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Interstitial del(13)(q21·3q31) associated with psychomotor retardation, eczema, and absent suck and swallowing reflex

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SUMMARY A patient with a deletion (13)(q21·3q31) showed only eczema and absent suck and swallowing reflex, in contrast to other well documented cases with a similar deletion. Apparently there is wide clinical variability in patients with deletions in this area.

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Cases with partial deletions of the long arm of chromosome 13 constitute a clinically and cytogenetically heterogeneous group. We describe a patient with a del(13)(q21·3q31) and compare our findings with well documented cases of deletions in the same region.

Case report

The proband, a boy, was the fourth child of healthy, unrelated parents. After an uneventful pregnancy the infant was born at term weighing 2635 g. In the neonatal period he was observed in another hospital for one week for possible sepsis. Three months later he was admitted because of failure to thrive and feeding difficulties.

He was a small boy (weight 3720 g, length 54 cm, head circumference 37 cm) with extensive eczema (fig 1). Retardation of psychomotor development, muscular hypotonia, weak tendon reflexes, absent suck and swallowing reflex, and inability to follow with the eyes were present. He had a partly depigmented right iris and low set, normal ears. No cardiac murmur was audible. There were recurrent respiratory and urinary tract infections. Radiography revealed no urinary tract anomalies. A CT scan of the brain was normal.

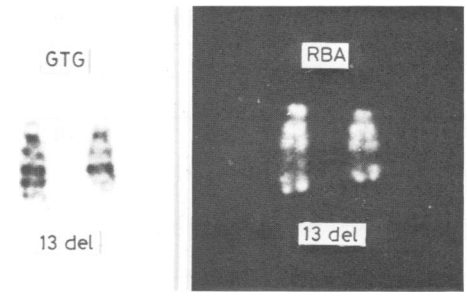

FIG 2 *GTG and RBA banded chromosome 13.*

CYTOGENETIC STUDIES

GTG and RBA banded chromosomes from blood lymphocytes and skin fibroblasts showed a 46,XY,del(13)(pter→q21·3::q31→qter) karyotype in all metaphases analysed (fig 2).

Both parents had normal chromosomes.

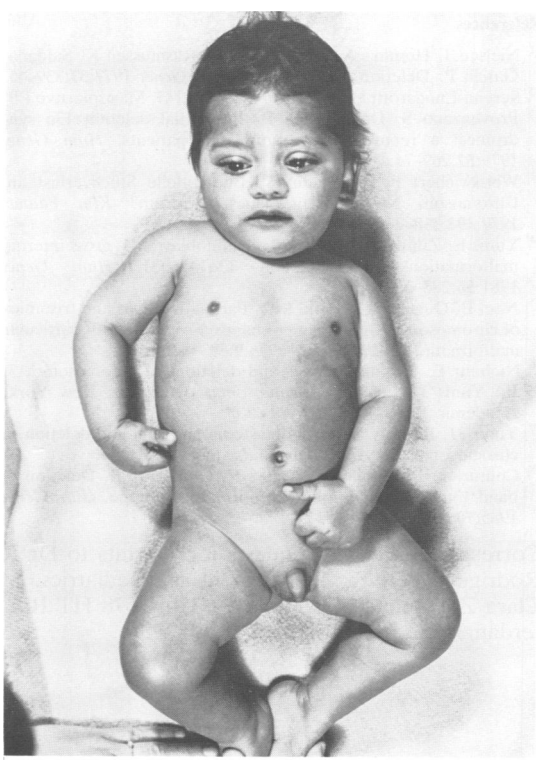

FIG 1 *The proband at five months.*

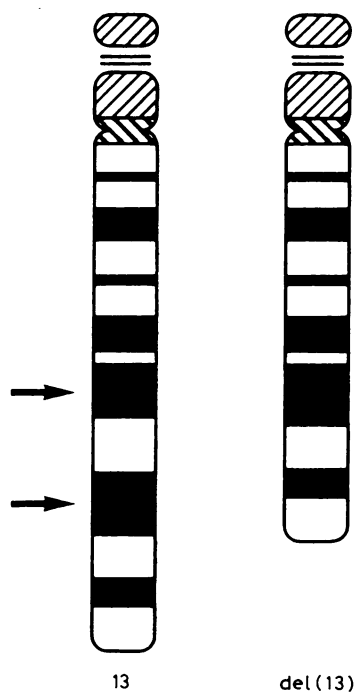

FIG 3 *Schematic diagram of del(13)(q21·3q31).*

TABLE *Clinical and cytogenetic features in 13q monosomy.*

	Present case	*Reference*				
		1	*2*	*3*	*4*	*5*
Extent of deletion	21·3→31	21→31	21→31	21·3→31·2	14→31	21→22
Sex	M	F	F	F	M	F
Birth weight <2800 g	+	+	+	+	+	
Psychomotor retardation	+	+	+	+		+
Hypotonia	+			+		+
Generalised dermatitis	+					
Microcephaly	+		+	+		+
Absent suck and swallowing reflex	+					
Partial iris depigmentation	+					
Low set or malformed ears	+	+	+	+	+	
Short neck	−	+	+	+		
Prominent nasal bridge	−		+	−		+
Broad nasal bridge	−		+	+	+	
Hypertelorism	−			+	+	
Microphthalmia	−			−		+
Epicanthus	−			+		
Retinoblastoma	−			−	+	−
Hypoplastic 5th digit + clinodactyly	−			+		−
Cardiac defect	−		+			−
Multiple intestinal malformations	−				+	−

Discussion

Clinical symptoms associated with 13q deletions are variable. Comparison of cases with deletions in the same region as our patient (table)[1–5] does not help to establish a definite clinical pattern[6]; psychomotor retardation seems to be the most constant finding.

Gross intestinal malformations are possibly associated with deletions in the region q13→q31,[4 7] and deletion of band 13q14 is associated with retinoblastoma.[7] Our case shows absence of suck and swallowing reflexes which has not previously been reported, and extensive eczema; dermatitis has been reported in one other case.[7]

More distal deletions may give a recognisable clinical picture. Deletion of band 13q34 is probably responsible for the craniofacial dysmorphism classically described in the 13q monosomy syndrome.[6] Thumb hypoplasia is variably present in deletion 13q34.[6] The wide phenotypical variability is also demonstrated by the normal phenotype in a case with a deletion of band 13q21.[8] This was explained by the fact that this band is one of the latest replicating, which may indicate that it carries genetic material which is not transcriptionally active.

Establishing a definite phenotype for deletions in this region, if at all possible, will certainly require more cases studied by high resolution banding techniques.

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