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

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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.

FIG 2 Partial karyotype of the proband using GTG (a), QFQ (b), and high resolution RBA banding (c). The deleted chromosome 1 is on the right. Arrow in idiogram indicates presumed breakpoint.

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