PARTIAL 8 TRISOMY BY 3 : 1 SEGREGATION OF BALANCED MATERNAL TRANSLOCATION $t(6q+; 8q-)$

Keiji KOGAME and Hajime KUDO

*Department of Central Clinical Laboratory, Hirosaki University
School of Medicine, Hirosaki 036, Japan*

Summary We describe a patient with partial trisomy 8 resulting from maternal translocation, $t(6;8)(q27;q11.23)$. The female newborn who died at the age of nine days had multiple congenital malformations including round face, short webbed neck, micrognathia, high arched palate, low set ears, and cardiac defects. The autopsy findings are also presented. Review of twenty-six cases with trisomy 8p revealed that the clinical features were different from case to case and there was little similarity in clinical features between trisomy 8 mosaicism syndrome and trisomy 8p. It is suggested that the break at 8p11 may be responsible for agenesis of the corpus callosum in at least 8p trisomy patients.

INTRODUCTION

The advent of banding methods led to delineation of some clinical syndromes due to the structural rearrangements of the autosomes in all groups. Trisomy 8 mosaicism is not rare in autosomal trisomies and these patients have been described to establish a clinically recognizable syndrome. The reports of the patient with 8p trisomy have been accumulated. However these patients are not a distinct clinically recognizable entity. The present paper describes an additional case of trisomy 8p and compares the clinical and cytogenetical findings in the present case to those of previously reported cases.

CASE REPORT

Y. S., a female, was born after an uneventful pregnancy of 41 weeks to 27-year-old mother. Her father was also 27-year-old. The mother's first pregnancy terminated by artificial abortion and the second and third pregnancies terminated in spontaneous abortion. The abortuses were not examined. Both parents were phenotypically normal and the family history was negative with regard to consanguinity, congenital anomalies or mental handicap. The birth weight was 3,116 g and apgar

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Fig. 1. The patient at 7 days of age.

score was eight in one minute. She was transferred six days after the birth from the delivery ward to the pediatric ward because she showed poor feeding, poor activity, continuous cyanosis, jaundice and bilious vomiting. She was noted to have round face, short webbed neck, micrognathia, high arched palate, and low set ears (Fig. 1). Gallop rhythm and systolic murmur were heard at the base of the heart. Bilateral rhonchi were audible at the lungs. In palpation radial pulse was palpable in both sides but the pulse of femoral artery was not palpable. The extremities and the external genitalia were normal. Dermatoglyphics and palmar and plantar creases were unremarkable.

Laboratory investigation data showed a respiratory acidosis and a hyperbilirubinemia. In liver function tests glutamic-oxaloacetic transaminase (GOT) revealed increased activity but the results of the other tests were normal. Activity of glutathione reductase and coagulation factor VII were not measured. Hematological investigations showed no abnormalities. ECG revealed right axis deviation and right ventricular hypertrophy. UCG revealed slight enlargement of right ventricle and left atrium, and pulmonary arterial dilatation. X-rays of the chest showed congestion of pulmonary vein and cardiomegaly. She died of congestive failure on the 9th day.

An autopsy revealed additional congenital anomalies. The cardiovascular anomalies included preductal coarctation of the aorta, ventricular septal defect, patent ductus arteriosus, and dextroposition of aorta. The left lung was divided into three lobes. There were annular pancreas, mesenterium commune and ileac diverticulum. Immaturity of organs including kidney, uterus and thyroid gland and atelectasis were observed. Congestion was found in liver, lungs, kidneys, stomach, and digestive tracts under the stomach. The remaining organs were apparently normal. Examinations of the brain and the skeleton showed no anomaly.

CYTOGENETIC STUDIES

Chromosome analysis was performed on peripheral blood from the patient and the parents. Sixty cells from the cultures were examined for each individual with

Fig. 2. Partial GTG karyotypes of the mother from four different cells.

