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. and Geurts van 
Kessel et al., no genes have been assigned to either of these bands.

The authors wish to thank Ms W de Korte-Lodder and S C E Schaminee-Main for technical help in the 
cytogenetic studies.

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

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Sciences, Los Angeles, California 90024, USA.

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 dysmorphi 
f features, which included microcephaly (31.5 cm), low set and posteriorly rotated ears, with 
poor formation of the upper auricular helix, short

FIG 1 Proband at 4 months of age.

(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 unde-
scended right testis. Palmar creases were normal. 
Palmatoglyphs 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
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 labiogingival 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 bro-
modeoxyuridine 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 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 and McConnell et al. 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 character-
istic of this syndrome were ptosis, cataracts, long

<table>
<thead>
<tr>
<th>TABLE Common features of the four patients with deletions of bands q23 and q24.</th>
</tr>
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<tbody>
<tr>
<td>Psychomotor retardation</td>
</tr>
<tr>
<td>Low birth weight and/or failure to thrive</td>
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<tr>
<td>Microcephaly</td>
</tr>
<tr>
<td>Cranial sutural irregularities</td>
</tr>
<tr>
<td>Low set ears</td>
</tr>
<tr>
<td>Small face</td>
</tr>
<tr>
<td>Downward slanting palpebral fissures</td>
</tr>
<tr>
<td>Microphthalmia</td>
</tr>
<tr>
<td>Cataracts</td>
</tr>
<tr>
<td>Cleft of soft palate</td>
</tr>
<tr>
<td>Clenched hands</td>
</tr>
<tr>
<td>Long fingers and toes</td>
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<tr>
<td>Short phalirum</td>
</tr>
<tr>
<td>Upper labiogingival frenulum</td>
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<tr>
<td>Short neck</td>
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</tbody>
</table>

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

References

