Factors predisposing to adjacent 2 and 3:1 disjunctions: study of 161 human reciprocal translocations

P. JALBERT AND B. SELE

From the Laboratoire de Cytogénétique du CHU de Grenoble, Hôpital des Sablons, F 38700 La Tronche, France

SUMMARY Reciprocal translocations produce imbalances by three types of disjunction which are, in decreasing frequency, adjacent 1, 3:1, and adjacent 2.

Adjacent 1 disjunction produces duplication deficiencies of inverse topography to those of adjacent 2. The imbalanced chromosome segments in one of these types are balanced in the other. The disjunction 3:1 produces pure trisomies and monosomies.

The following situations predispose to adjacent 2 disjunction: translocations between the long arms of two acrocentric chromosomes or between one of these and that of a No 9 chromosome; centric segments, either short or carrying a heterochromatic zone (9qh); a balanced translocation in the mother.

The factors predisposing to the disjunction adjacent 2 operate by selection, or directly on the meiotic configuration. Some of them (shortness of the interstitial segment, shortness of the short arms of translocation chromosomes) act in both these ways. Their influence is probably responsible for the repetitive and exclusive character of this disjunction.

The conditions for the occurrence of the 3:1 disjunctions seem less strict than those for adjacent 2, although they should be of the same nature (involvement of acrocentrics or a chromosome 9 in the translocation, maternal origin).

Imbalances of reciprocal translocations can result from three types of disjunction.

Type adjacent 1 (the most frequent) occurs when one of the two modified chromosomes is transmitted together with the normal homologue of the other. This situation results in the duplication of one of the translocated segments and the deficiency of the other.

Type adjacent 2 (exceptional) occurs when one of the modified chromosomes is transmitted together with its own homologue. This mechanism results in the duplication deficiency of the non-translocated centric segments of the modified chromosomes; the translocated acentric segments are therefore balanced.

The offspring of the above types of disjunction have 46 chromosomes.

Type 3:1 (rare) results in pure partial monosomies or trisomies. The subjects have 45 to 47 chromosomes.

Two mechanisms are possible.

- (1) The two chromosomes affected by the translocation are transmitted together with only one of their normal homologues ('interchange trisomy or monosomy').
- (2) A single affected chromosome is transmitted together with the two normal homologues ('tertiary trisomy or monosomy').

The incidence and the mechanisms bringing about these different types of chromosome imbalance have already been reviewed by Ford and Clegg (1969), Lejeune *et al.* (1970), Hamerton (1971), and Lindenbaum and Bobrow (1975).

The observation that two members of a family were born with the same type adjacent 2 constitution led us to investigate published reports regarding this phenomenon and to study the mechanisms involved. Data concerning 3:1 disjunction previously established by Lindenbaum and Bobrow (1975) are relevant to the present study.

Received for publication 7 February 1979

Case report

The proband is the third child of the family; the two elder sisters, identical twins (Fig. 1), are normal. There had been no previous miscarriages. A maternal uncle is mentally retarded. At the time of the child's birth the mother was aged 24 years and the father 27. Weight at birth (full term) was 2510 g.

At 9 months, examination of the infant showed evidence of hypotrophy, retarded mental development, and a dysmorphic syndrome characteristic of a 9p trisomy.

There was a chromosome anomaly derived from a balanced reciprocal translocation in the mother and the maternal grandmother $t(9;21)(q22;q22)$ (Fig. 2a). The child's karyotype was: $46,XX,-21,+der(9)$,

Fig. 1 Pedigree of the family. I.2 and II.3 are $46,XX,t(9;21)(q22;q22)$, II.4 and III.3 are $46,-21,+der(9)t(9;21)(q22;q22)$ (9p trisomy).

Fig. 2 Partial karyotypes of the proband (b) and her mother (a).

$t(9;21)(q22;q22)mat$ (Fig. 2b). The maternal uncle (case II.4) has the same imbalance.

These two subjects have received from their mother the chromosome $9q-$, as well as the normal chromosome 9, but neither of the two chromosome 21. They are therefore trisomic for the centromeric portion of chromosome 9 and monosomic for that of 21.

The twin sisters and the father of the patient have normal karyotypes.

Materials and methods

A study was carried out on 161 families with reciprocal translocations with an abnormal proband (158 came from published reports of the last 5 years, 3 originate from this laboratory). These families contain 221 imbalanced subjects. The translocated chromosomes and the breakpoints were identified by banding techniques.

The sample was divided into 2 groups.

Group I: translocations involving at least one acrocentric chromosome (reciprocal translocation between two acrocentrics or between one acrocentric and one non-acrocentric): 98 families (Table 1).

Group II: translocations involving no acrocentric: 63 families (Table 2).

The subjects of the two groups were listed according to the precise origin of their chromosome constitution: alternate, adjacent 1, adjacent 2, or 3:1.

The paternal or maternal origin of the disjunction was noted. Spontaneous abortions and stillbirths were not listed unless their karyotype was known.

Results

The resulting imbalances in decreasing order frequency were disjunctions adjacent 1 (66.5%, 3.2%), 3:1 (27.6% $\pm$ 3.0%), and adjacent 2 (5.9% $\pm$ 1.6%). The last type was only found 13 times in families (Table 3).

ADJACENT 2 DISJUNCTIONS

These were all found in group I (Table 4) and never in group II (Table 5). These translocations always occur between the long arms of two acrocentrics or between that of an acrocentric and that of a chromosome 9 (Table 6). Any acrocentric can be involved; the sample size is too small to evaluate randomness.

However, there have been reported (Raoul *et al.*, 1976; Kim *et al.*, 1977) two translocations not affected by adjacent 2 segregation, though involving two long arms of acrocentrics (the interstitial segments are in these cases particularly long). Similarly, 15 translocations have taken place between a No

