

Interstitial deletion of the long arm of chromosome 2: $\text{del}(2)(\text{q}31\text{q}33)$

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SUMMARY A child with a de novo interstitial deletion, $46,\text{XX},\text{del}(2)(\text{q}31\text{q}33)$, is described. Clinical features included psychomotor retardation, hypotonia, microcephaly, hypertelorism, downward slanting palpebral fissures, macrostomia, cleft palate, micrognathia, abnormal ears, overlapping fingers, simian creases, and rocker bottom feet.

Partial trisomies and monosomies of chromosome 2 are rare but several cases of balanced rearrangements involving this chromosome have been reported. We report a case of a de novo interstitial deletion of the long arm of chromosome 2 in a $2\frac{1}{2}$ -year-old girl.

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Case report

The proband was born to a 19-year-old primiparous mother and a 30-year-old father, who were first cousins from Bangladesh. There was no history of exposure to drugs or radiation. Pregnancy and delivery at 38 weeks were normal. The child weighed 2.09 kg at birth and required special care in the neonatal unit because of her low birth weight and multiple congenital abnormalities, including cleft palate which caused feeding difficulties. She had two episodes of febrile convulsions at the ages of 1 and 2 years.

On physical examination at the age of $2\frac{1}{2}$ years the child showed delayed developmental milestones and generalised hypotonia with inability to walk, stand alone, or say any words. Her height, weight, and

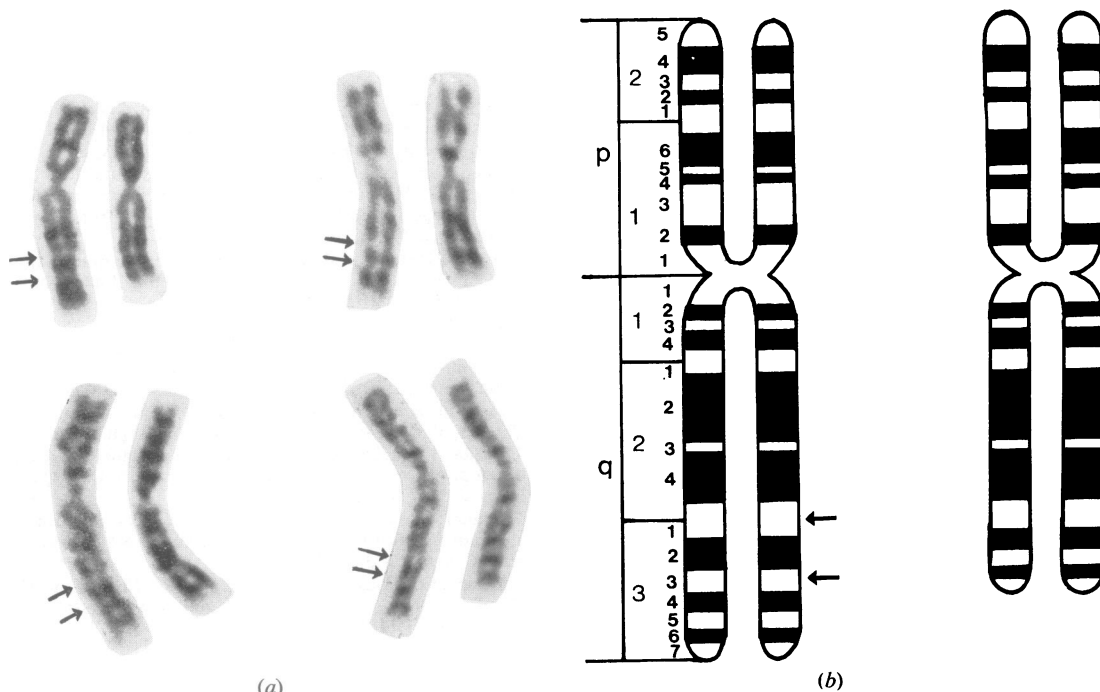


FIGURE (a) Four pairs of chromosome 2 from the proband (trypsin-Giemsa banding). (b) Diagrammatic representation of the normal and deleted chromosome 2. The deleted chromosomes $\text{del}(2)(\text{q}31\text{q}33)$ are on the right. The positions of the break-points are indicated by arrows on the normal chromosomes.

TABLE Clinical features in cases of interstitial deletion of 2q.

	Present case	German and Chaganti ¹	Warter <i>et al</i> ²	Fryns <i>et al</i> ³
Karyotype	46,XY,del(2)(q31q33)	46,XY,t(2;4)?del(12q14)	46,XX,t(2;6)del 2(q34;q36)	46,XX,del 2(q21q24)
Psychomotor and growth retardation	+	+	+	+
Microcephaly	+	+	+	—
Hypertelorism	+	—	+	—
Abnormal ears	+	—	+	—
Macrostomia	+	—	+	—
Dental malocclusion	+	—	+	—
Cleft palate	+	—	—	+
Micrognathia	+	—	—	+
Flexion deformity of fingers	+	—	—	+
Single palmar crease	+	—	+	—
Others	Prominent forehead, joint laxity, clinodactyly, rocker bottom feet, hypotonia	Musculoskeletal anomalies	Atrial septal defect	Microphthalmia, cataract, small face, relative macrocephaly

head circumference were all below the 3rd centile and her IQ was 50 on the Griffiths mental development scale. However, she appeared cheerful, responding with a broad smile when she was talked to. Her vision and hearing were apparently normal.

There was marked craniofacial dysmorphism including microcephaly, prominent forehead, round face, downward slanting palpebral fissures, sparse hair on the lateral half of the eyebrows, hypertelorism, flat nasal bridge, small nose, full upper lip, broad mouth, abnormal upper incisors, dental malocclusion, high arched cleft palate, narrow pointed tongue, maxillary hypoplasia, micrognathia, and small slightly low set ears with prominent antihelix. She had a short, broad neck but there was no webbing and the posterior hairline was normal. Her hands were clenched with bilateral overlapping fingers, clinodactyly of the fifth fingers, single palmar creases, and laxity of the interphalangeal, metacarpophalangeal, and wrist joints. She had rocker bottom feet and lax ankle joints. Her genitalia were female with hypoplastic labia minora and an anteriorly placed clitoris. There were no abnormalities of the chest, abdomen, and major organs.

Cytogenetic analysis using trypsin-G banding showed 46 chromosomes. There was an interstitial deletion in the long arm of chromosome 2 involving bands q31 to q33. The prominent dark band 2q32 was missing (figure a, b). The karyotype of the child was 46,XX,del(2)(pter→q31::q33→qter). The chromosomes of both parents were normal.

Discussion

Deletion of chromosome 2 is an extremely rare abnormality and no specific or tentative syndrome has been defined for it. Four cases with a deletion of the long arm of chromosome 2 have been reported (table). These, however, involved different segments so that it is not possible to make direct comparisons with their phenotypes. Nevertheless some phenotypic similarities are worthy of note, particularly between our case and that reported by Warter *et al.*²

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