25. *Elongated Long Arm of No. 9 Chromosome in Heavy Mental Retardates*

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(Communicated by Sajiro Makino, M. J. A., March 12, 1986)

In the recent reports using the methods of G- and C-banding techniques, it has been shown that the morphological variant of no. 9 chromosome as well as the variant of no. 1 chromosome is due to the addition of chromatin material in the area of the constitutive heterochromatin (Tüür, S. et al. 1974). The elongated long arm of no. 9 chromosome (9qh+) variant has been found in both normal persons and subjects with congenital abnormalities.

In a study on 72 cases with heavy mental retardation, it was found that 3 cases had 9qh+.

**Materials and methods.** All cases having 9qh+ were among 72 cases with heavy mental retardation, and they were admitted in the Kamo National Sanatorium.

The chromosome slides were prepared following the standard blood culture procedures. The karyotype analyses were made with the application of the conventional Giemsa staining and G- and C-banding differential staining.

**Case reports.**

*Case 1.* The patient was a 13-year-old boy. He suffered from cerebral palsy by perinatal cause which was asphyxia. His remarkable clinical findings were dolichocephaly, nystagmus, equino varus, seizure, right hemiplegia, inability in walking, behavior disorder; self injury, and speech disorders. His karyotype was given as 46,XY,9qh+. His parents and his younger sister were phenotypically normal and had the normal karyotypes.

*Case 2.* The patient was a 11-year-old girl. Her remarkable clinical findings were prominent by having the nasal bridge, seizure, speech disorders and behavior disorders; self injury, hyperactivity, impulsiveness. Her karyotype was given as 46,XX,9qh+. Her parents, who had been phenotypically normal, were already dead. The karyotype of her grandmother was 46,XX.

*Case 3.* The patient was a 18-year-old boy. His remarkable clinical findings were slight oxycephaly, speech disorders and behavior disorder; autistic tendency. His karyotype was given as 46,XY,9qh+. His parents were phenotypically normal. But the chromosome analyses of them were not cooperated.

**Results and remarks.** Fig. 1 showed no. 9 chromosomes of three cases. In those three cases, 9qh+ relatives were not found. Although the phenotype of those cases was variable, the common clinical findings were heavy mental retardation and behavior disorders.

In our report, the incidence of 9qh+ was 4.17%; three cases out of 72 cases. Nielsen et al. (1974) made the chromosomal examination on 8712 persons and reported that the incidence of 9qh+ was 0.1%.

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Palmer and Schroder (1971), and Nielsen et al. (1974) considered the possibility that persons with 9qh+ might have an increased risk of progeny with chromosome abnormalities and/or congenital abnormalities. Ghosh (1979) reported chromosome heteromorphism in the children with the congenital malformation and pointed out that the C-band heteromorphism was more common in children with congenital malformation as compared to normal persons. Soudek and Sroka (1979) mentioned in the report of chromosomal variants in mentally retarded and normal men that the increased size of 9qh+ seemed to be a factor with possible negative effects. Christian et al. (1980) reported a family in which a child of Ellis van-Creveld syndrome associated with the 9qh+ chromosomal marker was included. These reports described certain aspects of our present knowledge.

Recently several studies concerning the quantity and/or size of the constitutive heterochromatin of human chromosome 1, 9 and 16 have been reported. Balicek et al. (1977) used the method of the linear measurement of C-band according to Modan and Bobrow (1974) and demonstrated the results that the size of the heterochromatin portions decreased regularly with the increase of the degree of the euchromatin contraction. Verma et al. (1978) used the C-banding technique by barium hydroxide using Giemsa for the estimation of the size of the heteromorphisms of nos. 1, 9 and 16 chromosomes and classified the size of heteromorphisms into one of five sizes; very small, small, intermediate, large, and very large. And they reported that the majority of the number 9 chromosomes were classified as small ones. Azumi et al. (1979) presented the method of the numerical expression to express the size of C-positive qh regions of chromosomes in terms of the standard deviation. Lopetegui (1980) used a modular system for the semiautomatic quantitative evaluation of image for measurement of C-band areas, and distributed each area of nos. 1, 9 and 16 chromosomes according to a system of the classification proposed by Patil and Lubs (1977), and reported that the distribution of the C-band areas in parents of Down’s syndrome patients appeared to be significantly different from the distribution of these areas in a normal population chosen at random. Maes et al. (1983) measured the length of nos. 1, 9, 16 and Y chromosomes in couples with recurrent early abortions using a Hewlett Packard digitiser and recorded co-ordinates of four points; the start and the end of the chromosome, and the start and the end of the C-band. They reported that the results did not show a relationship between the C heterochromatin length and the occurrence of recurrent abortions.

Up to date, in spite of these previous studies, the significance of the variation in the quantity of the constitutive heterochromatin and the relationship between the variation of the area of the constitutive heterochromatin and mental
retardation are yet in the dark. It is a subject for further investigation.

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.

References