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ABO genotyping in leukemia patients reveals new ABO variant alleles

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ABSTRACT. The ABO blood group is the most important blood group system in transfusion medicine and organ transplantation. To date, more than 160 ABO alleles have been identified by molecular investigation. Almost all ABO genotyping studies have been performed in blood donors and families and for investigation of ABO subgroups detected serologically. The aim of the present study was to perform ABO genotyping in patients with leukemia. Blood samples were collected from 108 Brazilian patients with chronic myeloid leukemia (N = 69), chronic lymphoid leukemia (N = 13), acute myeloid leukemia (N = 15), and acute lymphoid leukemia (N = 11). ABO genotyping was carried out using allele specific primer polymerase chain reaction followed by DNA sequencing. ABO*O01 was the most common allele found, followed by ABO*O22 and by ABO*A103. We identified 22 new ABO* variant alleles in the coding region of the ABO gene in 25 individuals with leukemia (23.2%). The majority of ABO variants was detected in O alleles (15/60.0%). In 5 of 51 samples typed as blood group O (9.8%), we found non-deletional ABO*O alleles. Elucidation of the diversity of this gene in leukemia and in other diseases is important for the determination of the effect of changes in an amino
Acid residue on the specificity and activity of ABO glycosyltransferases and their function. In conclusion, this is the first report of a large number of patients with leukemia genotyped for ABO. The findings of this study indicate that there is a high level of recombinant activity in the ABO gene in leukemia patients, revealing new ABO variants.

**Key words:** ABO blood group; DNA polymorphism; Leukemia; Genetic polymorphism; ABO gene; Brazilian population