Case reports

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Partial monosomy 3q in a boy with short stature, developmental delay, and mild dysmorphic features

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SUMMARY We describe the clinical and cytogenetic findings in a boy with an unbalanced karyotype involving monosomy for 3q27-q29. He does not resemble other reported cases of del(3q). Deletions of the long arm of chromosome 3 are extremely rare, having been reported in five cases, only two of which had terminal 3q deletions.

healthy, non-consanguineous parents. The family history was negative and there is a healthy brother and sister. The pregnancy was uneventful and the child was born at term after a normal delivery, weighing 2610 g. He was a floppy baby and subsequent development was slow; he first walked at two and a half years.

He was referred to the Genetics Clinic at eight years of age for assessment. At this stage his length

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The proband, an eight year old male, was born to

FIG 1 Facial appearance of the proband at eight years.

FIG 2 G banded partial karyotypes of the proband with the deleted chromosome on the right. The idiogram of chromosome 3 illustrates the breakpoint at 3q27.

Received for publication 18 May 1989.
Accepted for publication 1 June 1989.

TABLE Cytogenetic and clinical findings in patients with terminal 3q deletion.

	Alvarez Arratia <i>et al</i> ⁴	Sargent <i>et al</i> ⁵	Present case
Deleted segment	3q28-qter	3q27-qter	3q27-qter
Gestation	40 weeks	37 weeks	40 weeks
Birth weight	1700 g	3170 g	2610 g
Failure to thrive	+	+	-
Short stature	+	-	+
Developmental delay	+	+	+
Hypotonia	+	+	+
Microcephaly	+	+	-
Skull shape	Dolichocephaly	Trigonocephaly	Normal
Sparse hair	+	-	-
Synophrys	-	+	-
Hypoplastic nails	+	-	-
Angiomata	-	-	+
Microphthalmia	+	-	-
Strabismus	-	-	+ Left eye
Broad nose	+	+	+
Telecanthus	+	+	-
Ears	Low set, malformed	Low set	Small, posteriorly rotated
Long, smooth philtrum	-	-	+
Lip and palate	Cleft lip and palate	-	High arched palate
Retromicrognathia	+	-	-
Short neck	+	+	-
Thoracic abnormalities	13 thoracic vertebrae and pairs of ribs	-	Kyphosis
Cardiac defect	Systolic murmur, cardiomegaly	Death from acute pericarditis	-
Recurrent infections	+	-	-
CT scan		Cerebral atrophy, absence of cerebellar vermis, ventricular dilatation, posterior fossa cyst	Normal
Survival	Died at 3 months	Died at 26 months	Alive and well

was 110 cm (below the 3rd centile), but his head circumference of 50.5 cm was on the 10th centile. He was noted to have several dysmorphic features (fig 1) including small, posteriorly rotated ears, a high arched palate, and a smooth philtrum. He had a squint in his left eye, mild thoracic kyphosis, and a resolving haemangioma on his thoracic spine (a capillary haemangioma on each eyelid had resolved previously). He had obvious developmental delay, with speech particularly retarded. A CT brain scan showed no abnormality.

Chromosome analysis showed a terminal deletion of the long arm of chromosome 3, with a breakpoint at 3q27, the unbalanced karyotype being 46,XY, del(3)(pter→q27:) (fig 2). The parental karyotypes were normal.

An alternative interpretation is that of de novo,

unbalanced translocation 46,XY,-3,+der(3),t(3;?) (q27;?) with monosomy 3q and simultaneous trisomy for a telomeric region of unknown origin.

Discussion

From the clinical viewpoint there is very little similarity between the five published cases of 3q deletion¹⁻⁵ and our patient. Since the three patients with terminal 3q deletion are cytogenetically similar, all being monosomic for bands q28 and q29, some phenotypic resemblance might be expected. However, the findings in common are non-specific, such as developmental delay, hypotonia, and ear abnormalities. The table summarises the clinical features of these patients. Features shared by the other two cases,⁴⁻⁵ but not by our patient, include microcephaly, abnormal skull shape, failure to thrive, telecanthus, and death from cardiac failure (? cause) before three years of age. The other three cases of known partial monosomy 3q are all interstitial deletions which overlap with one another but not the present case; the deleted segments were q22.1-q24,¹ q23-q26,² and q23-q25.³ These three had the following in common: growth retardation, developmental delay, telecanthus, a long, thin philtrum, and low set ears. The lack of phenotypic similarity, especially for more specific features such as microphthalmos, cleft lip and palate, retromicrognathia and skull shape, makes delineation of a 3q- syndrome difficult. Given the paucity of reported cases of 3q deletion and the clinical variability seen among these patients, recognition of a 3q- phenotype remains elusive.

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