a karyotype of 46,X,del(Y)(q11). The deleted Y had neither C positive nor brightly fluorescing bands (figure). The Y chromosome of the proband's father was normal in appearance.

Discussion

The amount of the missing part of the Yq in our case is difficult to judge. However, evaluating the cytogenetic findings, and comparing the patient's phenotype with the karyotype-phenotype correlations published so far,¹⁻³ indicate that some of the euchromatic Yq must have been deleted together with the heterochromatin. The patient has small stature and failure of spermatogenesis which indicates that growth promoting factors and genes for spermatogenesis are located close to each other on the deleted segment of Yq, adjacent to the heterochromatin. The patient's male phenotype indicates, in agreement with published cases, that the primary inducers of maleness exist on the non-deleted portion of the Y chromosome.

Very little is known about the psychotic character of patients with deleted Yq. Kato *et al*⁴ described a schizophrenic man of normal phenotype who had a small Y chromosome; however, neither C nor Q banding was done. Since psychotic disorder is not a common symptom in such cases, a direct connection between the Yq deletion and the schizoid character disorder observed by us is questionable. The hypothesis of Forssman,⁵ namely that gonosomal

'imbalance' in general causes cerebral dysfunction favourable to the development of psychosis, may offer an explanation. However, the neonatal resuscitation performed in our case must be taken into consideration as well, since perinatal asphyxia is known to cause 'minimal brain damage' which is also favourable to the development of psychosis. To decide whether or not psychotic disorders are accompanying features of Yq—cases, together with small stature and abnormal male sexual differentiation, further karyotype-phenotype correlations are needed.

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Complete trisomy 5p owing to de novo translocation t(5;22)(q11;p11) with isochromosome 5p associated with a familial pericentric inversion of chromosome 2, inv 2(p21q11)

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SUMMARY A boy with a de novo translocation (5;22) and isochromosome 5p associated with a pericentric inversion of chromosome 2 (p21q11) is described. The pericentric inversion was also present in the mother. The main clinical features of the 'complete trisomy 5p' syndrome were present in the proband.

Reports concerning short arm trisomies of chromosome 5 have been reviewed recently.¹ Most of them were partial short arm duplications. Only a few were complete short arm trisomies and a new case is reported here.

Case report

The proband was born at term after an uncomplicated pregnancy. Birthweight was 3000 g. He was

Received for publication 10 March 1983.
Accepted for publication 31 March 1983.

Case reports

the second child of a 28-year-old mother and a 26-year-old father, who were healthy and non-consanguineous. The older child was a healthy 19-month-old boy. The family history was negative.

One week after birth the child was admitted to our paediatric department because of feeding difficulties. His weight was 3050 g, length 51 cm, and head circumference 37.5 cm. Clinical examination (fig 1) revealed macrocephaly, widely patent fontanelle and prominent sutures, hypertelorism, mongoloid slant of the eyes, broad flat bridge of the nose, long philtrum, macroglossia, microretrognathia, and low set asymmetrical ears with a flat helix and a poorly formed lobule. The neck was short and there was cutis laxa. The hands were broad and the thumbs proximally implanted. There were club feet of the equinovarus type and short first toes. Except for wide set nipples, examination of the thorax and abdomen was normal. Normal male genitalia were present. The child showed generalised hypotonia. Auscultation of the heart revealed a holosystolic murmur.

Examination of the fundus and an electroencephalogram were normal, as was x-ray of the skull. Echography of the abdomen did not show any abnormalities of the liver, spleen, or kidneys.

X-ray of the thorax revealed cardiomegaly and echocardiography suggested transposition of the great vessels. Otorhinolaryngeal investigation suggested some abnormality of the larynx.

Further exploration of the cardiopathy and the possible larynx malformation was not possible as the child died at the age of 52 days. No necropsy was performed.

CYTOGENETIC INVESTIGATIONS

G and R banding analysis of peripheral blood mitoses from the proband revealed a 46,XY karyotype, with a translocation between chromosome

FIG 2 The normal and abnormal G banded chromosomes 5, 22, and 2 of two mitoses of the proband.

