

Allele-Specific PCR of the Human ABO Locus

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The ABO locus determines the most medically-important phenotype in humans, and genetics courses use it as a classic example of codominance. Previous work has focused on utilizing single nucleotide polymorphisms and linkage disequilibrium to differentiate among the five most common alleles, A1, A2, B, O1, and O2, in order to determine individual genotypes. This technique allowed us to use inexpensive restriction enzymes to produce restriction fragment length polymorphisms. In this study, we performed allele-specific PCR that was designed to amplify a single ABO allele. Two primers, differing only at the SNP, were paired with a common reverse primer in separate reactions in order to produce a product of unique length (when compared to the other allele-specific reactions). We have shown that A2, B, O1, and O2 alleles can be amplified with great specificity in a total of eight reactions to yield successful genotypic determination without the need for any restriction digests following amplification. This work will allow students to determine their genotype within two laboratory periods, and it is appropriate for students of all levels.

Keywords: ABO locus, PCR, blood typing

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