Fig. 3. GTG karyotype of the patient. Arrow indicates 8q— chromosome.

four kinds of banding techniques (GTG-, QFQ-, CBG-, and RBG-bandings). In the patient, 47 chromosomes were present in all cells except in three in which there was random loss of one or more chromosomes. The extra chromosome could be identified by the finding of a reciprocal translocation between chromosomes 6 and 8 in the mother: karyotype 46,XX,t(6;8)(q27;q11.23) (Fig. 2). The patient karyotype would be designated as 47,XX,+der(8),t(6;8)(q27;q11.23)mat, so that she had a partial trisomy for the segment at 8q11.23→pter (Fig. 3). The karyotype of the patient's father was normal.

DISCUSSION

Since trisomy 8p was first described by Yanagisawa (1973) in three relatives, twenty-six cases have been reported up to the present (Rosenthal *et al.*, 1973; Sonta *et al.*, 1974; Chiyo *et al.*, 1975; Funderburk *et al.*, 1976, 1978; Weleber *et al.*, 1976; Clark *et al.*, 1977; Rethore *et al.*, 1977; Taylor *et al.*, 1977; Hongell *et al.*, 1978;

Lazjuk *et al.*, 1979; Fineman *et al.*, 1979; Mattei *et al.*, 1980; Jones *et al.*, 1980; MorenoFuenmayor *et al.*, 1980; Jensen *et al.*, 1982; Allen and Hodgkin, 1983). Eleven among these examples were trisomy due to tandem duplication (Weleber *et al.*, 1976; Rethore *et al.*, 1977; Taylor *et al.*, 1977; Hongell *et al.*, 1978; Mattei *et al.*, 1980; Jensen *et al.*, 1982), and the others were due to the familial reciprocal translocations. In thirteen cases an abnormal chromosome was inherited from a carrier mother (Yanagisawa and Hiraoka, 1971; Yanagisawa, 1973; Rosenthal *et al.*, 1973; Sonta *et al.*, 1974; Clark *et al.*, 1977; Lazjuk *et al.*, 1979; Fineman *et al.*, 1979; Jones *et al.*, 1980; MorenoFuenmayor *et al.*, 1980), and in three cases the carriers were fathers (Chiyo *et al.*, 1975; Funderburk *et al.*, 1976, 1978; Allen and Hodgkin, 1983) (Table 1). All familial translocation cases except one (Lazjuk *et al.*, 1979) were due to the usual type of meiotic segregation 2 : 2. On the other hand, a case reported

Table 1. Cytogenetic findings in reported cases of trisomy 8p.

Case	Duplicated segment	Deleted segment	Origin	Reference
1	8q12 → pter; 15p11 → pter	—	mat	Lazjuk <i>et al.</i> (1979)
2	8q11.23 → pter; 6q27 → qter	—	mat	Present case
3, 4, 5	8p11 → pter ^a	21pter	mat	Yanagisawa (1973)
6	8p11 → pter ^a	22qter	mat	Rosenthal <i>et al.</i> (1973)
7	8p11 → pter	13q34 → qter	pat	Chiyo <i>et al.</i> (1975)
8	8p11 → pter	15p11 → pter	pat	Funderburk <i>et al.</i> (1978)
9	8p1 → pter	10p15 → pter	mat	Clark <i>et al.</i> (1977)
10	8p12 → pter	15q25 → qter	mat	Jones <i>et al.</i> (1980)
11	8p21 → pter	10q26 → qter	mat	Fineman <i>et al.</i> (1979)
12, 13, 14	8p21 → pter	12p13 → pter	mat	MorenoFuenmayor <i>et al.</i> (1980)
15	8p21 → pter	11q25 → qter	pat	Allen and Hodgkin (1983)
16	8p11.2 → p23.1	8p23.1 → pter	<i>de novo</i>	Weleber <i>et al.</i> (1976)
17	8p11 → p23.2	8p23.2 → pter	<i>de novo</i>	Mattei <i>et al.</i> (1980)
18	8p11 → p23.2	8p23.2 → pter	?	Mattei <i>et al.</i> (1980)
19	8p11 → p23.1	8p23.2 → p23.3, 14q12 → pter	?	Mattei <i>et al.</i> (1980)
20	8p12 → p23	8p23 → pter?	<i>de novo</i>	Taylor <i>et al.</i> (1977)
21, 22	8p11 → p22	8p23 → pter	<i>de novo</i>	Rethore <i>et al.</i> (1977)
23	8p21 → p23	8p23 → pter	<i>de novo</i>	Taylor <i>et al.</i> (1977)
24	8p21 → p23	8p23 → pter?	<i>de novo</i>	Hongell <i>et al.</i> (1978)
25	8p21.2 → p23.1	8p23.2 → p23.3	<i>de novo</i>	Jensen <i>et al.</i> (1982)
26	8p21 → p23.3	8p23.3	<i>de novo</i>	Jensen <i>et al.</i> (1982)
27	8q12 → p23	18q11 → p11	mat	Sonta <i>et al.</i> (1974)

^a The exact breakpoints were assumed from the published photograph.

