Short Arm Deletion of Chromosome 14

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Summary. 3 cases with a Dp-chromosome, designated by autoradiography as a No. 14, are presented by the authors. The first case was a mentally retarded boy with minor malformations. Cases 2 and 3 had normal phenotypes and were detected by cytogenetic investigation of family members of a mentally retarded boy with a ring G chromosome. The 14 p- was the only karyotype abnormality in the father (case 2). It was associated with other abnormalities in the daughter (case 3) who had a D/G translocation of the centric fusion type (46,XX,15--,21--,t(15p21p)+, t(15q21q)+).

Zusammenfassung. 3 Fälle mit einem Dp-Chromosom, das durch Autoradiographie als ein Nr. 14 identifiziert werden konnte, werden dargestellt. In dem ersten Fall bestanden Debilität und unbedeutende morphologische Anomalien. Fall 2 und 3 hatten einen normalen Phänotyp und wurden im Verlaufe von cytogenetischen Untersuchungen von Familienangehörigen eines debilen Jungen mit einem Ring 22 entdeckt. Das 14p-Chromosom war die einzige Anomalie im Karyotyp des Vaters (Fall 2). Bei der Tochter (Fall 3) bestand außerdem eine D/G-Translokation (46,XX,15--,21--,t(15p21p)+, t(15q21q)+).

A deletion of the short arm of a group D chromosome has been described by several authors (Grouchy et al., 1966; Lieber et al., 1967; Bias and Migeon, 1967; Starkman and Shaw, 1967; Emerit et al., 1968; Brogger, 1969; Parker et al., 1969). Autoradiographic studies could identify the abnormal chromosome as a number 13 in 5 observations (Bias and Migeon, 1967; Lieber et al., 1967; Emerit et al., 1968; Brogger, 1969, Parker et al., 1969). The present report concerns 3 cases with a deletion of the short arm of a chromosome 14, as could be concluded from radioactive thymidine labeling patterns.

Case No. 1

is a 7.8 years old boy (height 120 cm, weight 25 kg), with severe mental retardation (IQ 45) and epileptic seizures. He was noted to have only slight morphological anomalies such as a high arched palate and misshapen ears.

Dermatoglyphic prints were without any particularity.
Fig. 1. Caryotype of case 1

Fig. 2. Family tree and blood groups of case 1

Chromosome Analysis

Dividing blood lymphocytes were studied using standard cytogenetic techniques. The short arm of a group D chromosome was deleted in all the 38 mitoses examined (Fig. 1). Investigation of the family did not reveal chromosome aberrations in the mother and the 3 sisters of the propositus. The father was not available for study (Fig. 2).

Autoradiography was done according to the procedure of Schmid (1963). Tritiated thymidine (specific activity 5 Ci/millimol) was added to the culture medium in a final concentration of 0.5 μCi/ml during the last 6 hrs of incubation. The Kodak Stripping film AR10 was exposed during 20 days. The metaphases were first photographed with the silver grains and relocated after removal of the film. A total of 60 mitoses was available. The abnormal D chromosome could be identified as a No. 14 by qualitative analysis in 42 of these 60 mitoses (Fig. 3),