Table 1 Translocations involving at least one acrocentric chromosome

Authors	Chromosomes	Altern	Adj 1	Adj 2	3:1	Proband
Abuello <i>et al.</i> (1977)	(8;13) (q21;q34)mat		1			trisomy 8q
Alfi <i>et al.</i> (1973)	(7;14) (q31;qter)pat		1			trisomy 7q
Archidiacono <i>et al.</i> (1978)	(9;22) (cen;p11)mat		1			trisomy 9p
Armendares <i>et al.</i> (1975)	(12;21) (p11;p11)mat	4	1			trisomy 12p
Armendares and Salamanca-Gomez (1978)	(2;15) (p21;q26)pat		2			trisomy 2p
Aurias <i>et al.</i> (1975)	(11;22) (q231;q111)mat	6			1	trisomy 11q
	(11;22) (q231;q111)mat	3			1	trisomy 11q
Ayraud <i>et al.</i> (1976)	(11;22) (q231;q111)mat	2			1	trisomy 11q
Baccichetti <i>et al.</i> (1975)	(4;21) (q32;q22)mat	2	1			trisomy 4q
Balicek <i>et al.</i> (1975)	(9;15) (q12;p11)mat	4			1	trisomy 9p
Bass <i>et al.</i> (1978)	(5;14) (p11;q11)mat				1	monosomy 5p
Bergamo <i>et al.</i> (1977)	(9;15) (p22;q11)mat	1			1	monosomy 9p
Biederman <i>et al.</i> (1977)	(12;21) (p11;pter)pat		1			trisomy 12p
Blank <i>et al.</i> (1975)	(9;22) (q13;q11)mat	11		4		trisomy 9p
Borgaonkar <i>et al.</i> (1973)	(17;22) (p1;q1)mat				1	trisomy 22q
Brimblecombe <i>et al.</i> (1977)	(5;15) (p11;p12)mat	33	1			trisomy 5p
Cantu <i>et al.</i> (1975)	(10;21) (p11;p11)mat	2	2			trisomy 10p
Castel <i>et al.</i> (1976)	(7;15) (q35;q14)mat	1			1	trisomy 15q
Centerwall <i>et al.</i> (1976)	(9;14) (p22;q32)mat	4	6			trisomy 9p
Centerwall and Francke (1977)	(13;20) (q34;p11.2)mat		3			trisomy 20p
Chaganti <i>et al.</i> (1976)	(5;21) (p11;q11)mat	2			1	monosomy 5p
						monosomy 21q
de la Chapelle <i>et al.</i> (1973)	(4;21) (q21;q21)mat	6			1	trisomy 4q
Chiyo <i>et al.</i> (1975)	(8;13) (p11;q34)pat		2			trisomy 8p
Coco and Penchaszadeh (1977)	(2;14) (q12;q13)mat	6			1	trisomy 14q
Cohen <i>et al.</i> (1975a)	(20;22) (p11;q13)pat	11	2			trisomy 20p
Cohen <i>et al.</i> (1975b)	(4;15) (p16;q22)mat	3			2	trisomy 15q
Dallapiccola <i>et al.</i> (1977)	(4;22) (p11;p12)mat	2	1		1	trisomy 4p
Darmady and Seabright (1975)	(4;21) (p11;p12)mat	1	1			trisomy 4p
Forabosco <i>et al.</i> (1973)	(2;13) (q32;q33)mat	1	1			trisomy 2q
Fried <i>et al.</i> (1977)	(5;14) (p15;q13)mat	3			1	monosomy 5p
Fryns <i>et al.</i> (1977)	(11;14) (q25;q23)pat		1			trisomy 14q
Fu <i>et al.</i> (1976)	(11;22) (q25;q11)mat	3				
Garlinger <i>et al.</i> (1977)	(4;22) (p16;q12)mat	3			1	trisomy 22q
Genest <i>et al.</i> (1977)	(9;22) (p11;p11)mat	1	1			trisomy 9p
Giovannelli <i>et al.</i> (1974)	(4;22) (p11;p12)mat		2			trisomy 4p
Giraud <i>et al.</i> (1977)	(2;13) (q36;q13)mat	1	1			trisomy 13q
Habedank and Faust (1978)	(9;21) (p11;q11)mat	1			2	trisomy 9p
Hansteen <i>et al.</i> (1978)	(12;21) (p12;p11)mat	3	2		1	trisomy 12p
						trisomy 21q
Hobolth <i>et al.</i> (1974)	(12;21) (q24;p11)mat		1			trisomy 12q
Howard-Peebles <i>et al.</i> (1977)	(9;13) (q22;q12)mat	7		1		trisomy 9p
Hustinx <i>et al.</i> (1974)	(10;14) (cen;cen)mat	1	1			trisomy 10p
Hustinx <i>et al.</i> (1975)	(4;15) (p11;q11)mat	5			1	trisomy 4p
Jenkins and Boyd (1976)	(4;21) (q27;p11)mat				1	trisomy 21
Jotterand and Juillard (1976)	(9;13) (p21;q21)mat	1	1			trisomy 13q
Kajii <i>et al.</i> (1974)	(13;18) (q12;q21)pat	1	1		2	
Kessel and Pfeiffer (1977)	(11;22) (q23;q12)mat				1	trisomy 22
Kim <i>et al.</i> (1977)	(13;22) (q22;q12)mat				1	trisomy 13q
						trisomy 22q
Lange and Alfi (1976)	(19;22) (q13;p13)mat	2	2			trisomy 19p
Larson <i>et al.</i> (1977)	(7;22) (p21;q13)pat	8	1			trisomy 7p
Laurent <i>et al.</i> (1973)	(14;21) (q12;q22)pat			1		trisomy 14q
						monosomy 21q
Laurent <i>et al.</i> (1975a)	(11;22) (q231;q11)mat				1	trisomy 11q
Mulcahy and Jenkyn (1975)	(9;21) (p13;q22)mat	1	1			trisomy 9p
Nakagome and Kobayashi (1975)	(10;22) (p11;p11)mat	3	2			trisomy 10p
Niazi <i>et al.</i> (1978)	(18;21) (q21;q22)mat	1	1			trisomy 18q
Nielsen <i>et al.</i> (1976)	(4;13) (q25;q31)mat	2	2			trisomy 4q
Noel <i>et al.</i> (1976a)	(11;22) (q231;q111)mat	4			1	trisomy 11q
Noel <i>et al.</i> (1976b)	(13;22) (q12;p11)mat		1		1	trisomy 13q
	(5;13) (p13;q14)mat		2			trisomy 13q
Orye <i>et al.</i> (1975)	(9;15) (p22;q26)pat		1			monosomy 9p
Owen <i>et al.</i> (1974)	(4;21) (p14;p22)pat		1			trisomy 4p
Pena <i>et al.</i> (1976)	(13;14) (q13;q22)mat			2		trisomy 14a
Pfeiffer and Kessel (1976)	(8;15) (q24;q13) mat	3			2	trisomy 15q
Pfeiffer <i>et al.</i> (1977)	(19;21) (p13;q21)mat	1	1			trisomy 21q
	(4;21) (q27;q21)mat				1	trisomy 4q
Philippe <i>et al.</i> (1975)	(9;22) (p11;q11)mat	1			1	trisomy 9p
Podruch and Weisskopf (1974)	(9;15) (q13;p11)mat	4			2	trisomy 9p
Raoul <i>et al.</i> (1975)	(10;14) (p152;q22)mat	2			1	trisomy 14q
Raoul <i>et al.</i> (1976)	(15;21) (q262;q21)mat	3	1		1	trisomy 21q
Rethoré <i>et al.</i> (1973a)	(9;22) (q13;q13)mat				1	trisomy 9p
	(9;22) (p12;p11)pat				2	trisomy 9p

Table 1—continued

Authors	Chromosomes	Altern	Adj 1	Adj 2	3:1	Proband
Rethoré <i>et al.</i> (1973b)	(15;21) (q13;q221)mat			2		monosomy 21q
Rethoré <i>et al.</i> (1975)	(12;14) (q12;p11)mat	1			1	trisomy 15q
Rosenmann <i>et al.</i> (1978)	(13;18) (q32;q11)pat	1	1			trisomy 12p
Roux <i>et al.</i> (1974)	(10;22) (q24;q12)mat	2	1			trisomy 18q
Sanchez <i>et al.</i> (1977)	(20;21) (q13;q214)mat				1	trisomy 10q
Schinz <i>et al.</i> (1976)	(9;13) (p24;q22)pat	8	1			trisomy 20q
Schrott <i>et al.</i> (1974)	(4;13) (q26;q34)mat	9	1			trisomy 21q
Schrott <i>et al.</i> (1974)	(4;13) (q26;q34)mat	3	1			trisomy 13q
Schwanitz and Grosse (1973)	(4;22) (p14;p11)pat		2			trisomy 4p
Schwanitz <i>et al.</i> (1977)	(3;22) (q25;p11)mat	2	2			trisomy 4q
Short <i>et al.</i> (1972)	(9;14) (p2;q24)mat	1	2			trisomy 4p
Simpson and Zellweger (1977)	(3;14) (q23;q21)mat	10			1	trisomy 3q
Stoll <i>et al.</i> (1974)	(2;15) (p22;p11)mat	1	2		1	trisomy 14q
Subrt <i>et al.</i> (1976)	(9;21) (q13;q11)mat			1		trisomy 14q
Surana <i>et al.</i> (1977)	(3;22) (p2;q13)pat	1	1			trisomy 2p
Turleau <i>et al.</i> (1974)	(9;15) (q13;q25)mat	13			1	trisomy 9p
Turleau <i>et al.</i> (1975)	(12;14) (q244;q21)mat	3			1	trisomy 3p
Turleau and de Grouchy (1977)	(14;18) (p13;q21)mat		1			trisomy 9p
Turleau <i>et al.</i> (1977)	(14;18) (p12;q22)mat		2			trisomy 14q
Vogel <i>et al.</i> (1973)	(7;21) (q31;p13)mat		1			trisomy 18q
Wilroy (1977)	(13;17) (q13;p13)mat	1	2		1	monosomy 18q
Young <i>et al.</i> (1976)	(14;16) (q11;q24)mat				1	trisomy 7q
Yunis <i>et al.</i> (1976)	(10;21) (p11;p11)pat		1			trisomy 13q
Yunis <i>et al.</i> (1977a)	(15;16) (p12;q11)pat				1	trisomy 14q
Yunis <i>et al.</i> (1977b)	(4;15) (q11;p11)pat		1			trisomy 10p
Zabel and Baumann (1977)	(X;15) (p22;q15)mat		1			trisomy 16q
Zabel <i>et al.</i> (1978)	(5;22) (q33;q13)pat	2	1			trisomy 4q
Present case No 73.276	(9;21) (q22;q22)mat	4		2		trisomy 15q
						trisomy 5q
						trisomy 9p

