The (2;9) translocation carrier was 36 years of age and was ascertained after cytogenetic investigation for habitual abortion. A pedigree of his family is presented in Fig. 1. Two experiments were performed from the same ejaculate using TEST-yolk buffer to store the sperm (Martin 1988b).

46,XY,t(4;6)(q28;p23). The (4;6) translocation heterozygote was 25 years of age and was ascertained after cytogenetic investigation because of a family history of multiple spontaneous abortions and four children born with a similar constellation of multiple congenital abnormalities and developmental delay. The pedigree is presented in Fig. 2. The abnormal children lived approximately 2 years and had the following abnormalities: bilateral congenital cataracts, bilateral coloboma of the irides, club feet, hypospadias, laryngeal

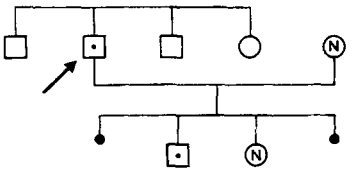

Fig. 1. Pedigree of t(2;9)(q21;p22). The *arrow* indicates the sperm donor. ⊡⊙ Translocation heterozygotes, Ⓝ normal karyotype, □○ chromosomes not studied, • spontaneous abortion

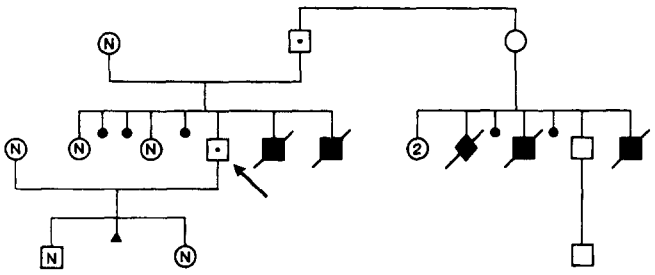

Fig. 2. Pedigree of t(4:6)(q28:p23). The *arrow* indicates the sperm donor. ⊡ Translocation heterozygotes, Ⓝ Ⓝ normal karyotypes, □ ○ chromosomes not studied, ■ multiple congenital abnormalities and developmental delay, ◆ stillbirth with multiple congenital abnormalities, • spontaneous abortion, ▲ spontaneous abortion with an unbalanced form of the translocation

stenosis and severe developmental delay. Cytogenetic investigation of a spontaneous abortion of the sperm donor's wife demonstrated an unbalanced segregation of the translocation: 46,XY, −6,+der(6),t(4:6)(q28:p23) pat. Seven experiments were performed from three ejaculates using TEST-yolk buffer to store the sperm (Martin 1988b).

46,XY,t(7:14)(q21:q13). The (7:14) translocation heterozygote was 55 years of age and was ascertained by chance when he volunteered to be a sperm donor for a study of the effect of age on the frequency of sperm chromosomal abnormalities (Martin et al. 1987). Balanced and unbalanced forms of the translocation were identified in sperm chromosome complements. Unfortunately only one experiment was performed as he was lost to follow-up. We have no family history except that he had fathered four normal children and had no spontaneous abortions.

Table 1. Segregation of sperm chromosomes in t(2:9)

Segregation type	Sperm chromosome complements[a]	No. of spreads	Percent
2:2 Segregations			
Alternate	23,X or Y	52	25
	23,X or Y,−2,−9,+der(2),+der+(9)	38	18.3
		90	43.3
Adjacent 1	23,X or Y,−2,+der(2)	23	11.1
	23,X or Y,−9,+der(9)	36	17.3
		59	28.4
Adjacent 2	23,X or Y,−2,+der(9)	34	16.3
	23,X or Y,−9,,+der(2)	15	7.2
	22,Y,−2,+9[b]	1	0.5
		50	24.1
3:1 Segregations	22,X,−2		
	22,X,−9		
	22,X,−2,−9,+der(9)		
	24,X or Y,+der(2)		
	24,X or Y,+der(9)(4 sperm)		
	24,X or Y,+ der(2),+der(9)	9	4.3

[a] Sperm with numerical or structural abnormalities unrelated to the translocation were disregarded when assigning karyotypes to the various segregations

[b] Crossing over within the interstitial segments of chromosome 9 or non-disjunction at anaphase II

