A malformed girl with duplication of chromosome 9q

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SUMMARY A case of a 1 year old girl with a duplication of the long arm of chromosome 9 is described: 46,XX,dir dup(9)(q12→q32). The clinical features of the child are strikingly similar to those of the two cases reported by Turleau *et al*¹ and Faed *et al*.² The similarity of the abnormalities may help in recognising this syndrome by its characteristic dysmorphic features.

Case report

The child, a girl, birth weight 2300 g, was born at 37 weeks' gestation after an uneventful pregnancy when her mother and her father were 33 and 38 years of age, respectively. A 3 year old sister was normal. There was no history of abortion, stillbirth, or neonatal death.

She was referred to us when she was 8 months old. Physical examination revealed delayed growth and mental retardation, her weight being 5565 g (−3 SD), length 62.5 cm (−2.5 SD), and head circumference 37.5 cm (−4.2 SD). Other features included pale eczematous skin, microdolichocephaly, a narrow slender face, deep orbits, horizontal palpebral fissures, slight hypotelorism, bilateral epicanthic folds, beaked nose, thin upper lip with a receding lower lip, low set ears, congenital dislocation of the left hip joint, and small hands with relatively long tapering fingers (fig 1). Both thumbs were held in the flexed position and pressed firmly against the palm.

Dermatoglyphic study revealed bilateral simian creases, low axial triradii, and the following fingertip pattern: WL, W, W, W, Ur, WL, U, W, WL, and UI.

CYTOGENETIC STUDIES

Chromosomes were obtained by standard lymphocyte culture. One chromosome 9 was unusually long. Chromosomes were examined further by C banding, high resolution banding,³ and DA-DAPI staining.⁴ The abnormal chromosome 9 had the direct duplication: dir dup(9) (q12→q32) (fig 2). Both parents had a normal karyotype.

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FIG 1 The patient at 14 months of age.

FIG 2 Partial karyotype of the patient. From left: two pairs of DA-DAPI (top) and DAPI (bottom) stained chromosomes 9. The latter was obtained after destaining of the former. Within each pair, dup (9) is placed on the left. Two pairs of G banded chromosomes 9 (top) as well as a high resolution banded dup (9) (bottom) are also shown.

TABLE Summary of clinical findings in three cases of trisomy 9q.

	Turleau et al ¹	Faed et al ²	Present case
Trisomic segment	q11→q33	q12→q32	q12→q32
Pregnancy (wk)	40	36	37
Birth weight (g)	2300	1730	2300
Length (cm)	50		44
Head circumference (cm)	32.5		29
Sex	M	M	F
Asphyxia or neonatal problems	+	+	—
Microdolichocephaly	+	+	+
Slender face	+	(+)	+
Hypotelorism	+	— (Wide set)	+
Horizontal palpebral fissures	+	+	+
Epicanthic folds	+	+	+
Strabismus	+	+	—
Nose	Beaked	Beaked	Beaked
Mouth	Small	Small	Small
Lips			Thin upper lip overlaps lower lip
Micrognathia	+	+	+
Abnormality of the thumbs	+		+
Hip joint		Flexion contracture	Unilateral dislocation
Feet	Long toes with abnormal implantation		Normal
Internal malformation	Ventricular septal defect		
Thriving	Poor	Poor	Poor
Mental retardation	+	+	+
Natural history	Alive at 5 years	Alive at 4 years	Alive at 2 years

Discussion

As far as we are aware, there have been only two reported cases of trisomy 9q in which most of the long arm is involved (table). The clinical features common to all three cases are: low birth weight, failure to thrive, mental retardation, narrow face, epicanthic folds, beaked nose, small mouth, micrognathia, and good life expectancy during infancy. These similarities should make it possible to recognise this abnormality on the basis of clinical findings. It is remarkable that the presence of such a large extra segment has a relatively mild effect on the patient.

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References

- 1 Turleau C, de Grouchy J, Chavin-Colin F, et al. Partial trisomy 9q: a new syndrome. *Humangenetik* 1975;**29**: 233–41.
- 2 Faed M, Robertson J, Brown S, Smail PJ, Muckhart R. Pure partial trisomy for long arm of chromosome 9. *J Med Genet* 1976;**13**:239–42.
- 3 Matsubara T, Nakagome Y. High-resolution banding by treating cells with acridine orange before fixation. *Cytogenet Cell Genet* 1983;**35**:148–51.
- 4 Schweizer D, Ambros P, Andrie M. Modification of DAPI banding on human chromosomes by prestaining with a DBA-binding oligopeptide antibiotic, distamycin A. *Exp Cell Res* 1978;**111**:327–32.

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Familial centric fission of chromosome 4

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SUMMARY A centric fission of chromosome 4 is described in the proband and his mother, both phenotypically normal. In addition, partial monosomy for the long arm or the short arm or

both of chromosome 4 may have been present in two of the proband's sibs who died in infancy and childhood, respectively.

Centric fission of a chromosome consists of a break in the centromeric region, which results in the