Table 2 Translocations involving no acrocentric chromosome

Authors	Chromosomes	Altern	Adj 1	Adj 2	3:1	Proband
Abrisqueta <i>et al.</i> (1976)	(5;18) (p13;p11)mat	3	2			monosomy 5p
Andrie <i>et al.</i> (1976)	(4;7) (p12;qter)pat	6	1			trisomy 4p
	(4;7) (p12;qter)mat	6	1			trisomy 4p
Back <i>et al.</i> (1978)	(5;10) (p15;p11)mat	5	1			trisomy 10p
Ballesta and Vehi (1974)	(3;4) (p23;p16)mat		1			trisomy 3p
Bartsch-Sandhoff and Liersch (1977)	(5;8) (q33;p23)pat		2			trisomy 5q
Bauknecht <i>et al.</i> (1976)	(4;16) (p12;p13)pat		1			trisomy 4p
Berger <i>et al.</i> (1977)	(7;9) (q31;p24)mat	3	1			trisomy 7q
Biederman and Bowen (1976)	(2;4) (p25;q21)pat	3	1			trisomy 4q
Breuning <i>et al.</i> (1977)	(6;20) (p21;p13)pat	16	1			trisomy 6p
Canki <i>et al.</i> (1977)	(4;18) (q26;q23)mat	6	1			trisomy 4q
Cervenka <i>et al.</i> (1976)	(X;4) (q27;q25)mat	1	1			trisomy 4q
Chiyo <i>et al.</i> (1976)	(2;3) (p25;q21)mat	4	1			trisomy 3q
Dallapiccola <i>et al.</i> (1977)	(3;4) (p26;q13)mat				1	trisomy 4p
Dutrillaux <i>et al.</i> (1975)	(4;18) (q28;q22)mat		1			trisomy 4q
Fraisse <i>et al.</i> (1977)	(9;10) (q34;q24)pat	2	2			trisomy 10q
Fried <i>et al.</i> (1978)	(12;18) (q24;q21)mat	6			1	trisomy 18q
Garrett <i>et al.</i> (1975)	(1;4) (q25;q35)mat	5	1			trisomy 1q
Grace <i>et al.</i> (1973)	(3;7) (q27;q31)mat	1	1			trisomy 7q
Hoo (1976)	(9;12) (p24;p11)mat	1	1			trisomy 12p
Issa <i>et al.</i> (1976)	(4;9) (q31;q34)mat	2	1			trisomy 4q
Johnson and Bachman (1976)	(12;18) (qter;p11)mat		1			monosomy 18p
Johnson <i>et al.</i> (1977)	(7;10) (p22;p12)pat		1			trisomy 10p
Krøyer and Niebuhr (1975)	(10;18) (q25;q23)pat	15	1			trisomy 10q
Laurent <i>et al.</i> (1975b)	(11;17) (q231;p13)mat	2	1			trisomy 11q
Laurent <i>et al.</i> (1978)	(2;8) (q34;p23)pat, mat		6 mat, 1pat			trisomy 2q
Lurie <i>et al.</i> (1978)	(10;20) (p12;p12)mat	1	2			trisomy 10p
McDermott <i>et al.</i> (1977)	(4;5) (q32;p14)pat	2	1			monosomy 5p
Mason <i>et al.</i> (1975)	(1;9) (p36;q21)mat	1			1	trisomy 9p
Moirot <i>et al.</i> (1977)	(2;9) (q36;q31)mat				2	trisomy 9p
Moric-Petrovic <i>et al.</i> (1976a)	(4;7) (q35;q32)pat	2	2			trisomy 7q
Moric-Petrovic <i>et al.</i> (1976b)	(10;18) (p13;q23)pat	2	2			trisomy 10p
Mulcahy and Jenkyn (1975)	(8;9) (p22;p12)mat	3	1			trisomy 9p
Nakagome <i>et al.</i> (1973)	(6;18) (q2;q1)mat	5	2			trisomy 18q
Nielsen <i>et al.</i> (1977)	(8;12) (q24;q12)pat	1			1	trisomy 12p
Osztovcics and Kiss (1975)	(2;5) (p23;q31)pat	4	2			trisomy 5q
Pencaszadeh and Coco (1975)	(7;9) (q36;q13)mat				1	trisomy 9

Table 2—continued

Authors	Chromosomes	Altern	Adj 1	Adj 2	3:1	Proband
Penchaszadeh and Coco (1977)	(3;10) (q28;q11)mat				1	trisomy 10p
Prieur <i>et al.</i> (1975)	(10;17) (q24;q25)pat	1	1			trisomy 10q
Rethoré <i>et al.</i> (1973a)	(6;9) (q25;q11)mat	6			1	trisomy 9p
	(9;19) (p12;pter)mat	3	1			trisomy 9p
Rethoré <i>et al.</i> (1974)	(4;9) (q34;q21)mat	8			1	trisomy 9p
Rolland <i>et al.</i> (1977)	(4;10) (p16;p14)pat		1			trisomy 10p
	(10;12) (p11;q24)pat		1			trisomy 10p
Saadi and Moghadam (1976)	(5;7) (q35;q31)pat	4	1			trisomy 7q
Schwanitz <i>et al.</i> (1974)	(8;9) (q24;q22)mat	4			1	trisomy 9p
Sills <i>et al.</i> (1976)	(2;10) (q31;q24)pat	3	1			trisomy 1q
						monosomy 2q
Sparkes <i>et al.</i> (1977)	(4;20) (q25;q13)mat	2	1			trisomy 4q
Stern and Murch (1975)	(16;18) (p12;q11)mat	18			1	trisomy 18
Stoll <i>et al.</i> (1975)	(9;20) (q12;p13)mat	3			1	trisomy 9p
Taylor <i>et al.</i> (1976)	(18;20) (q23;p11)mat	3	2			trisomy 20p
Turleau <i>et al.</i> (1974)	(7;9) (q35;p13)mat	2	1			trisomy 9p
Turleau <i>et al.</i> (1976)	(7;18) (q333;q113)mat	2	1			trisomy 7q
Turleau <i>et al.</i> (1976)	(4;10) (q35;p11)mat	1	1			trisomy 10p
Turleau and de Grouchy (1977)	(8;18) (p23;q12)mat	2	1			trisomy 18q
Turleau <i>et al.</i> (1977)	(X;2) (p22;q32)mat	6	1			trisomy 2q
Wright <i>et al.</i> (1974)	(6;11) (p23;q22)pat		1			trisomy 11q
Yunis <i>et al.</i> (1977b)	(4;18) (q31;q23)mat	3	2			trisomy 4q
Yunis <i>et al.</i> (1978)	(4;5) (q32;p13)pat	10	2			trisomy 5p
Zabel <i>et al.</i> (1976)	(2;12) (q31;q24)mat	2	1			trisomy 2q
Zabel <i>et al.</i> (1978)	(3;5) (p27;p13)mat	1	1			trisomy 5p
Our case No 72.270	(5;18) (p12;p11)mat	4	2			monosomy 5p
Our case No 77.225	(5;10) (p13;p15)pat	4	3			monosomy 5p

Table 3 Segregation of 161 reciprocal translocations; study of 121 probands and affected relatives

	No of families	No of affected members
Adjacent 1	105	147
3:1	54	61
Adjacent 2	7	13

Table 4 Segregation of 98 translocations involving at least one acrocentric

Type	No of families	No of affected members
Adjacent 1	54	76
Adjacent 2	7	13
3:1	42	48

Table 5 Segregation of 63 translocations involving no acrocentric

Type	No of families	No of affected members
Adjacent 1	51	71
Adjacent 2	0	0
3:1	12	13

and an acrocentric, even though the long arms (Rethoré *et al.*, 1973b; Turleau *et al.*, 1974) were involved.

None of the 30 families whose reciprocal translocation involved the short arm of an acrocentric showed the disjunction adjacent 2.

A family incidence was noted 4 times out of 7 (Table 6). The distinction between two classes of

family, according to whether they have one case or more than one case of the same type of disjunction, allows a comparison of the repetitiveness of the three types, adjacent 1, adjacent 2, and 3:1 (Table 7). The disjunction adjacent 1 is more repetitive than the type 3:1 ($P < 0.02$). The disjunction adjacent 2 seems more repetitive than adjacent 1. The difference is not, however, significant in this small sample.

In addition, there are never adjacent 1 or 3:1 disjunctions in the families with adjacent 2 disjunction.

Finally for 12 of the 13 subjects, the adjacent 2 disjunction was derived from a maternal balanced translocation.

3:1 DISJUNCTIONS

These disjunctions (subjects with 45 or 47 chromosomes) were more frequent in group I (Table 4) than in group II (Table 5). They affected more subjects and more families ($P < 0.01$) and the acrocentrics involved were random. In group II, they were more frequent when the translocation affected a chromosome 9 (8 of 16 as opposed to 5 of 68 translocations not involving No 9) ($P < 0.001$) and their frequency in this case did not differ from that in group I. In group I, the incidence of 3:1 disjunction seemed also to be higher when a No 9 was involved (12 of 24 as opposed to 35 of 101 in the absence of No 9). This difference was not, however, statistically significant ($0.2 > P > 0.1$). The 3:1 disjunction resulted more often from a maternal translocation (55 times out of 61) than the adjacent 1 disjunction (97 times out of 147). The difference was significant ($P < 0.001$).