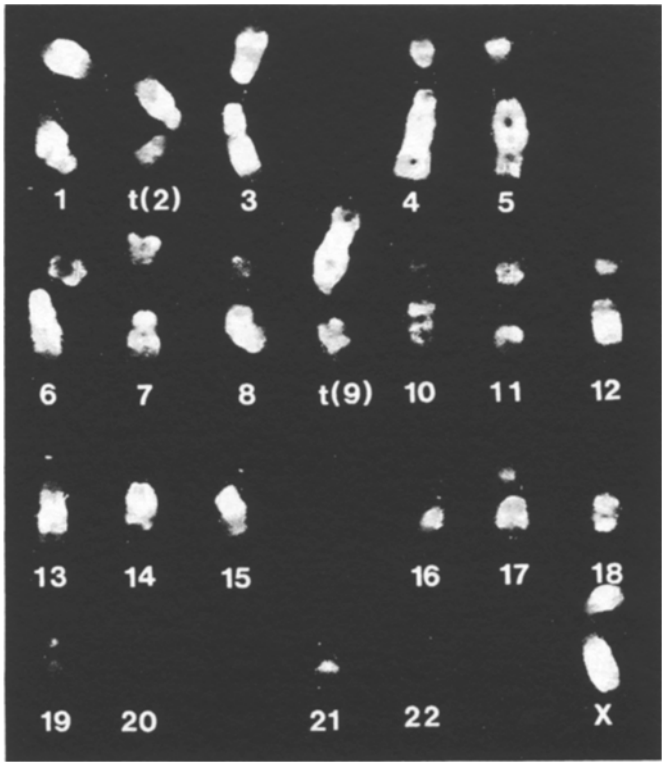

Fig. 3. A Q-banded karyotype of a sperm carrying the balanced form of the translocation 23,X,−2,−9,+der(2),+der(9)

Sperm chromosome complements

For t(2:9) and t(4:6) sperm was capacitated in TEST-yolk buffer (Martin 1988b) and for t(7:14) in BWW medium (Martin 1983). Sperm chromosome complements were obtained by in vitro penetration of golden hamster oocytes by human sperm.This technique, which has been described in detail elsewhere (Martin 1983, 1988b), allows analysis of Q-banded human pronuclear sperm chromosome complements.

Results

Reciprocal translocation t(2;9)

A total of 217 sperm chromosome complements was obtained of which segregations could be determined in 208. The relative proportions of alternate, adjacent 1, adjacent 2, and 3:1 segregation are presented in Table 1. The alternate segregation was the most frequent (43.3%), followed by adjacent 1 (28.4%), adjacent 2 (24.1%), and 3:1 segregation (4.3%). Within the alternate segregation, the number of normal sperm (52) did not differ significantly from the number of sperm carrying a balanced form of the translocation (38). An example of a sperm with a balanced form of the translocation is shown in Fig. 3. The frequency of sperm with an unbalanced form of the translocation was 56.7%. One sperm complement was observed that could be attributed to crossing over in

Table 2. Abnormal sperm chromosome complements unrelated to the translocation t(2;9)