Table 2. Clinical findings in 27 cases of trisomy 8p.

Cases	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19	20	21	22	23	24	25	26	27
Mental retardation	?	?	?	+	+	+	+	?	+	+	+	+	+	+	?	+	+	?	?	+	+	+	+	+	+	+	?
Growth retardation	?	?	?	+	+	+	+	?	+	+	+	+	+	+	+	+	+	±	?	+	+	+	+	+	+	+	+
Short stature			+	+	+	+	+	—	+	+	+	+	+	+	+	+	—	—	—	+	+	+	—	—	—	—	—
Microcephaly	—	—	—	—	—	—	—	—	+	+	+	+	+	+	—	—	+	—	—	—	+	+	+	—	—	—	—
Antimongoloid slant	+	—	—	—	+	+	+	—	+	+	—	+	+	+	+	+	+	—	—	—	+	+	+	—	—	—	—
Epicanthic folds	+	—	—	—	—	—	—	—	—	+	+	+	+	+	+	—	—	—	—	—	—	—	+	+	+	—	—
Hypertelorism	—	—	—	+	—	—	+	—	+	+	+	+	+	+	+	+	+	—	—	—	—	—	—	+	+	—	—
Strabismus	—	—	+	+	+	+	+	—	+	—	—	+	+	+	+	+	—	—	—	—	+	+	+	—	—	—	—
Large mouth	—	—	—	—	—	—	—	—	+	+	+	+	+	+	—	—	+	+	+	+	+	+	+	+	+	—	—
High arched palate	+	+	+	+	+	+	+	—	+	—	—	+	+	+	—	—	—	—	—	—	+	+	—	+	+	—	+
Prominent nasal bridge	—	—	+	+	+	+	+	—	+	+	—	—	—	—	—	—	—	—	—	—	—	—	+	—	—	—	—
Low set ears	+	+	—	+	—	—	—	+	+	+	+	—	—	—	+	+	—	—	—	—	—	—	—	+	+	—	—
Malformed ears	+	—	—	+	+	+	+	+	+	+	+	+	+	+	—	—	+	+	+	+	+	+	+	+	+	—	—
Short neck	+	+	—	—	—	—	—	—	—	—	—	—	—	—	—	—	+	+	+	+	+	—	—	—	—	—	—
Micrognathia	+	+	+	—	+	+	—	+	+	+	—	—	—	—	+	+	+	+	+	+	—	—	—	—	—	—	—
Hypoplastic nails	—	—	+	+	+	+	+	—	—	—	—	+	+	+	—	—	—	—	—	—	—	—	—	—	—	—	—
Genital anomaly	—	—	+	+	+	+	+	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—
Cardiac murmur or defect	+	+	+	+	—	—	—	+	+	—	—	+	+	—	+	+	+	—	—	—	—	—	—	—	—	—	—
Abnormal flexion of fingers	—	—	+	+	+	+	—	—	—	—	—	+	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—
Skull asymmetry	—	—	+	—	+	+	—	—	+	+	—	—	—	—	—	—	—	—	—	—	—	—	+	—	—	—	—
Agenesis of corpus callosum	—	—	—	—	—	—	—	+	+	—	—	—	—	—	—	+	+	+	+	+	—	—	—	—	—	—	—
Skeletal anomaly	—	—	—	—	—	—	—	—	—	+	—	+	—	—	—	—	—	—	—	—	—	+	+	—	—	+	—
Abnormal dermatoglyphics	?	+	+	+	+	+	+	—	—	—	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+

Reference for each case is cited in Table 1.

by Lazjuk *et al.* (1979) was due to a 3 : 1 segregation. To our knowledge, the present case is the second of the trisomy 8p due to maternal translocation 3 : 1.