Table 6 7 families with adjacent 2 disjunction

Authors	Translocation	Segregation			Resulting abnormalities
		Adj 2	Adj 1	3:1	
Rethoré <i>et al.</i> (1973a)	(15;21) (q13;q22)mat	2	0	0	Partial trisomy 15 and partial monosomy 21
Laurent <i>et al.</i> (1973)	(14;21) (q12;q22)pat	1	0	0	Partial trisomy 14 and partial monosomy 21
Pena <i>et al.</i> (1976)	(13;14) (q13;q22)mat	2	0	0	Partial trisomy 14
Blank <i>et al.</i> (1975)	(9;22) (q13;q11)mat	4	0	0	Trisomy 9pter→9q13
Subrt <i>et al.</i> (1976)	(9;21) (q13;q11)mat	1	0	0	Trisomy 9pter→9q13
Howard-Peebles <i>et al.</i> (1977)	(9;13) (q22;q12)mat	1	0	0	Trisomy 9pter→9q22
Present case No 73.276	(9;21) (q22;q22)mat	2	0	0	Trisomy 9pter→9q22

Table 7 Repetitiveness of the 3 types of disjunction

	Adj 1	Adj 2	3:1	Total
Families with one case	74	3	47	124
Families with more than one case	30	4	7	41
Total	104	7	54	165

Moreover, the relative increase of the maternal origin in the imbalance was highest in the two types of translocation which often segregate according to the type 3:1: in group I, 111 times out of 137, as opposed to 54 times out of 84 for group II ($P < 0.01$); in group II, with the involvement of a No 9, 14 times out of 16.

Discussion

CHARACTERISTICS OF THE ADJACENT 2 DISJUNCTION

This type of disjunction has never been the subject of an in-depth study because of its rarity. The true incidence of such a rare phenomenon cannot be fully appreciated from this retrospective investigation, the method of evaluation of which is biased. The present study shows its occurrence in only 7 of the 161 families recorded. It involves only 13 of the 221 abnormal subjects of these families. Before the advent of chromosome banding, Hamerton (1971) studied 75 families and only came across this phenomenon twice (his results are not included in this work). These two translocations show these common characteristics: at least one acrocentric is involved; no other type of disjunction (adjacent 1 or 3:1) occurs in the family; in spite of its rarity, adjacent 2 disjunction is repetitive (3 times in the family described by Uchida *et al.*, 1964, and twice in the family described by Lord *et al.*, 1967); these translocations are maternal.

These characteristics can now be stated more precisely.

The nature of the chromosomes involved

Translocations likely to result in adjacent 2 disjunction occur between the long arms of two acrocentric chromosomes or between that of an acrocentric and that of a No 9. The interstitial segments of the chromosomes involved are generally short; they are not, they carry an extended heterochromatic zone, the 9qh.

The exclusive nature of the disjunction

Neither adjacent 1 nor 3:1 disjunction has been found in the families with adjacent 2 disjunction. However, the mutual exclusiveness of adjacent 2 and 3:1 disjunctions may only be related to the small sample size. In comparison, the disjunctions adjacent 1 and 3:1 exist together in a limited number of cases (6 out of 54 families 3:1).

The repetitive nature of the disjunction

Adjacent 2 disjunction does recur; this is the case in 4 families out of 7. This repetitiveness is highest in the family described by Blank *et al.* (1975) which contains 4 disjunctions of the adjacent 2 type.

The parental origins of the imbalance

Adjacent 2 disjunction is nearly always maternal. This feature is shared with the 3:1 disjunction but not with adjacent 1. Also, the imbalance, regardless of its type, is more often maternal in origin each time that the translocation involves at least one acrocentric chromosome or a No 9 which, as previously mentioned, produces the disjunctions adjacent 2 and 3:1.

POSSIBLE MECHANISMS INDUCING ADJACENT 2 IMBALANCE

The selection

In the family reported here the adjacent 2 disjunction does not produce duplication or deficiency of the translocated segments 9q and 21q. It results, therefore, in a moderate imbalance in contrast to that which might result from a disjunction of the type

adjacent 1 or 3:1. As a general rule, the adjacent 2 disjunctions produce duplication deficiency of the centric segments (arms not involved in the translocation and interstitial segments situated between the centromeres and the breakpoints). They do not affect the translocated segments (except for crossing-over in the interstitial segments).

This feature is different from that in the adjacent 1 disjunctions where, under the same conditions (absence of crossing-over in the interstitial segment), the opposite is observed. Only the translocated segments are imbalanced. It is also different from that in the 3:1 disjunctions where the trisomy or monosomy involves both the centric segments and the translocated segments.

The long centric segments and the short translocated segments therefore predispose, by selection, to adjacent 1 disjunctions. Conversely, adjacent 2 disjunctions are more likely to produce a viable constitution when the translocated segments are important and the centric segments short. These observations recall those of Burnham (1950) who, in *Zea mays*, showed a proportional relationship between the shortness of the interstitial segments and the frequency of adjacent 2 disjunction. However, the interstitial segment is only a part of the centric segment; the other part (the arm not involved in the translocation) may have the same selective role.

Since the disjunctions adjacent 1 and 2 have an opposite effect, this may explain why they never co-exist. If the translocated segments are short, type 1 is probable and type 2 would result in a lethal imbalance; the converse is true if these segments are long. The translocations involving the long arms of acrocentric chromosomes are therefore predisposed to this type of disjunction when breakpoints are juxtacentromeric because only imbalance of their short arms is produced. Similarly, when a chromosome 9 is involved, it is by its long arms and breakpoints lie at q13 or q21-22, never beyond. In these conditions the disjunction results in trisomy of the region 9p and 9q proximal, segments whose duplication is not lethal.

These points do not explain why other chromosomes (X or Y), or other chromosome regions whose duplication or deficiency is not lethal, are never involved in these translocations. They also do not explain why adjacent 2 and 3:1 have not, as yet, been seen in the same family.

Meiotic configurations

Since this type of disjunction is determined at the time of the first meiotic division, the pachytene meiotic configurations are to be considered.

In this respect, two of the factors already mentioned as influencing the adjacent 2 disjunction by

selection appear to favour this segregation through the meiotic configuration: shortness of the interstitial segment and participation of the acrocentric chromosomes.

- (a) The shortness of the two interstitial segments, or rather the absence of crossing-over in them, may cause an error in tetravalent positioning which induces segregation to the same pole of the two homologous centromeres. This hypothesis is supported by the observation that the occurrence of crossing-over in the interstitial segments results in regular anaphase disjunction of homologous centromeres, thus preventing adjacent 2 disjunction (McClintock, 1945, on *Neurospora crassa*). The same observation has been made by Burnham (1950) with *Zea mays*. In four cases the adjacent 2 disjunction affects translocations with a long interstitial segment, but it is the heterochromatic 9qh which is likely to be achiasmatic, as suggested by the stability of the pericentric inversions of this region (Madan and Bobrow, 1974).
- (b) The involvement of at least a pair of acrocentrics at the meiotic quadrivalent seems to be a determinant. Besides, it is necessary that the acrocentric be translocated by its long arm.

Under these conditions the tetrad shows one very short terminal segment which corresponds to the short arms of the acrocentrics. In this region, probably achiasmatic (Hulten, 1974), the tetravalent opens. Therefore, the complex formed is not circular; it is a chain, the two ends of which are occupied by the short arms of the acrocentrics. Lewis and John (1963) have shown in *Chorthippus bruneus*, an orthoptera, that this configuration, called type II chain (to differentiate it from that of type I, where the achiasmatic segment is the translocated segment), produced adjacent 2 disjunctions with a high frequency but never adjacent 1. The analogy between this observation and that which takes place in man has already been suggested by Hamerton (1971).

CHARACTERISTICS OF THE 3:1 DISJUNCTION

The sampling is too biased to allow an evaluation of the frequency of this type of disjunction or to estimate, even roughly, a recurrence risk. For example, observations on 3:1 disjunctions, which are partially pure monosomies or trisomies, are probably more often published than those cases of adjacent 1 disjunction which are duplication deficiencies.

According to Lindenbaum and Bobrow (1975) 3:1 disjunction is rare and represents 6 to 25% of the total of the imbalances caused by reciprocal translocations.

The influencing factors on this type of disjunction

have been studied by Hamerton (1971) and Lindenbaum and Bobrow (1975). Only two points are considered here.

The nature of the chromosomes involved in the translocation

An acrocentric chromosome takes part most often in the translocation (50 out of 61 cases showing disjunction 3:1). Among these acrocentrics, those of G group are not more often involved than those of D group (22 G and 23 D). In 1975, Lindenbaum and Bobrow observed from their sample that 12 of the 25 C group chromosomes involved in 3:1 disjunctions were No 9, and they suggested a possible influence of this chromosome on the frequency of this type of disjunction. The present study confirms this hypothesis: in group II (translocations without acrocentrics) the incidence of 3:1 disjunction differs according to whether a chromosome 9 is or is not involved in the translocation. In the first case it is 50% and in the second only 7%.