Numerical abnormalities (9)	Structural abnormalities (47)
Hypohaploid (9)	
22,X,−2,−9,+der(2),+der(9),−8	23,Y,+ace
22,Y,−2,−9,+der(2),+der(9),−20	23,X,+ace,(?ctb(20)(p11))
22,X,−2,+der(2),−11	23,X,csb(X)(p11)
22,X,−2,+der(2),−17	23,X,csb(10)(p11)
22,Y,−9,+der(9),−18	23,X,csb(13)(q31)
22,Y,−2,+der(9),−16	23,X,csb(16)(q1),csb(19)(q13.1)
22,Y,−9,+der(2),−20	23,X,csb(22)q11.2 or 12)
23,X,+der(9),−5	23,X,csg(12)(q21)
21,X−9,−14	23,Y,csg(17)(q23)
	23,X,del(9)(q21 or 13)
	22,X,cte(1;16)(q12;q11)(qr,sym,complete),+ace
	23,Y,−16,+mar(3qter)q12 or 21::16q2 > pter),del(3)(q13 or 21),+ace(?16q2 >qter)
	23,Y,−2,−9,+der(2),+der(9),csb(der(2))(2q11 or 12 or 13),csb(8)(q21)
	23,X,−2,−9,+der(2),+der(9),del(1)(q12 or 21)
	21,X,−2,−9,+der(2),+der(9),cte(8;16)(q24;q23)(tr,incomplete),cte(der(9);13)(9p1;q3)(qr,sym, incomplete)
	22,X,−2,−9,+der(2),+der(9),cte(13;22)(p1;p1)(tr,incomplete)
	23,X,−2,+der(2),+ace
	23,X,−2,+der(2),csb(1)(p33 or 34)
	23,X,−2,+der(2),csb(1)(q12 or 21)
	23,Y,−2,+der(2),csb(7)(q22),csb(10)(q11)
	23,Y,−2,+der(2),csg(1)(q21)
	23,Y,−9,+der(9),+ace
	23,X,−9,+der(9),csb(6)(q16 or 21)
	23,X,−9,+der(9),csb(11)(cen)
	23,X,−9,+der(9),csb(15)(q22 or 23)
	23,X,−9,+der(9),csb(16)(q23)
	23,Y,−9,+der(9),csb(22)(q11)
	23,Y,−9,+der(9),ctg(4)(q28 or 31)
	23,X,−2,+der(9),csb(1)(p1)
	23,X,−2,+der(9),csb(der(9))(9q32)
	23,X,−2,+der(9),csb(19)(q13.1)
	23,X,−2,+der(9),csg(X)(q22 or 23 or 24),csg(4)(q31),csg(12)(q24),csg(17)(q22)
	23,Y,−2,+der(9),csg(14)(q31 or 32),ctg(7)(q21),+2aces
	23,Y,−2,+der(9),csg(19)(q13)
	22,Y,−2,+der(9),cte(1;7)(q42;p21 or 22)(tr,incomplete)
	22,X,−2,+ace
	24,Y,+der(9),csb(6)(q22),csb(12)(?cen),?del(18)(p)
	23,Y,+der(9),dic(3;6)(3qter > p21::6q16 > pter),+ace(3p21 > pter),+ace(6q16 > qter)
	. .MB
	. .MB + R(3 sperm)
	.Y.MB + R (5 sperm)

the interstitial segments, although nondisjunction at anaphase II could also account for the complement (Table 1).

The frequency of abnormalities unrelated to the translocation was 25.8%. The details of these abnormal complements are presented in Table 2. The frequency of numerical abnormalities was 4.3%, all of which consisted of hypohaploid complements. The frequency of structural chromosomal abnormalities was 21.7%. The frequencies of both the numerical and structural abnormalities were within the range of normal control donors (Martin et al. 1987). The percentage of X- and Y-bearing sperm was 53.5% and 46.5%, respectively, which does not differ significantly from the expected 50%.

Reciprocal translocation t(4;6)

A total of 164 sperm chromosome complements was obtained; of these 158 provided information on chromosome segregation. The relative proportions of alternate, adjacent 1, adjacent 2, and 3:1 segregation are presented in Table 3. The frequencies of alternate (45.6%) and adjacent 1 segregation (51.9%) were approximately equal, whereas adjacent 2 (1.9%) and 3:1 (0.6%) segregations were much rarer. Within the alternate segregation, the number of normal sperm (38) did not differ significantly from the number of sperm carrying a balanced form of the translocation (34). An example of a sperm with a balanced form of the translocation is shown in Fig. 4. The frequency of sperm with an unbalanced form of the translocation was 54.4%. Three sperm complements were observed that could be attributed to crossing over in the interstitial segments, although nondisjunction at anaphase II could also account for the complements (Table 2).

The frequency of abnormalities unrelated to the translocation was 26.8%. The details of these abnormal

Table 3. Segregation of sperm chromosomes in t(4;6)

Segregation type	Sperm chromosome complements[a]	No. of spreads	Percent
2:2 Segregations			
Alternate	23,X or Y	38	24.1
	23,X or Y,−4,−6,+der(4),+der(6)	34	21.5
		72	45.6
Adjacent 1	23,X or Y,−4,+der(4)	41	25.9
	23,X or Y,−6,+der(6)	41	25.9
		82	51.9
Adjacent 2	23,Y,−6,+der(4)		
	23,X,−4,+6[b]		
	23,X,−4,−6,+der(6),+der(6)		
		3	1.9
3:1 Segregations	24,Y,−4,+6,+der(4)[b]	1	0.6

[a] Sperm with numerical or structural abnormalities unrelated to the translocation were disregarded when assigning karyotypes to the various segregations

[b] Crossing over within the chromosomal interstitial segments or non-disjunction at anaphase II

complements are presented in Table 4. The frequency of numerical abnormalities was 7.9%, which mainly consisted of hypohaploid complements (6.7%). The frequency of structural chromosomal abnormalities was 17.7%. Two sperm complements had numerical and structural abnormalities (1.2%). The frequencies of both the numerical and structural abnormalities were within the range of normal control donors (Martin et al. 1987). The percentage of X- and Y-bearing sperm was 49.7% and 50.3%, respectively, which did not differ significantly from the expected 50%.

