148. *On the Ring 13 Chromosome in a Malformed Infant with Special Regard to the Break Point*

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This paper reports preliminarily the ring D chromosome in a one-year-old male infant having multiple malformations. The aberrant chromosome was analysed by the Giemsa banding technique, with particular concern to break points. No correlation between the development of retinoblastoma and the specific deleted band in the long arm of No. 13 chromosome was noted in this patient.

Case report. A one-year-old boy was the product of an uneventful course of pregnancy and normal delivery. The patient's mother and father were 21 and 23 years old at his birth and showed no physical anomaly. The family history showed no special disorders. The patient weighed 1770 g, measured 43 cm in length, and 30.1 cm in head circumference at birth. Imperforate anus and malrotation of the intestine were noted in his neonatal period. The patient received surgical operations. The postoperative course was uneventful, but somatic growth failure and mental retardation were remarkable.

His main clinical features were physical and mental retardation, somatic growth failure, microcephaly with flat occiput, large earlobes, protruding maxilla, micrognathia and short neck. The postoperative scars of anal atresia and intestinal malrotation were noted. The external genitalia were abnormal showing bilateral cryptorchidism and hypoplastic scrotum. Retinoblastoma was not recognized in this patient. Dermatoglyphics was within normal limits, except for absent *b* triradius.

Routine laboratory examinations, including hematological examinations, urinalysis, and blood chemistry, were all within normal limits. Hemoglobin-F was 6%, indicating a moderate elevation. The patient's haptoglobin was type Hp2-1, while it was also Hp2-1 in both parents. Thus, the haptoglobin locus seemed not to be deleted in this case. An intravenous pyelography failed to demonstrate the right kidney.

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Figs. 1-4. Trypsin-Giemsa banding patterns of normal (1-2) and ring 13 chromosomes (3-4). All 4 dark bands in the long arm of the normal 13 chromosomes (1-2) can be recognized in the rings (3-4). The break points are located at bands 13p11 and 13q34 (3-4).

Cytogenetic findings. Peripheral lymphocytes of the patient cultured by the ordinary 3-day-method indicated that in 40 cells examined, 31 cells (77.5%) were found to be 46, XY, r(D), and 9 cells (22.5%) were 45, XY,-D. By the 2-day-method, 39 cells (78%) were recognized as 46, XY, r(D) and 11 cells (22%) as 45, XY,-D.

The trypsin-Giemsa banding technique demonstrated the ring D chromosome as a ring 13. The break points were found to be located at bands 13p11 and 13q34. All four dark bands in the long arm of chromosome 13 were noticed. Thus, the loss of chromosome material was estimated to be very small.

Remarks. Clinical features of this case was consistent with the ring 13 syndrome so far reported. Since the chromosomal loss in the r(13) syndrome occurs in the long arm of the No. 13 chromosome, the 13q- syndrome and the r(13) syndrome share similar clinical fea-

tures. The clinical pictures characteristic of the two syndromes are mental retardation, microcephaly, trigonocephaly, large earlobes, protruding maxilla (or incisor), micrognathia and undescended testis. Imperforate anus and the abnormal inheritance of haptoglobin are frequently noticed in the r(13) cases.

Recently, Nakagome *et al.* (1973) investigated the break points in a ring 5 cat-cry-syndrome patient by the G-banding technique. In our r(13) case, break points were found to be located at 13p11 and 13q34 bands, resulting in a small amount of the chromosomal loss.

Orye *et al.* (1971) reported a case of the 13q- syndrome with retinoblastoma and showed that the missing portion of No. 13 chromosome was around the broadest band on its long arm. Willson *et al.* (1973) described a 13q- patient with retinoblastoma with the identification of the deleted band as 13q31.

The deleted portions of our case seemed to be some parts of 13p11 and 13q34, retaining the band 13q22 intact. It is commented that the present patient has no retinoblastoma at the age of one year, and may be free from developing the tumor in the future.

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