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

The following method was used to determine the proportion of A\(_1\) (or A\(_1\)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\(_1\) 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\(_1\) (or A\(_1\)B) cells to B cells in the blood of the propositus was approximately 1:5000.

<table>
<thead>
<tr>
<th>Cells of propositus</th>
<th>Titres of eluted anti-A</th>
</tr>
</thead>
<tbody>
<tr>
<td>1% A 99% B</td>
<td>16</td>
</tr>
<tr>
<td>0-1% A 99-99% B</td>
<td>512</td>
</tr>
<tr>
<td>0-01% A 99-99% B</td>
<td>64</td>
</tr>
<tr>
<td></td>
<td>8</td>
</tr>
</tbody>
</table>

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\(_{ab}\), A\(_{a}\), or A\(_{b}\). 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\(_1\) (or A\(_1\)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.

G. H. LONGSTER, E. ANGELA ROBINSON, AND D. I. NORTH

Regional Transfusion Centre, Bridle Path, Leeds

References


Requests for reprints to Mr G. H. Longster, Regional Transfusion Centre, Bridle Path, Leeds LS15 7TW.

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 trunk, 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 leptomeningitis 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 Phillipines. 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, encephalophaly, 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).
Case reports

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).

NAOMI FITCH, HERBERT SROLOVITZ, YVON ROBITAILLE, AND FRANK GUTTMAN

Lady Davis Institute for Medical Research, and Department of Pathology, Jewish General Hospital, 3755 Cote Saint Catherine Road, Montreal, Quebec, Canada H3T 1E2

Requests for reprints to Dr Naomi Fitch, Lady Davis Institute for Medical Research, Jewish General Hospital, 3755 Cote Saint Catherine Road, Montreal, Quebec, Canada H3T 1E2.

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.
Absent left hemidiaphragm, arhinencephaly, and cardiac malformations.

N Fitch, H Srolovitz, Y Robitaille and F Guttman

doi: 10.1136/jmg.15.5.399

Updated information and services can be found at:
http://jmg.bmj.com/content/15/5/399

*These include:*

**Email alerting service**
Receive free email alerts when new articles cite this article. Sign up in the box at the top right corner of the online article.

Notes

To request permissions go to:
http://group.bmj.com/group/rights-licensing/permissions

To order reprints go to:
http://journals.bmj.com/cgi/reprintform

To subscribe to BMJ go to:
http://group.bmj.com/subscribe/