

his red cells were examined for a minor population of Fy(a+) cells by absorption/elution techniques. Anti-Fy<sup>a</sup> was eluted from the red cells of the propositus after incubation with one anti-Fy<sup>a</sup> reagent, but not with another.

The following method was used to determine the proportion of A<sub>1</sub> (or A<sub>1</sub>B) cells in the blood of the propositus: the titre of anti-A eluted from his red cells was compared with that eluted from artificial mixtures of A<sub>1</sub> and B cells, using equal volumes of cells and anti-A for sensitisation in each case.

The results shown in the Table suggest that the ratio of A<sub>1</sub> (or A<sub>1</sub>B) cells to B cells in the blood of the propositus was approximately 1:5000.

Table *Titres of anti-A eluted from cells of the propositus and artificial mixtures of A<sub>1</sub> and B cells*

	<i>Titres of eluted anti-A</i>
Cells of propositus	16
1% A 99% B	512
0.1% A 99.9% B	64
0.01% A 99.99% B	8

## Discussion

In normal healthy adults the absence of ABO agglutinins in the serum to antigens the subject lacks on the red cell is extremely rare. It has been thought that this may be due to immune tolerance (Van Loghem *et al.*, 1965). However, it may occur when the antigen to the missing agglutinin is present in a weakened form or in small quantities, as in the present case.

The presence of a weak A antigen has several possible causes. It may be due to the inheritance of a weak variant of A such as A<sub>eb</sub>, A<sub>x</sub>, or A<sub>3</sub>. However, the ABO groups of the other members of the family indicate that this is not so in this case. The absence of A substance in the saliva of the propositus suggests that his unusual ABO group is not the result of modifying genes which suppress or inhibit the expression of A at the cell surface (Darnborough *et al.*, 1973).

The remaining possibility, that the propositus is a blood group chimera, seems most likely, considering that he had a stillborn twin, and that the presence of a small number of A<sub>1</sub> (or A<sub>1</sub>B) cells, resulting from a graft of haemopoietic tissue from the twin, has prevented him from producing anti-A. The presence of a small population of Fy(a+) cells demonstrable by absorption and elution techniques seems to confirm this. In view of the small proportion of the grafted cells it is not surprising that chimerism cannot be shown by 'mixed-field' agglutination, karyotype, or HLA type as in other cases described (Race and Sanger, 1975).

We wish to thank the propositus and his family for their co-operation. We also wish to thank Dr Mason for carrying out chromosome analysis, Dr Winifred Watkins for transferase studies, Mr M. Pepper for HLA typing, and Dr Carolyn Giles for confirming the blood groups.

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## Absent left hemidiaphragm, arhinencephaly, and cardiac malformations

**SUMMARY** An infant is reported with absent left hemidiaphragm, hydrocephalus, arhinencephaly, and cardiovascular anomalies. The parents are second cousins.

In this report, an infant with an unusual constellation of anomalies born to consanguineous parents is described.

### Case report

A male infant weighing 2180 g was delivered by caesarian section at 36 weeks' gestation because of pre-eclampsia and hydrocephalus. He died 2 hours after birth. The placenta weighed 410 g and had only 1 umbilical artery. The head (Fig.) was triangular, the nose bridge flat, and the forehead hirsute. The nails on the hands and feet were missing in both fifth fingers



Fig. Newborn with hydrocephalus and absent nails on fifth digits.

and hypoplastic in all the other digits. There was first degree penile hypospadias.

Necropsy showed that the left lobe of the liver, the stomach, and a portion of the small bowel were in the left pleural cavity owing to absence of the left hemidiaphragm. The right lung weighed 8 g and had no middle lobe. The left lung consisted of an upper lobe only and weighed 3 g. There was a low origin of both pulmonary arteries from the pulmonary truncus, and a high interventricular septal defect about 4 mm in diameter. There were fibrous pleural adhesions on the right side.

Neuropathological examination showed complete agenesis of the corpus callosum and anterior commissure; absence of olfactory bulbs and tracts; communicating hydrocephalus; moderately severe and chronic ependymitis and leptomenigitis of undetermined aetiology. The karyotype was normal.

The mother had had one previous pregnancy, 2 years before the birth of the propositus, which was terminated therapeutically. She was 27 years old and her husband was 19 years old. The pregnancy was complicated by a *Candida* urinary infection in the seventh month which was treated with nystatin. In the eighth month there was a bacterial urinary infection and mild pre-eclampsia. The mother had daily nosebleeds from the fifth to the seventh month of

pregnancy. In the last 2 months she also had daily diarrhoea of undetermined origin.

The family came to Canada from the Philippines. The mother's maternal grandfather and the father's paternal grandfather were brothers. A cousin of the paternal grandmother, who was also married to a cousin, had a child with ventricular septal defect and syndactyly of 2 fingers. (The informant did not know which digits were involved.)

