

Case reports

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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.

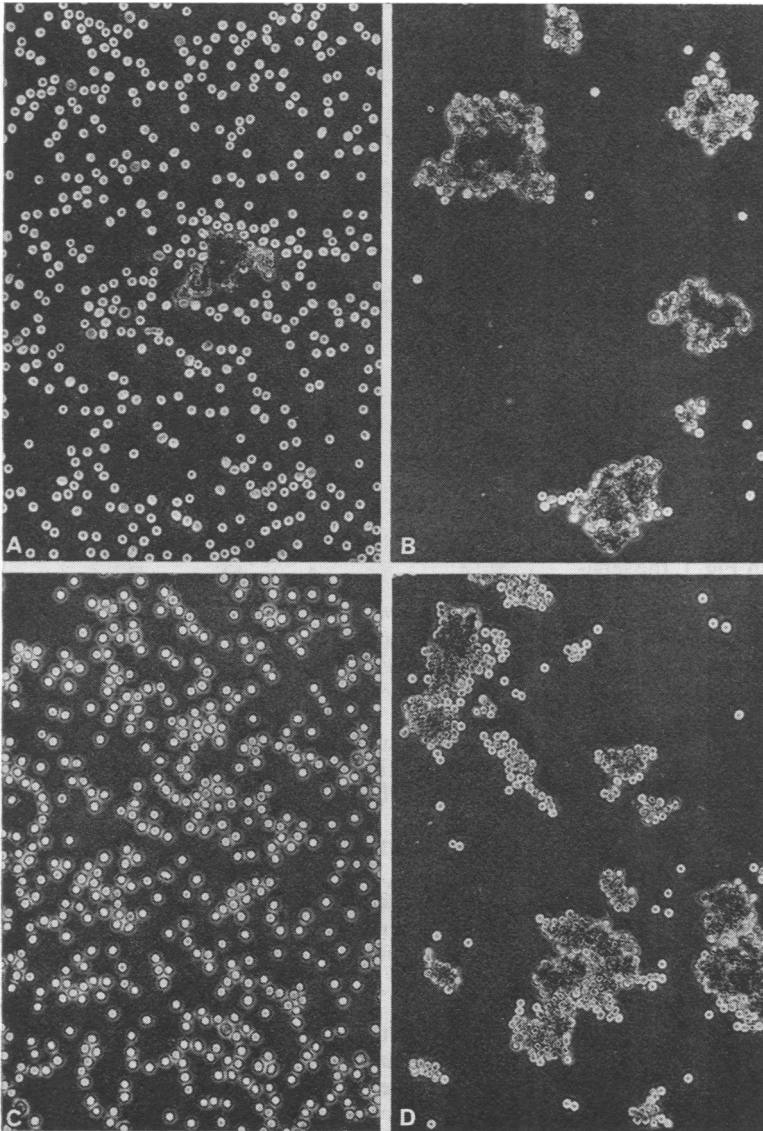


Fig. 1 Red cell suspensions prepared from whole blood of proband: (A) exposed to anti-A; (B) exposed to anti-A,B. Red cell suspensions prepared from O cells separated from whole blood of proband: (C) exposed to anti-A; (D) exposed to anti-A,B.

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, Km) did not reveal any mixtures.

Mrs Co.'s saliva contained A, H, and Le^a. Both populations of red cells were agglutinated by anti-Le^b and by anti-A₁Le^b. 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-A₁B 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-A₁Le^b. The red cells of the other family members (I.1, II.1, II.3, III.1, III.2) were not agglutinated by anti-A₁Le^b. The Table shows the serum levels of A₁- and A₂-specified N-acetylgalactosaminyltransferases.

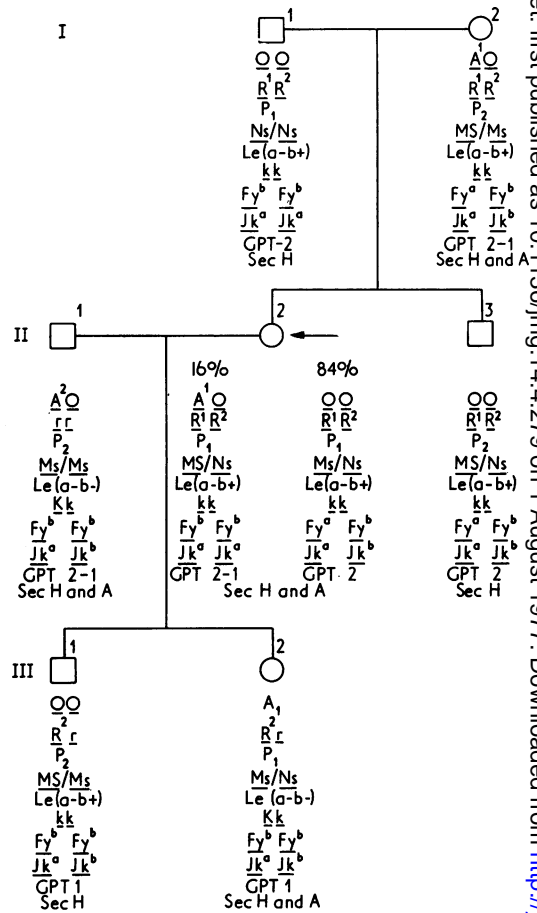


Fig. 2 Some genetic markers in the proband and her family. The presumed genotypes are underlined

Conclusions

The A₁ gene in the proband is not restricted to her haemopoietic tissue since: (1) her saliva contains A₁ substance; (2) her daughter (III.2) has inherited her A₁ gene; (3) the proband's plasma contains A₁ substance (detectable on her O cells) and A₁Le^b (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-acetylgalactosaminyltransferase (i.e. more than 20% of 16%)

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Table Serum N-acetylgalactosaminyltransferase levels in proband and family

Serum donor	Genotype	Counts per minute*
I.1 Father	OO	90
I.2 Mother	A ¹ O	2296
II.1 Husband	A ² O	464
II.2 Proband	A ¹ O	1089
II.3 Brother	OO	28
III.1 Son	OO	50
III.2 Daughter	A ¹ A ² (or A ¹ O)	1801
Controls C.T.	A ¹ O	3259
R.M.	A ²	403

*Radioactivity in product of an incubation mixture containing (in a final volume of 45 μ l) 0.063 μ mol LNF-1; 0.005 μ mol UDP-N-acetyl-D-galactosamine-1-¹⁴C (2×10^5 cpm per μ mol); 1.25 μ mol MES, pH 5.6; 1.25 μ mol MnSO₄; and 10 μ l 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 co-operation; 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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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