Case reports

Interstitial deletion of the long arm of chromosome 2 with normal levels of isocitrate dehydrogenase

I A Glass†, C A Swindlehurst*, D A Aitken*, W McCrea‡, and E Boyd*
*Duncan Guthrie Institute of Medical Genetics, and †Department of Child Health, Royal Hospital for Sick Children, Yorkhill, Glasgow G3 8SJ; and ‡Royal Scottish National Hospital, Larbert, Stirlingshire.

Summary We report a 16 year old boy with the abnormal karyotype 46,XY,del(2)(q32.2q33.1) who has mental retardation, microcephaly, epilepsy, craniofacial dysmorphism, distinctive scalloped skin pigmentation, and normal levels of isocitrate dehydrogenase.

Over 20 persons with interstitial deletions of the long arm of chromosome 2 have been described.1-3

We report a further patient with an interstitial deletion with breakpoints del(2)(q32.2q33.1) which have not previously been reported, who has distinctive clinical features including an unusual pattern of skin pigmentation. We compare his cytogenetic and clinical findings with those of eight previously reported cases in the largest subgroup of 2q deletions: del(2)(q31q33).2-8

Case report

The proband is the third child of healthy, unrelated parents, the mother being 23 years at delivery. The pregnancy was normal until 32 weeks of gestation.

FIG 1 The proband aged 16 years.
when premature delivery occurred following an antepartum haemorrhage. The birth weight was 2070 g. Global developmental retardation and epilepsy were apparent from early childhood and ultimately resulted in his admission to an institution for the mentally handicapped. Evaluation at 16 years showed mental retardation with no comprehensible speech and total dependency, microcephaly (OFC 47 cm, \(-5.3\) SD), short stature (149.5 cm, \(-3.2\) SD), a large beaked nose, bilateral corneal ectasia, divergent strabismus, bilateral ptosis, and a cleft palate (fig 1). He has a striking pattern of scalloped skin pigmentation, present from birth, which is approximately symmetrical on the trunk and proximal limbs and clearly demarcated from the normal skin (fig 2). His gait was slow and jerky but there were no other neurological signs.

An EEG showed diffuse abnormalities on an irregular polyrhythmic background.

A G banded karyotype of peripheral lymphocytes showed an interstitial deletion of the long arm of chromosome 2: del(2)(q32.2q33.1) (fig 3). Skin fibroblasts from both normal skin and pigmented skin showed the same karyotype: 46,XY del(2)(q32.2q33.1). The parents were unavailable for study.

Assay of the activity of the soluble form of isocitrate dehydrogenase (ICD-S, E.C.1.42) activity in red cells from the index case gave normal activity (1.13 IU/g Hb, mean of 18 controls 0.93 IU/g Hb, SD 0.28).
<table>
<thead>
<tr>
<th>Deletion</th>
<th>Sex</th>
<th>Mental retardation</th>
<th>Prenatal growth failure</th>
<th>Postnatal growth failure</th>
<th>Microcephaly</th>
<th>Prominent forehead</th>
<th>Seizures or abnormal EEG</th>
<th>Microphthalmia</th>
<th>Corneal abnormality</th>
<th>Peculiarities</th>
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<tbody>
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<td>2q32-3q33</td>
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<td>(Bilateral ectasia)</td>
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**Other features**

- Strabismus, skin pigmentation
- Tapering and overlapping digits
- Bilateral iris colobomata
- Macrostomia, joint laxity
- Father shown to have balanced intrachromosomal translocation
- 46,XY, t(2q32→2p13)

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**TABLE Clinical features in cases of interstitial deletion of 2q31-q33.**
Discussion

The most frequently received features of del(2)(q31q33) are cleft palate and mental retardation.

References


Correspondence and requests for reprints to Dr I A Glass, Duncan Guthrie Institute of Medical Genetics, Yorkhill, Glasgow G3 8SJ.

A terminal deletion (14)(q31.1) in a child with microcephaly, narrow palate, gingival hypertrophy, protuberant ears, and mild mental retardation

FU-SUN YEN, PHILIP E PODRUCH, AND BERNARD WEIsskOPF
Child Evaluation Center, Department of Pediatrics, University of Louisville School of Medicine, Louisville, Kentucky, USA.

SUMMARY A female child with a terminal deletion on the long arm of chromosome 14, 46,XX,del(14)(q31.1), presented with microcephaly, narrow palate, gingival hypertrophy, protuberant ears, and a small haemangioma on the back. She was mildly mentally retarded. Only a few patients with a partial deletion of 14q (14q-) have been reported, without consistent clinical findings. Although a clinical syndrome associated with ring chromosome 14, r(14), has been established, no distinct pattern has been so far reported in 14q-.

Five patients with 14q-- have been reported.1-4 Three patients had interstitial deletions (fig 1, cases 1, 2, and 3). One patient had a terminal deletion (fig 1, case 4).