mild mental retardation and relatively mild dysmorphic features, is striking. That only four patients with this deletion have been described until now could be due to the fact that in the past no chromosome analysis was done routinely in such cases. As bands q14 and q22 are of similar size and density it is not possible to determine which band is missing. Discrimination between these two bands based upon gene marker studies is not possible because, according to Shows *et al*⁶ and Geurts van Kessel *et al*,⁷ no genes have been assigned to either of these bands.

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Interstitial deletion 2q24·3: case report with high resolution banding

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SUMMARY Interstitial deletions of the long arm of chromosome 2, involving band 2q24, have been described on three occasions.¹⁻³ We report our findings in a further case, in which we mapped the deletion to band 2q24·3.

Case report

The proband was the second child of non-consanguineous parents. Both parents and a sib were in good health. Pregnancy was complicated by amniotic leak at about seven months, but this subsided spontaneously and did not require treatment. Delivery was at term and birth weight was 2300 g. The umbilical cord had three vessels, but the placenta was small. The baby was brought to our attention on the second day of life because of lethargy, hypotonia, feeding difficulties, and dysmorphic features, which included microcephaly (31·5 cm), low set and posteriorly rotated ears, with poor formation of the upper auricular helix, short

(1·5 cm) and downward slanting palpebral fissures, high nasal bridge, and a short philtrum (fig 1). Micrognathia was evident, the neck was short, and the genitalia were normal except for an undescended right testis. Palmar creases were normal. Dermatoglyphs of the right hand showed arches on the index and third fingers; all others, including the left hand, were ulnar loops. During the first four months of life he improved little. He was feeding well but with some occasional regurgitation. The

FIG 1 Proband at 4 months of age.

head circumference at 4 months was 37.5 cm (below the 3rd centile) and apart from the aforementioned, new findings were cranial asymmetry, prominent ears, upper midline labiokingival frenulum, clenched hands, a heart murmur, and laryngeal stridor. Shortly after 4 months, he presented in status epilepticus. CT scan showed mild ventricular enlargement with some degree of cortical atrophy. A two dimensional echocardiogram revealed no abnormalities. Ophthalmological examination showed no abnormalities. The stridor was attributed to laryngomalacia. The patient failed to thrive, his weight at 8 months being 6.1 kg, and had several respiratory tract infections, dying at 16 months after such an episode. Necropsy was performed but details were not available to us.

CYTOGENETIC STUDIES

High resolution G banding studies using RPMI 1640 medium with glutamine and fetal calf serum, methotrexate for cell synchronisation, and bromodeoxyuridine indicated that band 2q24.3 (fig 2) was missing. Parental chromosomes were studied with a standard trypsin-Giemsa technique and were normal. No high resolution was attempted on parental chromosomes.

Discussion

Shabtai *et al*¹ suggested that deletion of bands q23→31 of 2q may be associated with a specific syndrome. They compared their case to those reported by Fryns *et al*² and McConnell *et al*.³ The only bands that were uniformly involved in these patients were q23 and q24. Because our patient had only a small deletion of band q24, it is of interest to compare his phenotype with these other patients

(table). The features that our proband had in common with them were psychomotor retardation, low birth weight and failure to thrive, microcephaly, low set ears, clenched hands, and cranial sutural irregularities. Except for this last finding, all the others are features general to many chromosomal syndromes. Other features mentioned as characteristic of this syndrome were ptosis, cataracts, long

FIG 2 High resolution banding studies. The deleted chromosome is represented on the right. The diagram shows the deleted band to be 2q24.3.

TABLE Common features of the four patients with deletions of bands q23 and q24.

	Fryns <i>et al</i> ² 2q21→24	Shabtai <i>et al</i> ¹ 2q23→31	McConnell <i>et al</i> ³ 2q22→31	Present case 2q24.3
Psychomotor retardation	+	+	+	+
Low birth weight and/ or failure to thrive	+	+	+	+
Microcephaly	-	+	+	+
Cranial sutural irregularities	-	-	+	+
Low set ears	-	+	+	+
Small face	+	+	-	-
Downward slanting palpebral fissures	-	-	±	+
Microphthalmia	+	+	-	-
Cataracts	+	+	-	-
Cleft of soft palate	+	+	+	-
Clenched hands	+	-	+	+
Long fingers and toes	+	+	+	-
Short philtrum	-	-	-	+
Upper labiokingival frenulum	-	-	-	+
Short neck	+	-	-	+

fingers and toes, cleft of the soft palate, and cleft between the first and second toes. These were not present in our patient. Since our patient only had a small deletion of band q24, it is possible that the similar phenotype reported by Shabtai *et al*¹ was due to the deletion which included band q23. The studies reported before did not use high resolution banding analysis. We regard it as important to use this technique in order to map accurately small deletions. It could be that patients considered to have overlapping deletions when standard techniques are applied have in fact different segments involved. This could account for the different phenotype found in our patient. With more widespread use of high resolution techniques, a better delineation of clinical syndromes seems possible. Our proband died at 16 months following an upper respiratory tract infection. The ages of death in the other cases

were at 8 months,² 20 years,¹ and at birth.³ The survival up to 20 years in the patient reported by Shabtai *et al*¹ could be due to her mosaic state.

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Hyperinsulinaemic hypoglycaemia in an infant with mosaic trisomy 13

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SUMMARY An infant with mosaic trisomy 13, who was small for gestational age, became severely hypoglycaemic. For the first 19 days of life, glucose requirements to maintain normoglycaemia were high (up to 21.7 mg/kg/min) and at the same time the infant had high plasma insulin levels and low glucose insulin ratios. Treatment with hydrocortisone and susphrine was of questionable benefit. Hyperinsulinism abated by the third week of life. This case illustrates early remission of hyperinsulinaemic hypoglycaemia and raises the possibility of an association with trisomy 13.

Hypoglycaemia, although common in the premature or small for gestational age neonate, is rarely caused by hyperinsulinaemia in the neonatal period. Causes of neonatal hyperinsulinaemic hypoglycaemia are nesidioblastosis, adenomatoid dysplasia, islet cell adenoma, ectopic pancreatic tissue, and Beckwith-Wiedemann syndrome.¹

This paper describes a mosaic trisomy 13 infant, small for gestational age, who developed hyperinsulinaemic hypoglycaemia. This association has not hitherto been reported.

Case report

The patient was a 2400 g, 37 week female, small for gestational age, delivered to a 25 year old non-diabetic mother. The pregnancy was complicated by mild hypertension. The family history was negative for congenital malformations or mental handicaps. The infant was born by vaginal delivery and required endotracheal intubation and suction because of the presence of meconium beyond the vocal cords. The Apgar scores were 7 and 8 at one and five minutes, respectively. Because of meconium aspiration syndrome, the infant required mechanical ventilation for 12 hours followed by oxygen therapy for an additional three days.

The physical examination showed the following findings: malformed low set ears with small pinna, hypertelorism, elongated philtrum, micrognathia, elongated retroflexible thumbs held in a cortical position with clenched fists, and a two vessel cord.