An interstitial deletion of chromosome 7(q35)

Kerry Fagan, Christine Kennedy, Laurence Roddick, Alison Colley

Abstract
We describe a patient with developmental delay, mild dysmorphic features, and monosomy of 7q35. Only one other patient with an interstitial deletion of this band has been previously reported. A review of clinical features of these two children did not show similarities in dysmorphic features. Reports of patients with other 7q interstitial deletions are listed.

(Cytogenetic Studies
Chromosome studies performed on the patient at birth and at the age of 2 years showed an apparently normal female karyotype. On re-examination at 11 years using high resolution cytogenetic techniques, a small interstitial deletion of band q35 was detectable on one chromosome 7 (fig 2). This constitutes a loss of only 0.18% of HAL (haploid autosomal length). The karyotype can be described as: 46,XX, del(7)(pter→q34::q36→qter). The chromosomes, painted with a chromosome 7 specific probe (Cambio) by fluorescent in situ hybridisation, did not show evidence of a cryptic translocation or insertion. The parental chromosomes were normal.

Discussion
Deletions of interstitial segments of the long arm of chromosome 7 are still relatively rare. A review of published reports indicates several broad groups of 7q deletions which have breakpoints and phenotypic features in common (fig 3).

A deletion which includes the G band negative region of q11 results in severe growth and developmental retardation and a poor prognosis.1 Spasms and seizures are common and most patients remain non-ambulatory.

Four patients with interstitial deletions of chromosome 7 which included loss of band q21 have been reported.12 These patients all had split hand/split foot malformations among other dysmorphic features.

The most common interstitial deletion reported for chromosome 7, del(7)(q22→q31), has been described in 12 patients so far and a characteristic clinical picture is now emerging, including severe developmental retardation, an unusual cry in infancy, a large mouth, seizures, and feeding or gastro-esophageal difficulties.

Patients with interstitial deletions in the q3 region of chromosome 7 include three members of one family with monosomy for bands

Cytogenetics Unit, Hunter Area Pathology Service, The John Hunter Hospital, Locked Bag No 1, Newcastle Mall Exchange, NSW 2305, Australia
R Fagan
C Kennedy

Department of Paediatrics, The John Hunter Hospital, Newcastle, NSW, Australia
L Roddick

Regional Medical Genetics Unit, PO Box 84, Waratah, NSW, Australia
A Colley

Correspondence to Dr Fagan.
Received 11 February 1994
Revised version accepted for publication 8 April 1994
An interstitial deletion of chromosome 7(q35) 739

Figure 1  Facial appearance at 11 years.

Figure 2  G banded partial karyotype. Deleted chromosome 7 is on the right.

Figure 3  Diagram of 7q interstitial deletions in reported cases.

q32→q34.1 Their deletion resulted from malsegregation of an inversion carrier and the patients all had similar phenotypic features, including mental and developmental retardation, external ear malformation, hypertelorism, flat nasal bridge, a bulbous nose tip, and a wide mouth. A single case report of a deletion of bands q33 and q34 (patient 3) had microcephaly, prominent forehead, mongoloid slant of the eyes, bilateral simian creases, and cleft oralmegaly, as well as severe growth and developmental retardation. One patient described3 had a deletion of bands 7q33→q35. Her features included microcephaly, neurodevelopmental delay, a cleft lip and palate, conductive deafness, and protruding tongue.

One patient has monosomy for band 7q35, similar to our patient, but it resulted from a de novo translocation t(3;7)(q27;q35).2 This patient was severely hypotonic at birth with absent Moro reflex; she had microbrachycephaly, ptosis of the left eyelid, alternate squint, small nose with a flat nasal bridge, and microstomia. The growth rate had been below the 3rd centile. Seizures occurred shortly after birth and a profound neural hearing deficit was documented. By comparison, our patient shows only mild dysmorphism but has a squint in common with their patient.

Conclusions
The clinical features of patients described with interstitial deletions of different segments of the long arm of chromosome 7 vary. More case reports are needed with deletion breakpoints accurately defined using high resolution banding to confirm whether a pattern of phenotypic characteristics can be described. Our case reiterates the need to repeat testing when a chromosome anomaly is suspected, despite an initially normal karyotype.

EBV transformation of the patient’s lymphocytes is in process and will be available on request.