The parental origin of the translocation

Of the 61 subjects, 55 (90.2%) with a 3:1 imbalance are the offspring of heterozygous women. For 3:1 and adjacent 2 disjunctions together, a maternal origin is observed 67 times out of 74 (90.6%). By comparison, the origin of the adjacent 1 imbalance is maternal in only 97 out of 147 cases (65%).

This maternal origin is observed in 81% of the two types of translocation which often segregate according to type 3:1 (group I and group II with involvement of a No 9). This is significantly reduced when neither an acrocentric nor a No 9 are involved in the translocation (58%).

We thank Miss E. Blanc, Miss G. Chatain, Miss D. Chmara, and Mrs. C. Ostorero for cytological assistance, Mrs M. J. Bossan and Mrs F. Courty for typing the manuscript, and Mr J. Gilliotte for photographic assistance.

References

- Abrisqueta, J. A., Perez, A., Aller, V., Del Mazo, J., Goday, C., Martin, M. A., and De Torres, M. L. (1976). Cri du chat syndrome and translocation $t(5p-;18p+)$. *Journal de Génétique Humaine*, **24**, 173-182.
- Abuelo, D., Peri, D. P., Henkle, C., and Richardson, A. (1977). Partial trisomy 8 (trisomy $8q2106 \rightarrow 8qter$). *Journal of Medical Genetics*, **14**, 463-465.
- Alfi, O. S., Donnell, G. N., and Kramer, S. L. (1973). Partial trisomy of the long arm of chromosome no 7. *Journal of Medical Genetics*, **10**, 187-189.
- Andrie, M., Erlach, A., Killian, W., and Rett, A. (1976). Two cases of trisomy 4p with translocation $t(4p-;7q+)$ in several members of one family. *Human Genetics*, **33**, 155-160.
- Archidiacono, N., Rocchi M., de Vonderweid, U., and Filippi, G. (1978). $t(9/22)$ with centric fission and per centric translocation leading to a case of pure 9p trisomy in the offspring. *Human Genetics*, **40**, 325-331.
- Armendares, S., and Salamanca-Gomez, F. (1978). Partial 2p trisomy ($p21 \rightarrow pter$) in two siblings of a family with a $2p-;15q+$ translocation. *Clinical Genetics*, **13**, 17-24.
- Armendares, S., Salamanca-Gomez, F., Nava, S., Ramires, S., and Cantu, J. M. (1975). The 12p trisomy syndrome. *Annales de Génétique*, **18**, 89-94.
- Aurias, A., Turc, Cl., Michiels, Y., Sinet, P. M., Graveleau, D., and Lejeune, J. (1975). Deux cas de trisomie 11q ($q231 \rightarrow qter$) par translocation $t(11;22)(q231;q111)$ dans deux familles différentes. *Annales de Génétique*, **18**, 185-188.
- Ayraud, N., Galiana, A., Llyod, M., and Deswarte, M. (1976). Trisomie 11q ($q231 \rightarrow qter$) par translocation maternelle $t(11;22)(q231;q111)$. Une nouvelle observation. *Annales de Génétique*, **19**, 65-68.
- Baccichetti, C., Tenconi, R., Anglani, F., and Zacchello, F. (1975). Trisomy $4q32 \rightarrow 4qter$ due to a maternal 4p21 translocation. *Journal of Medical Genetics*, **12**, 425-427.
- Back, E., Vogel, W., Hertel, C., and Schuchmann, L. (1978). Trisomy 10p due to $t(5;10)(p15;p11)$ segregating in a large sibship. *Human Genetics*, **41**, 11-17.
- Balicek, P., Zizka, J., and Lichy, J. (1975). A case of trisomy 9p in a family with translocation $9/15$. *Humangenetik*, **23**, 353-358.
- Ballesta, F., and Vehi, L. (1974). Trisomie partielle pour la partie distale du bras court du chromosome 3. *Annales de Génétique*, **17**, 287-290.
- Bartsch-Sandhoff, M., and Liersch, R. (1977). Partial duplication 5q syndrome: phenotypic similarity in two sisters with identical karyotype (partial duplication $5q33 \rightarrow 5qter$ and partial deficiency $8p23 \rightarrow pter$). *Annales de Génétique*, **20**, 281-284.
- Bass, H. N., Sparkes, R. S., Crandall, B. F., Galos, R., and Howard, J. (1978). A 45,XX,-5,-14,+t(5q;14q)mat cri du chat child. *Annales de Génétique*, **21**, 56-59.
- Bauknecht, Th., Betteken, F., and Vogel, W. (1976). Trisomy 4p due to a paternal $t(4p-;16p+)$ translocation. *Human Genetics*, **34**, 227-230.
- Bergamo, F., Crosato, F., Francesconi, D., Pasquali, F., and Zuffardi, O. (1977). The 9p- deletion syndrome. A patient with a 45,XX,-9,-15,+t(9/15) constitution due to a maternal 3:1 meiotic disjunction. *Clinical Genetics*, **11**, 219-223.
- Berger, R., Turc, C., Wachter, H., and Begue, G. (1977). Partial 7q trisomy. *Clinical Genetics*, **11**, 39-42.
- Biederman, B., and Bowen, P. (1976). Partial trisomy 4q due to familial 2/4 translocation. *Human Genetics*, **33**, 147-153.
- Biederman, B., Bowen, P., Robertson, C., and Schiff, J. (1977). Partial trisomy 12p due to $t(12;21)$ pat translocation. *Human Genetics*, **36**, 35-41.
- Blank, C. E., Colver, D. C. B., Potter, A. M., McHugh, J., and Lorber, J. (1975). Physical and mental defect of chromosomal origin in four individuals of the same family. Trisomy for the short arm of 9. *Clinical Genetics*, **7**, 261-273.
- Borgaonkar, D. S., McKusick, V. A., and Farber, P. A. (1973). An inherited small extra chromosome: a mother with 46,XX,t(17;22)(p1;q1) and a son with 47,XY,+der(22)mat. *Journal of Medical Genetics*, **10**, 379-383.
- Breuning, M. H., Bijlsma, J. B., and de France, H. F. (1975). Partial trisomy 6p due to familial translocation $t(6;20)(p21;p13)$. A new syndrome? *Human Genetics*, **38**, 7-13.
- Brimblecombe, F. S. W., Lewis, F. J., and Vowles, M. (1977). Complete 5p trisomy: 1 case and 19 translocations in 6 generations. *Journal of Medical Genetics*, **14**, 271-275.