The clinical features of trisomy 8p patients are not distinctive enough to form a single disease entity (Funderburk *et al.*, 1978; Lazjuk *et al.*, 1979; Jensen *et al.*, 1982; Allen and Hodgkin, 1983). Our patient presents a few clinical features similar to those reported in cases with trisomy 8p (Table 2). From the review of 27 cases with trisomy 8p, it is considered that mental retardation, growth retardation, short stature, and abnormal dermatoglyphics are common clinical features. Hypertelorism, malformed and low set ears, and high arched palate are also frequently observed. However, the other anomalies are different from case to case. Rethore *et al.* (1977) mentioned that large mouth and broad nose are important characteristic features for clinical diagnosis. However, these are observed in only about 30–40% of the patients with trisomy 8p. The clinical features of trisomy 8p appear to consist of several dysmorphic features common to many other chromosome abnormalities. Therefore, as stated by Lazjuk *et al.* (1979), it seems that it is difficult to diagnose trisomy 8p based on the clinical features alone.

Trisomy 8 mosaicism syndrome (T8Ms) is characterized by mental retardation, distinctive facies, absent or dysplastic patellas, joint contractures, deep plantar furrows, vertebral defects, narrow pelvis, renal and ureteral abnormalities, distinctive toe posture, and so on (Riccardi, 1977). Among these characteristic anomalies for T8Ms, vertebral defects were present in 26% of the patients with trisomy 8p. However, absent patellas, narrow pelvis, renal and ureteral abnormalities, and distinctive toe posture were not present in all patients with trisomy 8p. Comparison of the clinical features of the patients with trisomy 8p and those of T8Ms suggests that there are a few common clinical features between them. From the above-mentioned facts it is not likely that the clinical features of T8Ms derive from only short arm.

Among the patients with trisomy 8p, there were 14 males and 13 females. The oldest patient is 25 years old (Hongell *et al.*, 1978) and the youngest one is 4.5 days old (Funderburk *et al.*, 1976, 1978). Cases of early neonatal death were reported in only four cases (Funderburk *et al.*, 1976, 1978; Lazjuk *et al.*, 1979; Jones *et al.*, 1980; present case). Therefore it seems that the lifespan is not shortened. Mean maternal age is 27.3 ± 5.22 years (ranged from 16 to 40), and paternal age is 31.6 ± 7.42 years (ranged from 23 to 56). Mattei *et al.* (1980) stated that the mean parental age is greater than in the general population. However, it is not significantly older in the data from the 27 reported cases.

The twenty-six cases with trisomy 8p can be divided into nine groups according to the type of the trisomic segment. Our case (type 2) is an additional new one for trisomic segment (Table 3). In these cases trisomy for the segments at p11→pter, p21→pter, p11→p23, and p21→p23 are relatively frequent. Three cases (type 1, 2 and 10) are trisomy which involves a part of long arm, and the other 24 cases are trisomy which involves only short arm. Fineman *et al.* (1979) reported that trisomy for varying amounts of chromosome 8 seems to play an important part in determining

Table 3. Comparison of clinical manifestations of patients carrying different 8p trisomies.