5 and 22 and the formation of an isochromosome 5p (fig 2). In addition, a pericentric inversion of chromosome 2 was present. The karyotype was 46,XY,-5,-22,+t(5;22) (5qter→5q11 :: 22p11→22pter)+i(5p),inv(2)(p21q11).

The father had normal chromosomes, but there was an inversion of chromosome 2 (p21q11) in the mother. The (5;22) translocation was not present in either parent. The older child was not available for investigation.

Discussion

The de novo (5;22) translocation with iso(5p) produced a trisomy for the whole of the short arm of chromosome 5, associated with a small short arm deletion of chromosome 22. Short arm deletions of acrocentric chromosomes are generally believed not to cause any phenotypic effect and therefore the abnormalities in the proband were probably the result of a pure 5p trisomy.

As far as we know, only three cases of complete trisomy 5p have been described up to now. Surprisingly, in two of them,^{2,3} the trisomy also originated from a de novo iso(5p) chromosome. In the third case there was a familial (5;15) (p11;p22) translocation. The clinical findings in cases of partial trisomy 5p vary considerably, but tend to become a recognisable clinical entity depending on the extent of the 5p duplication.^{1,2} A distinct clinical syndrome should exist only in

FIG 1 Front and lateral view of the proband.

complete 5p trisomy cases. The main features are normal weight and length at birth, macrocephaly, psychomotor retardation, hypotonia, postnatal growth failure, and facial dysmorphism involving mongoloid eye slant, epicanthus, hypertelorism, low set and dysplastic ears, and flat bridge of the nose. The lower extremities show short first toes and club feet.

The three reported cases with complete 5p trisomy had seizures. In our patient most of the main features mentioned were present but, in addition, he had a congenital heart defect, not seen in the three cases reported. However, seizures were not noted in our patient.

Tetrasomy 9p confirmed by GALT

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SUMMARY We report a boy aged 12 years 7 months with mental retardation, hydrocephalus, dysmorphic facial features, congenital heart disease, and skeletal and renal anomalies. The karyotype showed a mosaic tetrasomy 9p involving the secondary constriction. This result was confirmed by tetraplex gene dosage effect for galactose-1-P-uridylyltransferase (GALT). Comparing the clinical features of our case with those of previously reported patients, tetrasomy 9p appears to be a distinctive and clinically recognisable malformation syndrome.

Tetrasomy for the short arm of chromosome 9 is a very rare condition, first described by Ghymers *et al.*¹ Since then, tetrasomy 9p has been found in eight additional patients with variable phenotypic expression and cytogenetic findings. This report concerns a new case of tetrasomy 9p in a male aged 12 years 7 months.

Case report

The proband is the second child of healthy, unrelated parents. The mother was 38 and the father 36 years old at the time of his birth. The family history is

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unremarkable and the couple's first child is in good health.

The proband was born at term by normal delivery after a normal pregnancy. Birth weight was 3200 g. Asphyxia and skull asymmetry owing to a left parietal cephalohaematoma were noted at birth. At the age of 12 years 7 months he was admitted to hospital because of mental retardation and dysmorphic features.

PHYSICAL EXAMINATION

The following findings were present (fig 1): normal growth with a height of 157 cm (75th to 90th centile) and weight 39 kg (50th to 75th centile). He had a macrocephalic and asymmetrical skull, head circumference 58 cm (>97th centile), a low anterior hairline, multiple hair whorls, flat forehead, sunken eyes, moderate hypertelorism, antimongoloid slants, broad nasal root, high nasal bridge and bulbous nasal tip, short philtrum, down-turned corners of the mouth, retrognathia, cup-shaped, anteriorly rotated, and protruding ears, and a short neck. There was marked thoracic kyphosis and lumbar scoliosis with hyperlordosis, narrow chest widely spaced nipples, protruding abdomen, mild truncal obesity, and narrow pelvis. There was also an incurved radius and ulna, limited joint movements of the elbows, knees, and ankles, coxa and genu valgus, narrow hands and feet, bilateral pes planovarus, and dysplasia of the