Jpn. J. Human Genet. **28**, 209–213, 1983

DOWN SYNDROME WITH 45 CHROMOSOMES

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Summary A case of Down syndrome with 45 chromosomes was presented. Karyotype of the propositus was determined as 45,XY,−13,−14,−21, +t(13q14q), +t(21q21q). This is the fifth report of Down syndrome with 45 chromosomes. SOD-1 activity was as 1.5 times high as controls.

INTRODUCTION

There are some reports of Down syndrome with a familial D/D translocation and regular type 21 trisomy. But there was only one report of Down syndrome with familial D/D Robertsonian translocation and *de novo* G/G translocation. In this report, we present such an additional case.

CASE REPORT

The patient was the first product of healthy and unrelated parents after an uneventful course of pregnancy and normal delivery at 40th week of gestation. At the child's birth, the mother was 26 years old and the father 30. His paternal cousin suffers from congenital heart disease and his mentality is retarded, but further information is not available. His birth weight was 3,250 g, length 48 cm, head circumference 32 cm and chest circumference 32 cm. He was referred to us because of feeding difficulty and muscle hypotonia. No murmur was audible and his chest X-p was almost normal. He showed several features typical for Down syndrome; hypotonia, flat face, brachycephaly, broad hands with stubby fingers and depressed nasal bridge (Fig. 1).

Dermatoglyphic study revealed bilateral axial triradii in the t″ position, increased ulnar loop patterns on the fingertips (8/10), bilateral tibial arch in the hallucal areas, clinodactyly and single flextion crease of the 5th fingers and a right transverse palmar crease. Dermatoglyphic formulas of the propositus and the parents are shown in Table 1.

Cytogenetic examination revealed that the propositus had only 45 chromosomes in all observed cells. Examination by G-banding technique revealed that his karyo-

Received September 9, 1982

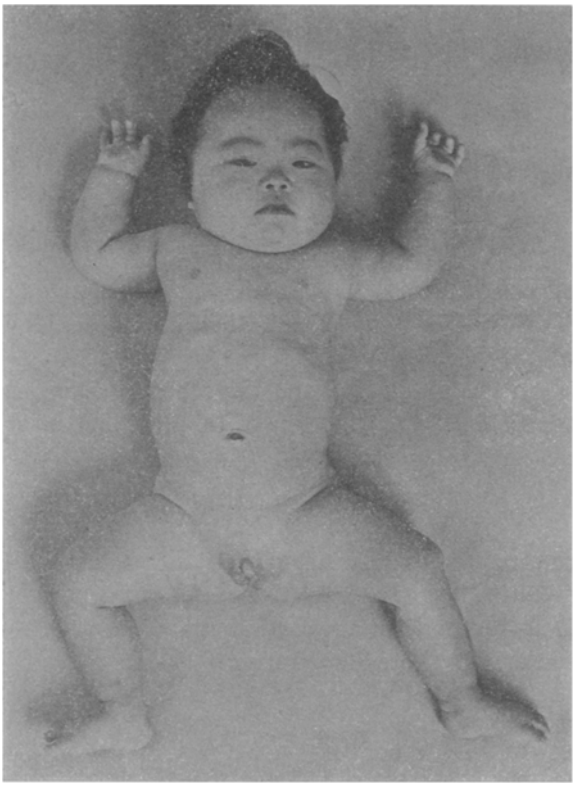

Fig. 1. Patient at 8 months of age.

Table 1. Dermatoglyphic findings of the proband and his parents.

	Finger patterns										TFRC
	L					*R*					
	5	4	3	2	1	1	2	3	4	5	
Proband	U	U	U	U	D	W	U	U	U	U	125
Father	D	W	W	W	W	W	W	W	W	W	187
Mother	U	U	U	U	D	D	D	U	U	U	135

	Palm formula
Proband	L 11.9.7.5′.13′. -tt″-L^{u}.O/O.O.L.O.
	R 11.9.7.5′.13′. -tt″-L^{u}.O/O.O.L.O.
Father	L 9.9.5″.5′.13′. -t- L^{r}.L^{r}/L.O.L.O.
	R 11.9.7.5′.13′. -tt′-L^{u}.O/O.O.L.O.
Mother	L 11.9.7.3.13″. -t- O.O/O.O.O.L.
	R 9.7.5′.3.13′. -tt″-L^{u}.O/O.O.L.O.

type was 45,XY,−13,−14,−21,+t(13q14q),+t(21q21q), his father's karyotype was 45,XY,−13,−14,+t(13q14q), and his mother's karyotype was 46,XX. Figure 2 showed G-banded and R-banded partial karyotypes of D and G group chromosomes. We attempted to trace the t(21q21q) chromosomes back to its origin using Q and C heteromorphism, but it was not successful.

