Discussion

The most frequently reported interstitial deletion of 2q involves the segments del(2)(q31q33) and the clinical features of the other eight reported cases are outlined in the table. It is apparent that in addition to the general features shared with other 2q deletions (mental retardation, microcephaly, growth failure, and congenital heart defects),[1-3] more specific features of del(2)(q31q33) deletions, as suggested by Schinzel,[9] include microphthalmia, corneal anomalies, ptosis, a beaked nose, micrognathia, cleft palate, large or low set ears, clinodactyly of the fifth finger, camptodactyly of the fingers, and syndactyly of the toes.

The present case shares some of the features of this subgroup of 2q deletions but he also shows distinctive skin pigmentation. The distribution of the skin abnormality did not follow Blaschko's lines and we found no evidence of chromosomal mosaicism by demonstrating the identical karyotypes in the fibroblasts derived from the pigmented and non-pigmented skin. The skin pigmentation may be related to the breakpoints of this deletion allowing expression of an otherwise suppressed gene, or it may represent a coincidental abnormality; further assignment of gene loci to 2q31–q33 may resolve this question.

The structural gene for the soluble form of isocitrate dehydrogenase (ICD–S, E.C.1.42) has been previously mapped to 2q33.3 by somatic cell hybridisation and gene dosage studies.[10] The presence of normal levels of ICD-S in the proband suggested that the deletion breakpoint in band q33 lies proximal to the ICD-S locus.

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A terminal deletion (14)(q31.1) in a child with microcephaly, narrow palate, gingival hypertrophy, protuberant ears, and mild mental retardation

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SUMMARY A female child with a terminal deletion on the long arm of chromosome 14, 46,XX,del(14)(q31.1), presented with microcephaly, narrow palate, gingival hypertrophy, protuberant ears, and a small haemangioma on the back. She was mildly mentally retarded. Only a few patients with a partial deletion of 14q (14q−) have been reported without consistent clinical findings. Although a clinical syndrome associated with ring chromosome 14, r(14), has been established, no distinct pattern has been so far reported in 14q−.

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Five patients with 14q− have been reported.[1-4] Three patients had interstitial deletions (fig 1, cases 1, 2, and 3). One patient had a terminal deletion (fig

1, case 4). The remaining patient had a chromosome mosaicism with two cell lines, one with a 46,XX karyotype. The other cell line had a complex inversion and a terminal deletion (fig 1, case 5). Patients with a similar phenotypic pattern associated with r(14) have been observed, establishing a clinical entity.[5]

We wish to report a patient with a terminal deletion (14)(q31.1) (fig 1, case 6). The patients with 14q− have so far not presented with similar clinical features.[1–4] Whether or not this is because of the deletion of different chromosome segments of 14q is not known (fig 1). Reported features vary and are noted in the table.

Case report

The proband (fig 2) was born on 17.4.79 and was seen in the Child Evaluation Center at five years three months and again at seven years four months

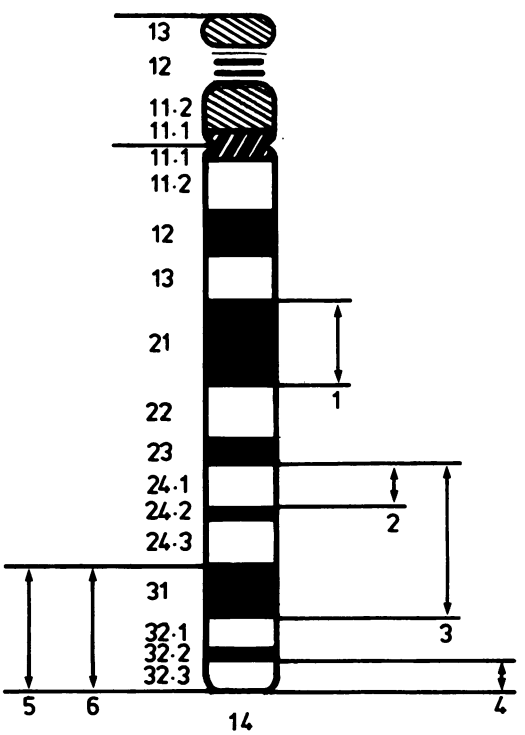

FIG 1 *Schematic representation of deleted chromosome segments of 14q in six patients. Case 1: del(14)(q13q22).[1] Case 2: del(14)(q23q24.2).[2] Case 3: del(14)(q23q32).[2] Case 4: del(14)(q32.3).[3] Case 5: mos 46,XX/46,XX,inv(14)(q21q31),del(14)(q31)=46%:54%.[4] Case 6: del(14)(q31.1), present case.*