Reciprocal translocation t(7;14)

A total of 19 sperm chromosome complements was obtained from the single experiment. The relative proportions of alternate, adjacent 1, adjacent 2, and 3:1 segregation are presented in Table 5. The alternate segregation was the most frequent (52.6%) followed by adjacent 1 (31.6%) and adjacent 2 (15.8%) segregations. Within the alternate segregation, the number of normal sperm (6) did not differ significantly from the number of sperm carrying a balanced form of the translocation (4). An example of a sperm with a balanced form of the translocation is shown in Fig. 5. The frequency of sperm with an

Table 4. Abnormal sperm chromosome complements unrelated to the translocation t(4;6)

Numerical abnormalities (13)	Structural abnormalities (29)
Hypohaploid (11)	
22,Y,−22	23,Y,csb(1)(q12),csb(6)(p21 or 22),csb(8)(q21)
22, ,(X or Y)	23,Y,csb(5)(q31)
22,X,−4,−6,+der(4),+der(6),−14	23,Y,csb(8)(q22)
22,Y,−4,−6,+der(4),+der(6),−19	23,Y,csb(12)(p12)
22,X,−4,−6,+der(4),+der(6),−D	23,Y,csb(19)(q13.1)
22,X,−4,+der(4),−10	23,X,csg(19)(q13.1),ctg(3)q13)
22,X,−4,+der(4),−17	20,Y,−4,−6,+der(4),+der(6),cte(Y;7;12)(p11;p15;p)(complex),cte(2;5)(q24 or 25;q31) (tr,incomplete)
22,X,−4,+der(4),−20	23,X,−4,−6,+der(4),+der(6),csb(9)(?cen)
20,Y,−4,+der(4),−8,−10,−16	23,Y,−4,−6,+der(4),+der(6),csb(9)(q13)
21,X,−6,+der(6),−D,−D	23,Y,−4,−6,+der(4),+der(6),csb(der(4))(4q1)
20, ,−6,+der(6),−(X or Y),−1,−15	23,X,−4,−6,+der(4),+der(6),csg(8)(q21)
Hyperhaploid (2)	23,X,−4,+der(4),csb(10)(p13 or 14)
	23,Y,−4,+der(4),csb(16)(q12)
24,X,−4,+der(4),+21	23,X,−4,+der(4),csb(19)(q13.1)
24,Y,−4,+der(4),+22	23,X,−6,+der(6),csb(5)(q32)
	23,X,−6,+der(6),csg(10)(cen)
	22,Y,−6,+der(6),cte(4;7)(q31;q31)(qr,asy,incomplete)
	22,Y,−6,+der(6),cte(4;der(6))(q35;6p23 or 4q28)(tr,complete)
	23,Y,−6,+der(4),csb(4)(p11 or 12)
	, ,MB+R(4 sperm)
	,Y,MB+R(2 sperm)
	,X,MB+R(normal 4 and 6)
	,Y,MB+R(normal 4 and 6)
	,Y,MB+R,−4,−6,+der(4),+der(6)
	, ,MB+R,−6,+der(6)
Numerical and structural (2)	
19,X,−4,−6,+der(4),+der(6),−8,−9,−17,−22,del(1)(p3),+3aces	
21,Y,−4,−6,+der(4),+der(6)−18,cte(3;20)(q27 or 28;q11)(qr,asy,incomplete)	

