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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References

- Buchanan P, Rao K, Doerr C, Aylsworth A. A complex translocation involving chromosomes 3, 11, and 14 with an interstitial deletion, del(14)(q13q22) in a child with congenital glaucoma and cleft lip and palate. *Birth Defects* 1978;XIV (6c):317-22.
- Turleau C, de Grouchy J, Chavin-Colin F, *et al*. Two patients with interstitial del(14q), one with features of Holt-Oram syndrome. Exclusion mapping of PI (alpha-1-antitrypsin). *Ann Genet (Paris)* 1984;27:237-40.
- Hreidarsson S, Stamberg J. Distal monosomy 14 not associated with ring formation. *J Med Genet* 1983;20:147-9.
- Nielsen J, Homma A, Rasmussen K, Ried E, Sorensen K, Saldana-Garcia P. Deletion 14q and pericentric inversion 14. *J Med Genet* 1978;15:236-8.
- Schmidt R, Eviatar L, Nitowsky HM, Wong M, Miranda S. Ring chromosome 14: a distinct clinical entity. *J Med Genet* 1981;18:304-7.

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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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Common clinical features include infants born at term with early feeding difficulties and jaundice, mild mental retardation, delayed development, hypertelorism, broad nasal bridge, prominent philtrum, thin, downturned upper lip, mild micrognathia and retrognathia, low set ears, short fifth finger, camptodactyly, clinodactyly, and dermatoglyphic abnormalities.

Family history

Three sibs (IV.1, IV.3, IV.4, fig 1) are trisomic for chromosome 8(q24.13→qter) as a result of inheriting a der(1) chromosome from a balanced reciprocal translocation $t(1;8)(q44;q24.13)mat$ (fig 2). The children have mild mental retardation, hypertelorism, bushy eyebrows, low set, dysplastic ears (unravelling helix), and mild micrognathia and retrognathia. The noses are quite distinctive. Impressive are the short columella and broad alae nasi, giving a snubbed appearance, as well as the wide root and bridge of the nose and blunt nasofrontal angle. All have a thin, downturned upper lip (fig 3). There is clinodactyly and camptodactyly of the fifth fingers and a wide space between the first and second toes (fig 3). The skin is dry and

coarse. Dentition, palate, tongue, and mucosa are normal.

Case reports

iv.1

IV.1 is a 12 year old girl. Birth weight was 3000 g at 39 weeks' gestation. Apgar scores were 9 and 10 at one and five minutes. No external abnormality was noted, other than mild talipes. At the age of 12 years, her height is 137 cm (10th centile) and weight 24.1 kg (3rd centile). She is hirsute, with hypertrichosis on the back and limbs. There are no ocular abnormalities. The neck is short but not webbed (fig 3). She has a long, slender trunk and normal external genitalia. Her general health is good.

iv.3

IV.3 is a four and a half year old boy. Birth weight was 3800 g at 41 weeks' gestation. Apgar scores were 8 and 9 at one and five minutes. Mild cerebral palsy was diagnosed at six months, but is now improving. General health has been good, apart from a single febrile convulsion at the age of three years. There is a lifelong history of short stature. At the age of four and a half years, he weighs 15.0 kg (3rd centile) and is 99 cm tall (3rd centile). There is hypertelorism and a right

FIG 1 Pedigree of family.

FIG 2 GTG banded partial karyotypes of III.1 showing $t(1;8)$. Broken dotted lines indicate breakpoints. Affected subjects have inherited the $der(1)$.

convergent squint (fig 3). There is an undescended right testis.

IV.4

IV.4 is a three year old boy. Birth weight was 3770 g at term. Fetal distress was noted with meconium staining of the liquor and poor Apgar scores (6 and 7 at one and five minutes). A symptomless cardiac murmur was noted on day four and he went into cardiac failure on day seven. He was not cyanosed at any time. Chest x ray showed generalised cardiomegaly and pulmonary plethora. ECG showed right axis deviation and right ventricular strain. A 2D echocardiogram was unremarkable. A clinical diagnosis of ventricular septal defect with moderate left to right shunt was made. Cardiac failure responded promptly to medical treatment with digitalisation and diuretics. He has had no subsequent cardiovascular symptoms and his murmur has become softer, presumably because of spontaneous partial closure of his VSD.

There is a lifelong history of short stature. At the age of two years three months he weighed 11.0 kg (3rd centile) and was 83.5 cm tall (3rd centile). He has a right undescended testis.

III.5

III.5 is a 28 year old female. She was jaundiced at birth and had early feeding problems, failure to thrive, talipes, and developmental delay. She has strabismus, poor speech, and is mildly mentally retarded. Chromosome studies show a $46,XX,-1,+der(1),t(1;8)(q44;q24.13)mat$ karyotype. No other information is available.