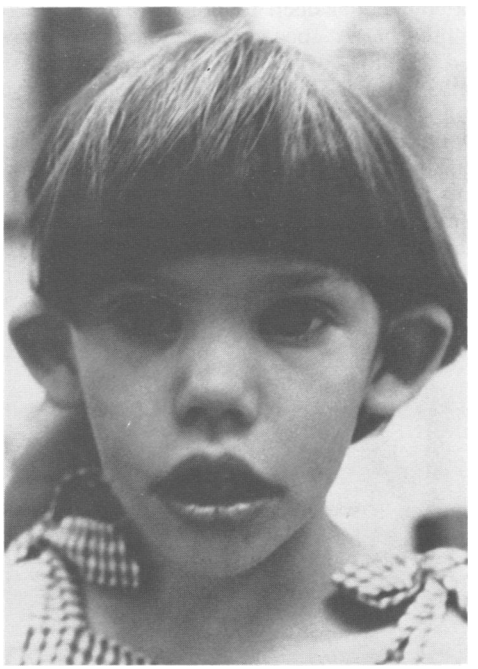

FIG 2 *The proband.*

of age. She was the younger of two living children. At the time of her birth, her father and mother were 32 and 29 years of age, respectively. The pregnancy, apart from excessive weight gain, and the delivery were uncomplicated. Birth weight was 4400 g, length was 52 cm, and head circumference was 34·3 cm (50th centile). She was noted to have a fractured right clavicle at birth. Her motor milestones were reported to be in the normal range. She used words at nine months of age, but sentences were not present until four years of age. She was reported to have had one febrile convulsion at 18 months. The EEG was normal.

Physical examination at seven years four months of age showed she was at the 40th centile for height (120 cm) and the 25th centile for weight (20 kg). Her head circumference of 46·2 cm was below the 2nd centile. She was a thin, slightly apprehensive, Caucasian female. Physical findings included microcephaly, protuberant ears, epicanthic folds, perforated left tympanic membrane, micrognathia, pointed chin, tight tongue frenulum, narrowed palate, gingival hypertrophy, open bite, and blue sclerae. She was myopic. A faded haemangioma was present on her back. Neurological examination indicated poor tongue control, with otherwise normal cranial nerves. There was incoordination with

brisk, but equal deep tendon reflexes. Hypotonia was present. A speech articulation problem was noted. Scoliosis was reported at seven years two months of age. The sensory, autonomic, and cerebellar systems were normal.

Dermatoglyphic studies were unremarkable. The urine amino acid screening chromatogram showed a normal pattern. The tests for reducing substances and mucopolysaccharides were normal. Skull *x* rays indicated that the calvarium was small compared to the size of the face, consistent with microcephaly. No abnormal findings were noted on the CT scan of the brain.

Psychological studies obtained when she was five years three months of age indicated an IQ of 68 on the Stanford-Binet Intelligence Scale and an IQ of 84 on the Leiter International Scale. When she was seven years four months, psychological testing using the Stanford-Binet test indicated an IQ of 64. When measured by the WISC-R, a verbal IQ of 52, a performance IQ of 57, and a full scale IQ of 50 was obtained. Her receptive and expressive language skills were depressed and relatively lower than the IQ scores. A mild speech articulation problem was noted.

Chromosome studies indicated that the child had a 46,XX,del(14)(q31.1) karyotype by GTG banding

