Deletion 14q and pericentric inversion 14

SUMMARY A woman with deletion 14q as well as inversion 14 is presented, and physical signs are compared with those of patients with deletion long arm 13. No previous case of deletion long arm 14 has been published.

Deletion Dq has been described in a number of cases. In most where banding was performed, it has, however, been found to be deletion 13q (Allerdice et al., 1969; Wilson et al., 1969; Gey, 1970; Orbeli et al., 1971; Grosse and Schwanitz, 1973; Wilson et al., 1973; Adámek and Kašpárková, 1974; Ikeuchi et al., 1974; Orye et al., 1974; Kučerová et al., 1975; Noel et al., 1976) (Table). Against the background of these findings and the findings of partial trisomy 13, Lewandowski and Yunis (1975) have produced a phenotypic map of chromosome 13.

Wilson et al. (1969) described a presumptive case of deletion 14q, but later found that it was actually deletion 13q (Wilson et al., 1973).

Very few cases of pericentric inversion D have been described previously; 4 cases of pericentric inversion 13 (Hauksdóttir et al., 1972; Taysi et al., 1973; McDermott and Parrington, 1975), and 2 cases of pericentric inversion 15 (Cohen et al., 1967; Crandall and Sparkes, 1970) have been described, but no cases of chromosome 14 pericentric inversion.

We wish to report one case of pericentric inversion 14 as well as deletion 14q.

Subjects and methods

The proband was found in the prevalence study of all cases of mental retardation in the Århus County with a population of approximately 560 000. Chromosome examination was made on 48-hour lymphocyte cultures, and staining was done with the BUDR-acridine-orange method.
was difficult to understand, but was co-operative, friendly, and quiet and was able to do simple needlework and to help with housework. She was also able to take care of her own needs.

Physical examination at the age of 14 and again at 62 (Fig. 1) showed bilateral congenital hip luxation, micrognathia, large nose, high palate, asymmetrical face, low-set ears, strabismus convergens, short neck, bilateral camptodactyly, and pes valgus. Electroencephalogram was of borderline type with no paroxysmal aberrations or signs indicating epilepsy.

**CYTOGENETIC EXAMINATION**

Cytogenetic examination showed 46,XX in 46% of the cells; in most of the cells we found one normal chromosome 14 and one metacentric chromosome consisting of 14 material. The banding pattern showed that the material normally situated between 14(q21) and 14(q31) had been inverted and placed on top of the short arms (14(q13)). The intense fluorescing band in R-banding normally located between 14(q31) and 14(qter) was missing and was not found elsewhere on the abnormal 14 or on any other chromosome and therefore must have been deleted.

The karyotype was 46,XX/46,XX,inv(14) (q21→q31),del(14)(q31) as shown in Fig. 2.

It was not possible to obtain blood samples from any relatives, but no relatives had mental retardation, and there was no evidence of any increased frequency of abortions or stillbirths in the family; the inversion as well as the deletion of chromosome 14 were thus most probably of de novo type.

**DERMATOGlyphS**

The dermatoglyphs of the proband were abnormal (Fig. 3).

The digital markings showed an excess of whorls and contained more ridges than is normal for women. As a result, the proband had a high finger pattern intensity index (18) and also a high total finger ridge count (182 ridges).
On both hands there was a transverse crease. On the left hand the courses of the main lines A and D were anomalous. The sum of the left and right maximal adt angles (75°) was well below the average figure of 85.9° (Penrose, 1954) for adult female controls. The mean ridge width in the a-b interval (0.402 mm) was low for a female of her age (0.504 mm: Saldaña-Garcia, 1975). On the right palm the pattern configurations were of common types, but on the left palm the association of radial loop I' with peripheral loops II and III was unusual. The pattern-type resulting from this association—I' II III e t 5—was not found in a sample of 900 English controls (450 females + 450 males).

Discussion

The Table shows the clinical signs in the previous 11 cases of del(13q), 4 of which were terminal deletions, 4 interstitial, and 3 unspecified.

The signs found in the present case of interstitial deletion long arm 14 were all found in from one to all of those with del(13q). Presentation of further cases of del(14q) is needed in order to be able to produce a phenotypic map of chromosome 14 as done by Lewandowski and Yunis (1975).

References


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*J Med Genet* 1978 15: 236-238
doi: 10.1136/jmg.15.3.236

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