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CONGENITAL ABNORMALITIES IN A CHILD WITH PRESUMPTIVE KARYOTYPE 46. XX, t (2q⁻; 17q+)

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IN an analysis of 243 identifiable break points from 129 cases of presumptive reciprocal translocation, Ford and Clegg (1) noted 32 breaks involving group A chromosomes and 32 breaks involving members of group E. The present report outlines the clinical, dermatoglyphic and cytogenetic features of a case with an apparent 46, XX, t (2q⁻; 17q+) karyotype.

Case Report:

B.N., a 2750 g. female infant was born four weeks before term. The pregnancy had been complicated by frequent severe headaches and labour was eventually induced because of pre-eclamptic toxaemia. At this time B.N.'s father was aged 28 years and her mother 33 years. Initial clinical findings were marked hypertelorism, hypotonia, mild bilateral talipes calcaneus and left hip subluxation. Head circumference was 33 cm.

In subsequent follow-up, persistent hypotonia with motor and mental retardation were noted. The results of urinary chromatography were normal. Psychological assessment at age 30 months revealed an estimated mental age of 14.2 months.

When 3 years old the child was admitted to hospital for full clinical evaluation. She was then able to walk somewhat unsteadily. Although apparently interested in her surroundings, she spoke only a few, mostly unintelligible, words.

Physical examination showed unusual facies with striking hypertelorism, depressed nasal bridge, long upper lip and small jaw (Fig. 1). Her left upper eyelid drooped slightly and lacked a skin crease. The eyes were otherwise normal. Internal examination of the nose and throat revealed no abnormality. The ears were low set and mis-shapen. Each ear had a prominent helix with poorly developed antihelix, tragus and antitragus. The auditory canals and middle ears were normal and deafness was excluded by audiometry. Examination of the skull showed cebocephaly and a cranial circumference of 43 cm. There were no abnormal findings in the heart, lungs or abdomen. Both limbs were normal; the mild hip and heel de-

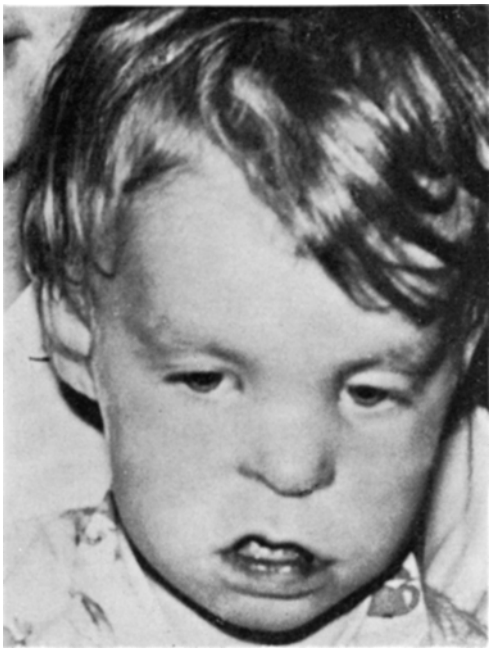

Fig. 1—B. N., aged 3 years, showing marked hypertelorism.

formities noted at birth had apparently been spontaneously corrected. Further psychological evaluation was difficult because of poor co-operation but the previous impression of moderate mental handicap was confirmed.

Dermatoglyphics

Examination of the patient's palm prints showed bilateral distal triradii. Due to preponderance of digital arch patterns the total ridge count was very low (Table 1). Tiny loop patterns were seen in the left second interdigital and right third interdigital areas. An ulnar loop was present in the left hypothenar area.

TABLE I

Patient's dermatoglyphics : Digital patterns and ridge counts.

	LEFT HAND					RIGHT HAND				
DIGIT	1	2	3	4	5	1	2	3	4	5
PATTERN	UL	A	A	UL	A	UL	A	UL	TA	TA
RIDGE COUNT	0-9	0	0	0-7	0	6-0	0	4-0	0	0
	TOTAL RIDGE COUNT 26									

Cytogenetic Investigation

Repeated chromosome analyses (leucocyte culture) on the patient showed apparent trisomy 3 with monosomy 2 and a group E chromosome with un-

