104. A Case with the Interstitial Deletion of the Long Arm of No. 5 Chromosome

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(Communicated by Sajiro MAKINO, M. J. A., Dec. 12, 1984)

During a chromosome survey of the heavy mental defectives, a 28-year-old man was found to have an interstitial deletion of the long arm of no. 5 chromosome with no hematologic disorder. The present article describes some clinical and cytogenetical features in this patient.

Case reports. The propositus was a 28-year-old man. He was born to a 32-year-old mother and a 33-year-old father as the youngest of four sibs. There was no history of abortion, still birth, congenital malformations, consanguinity, and the atomic bomb exposure in this couple. The clinical examination revealed that his mother, his elder sister, and three of elder brothers were phenotypically normal showing no evidence for the mental deficiency. His father and elder sister were dead.

The birth weight of the propositus was 3100 g at 40 weeks of gestation. His remarkable clinical findings were mental retardation: Idiot, short stature, square head, low set hairline, older facies, blepharophimosis, hypertelorism, flat nasal bridge, broad nose, weak in sight, short webbed neck, kyphosis, incomplete development of the external genitalia with a small penis, and abnormal behavior.

Cytological findings. Chromosome slides for this study were prepared from the leucocyte cultures established from the propositus, one of the elder brother, and his mother. The G-banding differential staining was applied for the chromosome identification. The chromosome numbers were made with 24 well-delineated metaphases. Karyotypes were analysed in 12 cells each by the conventional and G-band preparations.

The karyotype of the patient based on the conventional Giemsa method showed 46 chromosomes with a no. 5 chromosome having an unusually shorted long arm. The G-banding analyses revealed that unusually an element corresponded to the interstitial deletion of the

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Then the chromosome formula of the patient was given as 46,XY,del(5)(q22q31) (Fig. 2). His mother and the third brother were chromosomally normal with no slight evidence for the aberration, based on 12 cells karyotyped in each. The chromosomal examination of his second brother was not cooperated.

Remarks and conclusion. The deletion of the long arm of no. 5 chromosome has been described to occur very frequently in hematologic disorders, such as refractory anemia (Van den Berghe et al. 1974; Sokal et al. 1975; Verhest et al. 1977; Kaffe et al. 1978; Van den Berghe et al. 1979; Kadotani et al. 1979; Mahmood et al. 1979; DiBenedetto et al. 1979; Knuutila et al. 1980; Sidengo et al. 1981; Mecucci et al. 1981; Hartley and McCallum 1981), and preleukaemia and/or leukaemia (Van den Berghe et al. 1976; Rowley 1976; Verhest
et al. 1976; Cabrol and Abele 1978; Petit and Van den Berghe 1979; Pedersen-Bjergaard et al. 1980; Tomiyasu et al. 1980; Abe et al. 1979; Van den Berghe et al. 1979; Sadamori et al. 1981; Teerenhovi et al. 1981; Swolin et al. 1981) in addition to the other karyotypic abnormalities. On the other hand, only a few cases without hematological disorder were reported on interstitial deletion of the long arm of no. 5 chromosome (Felling and Kristoffersson 1980; Stoll et al. 1980; Rodewald et al. 1982; Harprecht-Beato et al. 1983; Fukuda et al. 1984).

The deletion found in our case without hematological disorder was also interstitial by showing the band q23. Our case showed mental retardation, short stature, square head, low set hairline, older facies, blepharophimosis, hypertelorism, flat nasal bridge, broad nose, weak in sight, short webbed neck, kyphosis, incomplete development of the external genitalia with a small penis, and abnormal behavior.

Summary. A man with mental retardation and dysmorphic features is found to have an interstitial deletion of the long arm of chromosome no. 5: 46,XY,del(5)(q22q31).

Acknowledgements. We are cordially obliged to Emeritus Professor Sajiro Makino, M. J. A., the senior director of the Kadotani Medical Research Foundation, for improvement of this manuscript. Financial aid from the Japan Academy is gratefully acknowledged here.

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