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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.

Table Physical signs in 11 previously published cases of del(13q) and in present case of del(14q)

Physical signs	Previous cases of del(13q)	Present case of del(14q)
Birthweight (mean)	2347 g	
Poor thriving	9/11	
Psychomotor retardation	10/11	+
Microcephaly	6/11	
Trigonocephaly	5/11	
Broad prominent nasal bridge	4/11	
Hypertelorism	3/11	
Microphthalmus	3/11	
Epicanthus	4/11	
Ptosis	3/11	
Coloboma of iris	2/11	
Retinoblastoma	3/11	
Micrognathia	5/11	
Strabismus	3/11	+
Protruding upper incisors	4/11	
Microstomia	4/11	
High-arched palate	2/11	+
Teeth anomalies	2/11	
Short neck (lateral neck fold)	3/11	+
Low-set (malformed) ears	7/11	+
Large (malrotated) ears	6/11	
Congenital heart disease	2/11	
Hypospadias (epispadias)	2/3	
Undescended testes	3/3	
Pelvic girdle anomalies	6/11	+
Foot and toe anomalies	8/11	+
Thumb absent (hypoplastic)	2/11	
Short fourth or fifth finger	5/11	
Simian crease	2/11	
Muscular hypotonia	5/11	

The cases of del(13q) were published by 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) and Noel *et al.* (1976).

Case history

The proband was born on 14 June 1914. The father was 30 and the mother 24 years old when the proband was born; both parents were healthy. Chromosome examination of the sibs and parents was not possible.

The mother had pronounced vaginal bleeding during the fourth month of pregnancy. The proband weighed 4000 g at birth and thrived well during the first 6 months of life. At the age of 6 months she had pneumonia, and she later suffered from severe rachitis; she stayed in bed for nearly a year. In 1917, at the age of 3 years, she got 'Spanish flu' and was ill for approximately 6 months. She was retarded in mental as well as motoric development, and she was not able to walk or talk till the age of 3 years.

She had epileptic fits of *grand mal* type from the age of 8 to 14 years when she was admitted to an institution for the mentally retarded. She has not had any epileptic fits since then, and she has not been treated with any antiepileptic treatment.

At admission at the age of 14 her IQ was between 35 and 41. During her first years at the institution she

Fig. 1 The proband at the age of 62.

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.

Fig. 2 *BUDR* acridine orange stain of chromosomes 13, 14, and 15 from karyotype 46,XX/46,XX,inv(14)(q21 → q31),del(14)(q31).

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).

Fig. 3 Dermatoglyphs.

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 *atd* 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).

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