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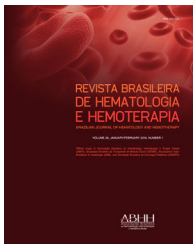

Scientific comment

Blood group polymorphisms in Brazil☆

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Blood group antigens are polymorphisms of proteins and carbohydrates on the surface of red blood cells (RBC) with the distribution of alleles varying between different ethnic groups and populations. By using the genetic information provided by the human genome project, the molecular basis of most blood group genes encoding blood group systems, antigens and phenotypes have been elucidated and nowadays the molecular basis is known for the 308 antigens clustered in the 36 blood group systems recognized by the International Society of Blood Transfusion (ISBT).[1] This knowledge has provided the means to predict red cell antigens and phenotypes in order to identify more extensively matched blood for chronically transfused patients. However, data on the prevalence of various blood group antigens in different populations are essential for estimating the availability of blood components.

The available wealth of serologically defined variants has contributed to the rapid rate with which the genetic diversity of blood group genes has been revealed. Initially, molecular information associated to each variant was obtained from only a small number of samples and applied to DNA analysis with the hopeful assumption that the molecular analysis would correlate with RBC antigen typing. By gathering more information, it became obvious that many molecular events result in the genotype and RBC phenotype being apparently discrepant.[2] Therefore, a large number of people from a variety of ethnic backgrounds need to be studied to determine the occurrence of particular genotypes and to establish more reliably the correlation between blood group genotype and phenotype.

Hemagglutination has identified many phenotypic variants and molecular analysis has revealed remarkable variations within variants. Investigations on the automation of SNP and DNA sequence analysis are ongoing and the potential for genotyping large numbers of samples has already been achieved.[3] Several assays for blood group genotyping have been developed to predict the blood group antigen profile of an individual and with the development of several commercial kits, molecular testing is being performed in many laboratories worldwide to solve clinical problems that cannot be addressed by hemagglutination.

The identification of the molecular basis of blood group antigens has also provided an insight into the generation of gene diversity in humans showing that blood group polymorphisms can be useful markers of genetic differentiation of human populations. The effects of mutations on protein expression and the relative ease of identifying individuals bearing variant alleles has led to the proposal that genes encoding blood group antigens are an important and unique resource for studies on human DNA variation.[4]

The Brazilian population is of heterogeneous ethnic origin. This diverse population is also unevenly distributed within a country of continental dimensions. Besides the Native-American population, Brazil received many immigrants from Portugal, Italy, Spain, Germany, Japan and the Middle East. During the slave trade from the 16th to the 18th century Brazil received approximately 4 million Africans, mainly from Angola, Congo and Mozambique, who settled in almost all regions of the country.[5] The intense process of miscegenation made the Brazilian population unique in its ethnic background. In a multi-ethnic population, such as that of Brazil, the frequencies of the different blood group antigens vary significantly. Genotyping studies in this population have shown that the blood group polymorphisms differ significantly from other populations[6,7] and studies inside the country have also

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shown a heterogeneous distribution of blood group alleles and variants between groups of people from different regions.[8,9] Moreover, the high degree of genetic mixture in the Brazilian population facilitates the generation of new alleles.[10,11] Therefore, studies on the distribution of blood group alleles and variants in donors and patients from different regions of Brazil can report the frequencies of defined polymorphic blood group antigens that function as receptors for parasites, map the rare phenotypes across the country and contribute in the selection of the most adequate blood component for chronically transfused patients.

Conflicts of interest

The author declares no conflicts of interest.

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