- Burnham, C. R. (1950). Chromosome segregation in translocations involving chromosome 6 in maize. *Genetics*, **35**, 446-481.
- Canki, N., Debevec, M., Rainer, S., and Rethoré, M. O. (1977). Trisomie 4q26→4qter par translocation t(4;18) (q26;q23)mat. *Annales de Génétique*, **20**, 195-198.
- Cantu, J. M., Salamanca-Gomez, F., Buentello, L., Carnevale, A., and Armendares, S. (1975). Trisomy 10p. A report of two cases due to a familial translocation rcp(10;21) (p11;p11). *Annales de Génétique*, **18**, 5-11.
- Castel, Y., Riviere, D., Boucly, J. Y., and Toudic, L. (1976). Trisomie 15q partielle par translocation maternelle t(7;15) (q35;q14). *Annales de Génétique*, **19**, 75-79.
- Centerwall, W., and Francke, U. (1977). Familial trisomy 20p. Five cases and two carriers in three generations. A review. *Annales de Génétique*, **20**, 77-83.
- Centerwall, W. R., Miller, K. S., and Reeves, L. M. (1976). Familial 'partial 9p' trisomy: six cases and four carriers in three generations. *Journal of Medical Genetics*, **13**, 57-61.
- Cervinka, J., Djavadi, G. R., and Gorlin, R. J. (1976). Partial trisomy 4q syndrome: case report and review. *Human Genetics*, **34**, 1-7.
- Chaganti, R. S. K., Morillo-Cucci, G., Friis, L., Degnan, M., and German, J. (1976). De novo appearance of a translocation t(5p;21q) and its transmission in both balanced and unbalanced forms to the next generation. *Annales de Génétique*, **19**, 43-48.
- de la Chapelle, A., Koivisto, M., and Schroder, J. (1973). Segregating reciprocal (4;21) (q21;q21) translocation with proposita trisomic for parts of 4q and 21. *Journal of Medical Genetics*, **10**, 384-389.
- Chiyo, H. A., Nakagome, Y., Matsui, I., Kuroki, Y., Kobayashi, H., and Ono, K. (1975). Two cases of 8p trisomy in one sibship. *Clinical Genetics*, **7**, 328-333.
- Chiyo, H. A., Kuroki, Y., Matsui, I., Niitsu, N., and Nakagome, Y. (1976). A case of partial trisomy 3q. *Journal of Medical Genetics*, **13**, 525-527.
- Coco, R., and Penchaszadeh, V. B. (1977). Partial trisomy 14q and familial translocation (2;14) (q12;q13). *Annales de Génétique*, **20**, 41-44.
- Cohen, M. M., Davidson, R. G., and Brown, J. A. (1975a). A familial F/G translocation t(20p-;22q+) observed in three generations. *Clinical Genetics*, **7**, 120-127.
- Cohen, M. M., Ornoy, A., Rosenmann, A., and Kohn, G. (1975b). An inherited translocation t(14;15) (p16;q22) leading to two cases of partial trisomy 15. *Annales de Génétique*, **18**, 99-103.
- Dallapiccola, B., Mastroiacovo, P. P., Montali, E., and Sommer, A. (1977). Trisomy 4p: five new observations and overview. *Clinical Genetics*, **12**, 344-356.
- Darmady, J. M., and Seabright, M. (1975). A family with an inherited translocation involving the No 4/No 21 chromosomes. *Journal of Medical Genetics*, **12**, 408-411.
- Dutrillaux, B., Laurent, C., Forabosco, A., Noel, B., Suerinc, E., Biemont, M. Cl., and Cotton, J. B. (1975). La trisomie 4q partielle. A propos de 3 observations. *Annales de Génétique*, **18**, 21-27.
- Forabosco, A., Dutrillaux, B., Toni, G., Tamborino, G., and Cavazzutti, G. (1973). Translocation équilibrée t(2;13) (q32;q33) familiale et trisomie 2q partielle. *Annales de Génétique*, **16**, 255-258.
- Ford, C. E., and Clegg, H. M. (1969). Reciprocal translocations. *British Medical Bulletin*, **25**, 110-114.
- Fraisse, J., Laurus, B., Laselve, A., and Freycon, F. (1977). Deux nouveaux cas de trisomie 10q24→10qter chez deux sœurs par translocation paternelle t(9;10) (q34;q24). *Annales de Génétique*, **20**, 128-131.
- Fried, K., Bar-Yochai, A., Rosenblatt, M., and Mundel, G. (1978). Partial 18 trisomy (with 47 chromosomes) resulting from a familial maternal translocation. *Journal of Medical Genetics*, **15**, 76-78.
- Fried, K., Tieder, M., Beer, S., Rosenblatt, M., and Krespin, H. I. (1977). Mental retardation with 45 chromosomes, 45,XX,-5,-14,+der(5)t(5;14) (p15;q13)mat due to familial balanced reciprocal translocation. *Journal of Medical Genetics*, **14**, 68-72.
- Fryns, J. P., Van Eygen, M., Tanghe, W., and Van Den Berghe, H. (1977). Partial trisomy 14q due to familial t(14q-;11q+)translocation. *Human Genetics*, **37**, 105-110.
- Fu, W. N., Borgaonkar, D. S., Ladewig, P. P., Weaver, J., and Pomerance, H. H. (1976). Structural aberrations of the long arm of chromosome no 22. Report of a family with translocation t(11;22) (q25;q11). *Clinical Genetics*, **10**, 329-336.
- Garlinger, P., McGeary, S. A., and Magenis, E. (1977). Partial trisomy 22: a recognizable syndrome. *Clinical Genetics*, **12**, 9-16.
- Garrett, J. H., Finley, S. C., and Finley, W. H. (1975). Fetal loss and familial chromosome 1 translocations. *Clinical Genetics*, **8**, 341-348.
- Genest, P., Roussy, J., Genest, F. B., and Dumas, L. (1977). La trisomie 9p. Un cas de translocation t(9;22) (p11;p11) mat chez une fillette. *Union Médicale du Canada*, **106**, 956-959.
- Giovannelli, G., Forabosco, A., and Dutrillaux, B. (1974). Translocation familiale t(4;22) (p11;p12) et trisomie 4p chez deux germains. *Annales de Génétique*, **17**, 119-124.
- Giraud, F., Mattei, J. F., and Mattei, M. G. (1977). Trisomie 13 partielle par translocation t(2;13) maternelle. *Annales de Génétique*, **20**, 203-208.
- Grace, E., Sutherland, G. R., Stark, G. D., and Bain, A. D. (1973). Partial trisomy of 7q resulting from a familial translocation. *Annales de Génétique*, **16**, 51-54.
- Habedank, M., and Faust, J. (1978). Trisomy 9p and unusual translocation mongolism in siblings due to different 3:1 segregations of maternal translocation rcp(9;21) (p11;q11). *Human Genetics*, **42**, 251-256.
- Hamerton, J. (1971). Chromosome translocations. In *Human Cytogenetics*, Vol. 1, pp. 247-272. Academic Press, New York.
- Hansteen, I. L., Schirmer, L., and Hestetun, S. (1978). Trisomy 12p syndrome. Evaluation of a family with a t(12;21) (p12.1;p11) translocation with unbalanced offspring. *Clinical Genetics*, **13**, 339-349.
- Hobolth, N., Jacobsen, P., and Mikkelsen, M. (1974). Partial trisomy 12 in a mentally retarded boy and translocation (12;21) in his mother. *Journal of Medical Genetics*, **11**, 299-302.
- Hoo, J. J. (1976). 12p trisomy: a syndrome? *Annales de Génétique*, **19**, 261-263.
- Howard-Peebles, P. N., Yarbrough, K. M., Stoddard, G. R., and Rary, J. M. (1977). Translocation 9q/13q resulting in duplication (trisomy 9pter→9q22) and deficiency (monosomy 13pter→13q12). *Clinical Genetics*, **11**, 46-52.
- Hulten, M. (1974). Chiasma distribution at diakinesis in the normal human male. *Hereditas*, **76**, 55-78.
- Hustinx, Th. W. J., Gabreels, F. J. M., Kirkels, V. G. H. J., Korten, J. J., Scheres, J. M. J. C., Joosten, E. M. G., and Ruten, F. J. (1975). Trisomy 4p in a family with a t(4;15). *Annales de Génétique*, **18**, 13-19.
- Hustinx, Th. W. J., Ter Haar, B. G. A., Scheres, J. M. J. C., and Ruten, F. J. (1974). Trisomy for the short arm of chromosome no 10. *Clinical Genetics*, **6**, 408-415.
- Issa, M., Potter, A. M., and Blank, C. E. (1976). Multiple congenital defects associated with trisomy for long arm of no 4. *Journal of Medical Genetics*, **13**, 326-329.