## Discussion

Diaphragmatic hernia occurs in about 0.45 in 100 births (Butler and Claireaux, 1962). It is usually on the left side and more common in females (David and Illingworth, 1976). The persistence of a posterolateral opening in the diaphragm produces one type of hernia, the diaphragmatic hernia of Bochdalek. Sometimes the defect can involve an entire half of the diaphragm, or even the whole diaphragm. The abdominal viscera project into the pleural cavity, and the resultant compression is probably the cause of the severe lung hypoplasia described in almost all cases of large diaphragmatic hernias.

About 50% of patients have other congenital malformations, most frequently of the central nervous system. CNS defects include spina bifida, hydrocephalus, anencephaly, encephalocele, iniencephaly, absent corpus callosum, and arhinencephaly (Yakovlev, 1959; David and Illingworth, 1976).

It is interesting to note that in 4 families who had infants with diaphragmatic hernia and CNS anomalies, sibs had been born who had only CNS anomalies (Butler and Claireaux, 1962; David and Illingworth, 1976). Landau *et al.* (1963) described an infant with arhinencephaly, absent corpus callosum, congenital heart defects, and a small diaphragmatic hernia, whose first cousin had anencephaly.

Cardiovascular anomalies are sometimes found in infants with diaphragmatic hernia. In David and Illingworth's series (1976) the incidence was 13.3%.

There have been several reports of diaphragmatic gaps in sibs (Mäkela, 1916; Mertins, 1952; Phillip and Skelton, 1952; Butler and Claireaux, 1962; Powell and Johnstone, 1962; Welch and Cooke, 1962; Passarge *et al.*, 1968; ten Kate and Anders, 1970; Feingold, 1971; Jensen and Altrogge, 1971; Daentl and Passarge, 1972; Harberg *et al.*, 1976). Almost all of these were on the left side. The exceptions were Jensen and Altrogge's report of 2 sibs, one of whom had a left sided aplasia and the other total aplasia. Feingold's patients included one with left sided aplasia, and the other with the same defect plus absence of two thirds of the right side. Additional anomalies in 2 of these families have been cardiovascular (Feingold, 1971) and urogenital (Mäkela, 1916).

There have been 2 reports of sibs, one of whom had a defect in the diaphragm and the other a diaphragmatic hernia with a sac (Scott and Paterson, 1966; Thomas *et al.*, 1976).

Diaphragmatic hernia has been reported in first cousins (Mosavy and Ashrafi, 1974) and in distant cousins (Turpin *et al.*, 1959).

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## Apical systolic click and murmur associated with neurofibromatosis

**SUMMARY** In this report we describe a child who had an apical systolic click and murmur, as well as widespread cutaneous neurofibromatosis. We were not able to show an anatomical basis for the click and murmur.

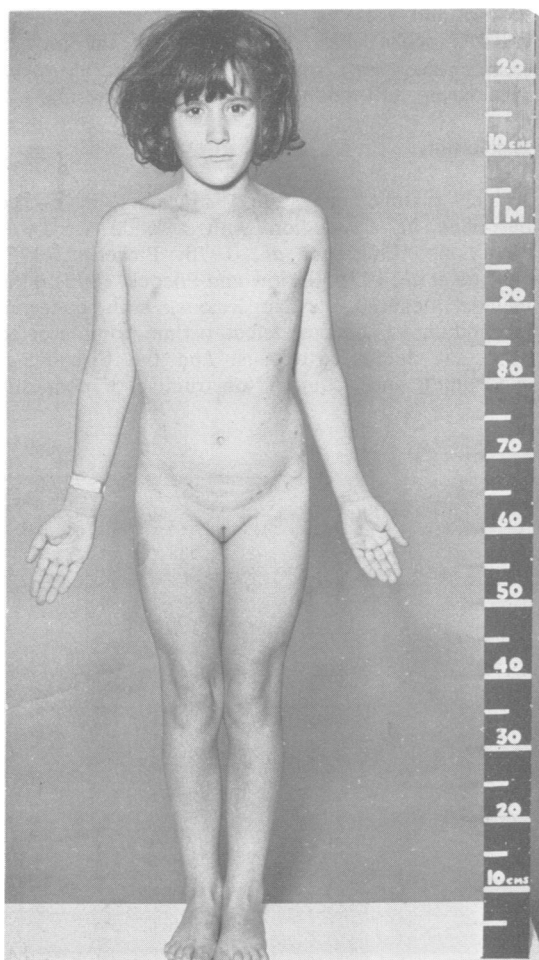


Fig. 1 Patient showing cutaneous lesions of neurofibromatosis.