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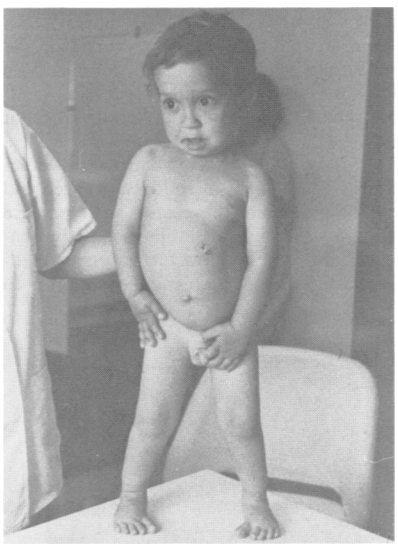
De novo 10q23 interstitial deletion

The proband (fig 1) was the product of the third pregnancy of a 25 year old mother and a 28 year old father. The first pregnancy ended in a spontaneous abortion and the second resulted in a healthy boy. The third pregnancy was uneventful. Delivery was at term with cephalic presentation. Birth weight was 3800 g (90th centile), length 51 cm (75th centile), and head circumference was 35 cm (75th centile). Apgar scores were 9 at one minute and 10 at five minutes.



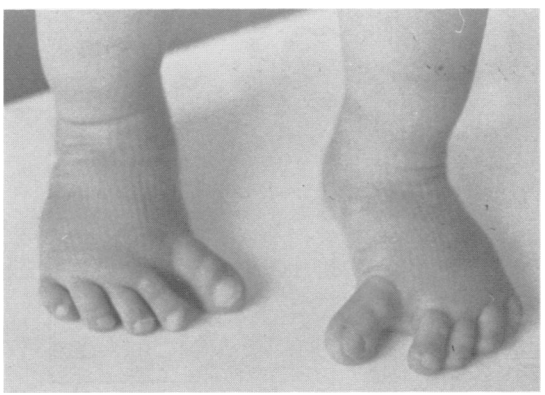


FIG 1 *The proband and his feet.*

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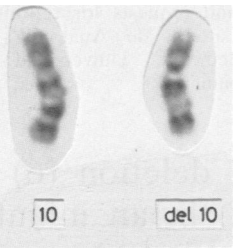

FIG 2 *Partial karotype of the proband.*

The clinical findings detected at birth were flat nasal bridge with anteverted nares, hypertelorism, a big mouth with downturned corners, low set ears, sacral dimple, equinovarus feet, fan shaped toes, and a gap between the first and second toes. The baby was hypotonic with psychomotor retardation during infancy: he sat at 12 months and began to walk at 31 months. A grade 1/6 systolic murmur was detected at six months. At that time he was also noted to have a bifid uvula, high arched palate, megalocornea, congenital glaucoma, macrocephaly (48 cm, 98th centile), broad forehead, dorsolumbar scoliosis, thoracolumbar lordosis, and a vesicoureteral reflux which was surgically corrected.

The last physical examination at 31 months showed the clinical features previously described and the systolic murmur persisting. At this time his weight was 10·6 kg and height 81 cm, which were both below the third centile for his age.

Chromosome analysis was performed on peripheral blood lymphocytes using GTG banding. The karyotype was found to be 46,XY,del(10)(pter→q22::q24→qter) (fig 2). Both parents and his brother had normal chromosomes.

To our knowledge there is only one other reported case with a 10q23 deletion.[1] The clinical features described in that report are very similar to those found in our case.

Since this is only the second report of del(10q23), more cases are needed before a syndrome can be delineated.

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Reference

[1] Shapiro SD, Hansen KL, Pasztor LM, *et al.* Deletions of the long arm of chromosome 10. *Am J Med Genet*1985;**20**:181–96.

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Interstitial deletion (6) (q11→q15) in an infant with congenital abnormalities

Deletions in the long arm of chromosome 6 are unusual, having been reported in only 14 cases to date,[1-3] including seven cases of interstitial deletion. The new case reported here displays a particularly large deletion (q11→q15) in a nine month old child with several congenital abnormalities.