- Jenkins, M. B., and Boyd, L. (1976). Reciprocal translocation, 4q-;21p+, giving rise to Down's syndrome. *Journal of Medical Genetics*, 13, 323-326.
- Johnson, G., and Bachman, R. (1976). A 46,XY,del(18)(pter→p1100) cebocephalic child from a 46,XX,t(12;18)(18pter→18p1100::12qter→12pter) normal parent. *Human Genetics*, 34, 103-106.
- Johnson, G., Bachman, R., Roed, T., and Riddervold, P. (1977). Partial trisomy 10p and familial translocation t(7;10)(p22;p12). *Human Genetics*, 35, 353-356.
- Jotterand, M., and Juillard, E. (1976). A new case of trisomy for the distal part of 13q due to maternal translocation t(9;13)(p21;q21). *Human Genetics*, 33, 213-222.
- Kajii, T., Meylan, J., and Mikamo, K. (1974). Chromosome anomalies in three successive abortuses due to paternal translocation t(13q-18q+). *Cytogenetics and Cell Genetics*, 13, 426-436.
- Kessel, E., and Pfeiffer, R. A. (1977). 47,XY,+der(11;22)(q23;q12) following balanced translocation t(11;22)(q23;q12)mat. Remarks on the problem of trisomy 22. *Human Genetics*, 37, 111-116.
- Kim, H. J., Hsu, L. Y. F., Goldsmith, L. C., Strauss, L., and Hirschhorn, K. (1977). Familial translocation with partial trisomy of 13 and 22: evidence that specific regions of chromosomes 13 and 22 are responsible for the phenotype of each trisomy. *Journal of Medical Genetics*, 14, 114-119.
- Krøyer, S., and Niebuhr, E. (1975). Partial trisomy 10q occurring in a family with a reciprocal translocation t(10;18)(q25;q23). *Annales de Génétique*, 18, 50-55.
- Lange, M., and Alfi, O. S. (1976). Trisomy 19q. *Annales de Génétique*, 19, 17-21.
- Larson, L. M., Wasdahl, W. A., and Jalal, S. M. (1977). Partial trisomy 7p associated with familial 7p;22q translocation. *Journal of Medical Genetics*, 14, 258-261.
- Laurent, C., Biemont, M. Cl., Bethenod, M., Cret, L., and David, M. (1975a). Deux observations de trisomie 11q (q231→qter) avec la même anomalie des organes génitaux externes. *Annales de Génétique*, 18, 179-184.
- Laurent, C., Biemont, M. Cl., Cognat, M., and Dutrillaux, B. (1975b). Studies of the meiotic behaviour of a translocation t(10;13)(q25;q11) in an oligospermic man. *Human Genetics*, 39, 123-126.
- Laurent, C., Biemont, M. Cl., Guibaud, P., Guillot, J., Noel, B., Quack, B., Genevieve, M., and Cressens, M. L. (1978). Sept cas de trisomie 2q34→2qter par transmission familiale d'une translocation t(2;8)(q34;p23). *Annales de Génétique*, 21, 13-18.
- Laurent, C., Dutrillaux, B., Biemont, M. Cl., Genoud, J., and Bethenod, M. (1973). Translocation t(14q-;21q+) chez le père, trisomie 14 et monosomie 21 partielles chez la fille. *Annales de Génétique*, 16, 281-284.
- Lejeune, J., Dutrillaux, B., and de Grouchy, J. (1970). Reciprocal translocations in human populations. A preliminary analysis. In *Human Population Cytogenetics*, pp. 81-87, ed P. A. Jacobs, W. H. Price, and P. Law. University of Edinburgh Press, Edinburgh.
- Lewis, K. R., and John, B. (1963). Spontaneous interchange in *Chorthippus brunneus*. *Chromosoma*, 14, 618-637.
- Lindenbaum, R. H., and Bobrow, M. (1975). Reciprocal translocations in man. 3:1 meiotic disjunction resulting in 47 or 45 chromosome offspring. *Journal of Medical Genetics*, 12, 29-43.
- Lord, P. M., Casey, M. D., and Laurance, B. M. (1967). A new translocation between chromosomes in the 6-12 and 21-22 groups. *Journal of Medical Genetics*, 4, 169-176.
- Lurie, I. W., Lazjuk, G. I., Gurevich, D. B., Kravtsova, G. I., Nedzved, M. K., and Shved, I. A. (1978). Partial trisomy 10p in two generations. *Human Genetics*, 41, 235-241.
- McClintock, B. (1945). Neurospora. I. Preliminary observations of the chromosomes of *Neurospora crassa*. *American Journal of Botany*, 32, 671-678.
- McDermott, A., Poulding, R., and Creery, D. (1977). Cri-du-chat syndrome in a child with a 46,XX,der(5),t(4;5)(q32;p14)pat karyotype. *Human Genetics*, 39, 109-112.
- Madan, K., and Bobrow, M. (1974). Structural variations in chromosome no 9. *Annales de Génétique*, 17, 81-86.
- Mason, M. K., Spencer, D. A., and Rutter, A. (1975). A case of partial (9p) trisomy in a family with a balanced translocation 46,XX,t(1p+9q-). *Journal of Medical Genetics*, 12, 310-315.
- Moirot, H., Fessard, Cl., and Hemet, J. (1977). Trisomie 2p par t(2;9)(q36;q31)mat. *Annales de Génétique*, 20, 45-51.
- Moric-Petrovic, S., Laca, Z., and Krajgher, A. (1976a). Translocation réciproque dans la famille de deux porteurs avec une trisomie partielle du chromosome 9. *Annales de Génétique*, 19, 133-136.
- Moric-Petrovic, S., Laca, Z., Krajgher, A., and Milosevic, J. (1976b). Deux cas de trisomie 10p partielle dus à une translocation paternelle t(10;18)(p13;q23). *Annales de Génétique*, 19, 195-197.
- Mulcahy, M. T., and Jenkyn, J. (1975). The 9p trisomy syndrome: two further cases arising from different familial translocations. *Clinical Genetics*, 8, 199-204.
- Nakagome, Y., Icnuma, K., and Matsui, I. (1973). Three translocations involving C or G group chromosomes. *Journal of Medical Genetics*, 10, 174-177.
- Nakagome, Y., and Kobayashi, H. (1975). Trisomy of the short arm of chromosome 10. *Journal of Medical Genetics*, 12, 412-414.
- Niazi, M., Coleman, D. V., and Saldana-Garcia, P. (1978). Partial trisomy 18 in a family with a translocation t(8;21)(q21;q22). *Journal of Medical Genetics*, 15, 148-151.
- Nielsen, J., Rasmussen, K., Lassen, L. B., and Christiansen, F. (1976). A family with a high risk of segregation for an autosomal unbalanced reciprocal translocation. *Human Genetics*, 32, 343-348.
- Nielsen, J., Vetter, M., Holm, V., Askjaer, S. A., and Reske-Nielsen, E. (1977). A newborn child with karyotype 47,XX,+der(12)(12pter→12q12::8q24→8qter), t(8;12)(q24;q12)pat. *Human Genetics*, 35, 357-362.
- Noel, B., Levy, M., and Rethoré, M. O. (1976a). Trisomie partielle du bras long du chromosome 11 par malsegregation d'une translocation maternelle t(11;22)(q231;q11). *Annales de Génétique*, 19, 137-139.
- Noel, B., Quack, B., and Rethoré, M. O. (1976b). Partial deletions and trisomies of chromosome 13; mapping of bands associated with particular malformations. *Clinical Genetics*, 9, 593-602.
- Orye, E., Verhaaren, H., and Van Den Bogaert-Van Heesvelde, A. M. (1975). The 9p- deletion syndrome. Report of a patient with a 46,XX,9p- constitution due to a paternal t(9p-;15q+) translocation. *Clinical Genetics*, 8, 349-357.
- Osztovcics, M., and Kiss, P. (1975). Familial translocation t(2;5)(p23;q31). *Clinical Genetics*, 8, 112-116.
- Owen, L., Martin, B., Blank, C. E., and Harris, F. (1974). Multiple congenital defects associated with trisomy for the short arm of chromosome 4. *Journal of Medical Genetics*, 11, 291-296.
- Pena, S. D. J., Ray, M., McAlpine, P. J., Ducasse, J., Briggs, J., and Hamerton, J. L. (1976). Tertiary trisomy 4: is there a syndrome? *Birth Defects: Original Article Series*, 12, 113-118.
- Penchaszadeh, V. B., and Coco, R. (1975). Partial 9 trisomy by 3:1 segregation of balanced maternal translocation (7q+;9q-). *Journal of Medical Genetics*, 12, 301-304.