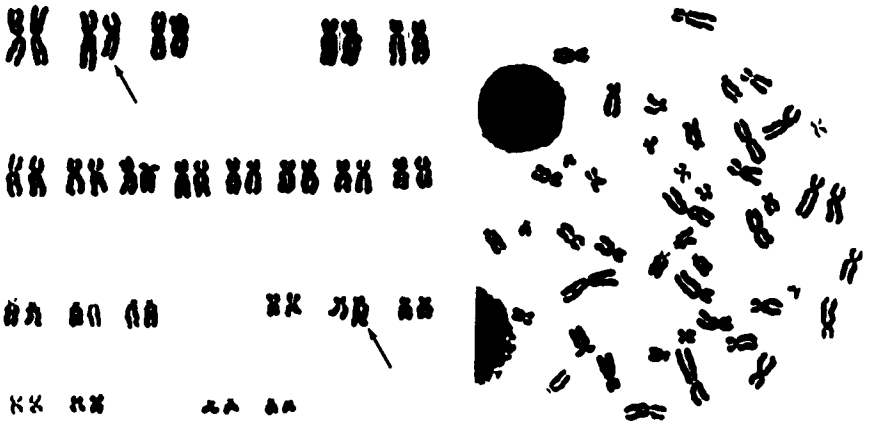

Fig. 2—Metaphase and corresponding karyogram of B. N.

usually long long arms (Fig. 2). These findings were interpreted as representing presumptive reciprocal translocation between 2 q and the long arm of a group E chromosome (Fig. 2). On morphological grounds the group E chromosome involved was provisionally identified as a no. 17. The karyotypes of the parents and of a normal sister born subsequently were unremarkable. Figure 3 shows three sets of group A and group E chromosomes from the patient.

Discussion

It has been estimated that about 0.5 per cent of the adult population are carriers of a structural chromosome rearrangement (2). In most instances, balanced reciprocal translocation is associated with a normal phenotype. Chromosomal findings in the patient described here are consistent with reciprocal exchange of material between a no. 2 and a no. 17 chromosome.

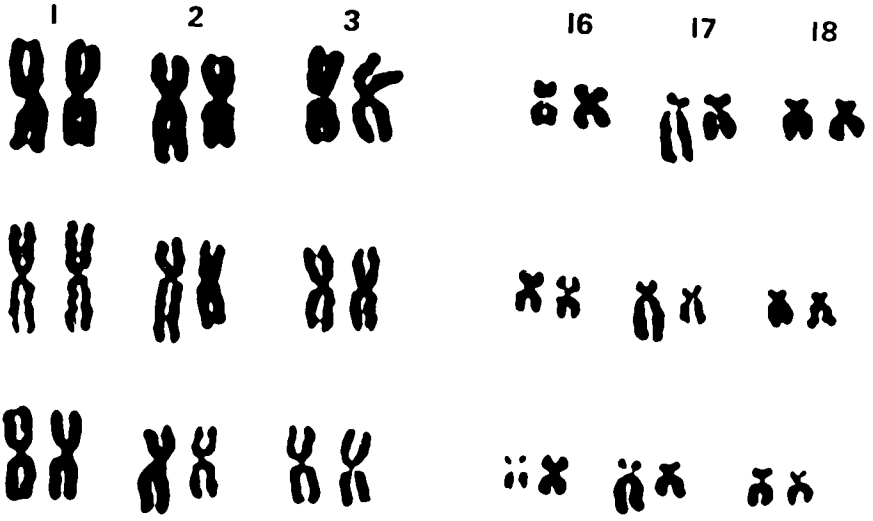

Fig. 3—Selection of group A and group E chromosomes from patient.

The parental karyotypes are normal and there is no evidence of mosaicism in either the propositus or the parents. The affected child is therefore presumably the carrier of a balanced translocation which arose in gametogenesis.

It is possible that the observed mental retardation and physical anomalies in our patient are merely co-incidental. Alternatively, however, the clinical abnormalities may be due to a position effect of the translocated fragments. Cowie *et al.* (3) have reported multiple congenital abnormalities in a child with an apparently balanced karyotype carrying a D/F reciprocal translocation. More recently Mantle *et al.* (4) have described a case of mental retardation and congenital anomalies with an apparently balanced translocation 46, XY, t ($2q^-$; Bq^+). The morphology of the affected no. 2 chromosome in their case is very similar to that of the child reported here. The importance of position effect in balanced reciprocal translocation is difficult to assess. Abundant evidence indicates that it has no direct phenotypic significance in most instances. Its relevance to the aetiology of mental retardation and congenital malformation may eventually be established by comparing the prevalence of apparently balanced reciprocal translocations in special institutions with that in the general population.

Summary

Clinical, dermatoglyphic and cytogenetic features of a child with apparently balanced karyotype carrying a presumptive 2/E translocation are presented. The relevance of the karyotypic anomaly to the clinical findings is briefly discussed.

Acknowledgment

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