

***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**^{variants} 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