Short report

An apparent de novo terminal deletion of chromosome 2 (pter→p24:)

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Deletions of 2p are rare and previous reports of isolated deletions have proven to be interstitial.[1–3] We report here the first instance of an apparent terminal deletion of 2p, confirmed by high resolution banding.

The male proband was delivered after a term pregnancy complicated by occasional use of alcohol and tobacco. The mother and father (20 and 23 years old respectively) were not related. Birth length was 47 cm (20th centile) and weight 2300 g (<10th centile). At the age of 3 months, dysmorphic features including highly arched palate, posteriorly rotated ears, single left transverse palmar crease, metatarsus adductus, Brushfield spots, micrognathia, depressed nasal bridge, and microcephaly were seen. Metabolic screening, thyroid indices, chest and hip radiographs, and banded metaphase chromosomes studied at that time were reported as normal, 46,XY. At 9 months of age, febrile seizures were treated with phenobarbital. Hearing impairment was evident by one year. He was profoundly developmentally delayed, sat at 13 months, walked at 5 years, did not talk until 7½ years, and was without bowel control at 8 years. Family history was negative.

At 8 years 5 months, physical examination showed microcephaly (47·2 cm, <5th centile), proportionate short stature (115 cm, <5th centile), weight of 19 kg (<5th centile), and a superficial resemblance to Down's syndrome (fig 1). Abnormal physical findings included brachycephaly with two occipital hair whorls, flat facial profile, redundant skin about the eyes, hypoplasia of the zygoma, underdeveloped columella, widened alveolar ridges, protruding ears with large lobes, bilateral short fifth fingers with single flexion creases and single transverse palmar creases, bilateral hallux valgus, and cutaneous syndactyly of the second and third toes. Global developmental delay (IQ 40) and profound hearing impairment were noted. An EEG showed an abnormal right sided spike discharge.

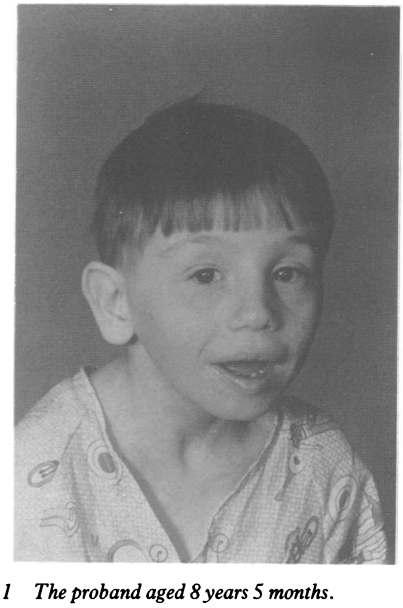

Figure 1 The proband aged 8 years 5 months.

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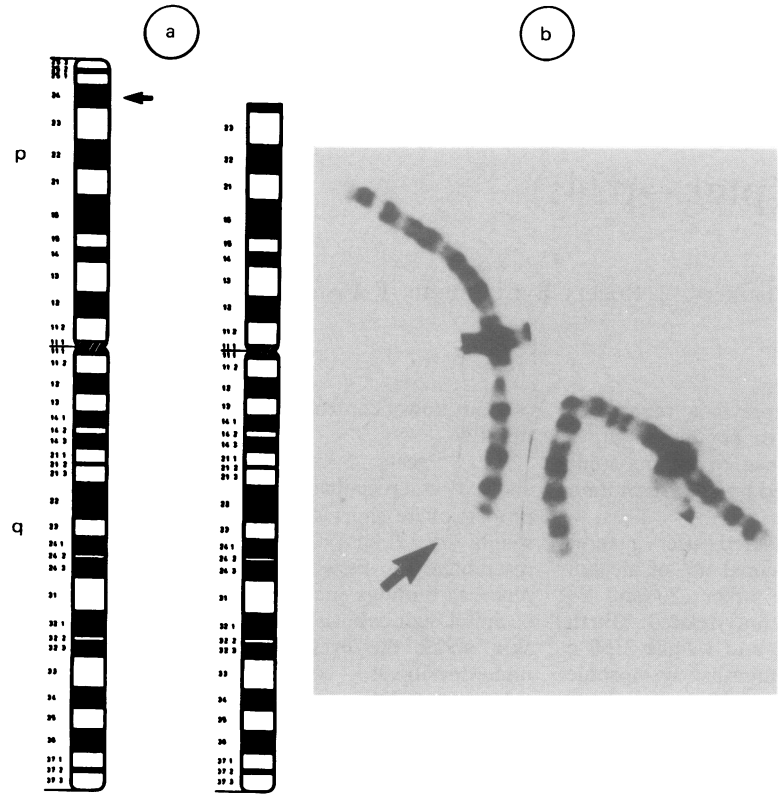

Figure 2 (a) Idiogram of chromosomes 2. Left: normal homologue. Arrow indicates breakpoint within band p24. Right: deleted homologue, del(2)(p24). (b) Photograph of chromosomes 2 showing a deletion at band p24 (arrow) in one homologue.

High resolution (approximately 600 bands) G banded lymphocyte chromosome studies showed an apparently terminal deletion in the short arm of chromosome 2 in all cells examined. The break appears to have occurred in band p24: 46,XY,del(2)(:p24→qter) (fig 2). High resolution banded karyotypes of both parents were normal.

Our case is the first instance of an apparent terminal deletion of 2p, confirmed by high resolution banding. Striking features include the superficial facial resemblance to Down's syndrome, microcephaly, short stature, profound developmental and language delay, hypoacusis, bilateral fifth finger clinodactyly with single flexion folds, and bilateral hallux valgus. A review of other cases identifies several features in common with more proximal deletions of 2p.[1-3] These include failure to thrive, developmental delay, seizures, microcephaly, short stature, highly arched palate, clinodactyly, single flexural crease of the fifth finger, rectangular face, bow shaped mouth, and micrognathia. Additional common features are inability to speak, hypotonia, prominent occiput and metopic suture, antimongoloid slant, strabismus, and low set ears. A clinical syndrome comprising failure to thrive, developmental delay, seizures, microcephaly, short stature, highly arched palate, clinodactyly with single flexion creases of the fifth finger, inability to speak, and variably dysmorphic facies associated with the common deleted segment (p24p25) is suggested.

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2 Neidich J, Zackai EH, Aronsom M, Emanuel BS. Deletion of 2p: a cytogenetic and clinical update. *Am J Med Genet* 1987;27: 707–10.
3 Penchaszadeh VB, Dowling PK, Davis JG, Schmidt R, Wapnir RA. Interstitial deletion of chromosome 2 (p23p25). *Am J Med Genet* 1987;27:701–6.