Types	1	2	3	4	5	6	7	8	9	10
Trisomic segment	q12→pter	q11→pter	p11→pter	p12→pter	p21→pter	p11→p23	p12→p23	p11→p22	p21→p23	q12→p23
Mental retardation	?	?	6/6(100)	?	4/4(100)	2/2(100)	+	2/2(100)	4/4(100)	?
Growth retardation	?	?	6/6(100)	+	2/2(100)	1/2(50)	+		2/4(50)	?
Short stature	?	—	5/6(83)	+	4/4(100)	0/2(0)	+		0/4(0)	+
Microcephaly	—	—	0/7(0)	+	1/5(20)	0/2(0)	+	2/2(100)	1/4(25)	+
Antimongoloid slant	+	—	3/7(43)	+	3/5(60)	2/4(50)	+	0/2(0)	1/4(25)	+
Epicanthic folds	+	—	1/7(14)		1/5(20)	0/4(0)	—		2/4(50)	—
Hypertelorism	—	—	3/7(43)	+	5/5(100)	2/4(50)	—		1/4(25)	—
Strabismus	—	—	3/7(43)		3/5(60)	0/4(0)	+		2/4(50)	
Large mouth	—	—	0/7(0)	+	3/5(60)	3/4(75)	+	2/2(100)	1/4(25)	
High arched palate	+	+	5/7(71)	+	3/5(60)	0/4(0)	—	0/2(0)	2/4(50)	+
Thick nose	+	—			3/5(60)		+	2/2(100)	1/4(25)	
Prominent nasal bridge	—	—	5/7(71)	+	0/5(0)	0/4(0)	+	0/2(0)	1/4(25)	
Low set ears	+	+	2/7(29)	+	2/5(40)	0/4(0)	—	0/2(0)	1/4(25)	
Malformed ears	+	—	5/7(71)	+	4/5(80)	3/4(75)	+	0/2(0)	2/4(50)	
Prominent helix	+	—	0/7(0)		2/5(40)	0/4(0)	—		0/4(0)	
Short neck	+	+	0/7(0)		0/5(0)	2/4(50)	—	0/2(0)	0/4(0)	+
Micrognathia	—	+	3/7(43)	+	1/5(20)	3/4(75)	—	0/2(0)	0/4(0)	+
Hypoplastic nails	—	—	5/7(71)		3/5(60)	0/4(0)	+	0/2(0)	1/4(25)	+
Genital anomaly	+	+	4/7(57)	+	1/5(20)	0/4(0)	+		1/4(25)	
Heart defects	+	+	3/7(43)	+	3/5(60)	1/4(25)	—	0/2(0)	0/4(0)	
Abnormal flexion of fingers	—	+	3/7(43)		1/5(20)					
Skull asymmetry	—	—	4/7(57)	+	0/5(0)	0/4(0)	—		1/4(25)	
Agenesis of corpus callosum	—	—	2/3(67)	—	0/4(0)	4/4(100)	?	1/2(50)	0/2(0)	?
Skeletal anomaly	—	—	0/7(0)		1/5(20)	0/4(0)	+	2/2(100)	3/4(75)	
Number of case	1	1	7	1	5	4	1	2	4	1

Figures in parentheses give percentages.

the severity of mental retardation and physical malformations, *i.e.*, patients who are trisomic for large chromosomal segments are mentally and physically more affected than patients who have smaller trisomic segments. As shown in Table 3, it seems that the length of trisomic segments does not correlate with the severity of clinical expression. Allen and Hodgkin (1983) suggested that the variable features may be the result of partial monosomy for genetic material on the other chromosomes involved in the translocations. Fineman *et al.* (1979) failed to recognize the phenotypic effect of the deleted genetic material in many cases. There are some cases which revealed similar clinical features in spite of having different deleted segments as shown in Tables 1 and 2. Judging from these findings, the suggestion of Allen and Hodgkin (1983) does not appear to be the case. All reported cases of 8p trisomy possess p21→p22 as a common trisomic region. This indicates that although the clinical features of 8p trisomy are variable, the cardinal features may be related to the trisomy of p21→p22.

Agensis of the corpus callosum has been reported in seven instances (Funderburk *et al.*, 1976, 1978; Weleber *et al.*, 1976; Clark *et al.*, 1977; Rethore *et al.*, 1977; Mattei *et al.*, 1980). However, absence of this anomaly was reported in many other cases including our case (Chiyo *et al.*, 1975; Rethore *et al.*, 1977; Lazjuk *et al.*, 1979; Fineman *et al.*, 1979; Jones *et al.*, 1980). Although it has been reported that this anomaly may have a high association with trisomy 8p (Weleber *et al.*, 1976; Lazjuk *et al.*, 1979; Mattei *et al.*, 1980), this anomaly is seen only in patients with trisomy for the segments at 8p11→p22, 8p11→p23, or 8p11→pter (Table 2). From this viewpoint, the break at 8p11 may be responsible for this anomaly in at least trisomy 8p patients.

Further reports of trisomy 8p with detailed descriptions of clinical and cytogenetical findings might well give more valuable knowledge of this abnormality.

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