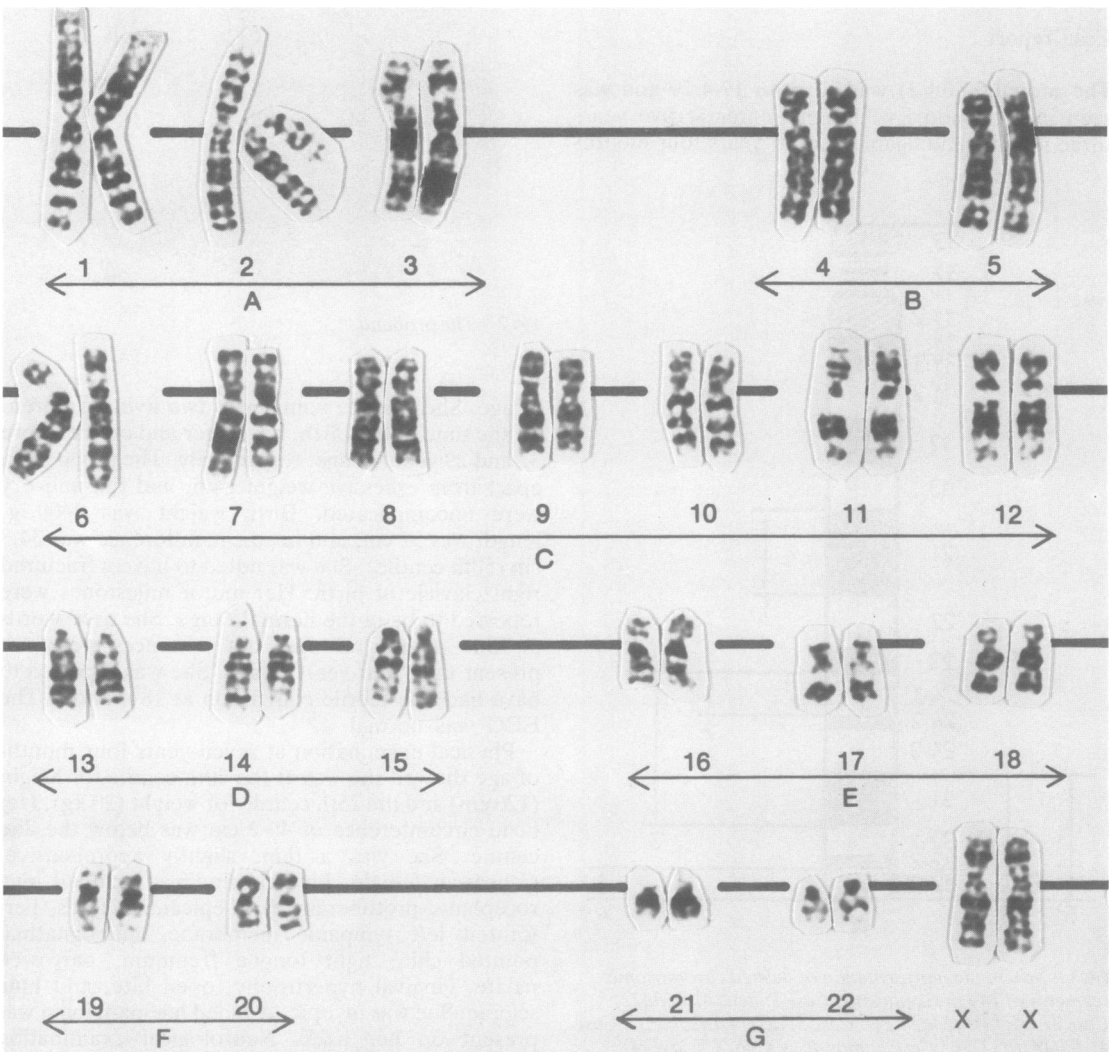

FIG 3 *GTG banded karyotype of the proband, 46,XX,del(14)(q31.1).*

TABLE *Comparison of clinical features in the proband with those with 14q− or r(14) in published reports.*

Dysmorphic features	*Features in 14q−*						*Features in r(14)*
	Case						
	1	*2*	*3*	*4*	*5*	*6*	
Head							
Dolichocephaly			+				+
Microcephaly						+	+
High forehead				+			+
Short neck					+		+
Eyes							
Small palpebral fissures			+				+
Ptosis			+				
Congenital glaucoma	+						
Epicanthic folds			+			+	+
Synophrys		+	+				+
Strabismus		+	+		+		+
Ears							
Large, protuberant		+	+			+	+
Folded over helix		+	+				
Poor helix formation				+			+
Preauricular skin tags		+					
Low set, malformed					+		+
Nose							
Short, bulbous		+	+				+
Depressed nasal bridge		+	+				+
Broad nasal bridge				+			+
Mouth							
Cleft lip and palate	+						
Micrognathia			+	+	+	+	+
High arched/narrow palate				+	+	+	+
Pointed chin		+				+	
Broad philtrum				+		+	+
Orthopaedic							
Leg and foot abnormalities					+		+
Arm and hand abnormalities		+	+		+		+
Hip luxation					+		+
Scoliosis		+				+	+
Other							
Congenital heart disease		+	+	+			+
Genitourinary abnormality		+	+				+
Seizures		+			+		+
Mental retardation		+	+		+	+	+

techniques from peripheral leucocyte cultures (fig 3). Chromosome studies on her parents showed normal karyotypes.

Discussion

Common findings in r(14) syndrome are noted in the table. There have been five cases of 14q− reported.[1–4] These are summarised in the table including findings in the case reported. Although some common features are reported, at present they are not consistent enough to establish a clinical entity. Some of the features seen in 14q− are seen in r(14), where deletions in both the short and the long arms of chromosome 14 have been reported.

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Familial distal trisomy 8(q24.13→qter)

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SUMMARY Trisomy for the distal part of the long arm of chromosome 8(q24.13→qter) is described in three sibs. The anomaly arose as an adjacent 1 meiotic segregation from a balanced reciprocal translocation t(1;8)(q44; q24.13)mat.

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