

region of the Y chromosome and to the distal heterochromatic region respectively. In situ autoradiographs showed two clusters of silver grains over the dicentric chromosome with probe pDP105 (fig 2) and none with probe pHY2.1. The karyotype was thus interpreted as 45, X/46, X dic (Y) (pter → q12:q12 → pter).

Parental blood samples were not available.

Discussion

The similarity between the phenotypic expression of Noonan's syndrome and Turner's syndrome can lead to confusion. The familial occurrence of Noonan's syndrome would indicate a dominant mode of inheritance for this condition and the coincident appearance of neurofibromatosis I in some cases may indicate that the Noonan's syndrome gene is also on chromosome 17.

The presence of features similar to those of Turner's syndrome in a phenotypic male specifically provokes a diagnosis of Noonan's syndrome. Some of these phenotypic males, with varying degrees of virilisation, have, however, been shown to be more appropriately called male Turner's syndrome, resulting from the presence of 45, X/46, XY mosaicism. Like the case reported here several of these have shown a non-fluorescent Y chromosome and it has been suggested that they may be dicentric chromosomes derived by sister chromatid reunion in band Yq1. The expected mitotic behaviour and unstable nature of such a dicentric chromosome would predispose to the development of a 45, X cell line resulting, in some cases, in the stigmata of Turner's syndrome.^{2 3}

We have been able to show in our case that the Y chromosome present, although not easily distinguishable from a normal Y on conventional trypsin/Giemsa banding, indeed did prove to be dicentric and to have lost the whole of the heterochromatic portion of the long arm when examined by C banding, Cd banding, and quinacrine fluorescence. This was confirmed by use of Y chromosome DNA probes that showed loss of the heterochromatic region recognised by the probe pY2.1 and duplication of the region

near the centromere recognised by the probe pDP105.

Although we were not able, probably for technical reasons, to demonstrate two centromeres in all cells no Y chromosomes with normal heterochromatin were observed. Unfortunately we were not able to examine the paternal chromosomes but the father was reported to be of normal phenotype.

It is clearly important from management and counselling aspects to ensure that Noonan's syndrome and Turner's syndrome are distinguished in patients. The latter are at risk of gonadoblastoma when Y chromosomal material is present and the reproductive expectations and recurrence risks are different for the two syndromes.

It has been suggested that gonadoblastoma is less likely to develop in dysgenic gonads when the Y chromosome lacks the fluorescent region.⁴ The number of cases for which long term surveillance is available and which contribute to this conclusion is small, however, and there should be caution in adopting a policy of non-interference. In males with descended testicles, such as this case, long term observation of the testes should be feasible.

Recent evidence in the use of recombinant growth hormone in female patients with Turner's syndrome has indicated the beneficial effect of this treatment in improving growth velocity and final height.⁵ The initial response to growth hormone in our patient suggests that such treatment may also have an important role in treatment of males with this syndrome.

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Congenital parvovirus infection

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Abstract

A case of congenital parvovirus (B19) viraemia with associated thrombocytopenic purpura and platelet antigen incompatibility in an infant is reported. Results of laboratory investigations indicated that the baby was infected in utero.

A recent report has shown evidence of the sequelae of congenital parvovirus (B19)

infection.¹ Others have shown haematological consequences of infection in an otherwise normal infant.² We report a confirmed intrauterine infection with B19 in an infant, which was associated with thrombocytopenic purpura and platelet antigen incompatibility.

Case report

An infant boy was born at 37 weeks' gestation after a normal delivery. Pregnancy had been

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Investigations for parvovirus B19

	Time sample taken after birth	B19 IgM*	B19 IgG*	B19 DNA†
Mother	2 weeks	>100	35	Negative
	8 weeks	31	>100	Negative
Baby	Birth	33	6.1	Positive
	2 weeks	>100	6.5	Negative
	8 weeks	6	19	Negative

*Radioimmunoassay units: <1 negative, 1-3 equivocal, and >3 positive.

†Dot blot hybridisation assay with digoxigenin labelled probe.

uneventful with no evidence of maternal illness. He weighed 2600 g and his Apgar scores were 8 at birth and 10 at five minutes. Three hours after birth the infant was noted to have a widespread petechial rash over his trunk, limbs, and palate and larger bruises were evident over bony prominences. There was no hepatosplenomegaly and examination was otherwise unremarkable. Initial investigations showed a haemoglobin concentration of 169 g/l, white cell count $20.7 \times 10^9/l$ (74% neutrophils), and platelets $19.0 \times 10^9/l$. Clotting screen and bacterial infection screen were normal. Recovery was uneventful except for moderate jaundice requiring four days of phototherapy. By day 8 the platelet count was $550 \times 10^9/l$. Cranial ultrasound scan was normal.

Further serological investigation showed the father to be platelet antigen (PI^{A1}) positive and the mother PI^{A1} negative. The mother also demonstrated anti-PI^{A1} antibody. The infant was PI^{A1} positive and his serum cross reacted with that of his mother.

Discussion

The results of B19 investigations (table) indi-

cated intrauterine infection with a high concentration of specific IgM and a rising concentration of specific IgG in mother and infant. In addition DNA studies demonstrated a viraemia in the first infant sample that cleared as the IgM rose. The recent survey by the Public Health Laboratory Service on B19 in pregnancy suggested a 30% intrauterine infection rate but no case showed the presence of the viraemia in the infant and no thrombocytopenia was noted.¹ A recent report showed viraemia in a 2 month old infant but this was persistent and associated with erythroid hypoplasia.² Maternal infection was not reported in this case. Others have demonstrated antiplatelet antibodies and thrombocytopenic purpura in previously normal children with viral infections, including parvovirus.^{3,4} This case shows not only confirmed intrauterine infection with B19 and transient viraemia in the newborn infant, but the association with thrombocytopenic purpura and platelet antigen incompatibility; this has not previously been demonstrated with congenital parvovirus B19 infection.

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