karyotype was therefore designated 46,XY,del(9) (pter→q22::q32→qter).

Both parents were subsequently studied and found to have normal karyotypes. The deleted chromosome 9 in the proband was also identified as being of maternal origin since its enlarged heterochromatin (C band region) corresponded to that in one of the mother’s chromosomes 9. This suggests that the breakage, giving rise to a deletion, took place during oogenesis.

Deletion mapping of the genes for ABO, adenylosuccinate kinase 1, and galactose-1-phosphate uridyl transferase was performed, but assays failed to localise any of these loci to the deleted segment.

Discussion

The present case is believed to be the second report of a chromosome long arm deletion consisting of only the euchromatic region. Since none of the other chromosomes was found to be involved in the aberration, this case is regarded as a bona fide 9q deletion syndrome. The clinical features present in the patient can thus be attributed to partial monosomy for the deleted segment. The phenotype seen in our case does not correspond to those reported previously, since different regions of the chromosome 9 were involved in each of the other cases. The findings in the extremities and the marked generalised hirsutism should aid in the clinical recognition of other similarly affected infants.

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References


A malformed baby with two separate de novo translocations

SUMMARY A case of two separate apparently balanced de novo reciprocal translocations 46,XX,t(2;4)(p25;q21), t(10;18)(p15;q12.2) in an infant girl with multiple malformations is reported.

Case report

The proband was the second child of a father aged 24 and a mother aged 25. She was born at 38 weeks' gestation, weight 2.06 kg, length 44.5 cm (both less than the 3rd centile), and head circumference 33.5 cm (25th centile). She had rather 'pixie' facies, a relatively large cranial vault, and malformed ears. No other malformations were detected. She had a weak cry and functional palatal incompetence, with considerable feeding difficulties. At 2 months, a

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FIGURE Partial karyotype showing translocation from 4q to 2p, and from 18q to 10p (GTG banding). Arrows indicate breakpoints.
Case reports

systolic murmur was noted but there was no evidence of heart failure. At 41 months, her weight and length were well below the 3rd centile, the head circumference continuing along the 25th. She was developmentally retarded, did not smile or fix or follow with her eyes, and head control was poor. She was found dead in her cot at 41 months. At necropsy, a 1 cm secondum type atrial septal defect, a bicornuate uterus with a double cervix, and a septate upper third of the vagina were found. No gross or histological abnormalities of the brain were noted apart from rather prominent gyri. The thymus was small (1.2 g) but histologically normal.

Metaphase preparations (GTG) on lymphocyte cultures consistently showed translocations involving chromosomes 2, 4, 10, and 18 (figure). The karyotype is interpreted as 46,XX,t(2;4)(p25;q21),t(10;18)(p15;q12-2). The parents' chromosomes were normal.

Discussion

Single apparently balanced reciprocal translocations are not uncommon, but the coincidence of two unrelated apparently balanced reciprocal translocations in one person is extremely rare. In previous reports, some have1-3 and some have not4 5 been associated with phenotypic abnormality. The phenotypic abnormality often associated with a single reciprocal translocation may be the result of a submicroscopic loss of chromosome material, or a gene function disturbance owing to a position effect.6 Where the phenotype is normal, the translocation is presumably indeed balanced, with no ill effect of the chromosomal shift. We may suppose the same principles apply to the double reciprocal translocation.

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A second patient with partial deletion of the short arm of chromosome 3: karyotype 46,XY,del(3)(p25)

SUMMARY. A child with monosomy for the distal part of the short arm of chromosome 3 is presented. Altered features include prenatal onset growth deficiency, postaxial polydactyly, ptosis, ear anomalies, and a triangular facial appearance. In addition to generalised delay in psychomotor development, specific problems in visual attention were present. Comparison with the previously reported case suggests that the phenotype observed constitutes a clinically recognisable pattern of malformation.

The purpose of this report is to present a patient with monosomy for the distal segment of the short arm of chromosome 3. One child with a similar chromosomal abnormality has been previously reported.1

Case report

Features of this child are depicted in fig 1. The patient was born after a 40 week gestation remarkable for decreased fetal activity. The mother smoked 20 cigarettes per day. Delivery was via caesarian section for frank breech presentation. Birth length, weight, and head circumference were 43·75 cm, 2552 g, and 32 cm, respectively. All are less than the third centile for 40 weeks' gestation. Craniofacial alterations included a small (1·5 x 1·5 cm) anterior fontanelle, overriding cranial sutures, a triangular face, micrognathia, short (1·6 cm) palpebral fissures.