III.6

III.6 was a female child who died at the age of 10 years of cardiac failure. There was jaundice, failure to thrive, developmental delay, and talipes. She had convergent strabismus and was mentally retarded. No chromosome studies were undertaken. No other information is available.

III.7

III.7 was a male child who lived for only 12 hours. He had hydrocephalus and spina bifida. No chromosome studies were performed. No other information is available.

III.8

III.8 is an intellectually handicapped male. Chromosome studies show a $46,XY,-1,+der(1),t(1;8)(q44;q24.13)mat$ karyotype. No other information is available.

Dermatoglyphics

The three affected sibs (IV.1, IV.3, and IV.4) show

very similar dermatoglyphic features. These features show considerable similarity to those thought to be typical of mosaic trisomy 8,^{1 2} although no patient had a simian crease, pattern intensity on palms and

soles was unremarkable, and arches were not a prominent fingertip pattern. The fingertip counts were arches 3, radial loops 2, ulnar loops 15, and whorls 10. However, the mean total finger ridge

FIG 3 Frontal and lateral facies and right hands and feet of the patients. (a) IV.1 at 12 years; (b) IV.3 at four years; (c) IV.4 at three years. Note hypertelorism, wide root and bridge of nose, thin, downturned upper lip, coarse, dry skin, clinodactyly, short little finger, and deviation of the hallux.

Case reports

count was reduced at 100. The mean *atd* angle was 60°. Arches on the big toes were present on three prints that could be clearly visualised. One patient had bilateral hallual whorls, in one patient a whorl was present on one foot, and the other patient had no hallual whorls.

Discussion

Distal trisomy for 8q24→qter has been reported on only four previous occasions.³⁻⁶ With the exception of the case reports by Soudek *et al*⁴ and Gódde-Salz *et al*,⁵ where the fullest of clinical descriptions is not given, the table shows the major features in patients reported so far with trisomy for 8q24→qter. It is most probable that in the cases of Sanchez and Yunis³ and in the patient of Gódde-Salz *et al*⁵ the extra secondary chromosomal material 22pter→q12 has little or no effect on phenotype. The same may be said for our patients and that of Barnes *et al*,⁶

where monosomy for the most terminal regions of 1q and 8p is involved.

Taking into account age differential and natural somatic variation, our three children (IV.1, IV.3, and IV.4) are remarkable for their striking similarity of triangular facies and distinctive noses. They have many features in common with other trisomy 8q24→qter patients and those of the two patients described by Schinzel⁷ with trisomy 8q23.3→qter, notably delayed development, hypertelorism, broad nasal bridge, micrognathia, retrognathia, low set ears, low hair line, short fifth finger, clinodactyly, camptodactyly, and mental retardation. They do not, however, have the more severe skeletal abnormalities and mental retardation as described in the cases of Sanchez and Yunis³ or as associated with trisomy for larger segments of 8q and complete trisomy 8 mosaicism.⁸

We thank Mrs H Dockery for analysis of the

TABLE Clinical manifestations in reported cases of 8q24→qter.

Clinical observations	Sanchez and Yunis ³		Soudek <i>et al</i> ⁴	Gódde-Salz <i>et al</i> ⁵	Barnes <i>et al</i> ⁶	Present cases		
	Case 1	Case 2				IV.1	IV.3	IV.4
Partial trisomy 8q segment	8q24.1→qter	8q24.1→qter	8q24.1→qter	8q24.1→qter	8q24.1→qter	8q24.13→qter	8q24.13→qter	8q24.13→qter
Secondary trisomic segment	22pter→q12	22pter→q12	14pter→q21	22pter→q12	—	—	—	—
Monosomy	—	—	—	—	8p23.3→pter	1q44→qter	1q44→qter	1q44→qter
Sex	Male	Female	Female	Male	Female	Female	Male	Male
Age (y)	18	29	4 2/12	4 10/12	3	12	4	3
Gestation	Term	—	—	43 weeks	38 weeks	39 weeks	41 weeks	Term
Normal birth weight	—	+	+	+	+	+	+	+
Physiological jaundice	—	—	—	—	+	+	+	+
Early feeding problems	—	—	+	—	+	+	+	+
Developmental delay	+	+	+	+	+	+	+	+
Growth retardation	+	+	+	+	+	+	+	+
Mental retardation	+	+	+	+	+	+	+	+
Face shape	Triangular	Triangular	—	Broad	Broad	Triangular	Triangular	Triangular
Prominent forehead	+	+	+	+	+	+	+	+
Hypertelorism	+	+	+	+	+	+	+	+
Coloboma	—	—	—	—	—	—	—	—
Strabismus	—	—	—	—	—	—	Convergent	+
Broad/depressed nasal bridge	Large	Large	—	+	+	+	+	+
Thick, everted lower lip	+	—	—	—	+	+	+	+
Downturned/thin upper lip	—	—	+	—	+	+	+	+
Prominent philtrum	—	—	+	—	+	+	+	+
Retrognathia	—	+	—	—	—	+	+	+
Micrognathia	—	—	—	+	+	+	+	+
Low set ears	+	+	+	+	+	+	+	+
Prominent ears	+	+	—	+	Small	+	+	+
Low hair line	+	+	—	—	+	+	+	+
Cleft palate	—	—	—	—	—	—	—	—
High arched palate	+	+	—	—	—	—	—	—
Short neck	Webbed	—	—	—	—	—	—	—
Widely spaced nipples	—	—	—	—	—	—	—	—
Slender trunk	+	+	—	—	—	+	+	+
Dry, coarse skin	—	—	—	—	—	+	+	+
Severe skeletal abnormalities	+	+	—	—	—	—	—	—
Genitalia	Normal	Normal	—	—	Cryptorchidism	Normal	Undescended R testis	Undescended R testis
Short fifth finger	+	+	—	—	+	+	+	+
Camptodactyly	+	—	—	—	—	+	+	+
Clinodactyly	+	—	—	—	+	+	+	+
Dermatoglyphic abnormalities	—	—	—	—	—	+	+	+
Foot abnormalities	+	+	—	—	—	+	+	+
Heart defects	—	—	+	—	—	—	—	+

