G-Deletion Syndrome II*

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Summary. A case with a ring G-chromosome is described as a further illustration of G-deletion Syndrome II.

An analysis of the published cases of syndromes associated with deleted or ring G chromosomes by Warren and Rimoin (1970) revealed that phenotypic differences exist that could be grouped into 2 distinct syndromes. It was suggested that these be designated G-deletion Syndrome I and G-deletion Syndrome II. 4 more cases having ring G chromosomes have since appeared (Armendares et al., 1971; Grosse et al., 1971; Richards et al., 1971); 3 of these patients presented as G-deletion Syndrome I and 1 as G-deletion Syndrome II. The purpose of this report is to describe another case with the phenotype of G-deletion Syndrome II.

Case Report

J. S. is the first male child of a non-consanguineous mating. His mother was 23 and father 29 years at the time of birth. Both are deaf. Parental karyotypes are normal. There is no further family history of deafness or other significant disorders. Pregnancy was uneventful apart from an automobile accident at the 13th week when the mother suffered a back injury, but this was not severe. The delivery was normal at the 37th week of gestation. Birth weight was 2950 g. The baby was a slow feeder, relatively inactive and floppy. At 10 months he could support his weight. He crawled at 11 months and sat supported at 13 months. On physical examination at 13 months, he appeared younger than his chronological age. His height was 71 cm (3rd percentile), weight 9 kg (10th—25th percentile) and head circumference 45.5 cm (10th percentile). There were no gross physical malformations of his head, limbs or digits. The fontanelle was closed. He had bilateral epicanthal folds with slight ptosis manifesting a dull expression (Fig. 1). The palate was slightly narrow and high. Muscle tone was decreased. Psychometric evaluation revealed an I.Q. of 67 on the Cattell-Infant Intelligence Scale. Urinalysis and urinary metabolic screening tests were negative. Skull x-rays and EEG were normal.

Dermatoglyphics showed 6 ulnar loops and 4 whorls. Both palms had an axial triradius in the t' position and large whorl patterns in the hypothenar area. The “b” triradius was displaced radially on the right palm. The “c” triradius was absent from both palms. A complete analysis is available on request.

Cytogenetic studies performed on peripheral lymphocytes and skin fibroblasts show a 45,XY,G-/46,XY,Gk karyotype (Table 1). A composite karyotype of the G group chromosomes from blood and skin is shown in Fig. 2.

Linkage studies, performed by Dr. Wilma Bias, revealed no deletions of blood group substances.

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Fig. 1. J. S. at age 10 mos

Fig. 2. Partial karyotype showing G group chromosomes