25. *A Case of a Ring Chromosome No. 22 with Mental Retardation*

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During a chromosome survey of congenital defectives, a 29-year-old man with mental retardation was found to have a ring chromosome no. 22. The present article describes the clinical and cytogenetic evidence in this man, and examined the relation between the symptoms and the ring chromosome no. 22.

**Case reports.** The propositus was a 29-year-old mentally retarded male, 54 kg in weight, 157 cm in height. He was born to a 21-year-old mother and a 27-year-old father as the first child. There was no history of abortion, stillbirth, congenital malformations, mental retardation, exposure to the atomic bomb, and consanguinity in this couple. His parents and his younger brother were phenotypically normal. The birth weight of the propositus was 2250 g at the 38th week of the eventless gestation. The remarkable clinical signs were ptosis of the left eyelid, strabismus, large and deformed auricles, saddle nose, thick lips, pigmentation on the dorsal region, kyphosis, severe mental retardation, behavior disorders, and speech disorders (Fig. 1).

**Cytological findings.** Chromosome slides for this study were prepared from the leucocyte cultures established from the propositus. The G-banding method and N-banding differential staining (Bloom and Goodpasture, 1976) were applied for the chromosome identification. The chromosome numbers were determined with 137 well-delineated metaphases. Karyotypes were analysed in 12 cells each following the conventional Giemsa, and the G- and N-banding procedures.

The karyotype of the propositus based on the conventional Giemsa specimen showed 46 chromosomes including three normal G group chromosomes, the forth being represented by a ring chromosome. On the analysis of the G-banding method, it was revealed that the ring chromosome was one of no. 22 chromosomes. On the silver staining (N-banding) analysis, the presence of the nucleolar organizing region on the ring chromosome no. 22 was present. This would suggest a break which occurred at p13 (Fig. 2). The G- and N-banding patterns of the ring chromosome no. 22 suggested that the distal break of the long arm was at q13 (Fig. 2).

All 137 cells had the ring chromosome no. 22, and in only 3 cells out of 137 cells the double sized ring chromosome was present (Fig. 3). Then, the chromosome formula of this propositus was given as 46,XY,r(22)(p13q13) (Fig. 2).

Both parents and his younger brother did not cooperate in chromosomal examinations.

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Remarks and conclusion. The ring chromosome no. 22 has been reported on at least 29 patients (Brookfield and Walker 1976; Rethore et al. 1976; Hunter et al. 1977; Howard-Peebles 1977; Palmer et al. 1977; Funderburk et al. 1979; Fryns and Berghe 1979; Fowler et al. 1980). Several patients manifested a distinctive features consisting of the mental retardation with verbal delay, epicanthal folds, full eyebrows, large ears, arched palate and/or high palate, large nose, low nasal bridge, thick lips, hypoplastic mandible, skeletal anomalies, syndactyly, ataxic gait and seizure (Table I). According to Table I, the characteristic features of the ring chromosome no. 22 syndrome were mental retardation (100%), epicanthal folds (83%), verbal delay (68%), abnormal ears (48%), nasal anomalies (45%), arched palate/high palate (44%), ataxic gait (38%), syndactyly (29%), full eyebrows (28%), skeletal anomalies (28%), seizure (28%), hypoplastic mandible (26%), thick lips (25%), and prominent mandible (17%). Our case manifested some frequent features in those characteristic features; mental retardation, verbal delay, abnormal ears, nasal anomalies, thick lips and skeletal anomalies.

On the cytogenetical studies of the ring chromosome no. 22, the absence of
the nucleolar organizing region on the ring chromosome no. 22 was reported by Hunter et al. (1977), Funderburk et al. (1979), and Fowler et al. (1980). But in the present case, the presence of the nucleolar organizing region on the ring chromosome no. 22 was shown.

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