dermatoglyphics, Mrs H Lawton for typing the manuscript, and the Audiovisual Unit of the Wellington Clinical School for help with the illustrations.

References

- ¹ Schauman B, Alter M. *Dermatoglyphics in medical disorders*. New York: Springer-Verlag, 1976:169-72.
- ² Rodewald A, Zankl H, Wischerath H, Borkowsky-Fehr B. Dermatoglyphic patterns in trisomy 8 syndrome. *Clin Genet* 1977;12:28-38.
- ³ Sanchez O, Yunis JJ. Partial trisomy 8(8q24) and the trisomy 8 syndrome. *Humangenetik* 1974;23:297-303.
- ⁴ Soudek D, Hunter P, O'Shaughnessy S, Simpson NE, Soudek V. Familial translocation t(8;14) with a case of tertiary trisomy, +14q. In: Summitt RL, Bergsma D, eds. *Sex differentiation and chromosome abnormalities*. New York: Alan R Liss. For the National Foundation—March of Dimes. Birth Defects 1978; XIV(6C):309-15.
- ⁵ Götde-Salz E, Oesinghaus S, Grote W. Meiotic segregation in familial reciprocal translocation t(8q;2q). *Am J Med Genet* 1982;11:241-7.
- ⁶ Barnes ICS, Kumar D, Bell RJM. A child with a recombinant of chromosome 8 inherited from her carrier mother. *J Med Genet* 1985;22:67-70.
- ⁷ Schinzel A. Partial trisomy 8q in half-sisters with distinct dysmorphic patterns not similar to the trisomy 8 mosaicism syndrome. *Hum Genet* 1977;37:17-26.
- ⁸ Walker AP, Bocian M. Partial duplication 8q12-q21.2 in two sibs with maternally derived insertional and reciprocal translocations. Case reports and review of partial duplications of chromosome 8. *Am J Med Genet* 1987;27:3-22.

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Is there an autosomal recessive form of the split hand and split foot malformation?

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SUMMARY A family is described in which four subjects in two sibships had typical split hand/foot malformation. The possibility of the existence of a rare autosomal recessive form of the malformation is discussed, as well as a two locus model. In the two locus model the dominant mutation leading to the split hand/foot malformation is controlled by a gene at another locus. A dominant mutation at the controlling locus leads to non-penetrance of the split hand/foot mutation.

Isolated split hand/split foot malformation is well known but rare. Many cases are sporadic, presumed to be new dominant mutations, since in most families with more than one affected subject autosomal dominant inheritance can be shown.¹ We present a family with four patients in two related sibships affected with the split hand/foot malformation.

Case report

The family (fig 1) originates from a small, very

inbred Muslim community. The parents of the proband are double first cousins and are healthy with no malformations. The maternal grandfather of the proband (III.4) as well as two of his sisters (III.2 and III.11) were also affected.

The proband was born after a pregnancy in which the mother suffered from depression and was treated with Halidol (haloperidol), Artane (trihexyphenidyl), and Largactil (chlorpromazine). The delivery was normal and the girl was normally developed for her age. At examination in our clinic at the age of 10 months, the only malformations were a left typical split hand and bilateral split foot (fig 2). There was complete syndactyly of the first and second fingers, the third and fourth fingers were fused into one finger with two nails, and the fifth finger was normal (fig 2a). In both feet (fig 2b, c), the second toe was absent and syndactyly of the third and fourth toes was noted. In the grandfather (III.4), there were only three fingers, the first, fourth, and fifth, on both hands. In III.2, there was split hand only on the right with similar findings as in the hand of the proband. Both hands were normal in III.11. In all these three affected subjects, bilateral split foot was present with a malformation similar to that observed in the proband.

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