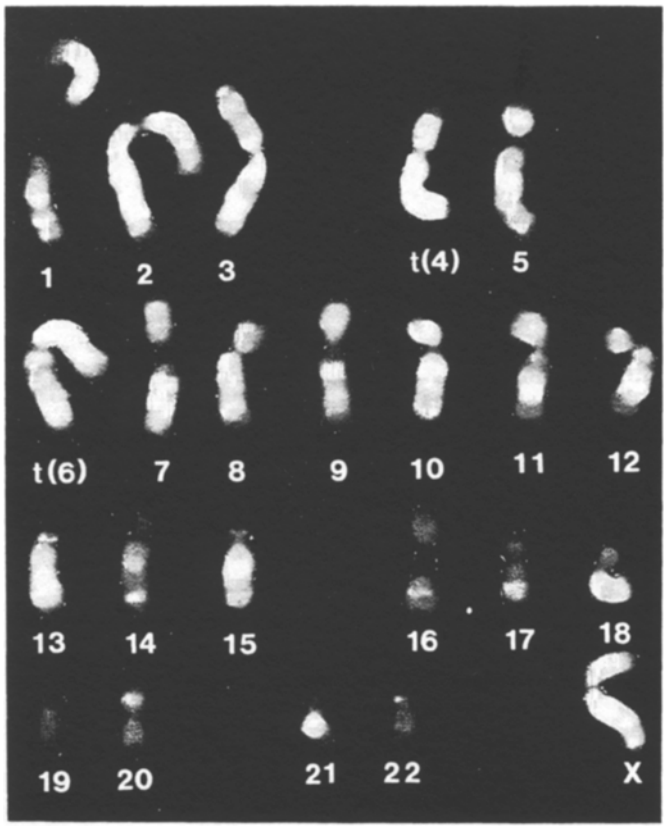

Fig. 4. A Q-banded karyotype of a sperm carrying the balanced form of the translocation 23,X,−4,−6,+der(4),+der(6)

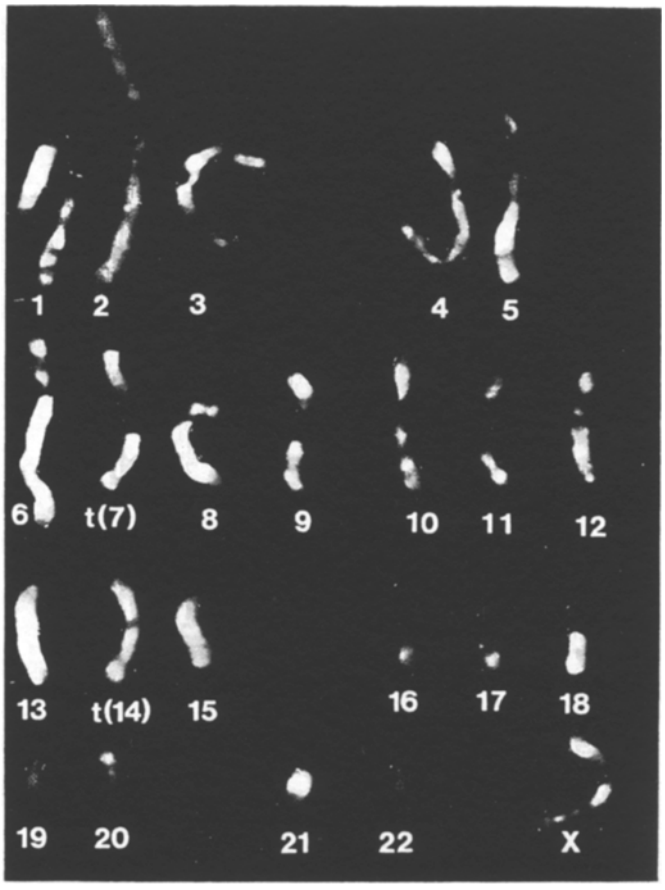

Fig. 5. A Q-banded karyotype of a sperm carrying the balanced form of the translocation 23,X,−7,−14,+der(7),+der(14)

unbalanced form of the translocation was 47.4%. One sperm complement was observed that could be attributed to crossing over in the interstitial segments, although nondisjunction at anaphase II could also account for the complement (Table 5). There were no chromosomal abnormalities unrelated to the translocation.

Discussion

For the two reciprocal translocations with large sample sizes t(2;9) and t(4;6) all theoretical 2:2 and 3:1 chromosomal segregations were observed. For t(7;14), all 2:2 segregations were observed. To date, a total of 17 reciprocal translocations from 16 men have been studied by sperm chromosome analysis. One male carried two translocations (Burns et al. 1986). The segregations observed in these translocations are detailed in Table 6. Alternate and adjacent 1 segregations were present in all of the translocations, with a mean of 49% and 38%, respectively. Adjacent 2 and 3:1 segregations were rarer with means of 8% in 11 translocations and 5% in 13 translocations, respectively. Adjacent 1 segregation was the most common segregation leading to chromosomal imbalance in all 17 translocations, even in those translocations known to have abnormal livebirths exclusively from another segregation such as the common t(11;12) segregation for which only 3:1 segregations have been reported in livebirths (Martin 1984). Thus it appears that the adjacent 1 segregation occurs preferentially during spermatogenesis. This is not surprising since both adjacent 2 and 3:1 segregations require nonseparation of homologues and are thus akin to nondisjunction.

