Reference

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Correspondence and requests for reprints to Dr M L Martínez-Frías, Càtedra de Anatomia II (ECEMC), Facultad de Medicina, Universidad Complutense, 28040 Madrid, Spain.

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.

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), inter nipple 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 karyotype 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 1 The patient at six months of age showing umbilical hernia.

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

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The case reported here is the only one we are aware of showing a deletion involving the region 6q11→q13.

H R SLATER*, A ROBB*, L A FORSYTH*,
D A HAMILTON*, M C CLARK*, AND
C A S GALLOWAY†

Departments of Pathology* and Paediatrics†, Raigmore
Hospital, Inverness IV2 3UJ.

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Correspondence and requests for reprints to Dr H R Slater, Pathology Department, Highland Area Laboratories, Raigmore Hospital, Inverness IV2 3UJ.

Terminal deletion of chromosome 1(q43) in a female infant

A sufficient number of cases with similar phenotypes corresponding to deletion of the distal band of the long arm of chromosome 1 has established the existence of a new chromosomal syndrome.¹ We have observed a case with additional dysmorphic features not previously described.

The proband, a female, was the only child of healthy, unrelated parents. At the time of birth the father was 28 and the mother was 25. She was born at 37 weeks' gestation by caesarean section because of intrauterine growth failure. Birth weight was 2080 g and length was 45 cm. The head was microcephalic with a circumference of 31 cm, 2 SD below the mean.

At the vertex of the head a tuft of long hairs delimiting a small area of skin aplasia (0.8×0.6 cm) was visible (fig 1). A high protruding forehead, pronounced epicanthus, a broad nose with flat root, and micrognathia with downturned corners of the mouth were observed at examination. The ears were low set and the neck was short and broad. A CT scan of the brain showed partial agenesis of the corpus callosum. An x ray of the chest showed the presence of a supernumerary rib. X ray of the spine showed abnormalities of the sacral vertebrae; in S4 the left transverse process was missing, while in S5 the right one

FIG 1 Posterior view of the head showing a small area of skin aplasia delimited by a tuft of long hairs.

was absent. She also had a ventricular septal defect, hydronephrotic kidneys, a grade III vesicoureteral reflux, and a neurogenic bladder.

At two days of age she had severe generalised seizures with a paroxysmal EEG pattern. She failed to thrive and her psychomotor development was markedly delayed. When last seen (at three years) her length and head circumference were below the 3rd centile and her weight was on the 10th centile. Her development was at a six month level and she was severely hypotonic.

Chromosome studies were performed on lymphocyte cultures using GTG, QFQ, and high resolution RBA banding. A deletion of the terminal segment of chromosome 1 was seen, with a breakpoint at 1q43: 46,XX,del(1)(q43→qter). The chromosomes of the parents were normal.

Our patient is strikingly similar in facial appearance and associated findings to the other published cases. In addition, however, she had a small area of skin aplasia and anomalies of the sacral vertebrae, which have not been previously described in this syndrome.

The significance of these minor anomalies which could represent further phenotypic variability in the 1q deletion syndrome is not known, but could be dependent on the precise breakpoints involved. This has already been suggested by Wright *et al.*,² who reported an unusual ocular finding associated with the 1q deletion syndrome.

In 11 of the 17 cases described so far the breakpoints started at 1q42, while we considered that the deletion in our case started at 1q43. It must be noted, however, that inadequate chromosome resolution can make the distinction between bands 1q42.2 or 1q42.3 and 1q43 extremely difficult and complicate the correct assignment of the breakpoints.³

A better understanding of monosomy of distal 1q and of its possible variability will therefore be possible only when additional cases, studied with high resolution banding and with deletions beginning at 1q42 or 1q43, are reported.