106. A Case of the Prader-Willi Syndrome having the Interstitial Deletion of No. 15 Chromosome

By Tetsuji KADOTANI,*) Yoko WATANABE,*) Suzue KANATA,**') Toshio KUMADA,**') and Ichiro TAKEMURA***

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Since nine cases with neonatal hypotonia, obesity, cryptorchism, mental retardation and short stature, have been reported by Prader, Labhart and Willi in 1956, the cases with these symptoms were called Prader-Willi syndrome or Prader-Labhart-Willi syndrome. The incidence of Prader-Willi syndrome (PWS) was estimated at one in 10000 live births (Wiedemann, 1985).

During a chromosome survey of the mental defectives, a 22-year-old man showing the Prader-Willi syndrome was found to have a tiny interstitial deletion of the long arm of no. 15 chromosome.

**Case reports.** The patient was a 22-year-old man and 162 cm in height, 74 kg in weight. He was born to a 28-year-old mother and a 27-year-old father as the first child of two sibs. There was no history of abortion, still birth, congenital malformations, and consanguinity in this couple. The patient’s mother was exposed by the atomic bomb at nine years old. The clinical examination revealed that his both parents and his younger sister were phenotypically normal showing no evidence for the mental deficiency.

The birth weight of the propositus was 3450 g at the full term gestation. His remarkable clinical findings were obesity, diabetes, light brown and sparse hair, impaired vision, female breast, incomplete development of external genitalia, cryptorchism, small penis, hyporeflexia, simianline at right hand, short III fingers in both feet, over sweating, and mental retardation; debile (Fig. 1).

**Cytological findings.** The blood cultures were performed by the medium MEM. Chromosome slides for this study were prepared from the patient, his younger sister and his parents. The G- and Q-banding differential stainings were applied for the chromosome identification. The chromosome numbers were made with 65 well-delineated metaphases. The karyotype was analysed in 15 cells each by the conventional and G- and Q-banding procedures.

The karyotype of the patient based on the conventional Giemsa procedures showed 46 chromosomes with one no. 15 chromosome having an unusually shortened long arm. The G-banding analyses revealed that an element unusually corresponded to the interstitial deletion of the segment q12 of no. 15 chromosome. Then, the chromosome formula of the patient was given as 46,XY,del(15)(q11q13). His parents and younger sister were chromosomally normal showing no slight evidence for the aberration, based on 10 cells karyotyped in each case. But no. 15 chromosomes of his father had the variants; one had the elongated short arm and another had somewhat larger and longer satellite. On the studies of the

*) The Kadotani Medical Research Foundation. 1248, Saijohigashi, Saijo, Higashihiroshima 724, Japan.

**) Psychiatric Department, Kamo National Sanatorium. Kurose-cho, Kamogun, Hiroshima, Japan.

*** Kojika-Gakuen. Awaya-cho, Miyoshi, Hiroshima, Japan.
origin of the abnormal no. 15 chromosome of the patient by the Q-bandning analyses, one no. 15 chromosome of the patient had somewhat larger and longer satellite than another chromosome, and one no. 15 chromosome of his father had the same variation (Fig. 2). Then, it was revealed that the origin of the abnormal no. 15 chromosome of the patient was from paternal.

Remarks and conclusion. The PWS patient having the proximal deletion of the long arm of no. 15 chromosome has been described. In PWS patients, the deleted segment of a 15(q11q13) was reported most frequently. Ledbetter et al. (1981) reported on 4 cases out of 5 PWS patients, Ledbetter et al. (1982) reported on 17 cases out of 40 cases, Cassidy et al. (1984) reported on all of the 12 patients, Golden et al. (1984) reported on 1 case out of 2 black females,
Fukushima et al. (1984) reported on 13 cases out of 14 patients, and Butler et al. (1986) reported on 21 cases out of 39 PWS patients. One case of this report had also the deleted segment of a 15(q11q13). Pauli et al. (1983) reported on one case showing the unbalanced translocation of the long arm of one chromosome no. 15 to the distal end of the long arm of one chromosome no. 11 and resulting the monosomy for the short arm and the more proximal portion of the long arm (through band q15) of the chromosome no. 15. On the parental studies to establish the origin of the chromosome deletion, Butler et al. (1986) described that the del(15q) was paternal in origin on the studies of 13 cases. On the results of our studies of the variants involving the short arm and the satellite region of the chromosome no. 15, it was revealed that the origin of the abnormal no. 15 chromosome of our case was from paternal.

In PWS, the origin of this disease has been problematic, and so we hope further investigation.

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References