A further example of human blood group chimaerism

SUMMARY Blood group chimaerism was detected in a healthy fertile woman, not known to be a twin. Her peripheral lymphocytes had a male karyotype (46/XY); fibroblasts cultured from her skin had a female karyotype (46/XX). The mechanism of chimaerism could not be established.

Fig. 1 Red cell suspensions prepared from whole blood of proband: (A) exposed to anti-A; (B) exposed to anti-A,B. Red cell suspensions prepared from O cells separated from whole blood of proband: (C) exposed to anti-A; (D) exposed to anti-A,B.
Since the first description by Dunsford et al. (1953) of blood group chimaerism in man, about 40 cases have been reported. In about half of these cases, the dual population of red cells was attributed to an exchange of haemopoietic tissue between twins in utero. In the remaining cases there was no history of twinning but other features supported the conclusion, first proposed by Gartler et al. (1962), that the chimaerism was the result of the fertilisation by two sperm of one or more egg nuclei. (See Race and Sanger, 1975.)

Sometimes the mechanism of chimaerism cannot be established. Such a case was described by Battey et al. (1974) and by Bird et al. (1976). The present report records a similar case.

Case report

Mrs Co., aged 30, was a blood donor in Boston, U.S.A. Discrepancies in ABO blood grouping led to further investigations and to the detection of blood group chimaerism. Her blood contained a major population of O cells and a minor population of A1 cells. Using the method of Booth et al. (1957), the two populations were separated and tested for many blood group antigens and red cell enzymes. The O cells were S-negative, Fy(a+), Jk(b+); the A1 cells were S-positive, Fy(a−), Jk(b−). In addition, the two red cell populations differed in their GPT (glutamic pyruvate transaminase) phenotype. The serum allotypes (Gc, Bf, C3, Hp, Gm, Kn) did not reveal any mixtures.

Mrs Co.’s saliva contained A, H, and Leb. Both populations of red cells were agglutinated by anti-Leb and by anti-A1Leb. As shown in Fig. 1, the separated O cells were not agglutinated by the anti-A in group B serum, but were agglutinated by the anti-A,B in group O serum.

Lymphocytes cultured from peripheral blood showed a normal male karyotype (46/XY); 200 interphase cells showed fluorescent Y-body in 90% of nuclei and analysis of 16 metaphases confirmed the presence of a Y-chromosome. Fibroblasts cultured from skin of the forearm showed a normal female karyotype (46/XX).

Family study

The results of testing blood and saliva from the proband and her family are shown in Fig. 2. The red cells of the proband (II.2) and of her mother (I.2) were agglutinated by anti-A1Leb. The red cells of the other family members (I.1, II.1, II.3, III.1, III.2) were not agglutinated by anti-A1Leb. The Table shows the serum levels of A1- and A2-specified N-acetylgalactosaminyltransferases.

Conclusions

The A1 gene in the proband is not restricted to her haemopoietic tissue since: (1) her saliva contains A2 substance; (2) her daughter (III.2) has inherited her A1 gene; (3) the proband’s plasma contains A2 substance (detectable on her O cells) and A1Leb (detectable on both cell populations); and (4) the level of N-acetylgalactosaminyltransferase in the proband’s serum is greater than would be expected if the haemopoietic tissue were the sole source of this enzyme.

It is known that the haemopoietic tissue contributes 20% of the blood-group transferases in serum (Schachter et al., 1971; Wrobel et al., 1974). Since only 16% of the cells in Mrs Co. are A1, the finding of significantly more A1-specified N-acetylgalactosaminytransferase (i.e. more than 20% of 16%)}
Case reports

Table Serum N-acetylglucosaminyltransferase levels in proband and family

<table>
<thead>
<tr>
<th>Serum donor</th>
<th>Genotype</th>
<th>Counts per minute*</th>
</tr>
</thead>
<tbody>
<tr>
<td>I.1 Father</td>
<td>O O</td>
<td>90</td>
</tr>
<tr>
<td>I.2 Mother</td>
<td>A1O</td>
<td>2296</td>
</tr>
<tr>
<td>II.1 Husband</td>
<td>A2O</td>
<td>464</td>
</tr>
<tr>
<td>II.2 Proband</td>
<td>A1O</td>
<td>1089</td>
</tr>
<tr>
<td>III.1 Son</td>
<td>O O</td>
<td>50</td>
</tr>
<tr>
<td>III.2 Daughter</td>
<td>A1A2</td>
<td>1801</td>
</tr>
<tr>
<td>Controls</td>
<td>C.T.</td>
<td>3259</td>
</tr>
<tr>
<td>R.M.</td>
<td>A O</td>
<td>403</td>
</tr>
</tbody>
</table>

*Radioactivity in product of an incubation mixture containing (in a final volume of 45 μl) 0.063 μmol LNaF-1; 0.005 μmol UDP-N-acetyl-D-glucosamine-1-14C (2 x 10⁵ cpm per μmol); 1.25 μmol MES, pH 5.6; 1.25 μmol MnSO₄; and 10 μl serum. The mixture was incubated at 37°C for 3 hours.

indicates that the A¹ gene is not restricted to her haemopoietic tissue.

If Mrs Co. were a twin, the presence of O red cells could be attributed to a graft in utero of haemopoietic tissue from a group O twin. However, Mrs Co. is not known to be a twin, and there is no history of twinning in her family. The mechanism of her blood group chimaerism remains unestablished.

We thank Mrs Co. and her family for their cooperation; Dr C. Alper (Boston) for determining the serum protein allotypes; Mr D. Della-Loggia (Boston) for technical assistance, Dr H. A. Gardner (Toronto) for provision of photographic equipment and advice; Dr P. S. Gerald (Boston) for analyses of Mrs Co.’s chromosomes; Dr E. R. Giblett (Seattle) for determining the red cell enzyme phenotypes; and Dr H. Schachter (Toronto) for provision of facilities to assay the A-specified transferases in serum.

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Referennces

Another human chimaera. Journal of Medical Genetics, 11, 283-287.


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The r(20) syndrome

SUMMARY A 21-year-old woman with a ring-20 chromosome is described. The clinical findings, behaviour problems, epilepsy, and low grade mental retardation are the same as in the 3 cases described earlier. It seems to be justified to speak of a specific ring-20 syndrome.

Abnormalities, involving F group chromosomes, are rare. Structural changes have been found in bone marrow cells associated with haematological disease and examples of aneuploidy have been reported in spontaneous abortions. It seems that a ring-20 chromosome has been described only once without mosaicism and twice with mosaicism. We present a case with r(20) with similar clinical findings to those in the 3 previous cases.

Case report

The patient was a girl born in 1955 when the father was 35 and the mother 37 years old. She had one