Short reports

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A case of malignant spinal cord ependymoma in association with a duplication of part of the long arm of chromosome 12

A 12 year old girl of Greek Cypriot origin presented with progressive gait disorder. There was no relevant family history. Her early development was rather slow and she made poor progress at a normal school. An assessment using the WISC had given her a full scale IQ of 65. Her early walking was reported as having a stamping quality tending to drag the left leg. However, no further problem was noticed until 2½ weeks before admission, when her gait had deteriorated progressively.

On admission she was found to be microcephalic with a head circumference of 47.5 cm, which is more than –3 SD below the mean. There were some dysmorphic features: arachnodactyly, hypermobile joints, proximally placed thumbs, and immobile first metacarpal-phalangeal joints. Forced eye closure produced an appearance reminiscent of a whistling face. The peripheral circulation was poor. There was marked weakness of the legs, in the left more than the right, with pathologically brisk tendon reflexes and bilateral extensor plantar responses, but she was still able to walk. In the upper limbs there was a mild degree of

pyramidal weakness, on the left more than the right, and an intention tremor. Sensory testing was difficult because of her poor cooperation. A CT scan was normal. A myelogram showed a large intramedullary tumour in the upper thoracic and cervical region. The CSF protein was markedly raised.

A laminectomy was performed by Mr Charles Polkey at the Guy's/King's/Maudsley Neurological Unit. Histology showed a malignant ependymoma, in which most of the cells had a perivascular concentration. The cells had moderately generally hyperchromatic nuclei and mitotic figures were present. She was then treated with a combination of steroids and radiotherapy with some improvement, but 8 months later her gait again deteriorated. A sub-total excision of tumour was then performed, which was followed by transient improvement. A year from the first presentation she developed raised intracranial pressure. A CT scan showed a mass in the brain stem and severe hydrocephalus. She died shortly after this and necropsy was not performed.

During the patient's first admission cytogenetic studies on peripheral blood cultures were carried out. Giemsa banded chromosome preparations showed an abnormality of chromosome 12, which was interpreted as a possible duplication of band q15, together with parts of the two adjacent bands (q14.3 and q21.1), in which case her chromosome complement would be written: 46,XX,dup(12)(q14.3→q21.1).

However, more than one interpretation is possible and instead of a duplication, the abnormality could be the result of an interstitial insertion of chromosome material of unknown origin.

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FIGURE (a) Partial karyotype showing the two chromosomes 12. The increase in length of the abnormal chromosome (shown on the right) between the bands q14.3 and q21.1 is thought to be due to a duplication. (b) Idiogram showing the position of the breakpoints and the region of duplication.

Interstitial deletion of chromosome 2

The proband described here was the first child of a 26 year old mother and 29 year old father. The mother had had one spontaneous abortion in the fourth month of a previous pregnancy. There was no family history of

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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 hyperaemia, 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*¹ 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*,² showed a 2q34→q36 deletion. Our proband's breakpoint was at 2q31→2q35.

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*³ 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→q36.²

FIG 1 Partial G banded karyotype of proband. Arrow indicates abnormal chromosome 2.

FIG 2 Diagram of chromosome 2, lost fragment, and deleted chromosome.

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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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