- Penchaszadeh, V. B., and Coco, R. (1977). Trisomy for the short arm of chromosome no 10. *Journal de Génétique Humaine*, **25**, 221-227.
- Pfeiffer, R. A., and Kessel, E. (1976). Partial trisomy 15q1. *Human Genetics*, **33**, 77-83.
- Pfeiffer, R. A., Kessel, E. K., and Soer, K. M. (1977). Partial trisomies of chromosome 21 in man. Two new observations due to translocations 19;21 and 4;21. *Clinical Genetics*, **11**, 207-213.
- Philippe, N., Requin, C., Germain, D., Licheron, A., and Hermier, M. (1975). Trisomie 9p par translocation maternelle (9;22) (p11;q11). *Journal de Génétique Humaine*, **23**, 309-317.
- Podruch, P. E., and Weisskopf, B. (1974). Trisomy for the short arms of chromosome 9 in two generations, with balanced translocations t(15p+;9q-) in three generations. *Journal of Pediatrics*, **85**, 92-95.
- Prieur, M., Forabosco, A., Dutrillaux, B., Laurent, C., Bernasconi, S., and Lejeune, J. (1975). La trisomie 10q24→10qter. *Annales de Génétique*, **18**, 217-222.
- Raoul, O., Carpentier, S., Dutrillaux, B., Mallet, R., and Lejeune, J. (1976). Trisomies partielles du chromosome 21 par translocation maternelle t(15;21) (q262;q21). *Annales de Génétique*, **19**, 187-190.
- Raoul, O., Rethoré, M. O., Dutrillaux, B., Michon, L., and Lejeune, J. (1975). Trisomie 14q partielle. I. Trisomie 14q partielle par translocation maternelle t(10;14) (p152;q22). *Annales de Génétique*, **18**, 35-39.
- Rethoré, M. O., Dutrillaux, B., and Lejeune, J. (1973a). Translocation 46,XX,t(15;21) (q13;q22,1) chez la mère de deux enfants atteints de trisomie 13 et de monosomie 21 partielles. *Annales de Génétique*, **16**, 271-275.
- Rethoré, M. O., Ferrand, J., Dutrillaux, B., and Lejeune, J. (1974). Trisomie 9p par t(4;9) (q34;q21)mat. *Annales de Génétique*, **17**, 157-161.
- Rethoré, M. O., Hoehn, H., Rott, H. D., Couturier, J., Dutrillaux, B., and Lejeune, J. (1973b). Analyse de la trisomie 9p par dénaturation ménagée. A propos d'un nouveau cas. *Humangenetik*, **18**, 129-138.
- Rethoré, M. O., Kaplan, J. C., Junien, C., Cruveiller, J., Dutrillaux, B., Aurias, A., Carpentier, S., Lafourcade, J., and Lejeune, J. (1975). Augmentation de l'activité de la LDH-B chez un garçon trisomique 12p par malségrégation d'une translocation maternelle t(12;14) (q12;p11). *Annales de Génétique*, **18**, 81-87.
- Rolland, M., Bourrouillou, G., Elana, G., Colombies, P., and Regnier, C. (1977). Trisomie 10p partielle d'origine paternelle. Deux nouvelles observations dans deux familles différentes. *Annales de Génétique*, **20**, 209-213.
- Rosenmann, A., Isacson, M., Cohen, R., Segal, M., and Cohen, M. M. (1978). Partial trisomy 18 (q11→qter) in an infant and aborted fetus resulting from a balanced paternal translocation t(13;18) (q32;q11). *Annales de Génétique*, **21**, 60-64.
- Roux, C., Taillemite, J. L., and Baheux-Morlier, G. (1974). Trisomie partielle 10q par translocation familiale t(10q-; 22p+). *Annales de Génétique*, **17**, 59-62.
- Saadi, A. A., and Moghadam, H. A. (1976). Partial trisomy of the long arm of chromosome 7. *Clinical Genetics*, **9**, 250-254.
- Sanchez, O., Mamunes, P., and Yunis, J. J. (1977). Partial trisomy 20 (20q13) and partial trisomy 21 (21pter→21q213). *Journal of Medical Genetics*, **14**, 459-462.
- Schinzel, A., Hayashi, K., and Schmid, W. (1976). Further delineation of the clinical picture of trisomy for the distal segment of chromosome 13. Report of three cases. *Human Genetics*, **32**, 1-12.
- Schrott, H. G., Sakaguchi, S., Francke, U., Luzzatti, L., and Fialkow, P. J. (1974). Translocation t(4q-;13q+) in three generations resulting in partial trisomy of the long arm of chromosome 4 in the fourth generation. *Journal of Medical Genetics*, **11**, 201-205.
- Schwanitz, G., and Grosse, K. P. (1973). Partial trisomy 4p with translocation 4p-, 22p+ in the father. *Annales de Génétique*, **16**, 263-266.
- Schwanitz, G., Schamberger, U., Rott, H. D., and Wieczorec, V. (1974). Partial trisomy 9 in the case of familial translocation 8/9 mat. *Annales de Génétique*, **17**, 163-166.
- Schwanitz, G., Schmid, R. D., Grosse, G., and Grahn-Liebe, E. (1977). Translocation familiale 3/22 mat avec trisomie partielle 3q. *Journal de Génétique Humaine*, **25**, 141-150.
- Short, E. M., Solitare, G. B., and Breg, W. R. (1972). A case of partial 14 trisomy. 47,XY,(14q-)+ and translocation t(9p+;14q-) in mother and brother. *Journal of Medical Genetics*, **9**, 367-373.
- Sills, J. A., Buckton, K. E., and Raeburn, J. A. (1976). Severe mental retardation in a boy with partial trisomy 10q and partial monosomy 2q. *Journal of Medical Genetics*, **13**, 507-510.
- Simpson, J., and Zellweger, H. (1977). Partial trisomy 14q- and parental translocation of no 14 chromosome. Report of a case and review of the literature. *Journal of Medical Genetics*, **14**, 124-127.
- Spaikes, R. S., Francke, U., Muller, H., and Toomey, K. (1977). Partial 4q duplication due to inherited der(20), t(4;20) (q25;q13)mat. *Annales de Génétique*, **20**, 31-35.
- Stern, L. M., and Murch, A. R. (1975). Pseudohermaphroditism with clinical features of trisomy 18 in an infant trisomic for parts of chromosomes 16 and 18: 47,XY,der (18),t(16;18) (p12;q11)mat. *Journal of Medical Genetics*, **12**, 305-308.
- Stoll, C., Levy, J. M., and Gardea, A. (1975). Trisomy 9p in a girl whose mother has a translocation t(9;20) (q12;p13). *Humangenetik*, **27**, 269-274.
- Stoll, C., Messer, J., and Vors, J. (1974). Translocation t(2;14) équilibrée chez une mère et trisomie partielle d'une partie du bras court d'un chromosome no 2 chez deux de ses enfants. *Annales de Génétique*, **17**, 193-196.
- Subrt, I., Blehova, B., and Pallova, B. (1976). Trisomy 9p resulting from maternal 9/21 translocation. *Human Genetics*, **32**, 217-220.
- Surana, R. B., Braudo, M. E., Conen, P. E., and Slade, R. H. (1977). 46,XY,t(3;22) (p2;q13) resulting in partial trisomy for the short arm of chromosome 3. *Clinical Genetics*, **11**, 201-206.
- Taylor, K. M., Wolfinger, H. L., Brown, M. G., Chadwick, D. L., and Francke, U. (1976). Partial trisomy 20p derived from a t(18;20) translocation. *Human Genetics*, **34**, 155-162.
- Turleau, C., Chavin-Colin, F., de Grouchy, J., Repesse, G., and Beauvais, P. (1977). Familial t(X;2) (p223;q323) with partial trisomy 2q and male and female balanced carriers. *Human Genetics*, **37**, 97-104.
- Turleau, C., and de Grouchy, J. (1977). Trisomy 18qter and trisomy mapping of chromosome 18. *Clinical Genetics*, **12**, 361-371.
- Turleau, C., de Grouchy, J., Bocquentin, F., Roubin, M., and Chavin-Colin, F. (1975). Trisomie 14q partielle. II. Trisomie 14q partielle par translocation maternelle (12;14) (q244;q21). *Annales de Génétique*, **18**, 41-44.
- Turleau, C., de Grouchy, J., Chavin-Colin, F., Roubin, M., and Langmaid, H. (1974). Trisomie 9p: deux nouvelles observations. *Annales de Génétique*, **17**, 167-174.
- Turleau, C., Doussau de Bazignan, M., Roubin, M., and de Grouchy, J. (1976). Trisomie 10p. Une observation ancienne précisée par marquage. *Annales de Génétique*, **19**, 61-64.

- Uchida, I. A., Wang, H. C., Laxdal, O. F., Zaleski, W. A., and Duncan, B. P. (1964). Partial trisomy deficiency syndrome resulting from a reciprocal translocation in a large kindred. *Cytogenetics*, **3**, 81-96.
- Vogel, W., Siebers, J. W., and Reinwein, H. (1973). Partial trisomy 7q. *Annales de Génétique*, **16**, 277-280.
- Wilroy, R. S. (1977). Partial monosomy and partial trisomy for different segments of chromosome 13 in several individuals of the same family. *Annales de Génétique*, **20**, 237-242.
- Wright, Y., Clark, W. E., and Breg, W. R. (1974). Craniorachischisis in a partially trisomic 11 fetus in a family with reproductive failure and a reciprocal translocation $t(6p+;11q-)$. *Journal of Medical Genetics*, **11**, 69-76.
- Young, S. R., Donovan, D. M., Greer, H. A., Burch, K., and Potter, D. C. (1976). Tertiary trisomy 47,XX,+14q-, resulting from maternal balanced translocation, 46,XX,t(14;16)(q11;q24). *Human Genetics*, **33**, 331-334.
- Yunis, E., Gonzales, J. T., and de Caballero, O. M. T. (1977a). Partial trisomy 16q-. *Human Genetics*, **38**, 347-350.
- Yunis, E., Giraldo, A., Zuniga, R., Egel, H., and Ramirez, E. (1977b). Partial trisomy 4q. *Annales de Génétique*, **20**, 243-248.
- Yunis, E., Silva, R., Egel, H., Zuniga, R., de Caballero, O. M. T., Ramirez, E., and de Ruiz, H. P. (1978). Partial trisomy 5p. *Human Genetics*, **43**, 231-237.
- Yunis, E., Silva, R., and Giraldo, A. (1976). Trisomy 10q. *Annales de Génétique*, **19**, 57-60.
- Zabel, B., and Baumann, W. (1977). Trisomie partielle pour la partie distale du bras long du chromosome 15 par translocation X/15 maternelle. *Annales de Génétique*, **20**, 285-289.
- Zabel, B., Baumann, W., Gehler, J., and Conrad, G. (1978). Partial trisomy for short and long arm of chromosome no 5. Two cases of two possible syndromes. *Journal of Medical Genetics*, **15**, 143-147.
- Zabel, B., Hansen, S., and Hartmann, W. (1976). Partial trisomy 2q and familial translocation $t(2;12)(q31;q24)$. *Human Genetics*, **32**, 101-104.

Requests for reprints to Professor P. Jalbert,
Laboratoire de Cytogénétique du CHU de Grenoble,
Hôpital des Sablons, F-38700 La Tronche, France.