Tandem Duplication (5q13→22) in a Mentally Deficient Girl

E. Kessel and R. A. Pfeiffer*

Abteilung für Humangenetik der Medizinischen Hochschule Lübeck, Ratzeburger Allee 160, D-2400 Lübeck, Federal Republic of Germany

Summary. A tandem duplication of 5q13→5q22, visualized using GTG- and RBA-banding, was found in a mentally defective girl with craniofacial dysmorphology.

“Tandem” duplications, i.e., successive repeats of the same chromosomal segment, either serial or inverted, are still rarely found in humans. We report on a mentally deficient child in which this type of aberration is suggested.

Case Report

SB 691010 is the first child of healthy unrelated parents who were both 33 years old at her birth. The second child is normal. The patient was born after an uneventful pregnancy 11 days before term. Her weight was 2800 g, and her length was 48 cm. Her psychomotor development was severely retarded, and her spontaneous movements were always very poor. When first examined at the age of 12 months, she was unable to sit and did not pursue objects or grasp them. Her muscles were hypotonic, and her muscular reflexes being symmetrical but weak. A moderate rigor was felt in the arms. Her weight was 9000 g, her length 78 cm, and her head circumference 44 cm. The intercanthal distance was 28 mm.

She had facial dysmorphology characterized by epicanthus, downward-slanting eyes, a bulbous nose, and micrognathia. Her ears were large and floppy. She had a high palate and the alveolar ridge appeared enlarged. Cardiac and renal malformations were ruled out by clinical examination. EEG results were not consistent with her chronological age but did not show distinctive abnormalities. Except for arches on both the first and second fingers and a twin loop in the hallucal area, dermatoglyphics showed unremarkable patterns.

The patient started walking at the age of 5 but never acquired speech. Her hearing and vision were considered normal. At the age of 9, her weight was 20 kg, her height 125.5 cm, and her head circumference 48 cm (all close to the tenth percentile). Her facial appearance, as shown in Figure 1 had remained unchanged. She had a funnel chest deformity and increasing kyphoscoliosis of the thoracic segment of the spine. X-ray examination did not reveal gross abnormalities of the skull, spine, long bones, and hands. Her bone age was within normal range.

* To whom offprint requests should be sent. Present address: Bismarckstr. 10, D-8520 Erlangen, Federal Republic of Germany
Cytogenetic Studies

The modal number of chromosomes from the patient's lymphocytes, examined at various occasions, was 46. Only one normal chromosome 5, identified first using $^3$H autoradiography, was seen. The long arm of the homologous chromosome was enlarged. QFQ- and GTG-banding and the replication pattern after incorporation of BrdU during the final 6 h of culture suggested a tandem duplication of 5q13→22 (Figs. 2 and 3). The karyotype of both parents was normal.