

Developmental delay and dysmorphic features associated with a previously undescribed deletion on chromosome 1

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Abstract

We report an 18 month old girl with developmental delay, dysmorphic features, and a karyotype 46,XX,del(1)(p32.1p32.3). To our knowledge the clinical features associated with this deletion have not been reported previously.

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Chromosomal abnormalities involving 1p are rare. We report the smallest deletion yet described, in a girl with developmental delay and multiple dysmorphic features.

Case report

This female subject was referred at the age of 18 months because of concern about her development. At that time she was unable to walk independently and had no identifiable speech. Her French father and English mother were unrelated and also have a healthy, 6 year old son. The pregnancy was complicated by recurrent bleeding but continued to term. Her birth weight, following a normal delivery, was 3550 g. A right preauricular skin tag was removed at birth but no other abnormalities were noted in the neonatal period.

On examination her appearance was rather cherubic (fig 1). Her ears were low set with a simple helical pattern. The nasal bridge and philtrum were prominent and her eyelashes

were long. A left divergent squint had previously been identified and hypermetropia of up to 3 dioptres was present. Fundoscopy was normal. There was no palatal defect and her palmar creases were normal. Her head circumference was just above the 50th centile for her age. Her nipples were widely spaced. Cardiovascular examination showed no abnormality and heart size was normal on chest x ray. A small umbilical hernia was present, there was no organomegaly, and her genitalia were normal. She was noted to be mildly hypotonic and had postural talipes calcaneovalgus affecting both feet.

A blood film showed no vacuolated lymphocytes. Routine chemistry, thyroid function, serum urate, creatine kinase, and a urine metabolic screen were all normal. A skeletal survey showed no bony abnormality. Serological testing for congenital infection was negative. High resolution G banded cytogenetic analysis of peripheral blood lymphocytes identified a small deletion from the short arm of one chromosome 1, 46,XX,del(1)(p32.1p32.3) (fig 2). Chromosome painting with a whole chromosome 1 paint showed no evidence of transfer of chromosome 1 material to another location. Both parents have normal chromosomes.

No fibroblast or lymphoblastoid cell line of the patient is available.

Discussion

Thirteen patients with short arm deletions of chromosome 1 were reviewed by Howard and Porteous¹ and further cases have been reported subsequently,^{2,3} but none has overlapped with the very small deletion identified in our patient. A prominent philtrum, low set ears, widely spaced nipples, and developmental delay were also reported by Yoshino *et al*³ in a girl with a del(1)(p32.3p34.1) immediately adjacent to that seen in our patient. These features, however, are frequently seen in chromosomal disorders and are unlikely to be specific for deletions of the p32 band.

More proximal deletions of the short arm of chromosome 1 between bands p22 and p31 have been associated with mental retardation, microphthalmia, colobomata, proximal insertion of the thumbs, and joint laxity in three patients,² while a high arched palate, hypertrichosis, and low set ears were observed with terminal short arm deletions (p34pter).¹ Nevertheless, the small number of cases, the limited overlap between reported deletions, and the non-specific nature of the dysmorphic fea-

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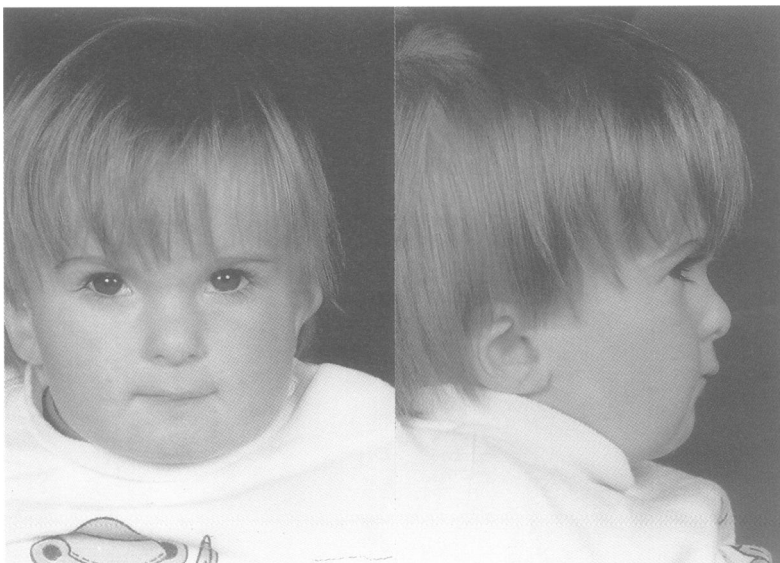


Figure 1 The patient's facial features at 18 months.

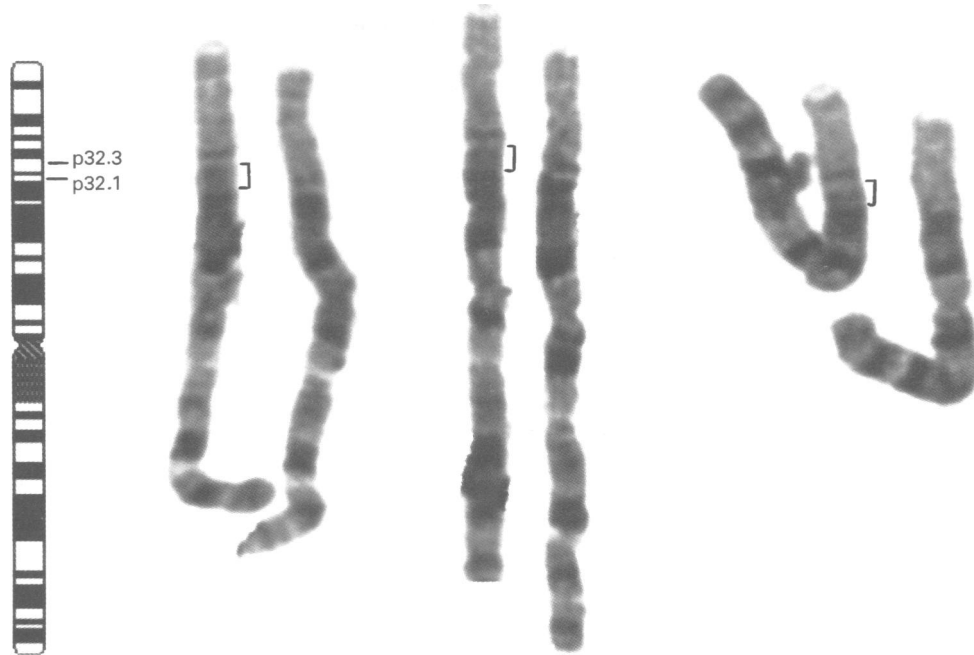


Figure 2 G banded homologues of chromosome 1 from three cells. The deleted chromosome, $del(1)(p32.1p32.3)$, is to the right, and the extent of the deletion is depicted by the bracket on the normal chromosome.

tures described still make it difficult to define a clinical phenotype related to specific interstitial 1p deletions.

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- 1 Howard PJ, Porteous M. Deletion of chromosome 1p: a short review. *Clin Genet* 1990;37:127-31.
- 2 Lai MMR, Robards MF, Berry AC, Fear CN, Hart C. Two cases of interstitial deletion 1p. *J Med Genet* 1991;28:128-30.
- 3 Yoshino M, Watanabe Y, Harada N, Abe K. De novo interstitial deletion of 1p (pter-p34::p32.2-qter). *J Med Genet* 1991;28:539-40.