The phenotype $A_e B$: a probable result of chimerism

**SUMMARY** An apparently normal healthy adult with the blood group phenotype $A_e B$ is described. The unusual ABO group is apparently the result of chimerism, the proportion of the minor population of cells being so small as to be only detectable by absorption and elution techniques.

**Case report**

The propositus was an apparently normal healthy blood donor, aged 24 years. During routine testing his red cells grouped as B, but no anti-A was detected in his serum. He had a brother and sister still living and also had a stillborn twin, sex unknown. His karyotype was 46,XY and all cells analysed showed a normal male pattern. His white cells typed as HLA A11, AW30: BW21, BW40.

**Serological findings**

Standard blood grouping methods were used throughout. Elution was carried out by the Landsteiner-Miller technique (as described by Dunstan and Bowley, 1967). In the ABO absorption and elution experiments, 0.5 ml packed cells were incubated with 2 ml anti-A for 2 hours at 4°C and the antibody was eluted into 1 ml physiological saline. In the anti-Fy* absorption and elution experiments, 4 ml packed cells were incubated with 8 ml anti-Fy* for 2 hours at 37°C and the antibody eluted into 1 ml antibody-free serum.

**Results**

The blood groups of the propositus and his family are as follows:

- **Propositus**, $A_e B$ Rh CcDee, M, S+s+, P+.
  - Lu(a−), K−, Le(a−b+) anti-A.
  - Fy(a+b−), Jk(a−b+).

- **Father**, $A_e B$ Rh CcDee, M, S+s+, P+.
  - Lu(a−), K−, Le(a−b+) anti-A.
  - Fy(a+b−), Jk(a−b+).

- **Mother**, $A_1 B$ Rh CcDee, M, S+s+, P+.
  - Lu(a−), K−, Le(a−b+) anti-A.
  - Fy(a+b+), Jk(a−b+).

- **Brother**, $B$ Rh ccDee, M, S+s+, P+.
  - Lu(a−), K−, Le(a−b+) anti-A.
  - Fy(a+b+), Jk(a−b+).

- **Sister**, $A_1 B$ Rh CcDee, M, S+s+, P+.
  - Lu(a−), K−, Le(a−b+) anti-A.
  - Fy(a+b+), Jk(a−b+).

The red cells of the propositus were not agglutinated by 10 different anti-A reagents or with a high titre anti-A+B that had been absorbed for anti-B. However, anti-A could be eluted from his red cells after they had been sensitised with anti-A. No anti-A was detected in his serum; however, on 2 occasions a weak anti-A, active at 4°C was found in his serum. He was a secretor of B and H substances, but not of A substance. His plasma contained B and H transferases within the normal range, but no A transferase was detected. The H antigen strength was similar to that of normal group B cells.

The blood groups of the family show that the only other antigen the propositus lacked, but his stillborn twin might have possessed, was Fy*. In view of this...
his red cells were examined for a minor population of Fy(a+) cells by absorption/elution techniques. Anti-Fy* was eluted from the red cells of the propositus after incubation with one anti-Fy* reagent, but not with another.

The following method was used to determine the proportion of A₁ (or A₂B) 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 A₁ 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 A₁ (or A₂B) cells to B cells in the blood of the propositus was approximately 1:5000.

Table  
<table>
<thead>
<tr>
<th>Cells of propositus</th>
<th>1% A</th>
<th>99% B</th>
<th>0-1% A</th>
<th>99-99% B</th>
<th>0-01% A</th>
<th>99-99% B</th>
</tr>
</thead>
<tbody>
<tr>
<td>Titres of eluted anti-A</td>
<td>16</td>
<td>512</td>
<td>64</td>
<td>8</td>
<td></td>
<td></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 A₀, A₁, or A₂. 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 A₁ (or A₂B) 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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References


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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.