The child (fig 1) was born normally to a healthy 16 year old mother. Birth weight was 2·26 kg (3rd centile) at 37 weeks' gestation. Several abnormalities were noted: head circumference on the 3rd centile, micrognathia, high arched palate, shallow philtrum, narrow vermilion border on upper lip, short neck, undescended testicles, long slender fingers, long flat feet with prominent heels, and a pigmented naevus on one thigh.

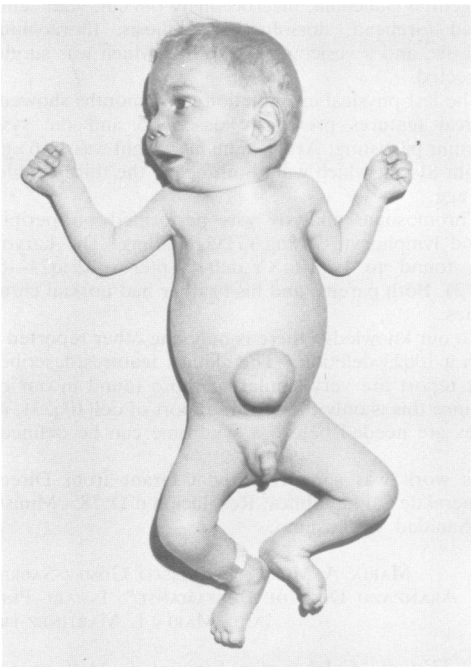

FIG 1 *The patient at six months of age showing umbilical hernia.*

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The child is now nine months old, feeds poorly, and his height, weight, and head circumference are all below the 3rd centile. There is obvious psychomotor retardation. A large umbilical hernia has required surgery. Current findings include OFC 42 cm (<2nd centile), horizontal palpebral fissures, interpupillary distance 3·8 cm (3rd centile), flat philtrum 1·4 cm long (55th centile), normal ears, 5 cm high (80th centile), hand length 8·2 cm (25th centile), internipple distance 8·5 cm (<3rd centile), small undescended testicles, and strange, laugh-like cry.

Chromosome analysis was performed on peripheral blood lymphocytes using GTG, CBG, and QFQ banding. A total of 50 cells was analysed and all showed an interstitial deletion in the long arm of one chromosome 6 homologue (fig 2). The karyotype was interpreted as 46,XY, del(6)(pter→q11::q15→qter). The deleted material did not appear to be translocated to other chromosomes in the karyotype. Chromosomes in skin fibroblasts derived from the patient showed the same deletion in all cells examined. The mother's karotype was normal; the father was not available for study.

Two other cases[1 3] have been reported which show deletions of chromosome 6 (q13→q15) in children of three and four years of age. Comparison of these cases with the case reported here shows several clinical similarities with mental retardation/psychomotor delay, low birth weight, feeding problems, and hernias being conspicuously common to all.

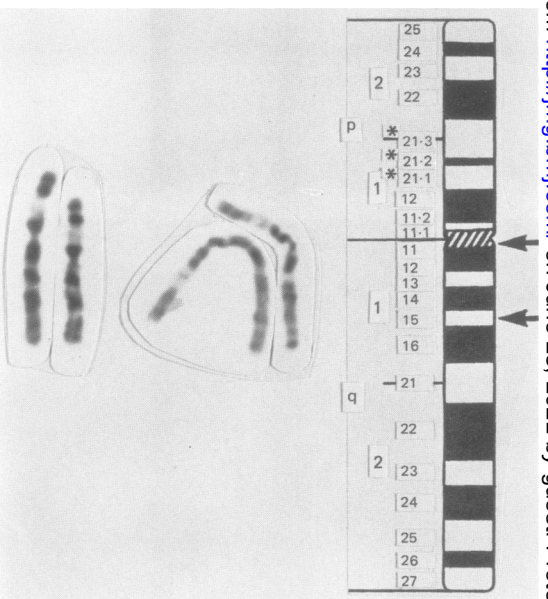

FIG 2 *Partial GTG banded karyotypes showing deletion in chromosome 6 with idiogram showing suggested breakpoints.*