Table 5. Segregation of sperm chromosomes in t(7;14)

Segregation type	Sperm chromosome complements	No. of spreads	Percent
2:2 Segregations			
Alternate	23,X or Y	6	31.6
	23,X or Y,−7,−14, +der(7),+der(14)	4	21.1
		10	52.6
Adjacent 1	23,X or Y,−7,+der(7)	2	10.5
	23,X or Y,−14,+der(14)	4	21.1
		6	31.6
Adjacent 2	23,X,−14,+der(7)	2	10.5
	23,X,−7,+14[a]	1	5.3
		3	15.8

[a] Crossing over within the interstitial segment of chromosome 9 or nondisjunction at anaphase II

Alternate segregation yields normal offspring: theoretically one-half should have normal chromosomes and one-half, the two balanced translocated chromo-

Table 6. Segregation of reciprocal translocations

Translocation	Segregations (%)				
	Balanced (alternate)	Unbalanced			Total unbalanced
		Adjacent 1	Adjacent 2	3:1	
t(5;18)[a]	78	16		3	19
t(5;13)[b]	77	21	2		23
t(5;11)[c]	70	26		4	30
t(6;14)[a]	68	32			32
t(2;5)[d]	71[d]		4	25	32[d]
t(4;7)[b]	57	35	7	2	43
t(7;14)[e]	53	32	16		47
t(6;7)[b]	51	49			49
t(12;20)[f]	47	42	10	2	53
t(4;6)[e]	46	52	2	1	54
t(2;9)[e]	43	28	24	4	57
t(9;10)[g]	41	48	5	6	60
t(3;16)[h]	37	41	16	5	63
t(8;15)[h]	37	38	21	4	63
t(9;18)[b]	34	63		2	66
t(7;14)[c]	30	48		17	70
t(11;22)[i]	23	39	23	15	77
Mean (SD)	49 (16)	38 (12)	8 (9)	5 (7)	51 (17)

[a] Balkan and Martin (1983)
[b] Pellestor et al. (1989)
[c] Burns et al. (1986); one male carried both translocations but they appeared to segregate independently
[d] Templado et al. (1988); some alternate and adjacent segregations could not be distinguished so the 32% frequency of unbalanced sperm is a minimal estimate
[e] This report
[f] Martin et al. (1990)
[g] Martin (1988)
[h] Brandriff et al. (1986)
[i] Martin (1984)

somes. This expectation was met in all three translocations studied in this report. Similarly in 16 of the 17 reciprocal translocations studied by sperm chromosomal analysis, the theoretical 1:1 ratio of normal and balanced complements was observed, which corroborates the validity of the system (Pellestor et al. 1989). The only exception was one translocation (5;11) in the male carrying two reciprocal translocations and this might have been caused by the small sample size or interference from the second translocation (Burns et al. 1986).

The frequency of chromosomally unbalanced sperm varied from 19% to 77% with a mean of 51% in the 17 translocations (Table 6). In fact most of the translocations, including the three studied in this report, have had approximately one-half of the sperm chromosomally unbalanced. This frequency of imbalance observed in sperm is much higher than seen in studies of human fetuses from translocation heterozygotes (Boue and Gallano 1984). The most likely explanation for this difference is that many chromosomally unbalanced embryos are lost early in embryological development. It is also possible that sperm selection in vivo might account for the lower frequency of unbalanced fetuses. However, this is unlikely since paternal reciprocal translocation carriers were found to have the same frequency of chromosomally unbalanced fetuses as maternal carriers (11.4%) in the European collaborative study of prenatal diagnosis (Boue and Gallano 1984).

There have been reports of children born with trisomies (such as Down syndrome) unrelated to the translocation carried in the family (Aurias et al. 1978). A number of researchers have suggested that there is an increased frequency of chromosomal abnormalities unrelated to the specific translocation and have termed this an "interchromosomal effect." In this study we had no evidence for an interchromosomal effect since the frequency of numerical abnormalities was within the normal range of control donors for all three translocations. Similarly, the frequency of unrelated numerical abnormalities has not been significantly increased for the other translocations studied by sperm chromosomes (Martin 1989), with the exception of the man heterozygous for two translocations (Burns et al. 1986), who had an extremely high frequency of aneuploid sperm (70%). The evidence, to date, does not support the concept of an increased risk of trisomy unrelated to the translocation for men carrying a single translocation. However significant samples of sperm chromosome complements need to be studied in more translocation carriers to assess the possibility of a small interchromosomal effect.

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