and low set ears, clinodactyly and other abnormalities of fingers and toes, and cryptorchidism or micropenis or both.

In most cases with 12p- syndrome a larger deletion than in our patient was present, mostly including band 12p12. It is quite clear, however, that our proband has several features in common with the 12p deletion syndrome. Patients with a ring chromosome 12 have also been reported.^{5,6} They have several features in common with the 12p deletion syndrome and with our patient. The clinical features in these patients are⁵: failure to thrive, mental retardation, epicanthus, 'cupped', low set ears, ogival palate, short webbed neck, low hairline, and cryptorchidism. In the ring chromosome cases a partial deletion of 12p13, similar to that in our patient, is present.

Regarding the enzyme lactate dehydrogenase B (LDHB), no reduction in activity was noted in our patient. Decreased activity was found in the 12p deletion cases⁴ but not in the ring chromosome 12 case of Zuffardi *et al.*⁶ This seems to be in accordance with the gene localisation of LDHB at 12p12 and points to a similar 12p deletion in our

patient as in the ring chromosome 12 case of Zuffardi *et al.*⁶

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Interstitial deletion of the long arm of chromosome 11

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SUMMARY A girl with an interstitial deletion of the long arm of chromosome 11 is described. The patient was mildly mentally retarded and showed some facial dysmorphic features, including hypertelorism, ptosis, and cleft palate.

Since Jacobsen *et al.*¹ reported a family with a terminal deletion 11q, about 30 other cases have been published. These cases were all severely mentally retarded and presented with characteristic craniofacial anomalies: trigonocephaly, epicanthus, ptosis, flat nasal bridge, short bulbous nose with anteverted nostrils, triangular shaped mouth, retrognathia, and low set malformed ears. In all but one of the reported cases a de novo deletion of 11q, with loss of at least the terminal region q24→qter, was present. De novo interstitial deletions of the long arm of chromosome 11 have been reported in four infants by Taillemite *et al.*,² Sørensen *et al.*,³

McPherson and Meissner,⁴ and Bateman *et al.*⁵ Two of these cases were mildly mentally retarded and had trigonocephaly, hypertelorism, short bulbous nose, and high and narrow palate. In three cases low set ears were also present. In the case of Bateman *et al.*,⁵ who presented with Peter's anomaly, neither mental retardation nor dysmorphic signs were noted.

We report a girl with only mild mental retardation and some of the above mentioned phenotypic features in whom an interstitial deletion of the long arm of chromosome 11 was found.

Case report

The proband was the only child born to a young woman, who was reported to take drugs and drank alcohol in unknown quantities throughout pregnancy. The proband was removed from the custody of her mother and placed under the care of the juvenile court. According to the rules of this organisation no further information on the mother can be published

here. The father is unknown. Birth weight and length were not noted. Five days after birth the child was admitted to the Department of Paediatrics of a local hospital because of dyspepsia. Clinical examination at that time revealed a bifid uvula and a submucous palatal cleft. Her weight was 2350 g.

Re-examination at the age of 7.9 years showed a normally sized girl, height 123 cm (10th centile), weight 22.0 kg (10th centile), and head circumference 51 cm (50th centile) with hypertelorism, right ptosis, a bulbous nose with anteverted nostrils, large mouth, bifid uvula, and high arched and narrow palate (fig 1). Moreover, bilateral pes planovalgus and an increased space between the first and second toes were noted. Neurological examination revealed mild hypotonia with low reflexes. Electroencephalogram showed a diffuse non-specific abnormal pattern; CT scan revealed no cerebral abnormalities. On routine biochemical investigations no abnormalities were found. There was mild mental retardation (IQ=88) and speech development was

FIG 1 The proband at 7.9 years of age.

FIG 2 Chromosomes 11 of the proband (GTG banding). The right hand chromosome of each pair is the deleted 11q.

distinctly retarded. Audiological investigations showed a slight hearing loss (45 dB). Dermatoglyphic analysis revealed a simian crease in the right palm and bilateral absence of the *c* triradius.

CYTOGENETIC INVESTIGATIONS

Using GTG and RBA banding an interstitial deletion of the long arm of one of the chromosomes 11 was found, both in cultured lymphocytes (60 metaphases) and fibroblasts (100 metaphases). No metaphases with a normal karyotype were found and therefore mosaicism of del(11) seems improbable. GTG banding showed that one of the two dark bands corresponding to either q14 or q22 was lost. The karyotype thus was: 46,XX,del(11)(q13q21) or 46,XX,del(11)(q21q23).

Discussion

The partial deletion 11q syndrome (Jacobsen syndrome) due to terminal deletions of various size (q21, q22, q23, or q24) shows, besides mental retardation, a characteristic picture consisting of trigonocephaly presenting with keel shaped forehead, ptosis, bulbous nose, and triangular shaped mouth. In the five patients with an interstitial deletion of the long arm of chromosome 11 (including our case) these signs are less pronounced or even absent (table). This finding, loss of a part of an autosome associated with normal intelligence or

TABLE Clinical findings in the 11q- syndrome.

	Terminal deletion	Interstitial deletion	Present case
Sex (F/M)	22/7	2/2	F
Mean maternal age	27.6	25	NK
Mean paternal age	29.9	28	NK
Mean birth weight (g)	2692	3580	2350
Mean birth length (cm)	47.3	52.5	NK
Psychomotor retardation	22/22	2/4	(+)
Speech retardation	9/9	1/1	+
Hypertonia	4/11	1/1	—
Trigonocephaly	20/25	2/4	—
Antimongoloid slant	10/23	1/1	—
Strabismus	4/5	2/2	—
Epicanthus	15/23	1/1	—
Hypertelorism	19/24	2/2	+
Ptosis	11/15	0/1	+R
Flat nasal bridge	14/15	—	—
Short bulbous nose	21/21	2/2	+
Anteverted nostrils	7/7	—	+
High, narrow/cleft palate	14/14	2/2	+
Triangular shaped mouth	18/19	0/1	—
Long thin upper lip	12/14	—	—
Micro/retrognathia	20/23	1/2	—
Low set abnormal ears	24/26	3/3	—
Abnormal fingers/toes	23/24	1/1	—
Simian crease	19/24	1/2	+R
Cardiopathy	16/23	2/2	—
Hearing loss	2/2	—	+

Number informative/total number.

NK=not known.

(+)=mild.

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.