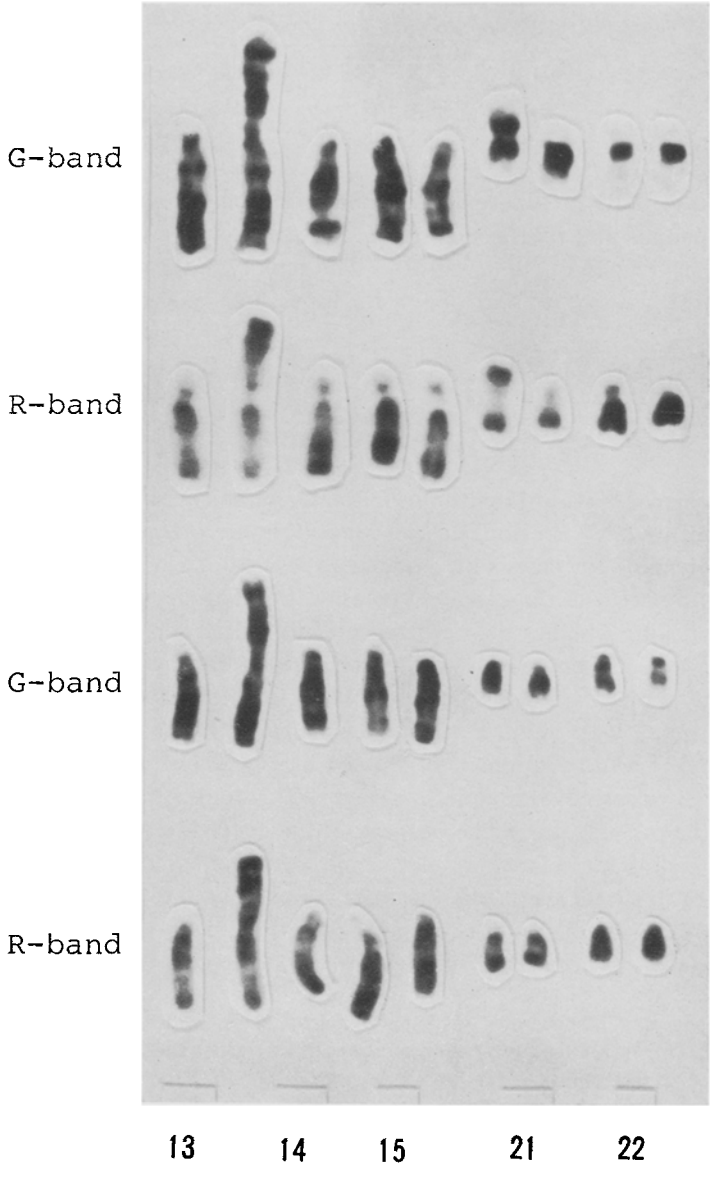

Fig. 2. Partial G and R banded karyotypes of the propositus (top) and of the father (bottom).

Table 2. Down syndrome with 45 chromosomes.

Authors	Karyotype
Marsden *et al.* (1966)	45,XX,−D,−D,−G,+t(DqDq)mat.,+t(GqGq)
Orye and Delire (1967)	45,XY,−D,−D,−D,+t(DqDq)mat.,+t(DqGq)pat.
Lurie (1979)	45,XX,−14,−21,−21,+t(14q21q)pat.,+t(21q21q)
Emberger *et al.* (1980)	45,XX,−14,−21,+t(14q21q21q)
Zellweger and Abbo (1965)	45,X,−D,+t(DqGq)/45,XX,−D,−D,+(DqDq)/ 44,X,−D,−D,−D,+(DqDq),+(DqGq)/46,XX

Erythrocyte SOD-1 (superoxide dismutase-1) activity was studied in the propositus, the parents and fourteen controls. It was assayed by the method described in the report by Yamamoto *et al.* (1979). The activity in the propositus was 23.67± 0.65 units/mgHb, which was as about 1.5 times high as 14 controls (16.35±1.06 units/mgHb).

DISCUSSION

Down syndrome with 45 chromosomes are extremely rare. Only 4 cases have been reported, so far. Previous cases in the literature are summarized in Table 2. As the karyotype of our case is the same as that of Marsden's case, the present case is the second report with familial D/D translocation and *de novo* G/G chromosome.

It is interesting whether the occurrence of t(21q21q) was influenced by paternal t(13q14q) or not. There were some reports about the occurrence of regular 21 trisomy from familial D/D translocation (Hamerton *et al.*, 1963; Yunis *et al.*, 1964; Zergollern *et al.*, 1964). Dutrillaux and Lejeaune (1970) analyzed 67 families with familial t(DqDq). In the progeny of women having D/D translocation, trisomy 13 and trisomy 21 occurred with equal frequency, 2%, suggesting "interchromosomal" effect. On the other hand, in the progeny of males having t(DqDq), the frequency of chromosomal abnormalities was not significantly increased. In the present case, the occurrence of two Robertsonian translocations in an individual might have been fortuitous. As far as the authors are aware of, there is no reason to believe that a Robertsonian translocation predisposes another one.

Acknowledgments This study was supported by a grant from the Ministry of Health and Welfare of Japan for research on prevention of physical and mental disabilities project on the monitoring of congenital disorders.

We also express our thanks to Dr. N. Ogasawara (Department of Biochemistry, Institute for Developmental Research, Aichi Prefectural Colony) for the assay of SOD-1 activity.

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