congenital malformations. Ultrasonic scans during the 38th week of pregnancy showed fetal growth retardation corresponding to the 34th week. No other anomalies were observed.

The infant was born at term. His weight was 2500 g and length 47 cm. He showed marked hypertelorism, deformed low set ears, short neck, and cleft of hard and soft palates. The mouth was small. The right leg was less well developed and hypoplastic, and there was pes equinovarus. He had hypoxia, had difficulties in adapting to extrauterine life, and died 4 days later. The findings were acute bilateral intra-alveolar bronchopneumonia, massive atelectasis, pulmonary oedema, compensating emphysema, acute hypoxaemia, and mild internal hydrocephalus.

Analysis of blood lymphocyte chromosomes showed a modal number of 46. However, in all cells analysed, an interstitial deletion (q31→q35) of the long arm of chromosome 2 was observed. The karyotype of the infant was: 46,XY,del(2)(pter→q31::q35→qter). Both parents had a normal chromosome complement. Taysi et al. compared the clinical findings of four reported patients with 2q deletion including their own case. Different regions of the long arm of chromosome 2 were involved. One of these probands was found to have the deletion at 2q31→q33 and another, described by Warter et al., showed a 2q34→q36 deletion. Our proband’s breakpoint was at 2q31→q35.

The common features were: intrauterine growth retardation, large malformed low set ears, and abnormalities of the central nervous system. While the CNS anomalies in the reported cases included microcephaly, our proband was found to have internal hydrocephalus of a mild degree, in addition to small head size. The case described by McConnell et al. showed a partial deletion of the long arm of chromosome 2 (q22→q31) with anomalies usually associated with trisomy 18. The phenotype of our proband was very similar to that of the patient with interstitial deletion 2q31→q36.

The exact site of the breaks, however, is always difficult to determine, even in well banded preparations. This limitation must be kept in mind when comparing clinical features with apparently similar chromosomal rearrangements.


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References


The proband, a newborn female, was referred for cytogenetic analysis because of multiple congenital anomalies. Chromosome analysis was performed on peripheral blood lymphocyte cultures. GAG, RHG, CBG, and Ag-NOR staining procedures were used according to standard techniques. One of the chromosomes 21 showed a considerably elongated short arm. This chromosome 21 carried two active nucleolus organiser regions (NOR) and

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Familial occurrence of a pseudodicentric chromosome 21

The proband, a newborn female, was referred for cytogenetic analysis because of multiple congenital anomalies. Chromosome analysis was performed on peripheral blood lymphocyte cultures. GAG, RHG, CBG, and Ag-NOR staining procedures were used according to standard techniques. One of the chromosomes 21 showed a considerably elongated short arm. This chromosome 21 carried two active nucleolus organiser regions (NOR) and
two centromeres indicated by two distinct C bands but only one primary constriction at the proximal C band. The two C bands were separated by chromosomal material staining pale in G banding and intensely dark in R banding (fig 1). Both NORs could be observed in satellite associations (fig 2). The chromosome was therefore defined as pseudodicentric chromosome 21 (pseudic 21). The same chromosome was found in the proband’s father and paternal grandmother.

Acrocentric chromosomes with a short arm morphology similar to that presented here have been reported by Balicek and Zikan. These authors paid no attention to the activity of the centromeres. The suppression of additional centromeres is indicated by the presence of only one primary constriction as shown by Ing and Smith in a dicentric (Y;13) translocation. Variants of acrocentric chromosomes are often observed in patients with congenital anomalies. The occurrence of the pseudodicentric chromosome 21 in the proband and her phenotypically normal father and grandmother indicates that there is no association between the chromosomal variant and the proband’s congenital anomalies.

![Chromosomes 21 of the proband (a) and her father (b) by G (GAG), R (RHG), C (CBG) banding, and silver staining (Ag-NOR).](image)

**FIG 1** Chromosomes 21 of the proband (a) and her father (b) by G (GAG), R (RHG), C (CBG) banding, and silver staining (Ag-NOR).

![Pseudodicentric chromosome 21 in satellite associations. (a) G bands, (b) R bands.](image)

**FIG 2** Pseudodicentric chromosome 21 in satellite associations. (a) G bands, (b) R bands.

References


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**Short reports**

Extra euchromatic band in the qh region of chromosome 9

Chromosome 9 variants with a small extra G band located within a large heterochromatic region in the long arm were first described by Madan, the extra band being detected in 3 to 50% of cells in various subjects, and similar variants have since been reported by other authors. We describe here an unusual variant 9 in which the extra band is large and easily identified in all the cells examined.

The anomalous chromosome 9, first detected in a child with primary trisomy 21, is also present in the mother and in one of two phenotypically normal brothers. The cytogenetic characteristics are illustrated in the figure. The extra band exhibits medium fluorescence with Q and separate Q/C staining. Three C bands were seen in the variant, interpreted as representing the heterochromatin of the centromere region, and the proximal and distal parts of the heterochromatin of the secondary constriction flanking the extra negatively stained band. In contrast, the C positive region in the homologue has two subunits.

The extra band gave a negative reaction with a silver staining method (Goyanes, first step), which shows a staining affinity roughly coinciding with the loci rich in satellite III DNA. This method stains the secondary constriction heterochromatin leaving the centromeric heterochromatin unstained. In the variant, two dark segments were seen with a negatively stained band between them, indicating that the extra band is located within 9q12. This was confirmed with distamycin A/DAPI and G11 staining.

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Three new cases of oculodentodigital (ODD) syndrome: development of the facial phenotype


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Correction
In the article ‘Familial occurrence of a pseudodicentric chromosome 21’ by Hancke and Miller (J Med Genet 1985;22:155–6), we regret that a chromosome was missing from fig 1a. The full, correct figure is printed below.